JOHN M. GRAHAM, JR., M.D., SC.D.

CURRICULUM VITAE

Date of Birth March 8, 1947

Place of Birth Wilmington, DE, USA

Business Address Medical Genetics Institute
Cedars-Sinai Medical Center
8700 Beverly Blvd, PACT Suite 400
Los Angeles, CA 90048

Business Phone 310-423-9909
Fax 310-423-9752
E-mail john.graham@cshs.org

BOARD CERTIFICATIONS

1982	American Board of Medical Genetics (No. 1176)
1982	American Board of Pediatrics (No. 039677)
1993	American College of Medical Genetics (Founding Fellow)

EDUCATION

1969	B.A., Natural and Behavioral Sciences	Johns Hopkins University
1975	M.D., Medicine	Medical University of South Carolina
1981	Sc.D., Public Health Administration and Communicative Disorders	Johns Hopkins University School of Hygiene and Public Health

POSTGRADUATE TRAINING

Pediatric Intern - Children's Hospital Medical Center, Boston MA, 1975-76.

Pediatric Resident - Children's Hospital Medical Center, Boston MA, 1976-77.

Fellow in Developmental Disabilities with Dr. Allen Crocker, Children's Hospital Medical Center, Boston, MA, 1977-78.

Fellow in Dysmorphology with Dr. David W. Smith and Instructor in Pediatrics, University of Washington, School of Medicine, Seattle WA, 1978-80.

PROFESSIONAL EXPERIENCE

Consultant Pediatrician and Medical Geneticist, Harbor-UCLA Medical Center,
Teaching Appointment, Torrance CA 2014-present

Director, Division of CSMC Clinical Genetics and Dysmorphology, retired 6/30/13 1988-2013
Director, CSMC Medical Genetics Birth Defects Center Outpatient Facility, retired 6/30/13 1988-2013
Director Clinical Training, CSMC Medical Genetics Training Program, retired 6/30/13 1990-2013
Director, CSMC Craniofacial Clinic, 1990-2005
Director, CSMC Fetal Dysmorphology/Pathology Service, 1990-2000
Cedars-Sinai Medical Center, Los Angeles, CA
Co-Director, CSMC Prenatal Diagnosis Center, 1988-1993
Associate Professor of Pediatrics in Residence, UCLA School of Medicine 1988-1990
Professor of Pediatrics in Residence, UCLA School of Medicine, Los Angeles, CA 1990-2013
Step I, 1990-1993; Step II, 1993-1996; Step III, 1996-1999; Step IV, 1999-2002;
Step V, 2002-2005; Step VI, 2005-2008; Step VII, 2008-2011; Step VIII, 2011.
Professor Emeritus in Pediatrics, David Geffen School of Medicine at UCLA Lifetime Appointment
Professor of Pediatrics and Biomedical Sciences, CSMC 2010-2013
Director of Clinical Genetics and Dysmorphology Program, 1981-1988
Dartmouth Medical School, Hanover, NH.
Medical Director of New Hampshire Genetic Services Program, 1981-1988
Bureau of Special Medical Services, Division of Public Health Services, Concord, NH.
Assistant Professor of Maternal and Child Health, 1981-1986
Associate Professor of Maternal and Child Health, 1986-1988
Adjunct Professor of Maternal and Child Health, 1988-1998
Dartmouth Medical School, Hanover, NH.

MEDICAL LICENSURE

1977	Massachusetts (Certificate No. 40798), inactive
1978	Washington (Certificate No. 16789), inactive
1980	Vermont (Certificate No. 6590), inactive
1980	New Hampshire (Certificate No. 6276), inactive
1988	California (Certificate No. G64797), active

PROFESSIONAL ACTIVITIES AND COMMITTEES

Steering Committee Member and Co-Founder, David W. Smith Morphogenesis and Malformations Workshop, 1980-2012.

Steering Committee Member, New England Regional Genetics Group, 1981-1988.

Chairman, Health Professional Advisory Committee, NH Chapter, March of Dimes, 1985-1988.

Member, Section on Child Development, American Academy of Pediatrics, 1986-present.

Member, Section on Genetics, American Academy of Pediatrics, 1995-present.

Member, American Society of Human Genetics Information and Education Committee, 1986-1990.

Member, Social Issues Committee of the American Society of Human Genetics, 1991-1993.

Chairman, American Society of Human Genetics Task Force on Teaching Human Genetics in North American Medical Schools, 1988.

Chairman, Genetic Services Triage Committee, Cedars-Sinai Medical Center, 1988-2013

Co-Chairman, Prenatal Diagnosis Case Review Conference, Cedars-Sinai Medical Center, 1988-1993.

Chairman, Medical Genetics Educational Mission Planning Task Force, Cedars-Sinai Medical Center, 1990-2013.

Chairman, Clinical Genetics Review Conference, Cedars-Sinai Medical Center, 1988-present.

Member, Review and Appraisal Committee, Department of Pediatrics, UCLA School of Medicine, 1989-1992 and 1999-2002

Co-Chairman, Year 2 Course on Medical Genetics (Genetics 201), 1989-1993, Chairman, Genetics 201 Planning Committee, UCLA School of Medicine, 1991-1993.

Executive Committee, UCLA Intercampus Fellowship Training Program in Medical Genetics, l991-2015.

Chairman of Genetics Curriculum Working Group, Curriculum Review Subcommittee, Medical Education Committee, UCLA School of Medicine, 1991-1993.

Chairman, Cedars-Sinai Medical Center Department of Pediatrics Course on Human Genetics, 1988-2013.

Member, Pediatric Advisory Committee, Cedars-Sinai Medical Center, 1988-1990.

Bioethics Committee, Cedars-Sinai Medical Center, 1990-1996.

Southern California March of Dimes Health Professional Advisory Committee, 1991-1994.

Liaison to American Society of Human Genetics from Teratology Society, 1990-2002; Liaison to Dysmorphology Societies (U.K., Europe, U.S.A.) from Teratology Society, 2002-2013.

Teratology Society, Publications Committee, 1989-1995; 2011-2012; Public Affairs Committee, 1997-2000; Finance Commitee 2010-2012; Education Commitee 2010-2012; Program Commitee 2010-2012, Council, 2002-2005; 2009-2013; Vice President Elect, 2009-10; Vice President 2010-11; President 2011-12.

Dysmorphology Sub-Committee, Clinical Practice Committee, American College of Medical Genetics, 1993-97.

Consulting Developmental Pediatrician, UCLA University Affiliated Program, 1995-1999.

President, Society of Craniofacial Genetics, 1998-2000.

Member of Gorlin Dysmorphology Meeting Organizing Committee, 1999-2005.

Guest Editor, American Journal of Medical Genetics, Special Issue on Gastrointestinal Disorders, 122A:281-353, November 1, 2003.
Guest Editor, European Journal of Medical Genetics, Special Issue on Epilepsy and Genetics, European Journal of Medical Genetics, 55(5): 279-280, 2012.
Guest Editor, European Journal of Medical Genetics, Special Issue on Genetics of Common Malformations, European Journal of Medical Genetics, January 2014.

Associate Editor of Clinical Teratology, Teratology, 1983-87, and 1989-95.

Editorial Board Member, Annales de Génétique (France), 2000-2004.

Editorial Board Member: Clinical Pediatrics, 1983-present.

Editorial Board Member, American Journal of Medical Genetics, 1995-2001; 2009-present

Editorial Board Member, Congenital Anomalies (Japan), 2000-present

Editorial Board Member, European Journal of Medical Genetics, 2004-present.

Editorial Board Member, Global Pediatric Health, 2014-present.

International Advisory Board for Indian Academy of Medical Genetics, 2012-present

Cedars-Sinai Medical Center Internal Review Board Committee, 2001-2004, 2010-2013.

SCHOLARLY SOCIETIES AND PROFESSIONAL ASSOCIATIONS

American Society of Human Genetics
European Society of Human Genetics
American College of Medical Genetics
American Board of Medical Genetics
Society of Craniofacial Genetics
Society for the Study of Behavioral Phenotypes
International Society for Prenatal Diagnosis
American Cleft Palate-Craniofacial Association
American Academy of Pediatrics
Society for Pediatric Research
American Pediatric Society
Western Society for Pediatric Research
European Society of Pediatric Research
Teratology Society

HONORS AND SPECIAL AWARDS

Distinguished Accomplishment, National Youth Science Center, Nasson College, Springvale ME, 1964.

First Place in Zoology, International Science Fair, St. Louis MO, 1965.

First Place, 7th Annual Student Research Competition, Medical University of SC, Charleston SC, 1972.

First Place, 8th Annual Student Research Competition, Medical Univ. of SC, Charleston SC, 1973.

Gold Medal and Grand Award, Student A.M.A. Squibb Scientific Exhibit Competition, National Student Research Forum, Galveston TX, 1973.

Alpha Omega Alpha Honorary Medical Fraternity, 1973, Lange Award for Scholarship and Student Community Service, 1973, Medical University of South Carolina, Charleston SC.

Gold Medal, Student A.M.A. Squibb Scientific Exhibit Competition, National Student Research Forum, Galveston TX, 1974.

Mosby Scholarship Book Award for Scholastic Excellence, Medical Univ of South Carolina, 1974 and 1975.

Poncin Scholarship Award, University of Washington School of Medicine, Seattle WA, 1979.

Certificate of Appreciation, United Leukodystrophy Foundation, 1986.

Saul Blatman Clinical Scholar Award (1986), and Saul Blatman Excellence in Teaching Award (1987), Department of Maternal and Child Health, Dartmouth Medical School.

Award for Excellence in Education, UCLA School of Medicine, Los Angeles CA, 1993.

Elected to Delta Omega, Honorary Public Health Society, Alpha Chapter, Johns Hopkins University, School of Hygiene and Public Health, 1994.

CSMC Medical Genetics Institute Paper of the Year Award: Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 36:405-410, 2004. Awarded 4/28/04.

Scroll of Appreciation from Brigadier General of the European Medical Command for CME lectures given to U.S. Army medical service providers, 5/21/09

Thank You Doctor Award from ALO Cultural Foundation for service to Lebonese Boy with Fraser Cryptophthalmos Syndrome, June 2009.

Listed by U.S. News and World Report as being among the best doctors and in the top 1% of Clinical Geneticists in the U.S.A. in 2012.

Cedars-Sinai Medical Center Golden Apple Award for Excellence in Teaching Medical Genetics, June 2012.

Cedars-Sinai Medical Center, Medical Genetics Institute, Lifetime Achievement Award in Recognition of Extraordinary Achievement in Teaching and Mentoring, June 27, 2013.

PREVIOUS AND CURRENT GRANTS AND CONTRACTS

"The role of maternal hyperthermia as a teratogen-potentiating factor." 4/1/81-3/25/83: $4000.00. BRSG Grant 2S07-RR05392-21, 6/10/82-7/1/83: $1000.00, Hitchcock Foundation Research Project #51(PI)

"Investigation into the biochemical and genetic basis for X-linked ocular albinism." 4/1/86-3/31/87: $5000.00. Gilman Fund. (PI)

"Investigation into the molecular basis of Beckwith-Wiedemann syndrome." 6/1/8512/31/86: $500 Hitchcock Foundation; $1000 March of Dimes Research Program; $3000.00 BRSG Grant 2S07-RR05392-25, 4/1/87 - 3/31/88. (PI)

"New Hampshire Genetic Services Program." 2/1/81-6/30/88: $283,117.00. New Hampshire Bureau for Special Medical Services, New Hampshire Division of Public Health Services. (PI)

"Multidisciplinary care for children with birth defects and inherited disorders that cause developmental disabilities." 7/1/86-6/30/89: $211,772.00. Jessie B. Cox Charitable Trust Development Program. (CI)

"Genetic counseling learning system: Part 1. Down syndrome, Part 2. Spina bifida." 10/1/87- 9/30/88: $13,200.00, New England Regional Genetics Group Special Project. (PI)

"Collaborative medical and developmental support services project for children with genetic and prenatally determined disorders." 10/1/88 - 9/30/91: $368,062.00, U.S. Department of Education, Bureau of Special Education and Rehabilitative Services. (CI)

"California AFP Screening Program Prenatal Diagnosis Center Follow-Up" Contract No. 88-93587. 7/1/88 - 6/30/92: $440,111.00, California Department of Health Services. (Program Director) Vendorized contract after 6/30/92.

"Longitudinal follow-up in 15 probands with Beckwith-Wiedemann syndrome." 5/23/92-8/31/92: $2,000.00, March of Dimes Summer Science Research Program for Medical Students (recipient Ms. Elaine Weng), UCLA Medical School, Class of 1995): #8-FY9280. (PI) plus Short Term Training Program Summer Research Grant: 5/24/93-8/13/93, $2,700.00, UCLA School of Medicine.

"Mosaic chromosome aneuploidy diagnosed prenatally: a prospective study." 1/1/94-6/30/94: $14,325. Feintech Foundation. (CI)

"Pallister Hall syndrome: genetic linkage studies", 10/1/94-9/30/95: $5,000. NIH National Center for Human Genome Research.

Consulting Geneticist and Developmental Pediatrician, UCLA University-Affiliated Program (MCH Grant): 1995-1999; total contractual award $68,000.

The Incidence and Prevalence of CHARGE Association/Syndrome. Canadian Pediatric Surveillance Program (10/1/01-9/30/04). Kim Blake (PI), John Graham (CI).

The Incidence and Prevalence of CHARGE Association/Syndrome. Kim Blake (PI), John Graham (Consultant). Start-up grant for $10,000 from CHARGE Syndrome Foundation (2001).

E. A. Baker Foundation (Canadian National Institute for the Blind) $23,000, The Incidence and Prevalence of CHARGE Association/Syndrome. Kim Blake (PI), John Graham (CI). (2002-2003).

Genesis Fund Grant for CSMC Craniofacial Clinic for $25,000 (2003), John Graham (PI) Boston MA.

Larsen Syndrome Grant from National Organization for Rare Diseases, $30,000, 10/1/03-9/30/04. John Graham (PI) Danbury CT.

CSMC Infant Progress Clinic; Contract for 20% salary to provide developmental and dysmorphology follow-up services to NICU graduates: (7/1/01-6/30/06).

CSMC Telepsychiatry Grant for Outreach Services; CA Dept of Developmental Services, Sacramento CA, Contract for 10% salary: (10/1/01-9/30/05; 10/1/06-9/30/07).

Identification of Autism Susceptibility Loci – The AGRE consortium. 5% salary to provide dysmorphology consultation. NIMH (3/15/02-2/28/07; renewal pending). $1,812,646. Dan Geschwind (PI). (CSMC IRB has approved my submission to participate in this contract.)

Cat Eye Syndrome Grant from National Organization for Rare Diseases, $30,000, 10/1/05-9/30/07. John Graham (PI) Danbury CT.

The Skeletal Dysplasias. NIH/NICHD Grant HD22657-11. Project period: 12/1/01 to 11/30/06; 12/1/06-4/30/12; Department of Health and Human Services, Public Health Service. David L. Rimoin, M.D., Ph.D. (PI), John M. Graham, Jr., MD, ScD (CI).

Medical Genetics UCLA Intercampus NIH/NIGMS Training Program Grant. GM08243-16". 7/1/02-6/30/07; 7/1/07-6/30/12; 7/1/12-6/30/17). David L. Rimoin, M.D., Ph.D.; Bill Wilcox (PIs), John M. Graham, Jr., M.D., Sc.D., (Member Faculty Executive Committee).

BIBLIOGRAPHY

PEER-REVIEW RESEARCH PUBLICATIONS

1. Graham J.M. Jr., Schreiber R.A., and Zemp J.W.: Effect of d-amphetamine sulfate on susceptibility to audiogenic seizures in DBA/2J mice. Behavioral Biology, 10:183-190, 1974.

2. Schreiber R.A. and Graham J.M. Jr.: Audiogenic priming in DBA/2J and C57BL/6J mice: Interactions between age, prime-to-test interval and index of seizure. Dev. Psychobiology, 9:57-66, 1976.

3. Wertelecki W., Graham J.M. Jr., and Sergovich F.: Clinical recognition of triploidy. Obstetrics and Gynecology, 47:69-76, 1976.

4. Graham J.M. Jr. and Smith D.W.: Parietal craniotabes in the neonate: Its origin and relevance. Journal of Pediatrics, 95:114-116, 1979.

5. Graham J.M. Jr., de Saxe M., and Smith D.W.: Sagittal craniostenosis: Fetal head constraint as one possible cause. Journal of Pediatrics, 95:747-750, 1979.

6. Graham J.M. Jr., Badura R.J., and Smith D.W.: Coronal craniostenosis: Fetal head constraint as one possible cause. Pediatrics, 65:995-999, 1980.

7. Graham J.M. Jr. and Smith D.W.: Metopic craniostenosis as a consequence of fetal head constraint: Two interesting experiments of nature. Pediatrics, 65:1000-1002, 1980.

8. Graham J.M. Jr., Miller M.E., Stephan M.J., and Smith D.W.: Limb reduction anomalies and early in-utero limb compression. Journal of Pediatrics, 96:1052-1056, 1980.

9. Miller M.E., Graham J.M. Jr., Higginbottom M.C., and Smith D.W.: Compression-related defects from early amnion rupture: Evidence for mechanical teratogenesis. Journal of Pediatrics, 98:292-297, 1981.

10. Graham J.M. Jr., Hoehn H., Lin, M.S., and Smith D.W.: Diploid-triploid mixoploidy: Clinical and cytogenetic features. Pediatrics, 68:23-28, 1981.

11. Graham J.M. Jr., Higginbottom M.C., and Smith D.W.: Preaxial polydactyly of the foot associated with early amnion rupture: Evidence for mechanical teratogenesis. Journal of Pediatrics, 98:943-945, 1981.

12. Pleet H., Graham J.M. Jr., and Smith D.W.: Central nervous system and facial defects associated with maternal hyperthermia at 4 to 14 weeks gestation. Pediatrics, 67:785-789, 1981.

13. Pauli R.M., Graham J.M. Jr., and Barr M.: Agnathia, situs inversus, and associated malformations. Teratology, 23:85-93, 1981.

14. Pagon R.A., Graham J.M. Jr., Zonana J., and Yong S.L.: Coloboma, congenital heart disease and choanal atresia with multiple anomalies: CHARGE Association. Journal of Pediatrics, 99:223-227, 1981.

15. Graham J.M. Jr., Stephens T.D., Siebert J.R., and Smith D.W.: Determinants in the morphogenesis of muscle tendon insertions. Journal of Pediatrics, 101:825-831, 1982.

16. Stephens T.D., Siebert J.R., Graham J.M. Jr., and Beckwith J.B.: Parasitic conjoined twins, two cases, and their relation to limb morphogenesis. Teratology, 26:115-121, 1982.

17. Hersh J.H., Graham J.M. Jr., Destrempes B.S., and Greenstein R.M. Teschler-Nicola Killian syndrome: A case report. Journal of Clinical Dysmorphology, 1:20-24, 1983.

18. Graham J.M. Jr., Marin-Padilla M., and Hoefnagel D.: Jejunal atresia associated with cafergot injestion during pregnancy. Clinical Pediatrics, 22:226-228, 1983.

19. Graham J.M. Jr., Stephens T.D., Shepard T.H.: Nuchal cystic hygroma in a fetus with presumed Roberts syndrome. American Journal of Medical Genetics, 15:163-167, 1983.

20. Jung J.H., Graham J.M. Jr., Schultz N., and Smith D.W.: Congenital hydranencephaly/porencephaly due to vascular disruption in monozygotic twins. Pediatrics, 73:467-469, 1984.

21. Graham J.M. Jr., Crow H.C., Rawsley E.F., Simmons G.M., and Hoefnagel D.: Enhanced visualization of soft tissues in the study of aborted fetuses through the use of xeroradiography. Teratology, 30:11-24, 1984.

22. Walzer S., Bashir A.S., Graham J.M. Jr., and Silbert A.R.: Communication disorders, learning disorders, learning difficulties and temperamental style in XXY boys. Journal of Developmental and Behavioral Pediatrics, 5(3):147-149, 1984.

23. Graham J.M. Jr. and Ferm V.H.: Heat and alcohol induced neural tube defects: Interactions with folate in a golden hamster model. Pediatric Research, 19:247-251, 1985.

24. Smith D.W., Marokus R., Graham J.M. Jr.: Tentative evidence of Y-linked statural gene(s) from growth in the testicular feminization syndrome. Clinical Pediatrics 24:189-192, 1985.

25. Siebert J.R., Graham J.M. Jr., and MacDonald C.: Pathologic features of the CHARGE Association: Support for involvement of the neural crest. Teratology 31:331-336, 1985.

26. Graham J.M. Jr., Brown F.E., Saunders R.L., Hinkle A.J., Frank J.E., Harris M.S., Klein R.Z. Bifid epiglottis, hand anomalies and congenital hypopituitarism, Lancet 2:443, 1985.

27. Park J.P., Graham J.M. Jr., Wurster-Hill D.H.: Familial t(4;21) (q2.4;q2.2) leading to an unbalanced offspring with the Down syndrome. American Journal of Medical Genetics, 25:399-402, 1986.

28. Wertelecki W., Breg U.R., Graham J.M. Jr., Iinuma K., Puck S.M., Sergovich F.R.: Trisomy 22 mosaicism syndrome and Ullrich-Turner stigmata. American Journal of Medical Genetics 23:739-749, 1986.

29. Graham J.M. Jr., Brown F.E., Struckmeyer C.L., and Hallowell C.: Dominantly inherited unilateral terminal transverse defects of the hand (adactylia) in twin sisters and one daughter, Pediatrics 78:103-106, 1986.

30. Graham J.M. Jr., Saunders R., Fratkin J., Spiegel P., Harris M., and Klein R.Z.: A cluster of Pallister-Hall syndrome cases (congenital hypothalamic hamartoblastoma syndrome), American Journal of Medical Genetics, Supplement 2, The Developmental Field Concept, Part 2:53-63, 1986.

31. Brown F.E., Cohen L.B., Addante R.R., and Graham J.M. Jr.: Correction of congenital auricular deformities by splinting in the neonatal period. Pediatrics, 78:406-411, 1986.

32. Disteche C.M., Casanova M., Saal H., Friedman C., Sybert V., Graham J.M. Jr., Thuline H., Page D.C., and Fellous M.: Small deletions of the short arm of the Y chromosome in 46,XY females, Proceedings of the National Academy of Sciences, 83:7841-7844, 1986.

33. Graham J.M. Jr., Brown F.E., and Hall B.D.: Thumb polydactyly as part of the range of genetic expression for thenar hypoplasia. Clinical Pediatrics, 26:11-17, 1987.

34. Graham J.M. Jr., Boyle U., Troxell J., Cullity G.J., Sprague P., and Beckwith J.B.: Cystic hamartomata of the lung and kidney: A spectrum of developmental abnormalities, American Journal of Medical Genetics, 27:45-59, 1987.

35. Morse R.P., Rawsley B.E., Crow H.C., Marin-Padilla M., and Graham J.M. Jr.: Bilateral renal agenesis in three consecutive siblings, Prenatal Diagnosis, 7:573-579, 1987.

36. Morse R.P., Rawsley B.E., Sargent S.K., and Graham J.M. Jr.: Prenatal diagnosis of a new syndrome: holoprosencephaly with hypokinesia, Prenatal Diagnosis, 7:631-638, 1987.

37. Park J.P., Wurster-Hill D.H., Graham J.M. Jr., Andrews P.A., and Cooley C.: Free partial trisomy 21 without the Down Syndrome. Clinical Genetics, 32:342-348, 1987.

38. Park J.P., Wurster-Hill D.H., Berg S.Z., and Graham J.M. Jr.: A denovo interstitial deletion of chromosome 6 (q22.2 q23.1), Clinical Genetics, 33:65-68, 1988.

39. Park J.P., Graham J.M. Jr., Andrews P.A., and Wurster-Hill D.H.: Ring chromosome 12. American Journal of Medical Genetics, 29:437-440, 1988.

40. Graham J.M. Jr., Hanson J.W., Darby B.L., Barr H.M., and Streissguth A.P.: Independent dysmorphology evaluations at birth and four years of age for children exposed to variable amounts of alcohol in utero, Pediatrics, 81:772-778, 1988.

41. Graham J.M. Jr., Bashir A.S., Stark R.E., Silbert A., Walzer S.: Oral and written language abilities of XXY boys: Implications for anticipatory guidance. Pediatrics, 81:795-806, 1988.

42. Moeschler J.B., Charman C.E., Berg S.Z., and Graham J.M. Jr.: Rett syndrome: Natural history and management. Pediatrics, 82:1-10, 1988.

43. Knoll J.H.M., Nicholls R.D., Magenis R.E., Graham J.M. Jr., Lalande M., Latt S.A.: Angelman and Prader-Willi syndromes share a common chromosome 15 deletion but differ in parental origin of the deletion. American Journal of Medical Genetics, 32:285-290, 1989.

44. Thomas I.T., Frias J.L., Cantu E.S., Lafer C.Z., Flannery D.B., and Graham J.M. Jr.: The association of pigmentary anomalies with chromosomal and genetic mosaicism and chimerism. American Journal of Human Genetics, 45:193-205, 1989.

45. Moeschler J.B., Pober B., Holmes L.B., and Graham J.M. Jr.: Acrocallosal syndrome: New findings. American Journal of Medical Genetics, 32:306-310, 1989.

46. Tantravahi U., Nicholls R.D., Shroh H., Ringer S., Neve R.L., Kaplan L., Wharton R., Wurster-Hill D., Graham J.M. Jr., Cantu E., Frias J., Kousseff B., and Latt S.: Quantitative calibration and use of DNA probes for investigating chromosomal abnormalities in the Prader-Willi syndrome. American Journal of Medical Genetics, 33:78-87, 1989.

47. Nicholls R.D., Knoll J.H., Glatt K., Hersh J., Brewster T., Graham J.M. Jr., Wurster-Hill D., Wharton R., Latt S.: RFLPs within proximal 15q and their use in molecular cytogenetics and the Prader-Willi syndrome. American Journal of Medical Genetics, 33:66-77, 1989.

48. Graham J.M. Jr., Rawnsley E.F., Wurster-Hill D.H., Park J.P., Marin-Padilla M., Crow H.C.: Triploidy: pregnancy complications and clinical findings in seven cases. Prenatal Diagnosis, 9:409-419, 1989.

49. Iafolla A.K., Fratkin J.D., Spiegel P.K., Cohen M.M., and Graham J.M. Jr.: Case report and delineation of the congenital hypothalamic hamartoblastoma syndrome (Pallister-Hall Syndrome). American Journal of Medical Genetics, 33(4):489-499, 1989.

50. Starman B.J., Eyre D., Charbonneau H., Harrylock M., Weis M.A., Weiss L., Graham J.M. Jr., and Byers P.H.: Osteogenesis imperfecta: the position of substitution for glycine by cysteine in the triple helical domain of the pro alphal(I) chains of type I collagen determines the clinical phenotype. J. of Clinical Investigation, 84:1206-1214, 1989.

51. Stern H.J., Graham J.M. Jr., Lachman R.S., Horton W.A., Bernini P.M., Bodurtha J., Bocian M, Ives E., Spiegel P.K., and Rimoin D.L.: Atelosteogenesis type III: a distinct skeletal dysplasia with features overlapping atelosteogenesis and otopalato-digital syndrome type II. American Journal of Medical Genetics, 36(2):183-195, 1990.

52. Pulst, S.M., Graham, J.M., Jr., Fain, P., Barker, D., Pribyl, T., and Korenberg, J.R.: The achondroplasia gene is not linked to the locus for neurofibromatosis 1 on chromosome 17. Human Genetics, 85:12-14, 1990.

53. Edwards M.J. and Graham J.M., Jr.: Studies of type 1 collagen in osteogenesis imperfecta. Journal of Pediatrics, 117:67-72, 1990.

54. Lin, A.E., Morter, G., Siebert, J., and Graham, J.M. Jr.: Central nervous system malformations in the CHARGE Association. American Journal of Medical Genetics, 37(3):304-310, 1990.

55. Morse R.P., Rockenmacher S., Pyeritz R., Sanders S., Bieber F., Lin A., MacLeod P, Hall B., Graham J.M. Jr.: Diagnosis and management of infantile Marfan's syndrome. Pediatrics, 86(6)888-895, 1990.

56. Cooley W.C., Graham E.S., Moeschler J.B., and Graham J.M. Jr.: Reactions of mothers and medical professionals to a film of parents of persons with Down syndrome. American Journal of Diseases of Children, 144(10):1112-1116, 1990.

57. Donnenfeld A.E., Graham J.M. Jr., Packer R.J., Aquino R., Berg S.Z., and Emanuel B.S.: Microphthalmia and chorioretinal lesions in a girl with an Xp22.2-pter deletion and partial 3p trisomy; clinical observations relevant to Aicardi syndrome gene localization. American Journal of Medical Genetics, 37(2):182-186, 1990.

58. Knoll J.H.M., Nicholls R.D., Magenis R.E., Glatt K., Graham J.M. Jr., Kaplan L., and Laland M.: Angelman syndrome: three molecular classes identified with chromosome 15qll-ql3 specific DNA markers. American Journal of Human Genetics, 47:149-155, 1990.

59. Falik-Borenstein T.C., Korenberg J.R., Davos I., Gans S., Goodman B., Schreck R., Graham J.M. Jr.: Congenital gastric teratoma in Wiedemann-Beckwith syndrome. American Journal of Medical Genetics, 38:52-57, 1991.

60. Finkelstein J.E., Doege K., Yamada Y., Pyeritz R.E., Graham J.M. Jr., Moeschler J.B., and Francomano C.A.: Analysis of the chondroitin sulfate proteoglycan core protein (CSPGCP) gene in achondroplasia and pseudoachondroplasia. American Journal of Human Genetics, 48(1):97-102, 1991.

61. Shohat M., Herman V., Melmed S., Neufeld N., Schreck R., Pulst S., Graham J.M. Jr., Rimoin D.L., Korenberg J.R.: Deletion of 20p 11.23→pter With Normal Growth Hormone-Releasing Hormone Genes. American Journal of Medical Genetics, 39(1):56-63, 1991.

62. Cooley W.C., Graham J.M. Jr.: Down syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 30(4):233-253, 1991.

63. Wagstaff J., Knoll J.H.M., Fleming J., Kirkness E.F., Martin-Gallardo A., Greenberg F., Graham J.M. Jr., Menninger J., Ward D., Venter J.C., Lalande M.: Localization of the gene encoding the GABAA receptor B3 subunit to the Angelman/Prader-Willi region of human chromosome 15. American Journal of Human Genetics, 49:330-337, 1991.

64. Christian C.L., Lachman R.S., Aylsworth A.S., Fujimoto A., Gorlin R.J., Lipson M.H., Graham J.M. Jr.: Radiological findings in Hallermann-Streiff syndrome: report of five cases and a review of the literature. American Journal of Medical Genetics, 41(4):508-514, 1991.

65. Lachman R.S., Tiller G.E., Graham J.M. Jr., Rimoin D.L.: Collagen, genes and the skeletal dysplasias on the edge of a new era: a review and update. European Journal of Radiology, 14:1-10, 1992.

66. Fischel-Ghodsian N., Bohlman M.C., Prezant T.R., Graham J.M. Jr., Cederbaum S.D., Edwards M.J.: Deletion in blood mitochondrial DNA in Kearns-Sayre syndrome. Pediatric Research, 31:557-560, 1992.

67. Cantrell M.A., Bogan J.S., Simpson E., Bicknell J.N., Goulmy E., Chandler P., Pagon R.A., Walker D.C., Thuline H.C., Graham J.M. Jr., de la Chapelle A., Page D.C., and Disteche C.M.: Deletion mapping of H-Y antigen to the long-arm of the human Y chromosome. Genomics 13,1255-1260, 1992.

68. Perez-Aytes A., Graham J.M. Jr., Hersh J.H., Hoyme H.E., Aleck K., Carey J.C.: The urethral obstruction sequence and lower limb deficiency: Evidence for the vascular disruption hypothesis, Journal of Pediatrics 123(3):398-405, 1993.

69. Cunniff C., Curry C.J.R., Carey J.C., Graham J.M. Jr., Williams C.A., Stengel-Rutkowski S., Luttgen S., Meinecke P.: Congenital diaphragmatic hernia in the Brachmann-deLange syndrome. American Journal of Medical Genetics 47(7):1018-1021, 1993.

70. Braddock S.R., Lachman R.S., Charman C., Carey J.C., Ireland M., Moeschler, J.B., Cunniff C., and Graham J.M. Jr.: The radiological features in Brachmann-deLange syndrome. American Journal of Medical Genetics 47(7):1006-1013, 1993.

71. Moeschler J.B., Graham J.M. Jr.: Mild Brachmann-de Lange syndrome: phenotypic and developmental characteristics of mildly affected individuals. American Journal of Medical Genetics 47(7):969-976, 1993.

72. Sharony R., Garber A., Viskochil D., Schreck R., Platt L.D., Ward R., Buehler B.A., Graham J.M. Jr.: Preaxial ray reduction defects as part of valproic acid embryopathy. Prenatal Diagnosis, 13:909-919, 1993.

73. Bacino C.A., Platt L.D., Carlson D., Garber A., Pepkowitz S., Lachman R.S., Sharony R., Rimoin D.L., Graham J.M. Jr.: Fetal akinesia/ hypokinesia sequence: Prenatal diagnosis and intra-familial variability. Prenatal Diagnosis, 13:1011-1019, 1993.

74. Braddock S.R., Braddock B.A., Graham JM Jr.: Rett syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 32:613-626, 1993.

75. Skovby F., Graham J.M. Jr., Sonne-Holm S., Cohen M.M. Jr.: Compromise of the spinal canal in Proteus syndrome. American Journal of Medical Genetics 47:656-659, 1993.

76. Garber A., Carlson D., Schreck R., Fischel-Ghodsian N., Wei-Tong H., Klein J., Oeztas S., Pepkowitz S., Williams J. III, Graham J.M. Jr.: Prenatal diagnosis and dysmorphic findings in mosaic trisomy 16. Prenatal Diagnosis 14:257-266, 1994.

77. Klein J., Graham J.M. Jr., Platt L.D., Schreck R.: Trisomy 8 mosaicism in chorionic villi sampling: case report and counseling issues. Prenatal Diagnosis 14:451-454, 1994.

78. Edwards M.J., Park J., Wurster-Hill D.H., and Graham J.M. Jr.: Mixoploidy in humans: two surviving cases of diploid tetraploid mixoploidy and comparison with diploid triploid mixoploidy. American Journal of Medical Genetics 52:324-330, 1994.

79. Korenberg J.R., Chen X.-N, Schipper R., Sun Z., Gonsky R., Gerwehr S., Carpenter N., Daumer C., Dignan P., Disteche C., Graham J.M. Jr., Hudgins L., McGillivray B., Miyazaki K., Ogasawara N., Park J.P., Pagon R., Peuschell S., Sack G., Say B., Schuffenhauer S., Soukup S., and Yamanaka T.: Down syndrome phenotypes: the consequences of chromosomal imbalance. Proceedings of National Academy of Sciences, 91:4997-5001, 1994.

80. Bacino C.A., Schreck R., Fischel-Ghodsian N., Pepkowitz S., Prezant T.R., Graham J.M. Jr.: Clinical and molecular studies in full trisomy 22: Further delineation of the phenotype and review of the literature. American Journal of Medical Genetics, 56:359-365, 1995.

81. Weng E.Y., Moeschler J.B., and Graham J.M. Jr.: Longitudinal observations of 15 children with Beckwith-Wiedemann syndrome. American Journal of Medical Genetics, 56:366-373, 1995.

82. Weng E.Y., Mortier G.R., Graham J.M. Jr.: Beckwith-Wiedemann syndrome: An update and review for the primary pediatrician. Clinical Pediatrics, 34:317-326, 1995.

83. Penny L.A., Dell'Aquila M., Jones M.C., Bergoffen J., Cunniff C., Fryns J.P., Grace E., Graham J.M. Jr., Kousseff B., Mattina T., Syme J., Voullaire L., Zelante L., Zenger-Hain J., Jones O.W., and Evans G.A.: Clinical and molecular characterization of patients with distal 11q deletions. American Journal of Human Genetics, 56:676-683, 1995.

84. Bhatt S., Schreck R., Graham J.M. Jr., Korenberg J.R., Hurvitz C.H., Fischel-Ghodsian N.: Transient leukemia with trisomy 21: description of a case and review of the literature. American Journal of Medical Genetics, 58:310-314, 1995.

85. Meyers G.A., Day D., Goldberg R., Daentl D., Przylepa A., Abrams L.J., Graham J.M. Jr., Feingold M., Moeschler J.B., Rawnsley E., Scott A.F., Jabs E.W.: FGFR 2 Exon Illa and Illx mutations in Crouzon, Jackson-Weiss, and Pfeiffer syndromes: Evidence for missense changes, insertions, and a deletion due to alternative RNA splicing. American Journal of Human Genetics 58:491-498, 1996.

86. Ming J.E., Stiehm E.R., Graham J.M. Jr.: Immunodeficiency as a component of recognizable syndromes. American Journal of Medical Genetics, 66:378-398, 1996.

87. Biesecker L.B., Graham J.M. Jr.: Syndrome of the month: Pallister-Hall syndrome. Journal of Medical Genetics, 33:585-589, 1996.

88. Ruder R.O. and Graham J.M. Jr.: Evaluation and treatment of the deformed and malformed auricle. Clinical Pediatrics, 35:461-465, 1996.

89. Goldberg, Y., Dibbern K., Klein J., Riccardi V.M., Graham J.M. Jr.: Neurofibromatosis Type 1 - An update and review for the primary pediatrician. Clinical Pediatrics, 35:545-561, 1996.

90. Biesecker L.G., Abbott M., Allen J., Clericuzio C., Feuillan P., Graham J.M. Jr., Hall J., Kang S., Haskins-Olney A., Lefton D., Neri G., Peters K., Verloes A.: Report from the workshop on Pallister-Hall syndrome and related phenotypes. American Journal of Medical Genetics, 65:76-81, 1996.

91. Biesecker L.G., Kang S., Schaffer A.A., Abbott M., Kelley R.I., Allen J. C., Clericuzio C., Grebe T., Olney A., and Graham J.M. Jr.: Exclusion of candidate loci and cholesterol biosynthetic abnormalities in familial Pallister-Hall syndrome. Journal of Medical Genetics, 33:947-951, 1996.

92. Kang S., Graham J.M. Jr., Olney A.H., and Biesecker L.G.: GLI3 frameshift mutations cause autosomal dominant Pallister-Hall syndrome. Nature Genetics, 15:266-268, 1997.

93. Thomas J.A., Graham J.M. Jr.: Chromosome 22q11 deletion syndrome: An update and review for the primary pediatrician. Clinical Pediatrics, 36:253-266, 1997.

94. Krantz I., Rand E.B., Genin A., Hunt P., Jones M., Louis A.A., Graham J.M. Jr., Piccoli D.A., Spinner N.B.: Deletions of 20p12 in Alagille syndrome: Frequency and molecular characterization. American Journal of Medical Genetics, 70:80-86, 1997.

95. Dibbern K., Rabinowitz Y.S., Shorr N., and Graham J.M. Jr.: Surgical correction of incomplete cryptophthalmos in Fraser syndrome. American Journal of Ophthalmology, 124:107-109, 1997.

96. Kang S., Allen J., Graham J.M. Jr., Grebe T., Clericuzio C., Patronas N., Ondrey F., Green E., Schaffer A., Abbott M., and Biesecker L.G.: Linkage mapping and phenotypic analysis of autosomal dominant Pallister-Hall syndrome. Journal of Medical Genetics, 34:441-446, 1997.

97. Curry C.J., Stevenson R.E., Cunniff C., Aughton D., Byrne J., Carey J.C., Cassidy S., Cunniff C., Graham J.M. Jr., Jones M.C., Kaback M.M., Moeschler, J., Schaefer G.B., Schwartz S., Tarleton J., Opitz J., and the American College of Medical Genetics: Evaluation of mental retardation: recommendations of a consensus conference. American Journal of Medical Genetics, 72:468-477, 1997.

98. Sampson, P.D., Streissguth A.P., Bookstein F.L., Little R.E., Clarren S.K., Dehaene P., Hanson J.W., Graham J.M. Jr.: The incidence of fetal alcohol syndrome and the prevalence of alcohol-related neurodevelopmental disorder. Teratology, 56:317-325, 1997.

99. Blake K., Davenport S.H., Hall B.D., Hefner M.A., Pagon R., Williams M.S., Lin A.E., Graham J.M. Jr.: CHARGE Association - An update and review for the primary pediatrician. Clinical Pediatrics, 37:159-174, 1998.

100. Alkalay A.L., Graham J.M. Jr., Pomerance J.J.: Evaluation of neonates born with intrauterine growth retardation: Review and practice guidelines. Journal of Perinatology, 18:142-151, 1998.

101. Kerner B., Flaum E., Mathews H., Carlson D.E., Pepkowitz S.H., Hixon H., and Graham J.M. Jr.: Cervical teratoma: Prenatal diagnosis and long-term follow-up. Prenatal Diagnosis, 18:51-59, 1998.

102. Graham J.M. Jr., Braddock S.R., Mortier G.R., Lachman R., Van Dop C., and Jabs E.W.: Syndrome of coronal craniosynostosis with brachydactyly and carpal-tarsal coalition due to Pro250Arg mutation in FGFR3 gene. American Journal of Medical Genetics, 77:322-329, 1998.

103. Graham J.M. Jr., Edwards M.J., and Edwards M.J.: Teratogen Update: Gestational effects of maternal hyperthermia due to febrile illnesses and resultant patterns of defects in humans. Teratology, 58:209-221, 1998.

104. Malzac P., Webber H., Moncla A., Graham J. M. Jr., Kukolich M., Williams C., Pagon R., Ramsdell L., Kishino T., and Wagstaff J.: Mutation analysis of UBE3A in Angelman syndrome patients. American Journal of Human Genetics, 62:1353-1360, 1998.

105. Graham J.M. Jr., Tackels D., Dibbern K., Superneau D., Rogers C., Corning K., Schwartz C.E.: FG syndrome: Report of three new families with linkage to Xq12-q21.1. American Journal of Medical Genetics, 80:145-156, 1998.

106. Dibbern K.M., Graham J.M., Jr., Lachman R.S., Wilcox W.R.: Cumming syndrome: report of two additional cases. Pediatric Radiology 28:798-801, 1998.

107. Fang P., Lev-Lehman E., Tsai T.-F., Matsuura T., Benton C.S., Sutcliffe J.S., Christan S.L., Kubota T., Halley D.J., Meijers-Heijboer H., Langlois S., Graham J.M. Jr., Beuten J., Willems P.J., Ledbetter D.H., Beaudet A.L.: The spectrum of mutations in UBE3A causing Angelman syndrome. Human Molecular Genetics, 8:129-135, 1999.

108. Graham J.M. Jr., Jones K.L., Brent R.L.: Contribution of clinical teratologists and geneticists to the evaluation of the etiology of congenital malformations alleged to be caused by environmental agents: ionizing radiaiation, electromagnetic fields, microwaves, radionuclides and ultrasound. Teratology, 59:307-313, 1999.

109. Laskari A., Smith A.K., Graham J.M. Jr.: Williams-Beuren Syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 38:189-208, 1999.

110. Biesecker L.G., Happle R., Mulliken J.B., Weksberg R., Graham J.M. Jr., Viljoen D.L., Cohen M.M. Jr.: Proteus syndrome: Diagnostic criteria, differential diagnosis, and patient evaluation. American Journal of Medical Genetics, 84:389-395, 1999.

111. Graham J.M. Jr., Superneau D., Rogers R.C., Corning K., Schwartz C.E., Dykens E.M.: Clinical and behavioral characteristics in FG syndrome. American Journal of Medical Genetics, 85:470-475, 1999.

112. Marsh D. J., Kum J. B., Lunetta K. L., Bennett M. J., Gorlin R. J., Bodurtha J., Ahmed S.F., Bodurtha J., Crowe C., Curtis M.A., Dasouli M., Dunn T., Felt H., Geraghty M. T., Graham J. M. Jr., Hodgson S. V., Hudgins L., Hunter A., Korf B. R., Manchester D., Miesfeldt S., Murday V. A., Nathanson K., Parisi1 M., Pober B., Romano C., Tolmie J. L., Trembath R., Winter R. M., Zakai E., Zori R. T., Weng L.-P., Dahia1 P. L.M., and Eng C.: PTEN mutation spectrum and genotype-phenotype correlations in Bannayan-Riley-Ruvalcaba syndrome suggest a single entity with Cowden syndrome. Human Molecular Genetics, 8:1461-1472, 1999.

113. Kerner B., Graham J.M. Jr., Golden J.A., Pepkowitz S.H., Dobyns W.B.: Familial lissencephaly with cleft palate and severe cerebellar hypoplasia. American Journal of Medical Genetics, 87:440-445, 1999.

114. Meira L.B., Graham J.M. Jr., Greenberg C.R., Busch D.W., Doughty A.T.B., Ziffer D.W., Coleman D.M., Savre-Train I., Friedberg E.C.: Manitoba aboriginal kindred with original Cerebro-Oculo-Facio-Skeletal syndrome has a mutation in the Cockayne syndrome Group B (CSB) gene. American Journal of Human Genetics, 66:1221-1228, 2000.

115. Naqvi S., Cole T., Graham J.M. Jr.: Cole-Hughes syndrome and associated autistic features. American Journal of Medical Genetics, 94:149-152, 2000.

116. Kayser M., Henderson L.B., Kreutzman J., Schreck R., and Graham J.M., Jr.: Blaschkolinear skin pigmentary variation associated with trisomy 7 mosaicism. American Journal of Medical Genetics, 95:281-284, 2000.

117. Graham J.M. Jr., Krakow D., Tolo V.T., Smith A.K., Lachman R.S.: Radiographic findings and Gs-alpha bioactivity studies and mutation screening in acrodysostosis indicate a different etiology from pseudohypoparathyroidism. Pediatric Radiology, 31:2-9, 2001.

118. Schweitzer D.N., Graham, J.M. Jr., Lachman R.S., Jabs E.W., Okajima K., Przylepa K.A., Shanske A., Chen K., Neidich J.A., and Wilcox W.R.: Subtle radiographic findings of achondroplasia in patients with Crouzon syndrome with acanthosis nigricans due to an Ala391Glu substitution in FGFR3. American Journal of Medical Genetics, 98:75-91, 2001.

119. Bar-Cohen, Y., Ferry D.A., Graham, J. M., Jr.: Cardiac surgery in an infant with CHARGE association from the Gaza strip. Medicine of the Americas, 2:103-108, 2001.

120. Graham J.M., Jr., Anyane-Yeboa K., Raams A., Appeldoorn E., Kleijer W.J., Garritsen V.H., Busch D., Edersheim T.G., and Jaspers N.G.J.: Cerebro-Oculo-Facio-Skeletal syndrome with a nucleotide excision-repair defect and a mutated *XPD* gene, with prenatal diagnosis in a triplet pregnancy. American Journal of Human Genetics, 69:291-300, 2001.

121. Wang R., Earl D.L., Ruder, R.O., Graham, J.M. Jr.: Syndromic ear anomalies and renal ultrasounds. Pediatrics, 108:e32, http://pediatrics.aappublication.org/content/108/2/e32.full.html, 2001.

122. Visootsak, J., Aylestock, M., Graham, J.M., Jr.: Klinefelter syndrome and its variants: an update and review for the primary pediatrician. Clinical Pediatrics, 40:639-691, 2001.

123. Graham J.M., Jr.: Editorial Comment: A recognizable syndrome within CHARGE association: Hall-Hittner syndrome. American Journal of Medical Genetics, 99:120-123, 2001.

124. Rosenberg M.J., Killoran C., Dziadzio L., Chang S., Stone D.L., Meck J., Aughton D., Bird L.M., Bodurtha J., Cassidy S.B., Graham, J.M., Jr., Grix A., Guttmacher A.E., Hudgins L., Kozma C., Michaelis R., Pauli R., Peters K.F., Rosenbaum K,N., Tifft C.J., Wargowski D., Williams M.S., and Biesecker, L.B.: Scanning for telomeric deletions and duplications and uniparental disomy using genetic markers in 120 children with malformations. Human Genetics, 109:311-318, 2001.

125. Villard L., Nguyen K., Cardoso C., Martin C.L., Weiss A.M., Silfry-Platt M., Grix A.W., Graham, Jr., J.M., Winter R.M., Leventer R.J., and Dobyns W.B.: A locus for bilateral perisylvian polymicrogyria maps to Xq28. American Journal of Human Genetics, 70:1003-1008, 2002.

126. Ming, J.E., Stiehm, J.R., Graham, Jr. J.M.: Genetic syndromes associated with immunodeficiency. Immunol Allergy Clin N Amer, 22:261-280, 2002.

127. Adams J., Bittner P., Buttar H.S., Chambers C.D., Collins T.F.X., Daston G.P., Filkins K., Flynn T.J., Graham, Jr., J.M., Jones K.L., Kimmel C., Lammer E., Librizzi R., Mitala J., Polifka J.E.: Statement of the Public Affairs Committee of the Teratology Society on the Fetal Alcohol Syndrome. Teratology 66:344–347, 2002.

128. Elliott, A.M., Graham, J.M., Jr., Curry, C.J.R., Pal, T., Rimoin, D.L., Lachman, R.S.: The spectrum of dolichospondylic dysplasia - two new patients with distinctive findings. American Journal of Medical Genetics113:351-361, 2002.

129. Wang, R. Martinez-Frias M.L., Graham, Jr. J.M.: Infants of diabetic mothers are at increased risk for the oculo-auriculo-vertebral sequence: a case-based and case-control approach. Journal of Pediatrics, 141:611-617, 2002.

130. Everman D.B., Bartels C.F., Yang Y., Yanamandra N., Goodman F.R., Mendoza-Londono J.R., Savarirayan R., White S.M., Graham, Jr. J.M., Gale R.P., Svarch E., Newman W.G., Kleckers A.R., Francomano C.A., Govindaiah V., Singh L., Morrison S., Thomas J.T., Warman M.L.: The mutational spectrum of Brachydactyly type C. American Journal of Medical Genetics, 112:291-296, 2002.

131. O'Connor M.J., Shah B., Whaley S., Cronin P., Gunderson B., Graham, Jr., J.M.: Psychiatric illness in a clinical sample of children with prenatal alcohol exposure. The American Journal of Drug and Alcohol Abuse, 28:743-754, 2002.

132. De Mollerat X.J., Everman D.B., Morgan C.T., Clarkson K.B., Rogers R.C., Colby R.S., Aylsworth A.S., Graham J.M., Jr., Stevenson R.E., Schwartz C.E.: p63 mutations are not a major cause of non-syndromic split hand/foot malformation. Journal of Medical Genetics, 40:55-61, 2003.

133. Savarirayan R., White S. M., Goodman F. R., Graham Jr., J. M., Delatycki, M. B., Lachman, R. S. Rimoin, D. L., Everman, D. B., Warman, M. L.: Broad phenotypic spectrum caused by an identical heterozygous CDMP-1 mutation in three unrelated families. American Journal of Medical Genetics, 117A:136-142, 2003.

134. Schweitzer, D.N., Earl, D.L., Lachman, R.S. Graham, J.M., Jr.,: Van Den Ende-Gupta syndrome of blepharophimosis, arachnodactyly, and congenital contractures: clinical delineation and recurrence in brothers. American Journal of Medical Genetics, 118A:267-273, 2003.

135. Lalani S.R., Stockton D.W., Bacino C, Molinari L.W., Glass N.L., Fernbach S.D., Towbin J.A., Craigen W.J., Graham Jr., J.M., Hefner M.A., Lin A.E., McBride K.L., Davenport S.L., Belmont J.W.: Toward a genetic etiology of CHARGE association: I. A systematic scan for submicroscopic deletions. American Journal of Medical Genetics, 118A:260-266, 2003.

136. Schweitzer D.N., Yano S., Earl D.L., Graham J.M., Jr.: Johnson-McMillin syndrome, a neuroectodermal syndrome with conductive hearing loss and microtia: report of a new case. American Journal of Medical Genetics, 120A:400-405, 2003.

137. Ming J.E., Stiehm E.R., Graham, Jr. J.M.: Genetic syndromes associated with immune abnormalities. Critical Reviews in Clinical Laboratory Sciences, 40(5):1-56, 2003.

138. Graham, J.M. Jr., Wheeler P., Tackels-Horne D., Lin A.E., Hall B.D., May M., Short K.M., Schwartz C.E., Cox T.C.: A new X-linked syndrome with agenesis of the corpus callosum, mental retardation, coloboma, micrognathia, and a mutation in the Alpha 4 gene at Xq13. American Journal of Medical Genetics, 123A:37-44, 2003.

139. Simpson J.L., de la Cruz F., Swerdloff R., Samango-Sprouse C., Skakkeback N., Graham, Jr., J.M., Hassold T., Aylstock M., Meyer-Bahlburg H.F.L., Willard H.F., Hall J.G., Salameh W., Boone, K., Staessen C., Geschwind D., Giedde J., Dobs A.S., Rogol A., Brinton B., Paulsen C.A.: Klinefelter syndrome: expanding the phenotype and identifying new research directions. Genetics in Medicine, 5:460-468, 2003.

140. Doyle T.F., Bellugi U., Korenberg J.R., Graham, Jr., J.M.: "Everybody in the world is my friend" hypersociability in young children with Williams syndrome. American Journal of Medical Genetics, 124A: 263-273, 2004.

141. Gleeson J.G., Keeler L.C., Parisi M.A., Marsh, S.E., Chance, P.F., Glass I.A., Graham J.M., Jr., Maria B.L., Barkovitch A.J., Dobyns W.B.: The molar tooth malformation of the midbrain-hindbrain junction: occurrence in multiple distinct syndromes. American Journal of Medical Genetics, 125A: 125-134, 2004.

142. Havens J.M., Visootsak J., Phelan M.C., Graham, Jr., J.M.: 22q13 deletion syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 43:43-54, 2004.

143. Krakow D, Robertson SP, King LM, Morgan T, Sebald ET, Bertolotto C, Wachsmann-Hogiu S, Acuna D, Shapiro SS, Takafuta T, Aftimos S, Kim CA, Firth H, Steiner CE, Cormier-Daire V, Superti-Furga A, Bonafe L, Graham JM Jr., Grix A, Bacino CA, Allanson J, Bialer MG, Lachman RS, Rimoin DL, Cohn DH. Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Nature Genetics, 36(4): 405-410, 2004.

144. Graham, J.M., Jr., Hennekam, R., Dobyns W.B., Roeder E., Busch D.: MICRO syndrome: an entity distinct from COFS syndrome. American Journal of Medical Genetics, 128A(3):235-245 2004.

145. Visootsak J., Rosner B., Dykens E., Schwartz C., Hahn K., White S., Szeftel R., Graham J.M., Jr.: Clinical and behavioral features of Borjeson-Forssman-Lehmann Syndrome patients with mutations in *PHF6*. Journal of Pediatrics, 145: 819-825, 2004.

146. Graham, J.M., Jr., Rosner B., Dykens, E., Visootsak J.: Behavioral features of CHARGE syndrome (Hall-Hittner syndrome): comparison with Down syndrome, Prader-Willi syndrome and Williams syndrome. American Journal of Medical Genetics, 133A: 240-247, 2005.

147. Issekutz K.A., Graham J.M., Jr., Prasad C., Smith I.M., Blake K.D.: An epidemiological analysis of CHARGE syndrome: preliminary results from a Canadian study. American Journal of Medical Genetics, 133A: 309-317, 2005.

148. Searle L.C., Graham J.M., Jr., Prasad C., Blake K.D.: CHARGE syndrome from birth to adulthood: an individual reported on from 0 to 33 years. American Journal of Medical Genetics, 133A: 344-349, 2005.

149. Graham J.M., Jr., Kreutzman J., Earl D., Halberg A., Samayoa C., Guo X.: Deformational brachycephaly in supine-sleeping infants. Journal of Pediatrics, 146:258-262, 2005.

150. Graham J.M., Jr., Gomez M., Halberg A., Earl D., Kreutzman J., Cui J., Guo X.: Management of deformational plagiocephaly: repositioning versus orthotic therapy. Journal of Pediatrics, 146:258-262, 2005.

151. Wang R., Visootsak J., Danielpour, M., Graham J.M. Jr.: Midline defects in FG syndrome: Does tethered spinal cord contribute to the phenotype? Journal of Pediatrics, 146:537-541, 2005.

152. Visootsak, J., Warren, S.T., Anido, A., Graham, Jr., J.M.: Fragile X syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 44:371-382, 2005.

153. Aligianis I.A., Johnson C.A., Gissen P., Chen D., Hampshire D., Hofmann K., Maina E., Morgan N.V., Tee L., Morton J., Ainsworth J.R., Horn D., Rosser E., Cole T., Stolte-Dijktra I., Fieggen K., Clayton Smith J., Megarbane A., Shields J., Newbury-Ecob R., Dobyns W.B., Graham J.M., Jr., Kjaer K.W., Warburg M., Bond J., Trembath R., Harris L.W., Takai Y., Mundlos S., Tannahill D., Woods C.G., Maher E.A.: Mutations in Rab3 GTPase activating protein (RAB3GAP) catalytic subunit cause Microphthalmia, Cataract, Microcephaly and Micropenis (Warburg MICRO syndrome). Nature Genetics, 37:221-224, 2005.

154. Johnston J.J., Olivos-Glander I., Killoran C., Elson E., Turner J., Peters K., Abbott M.H., Aughton D.J., Aylsworth A.S., Bamshed M.J., Booth, C., Curry C.J., David A., Dinulos M.B., Flannery D.B., Fox M.A., Graham, Jr., J.M., et al.: Molecular and clinical analyses of Greig cephalopolysyndactyly and Pallister Hall syndromes: Robust phenotype prediction from the type and position of GLI3 mutations. American Journal of Human Genetics, 76:609-622, 2005.

155. Armstrong L, Moneim AAE, Aleck, K, Aughton DJ, Baumann C, Braddock SR, Gillessen-Kaesbach G, Graham JM, Jr, et al.: Further delineation of Kabuki syndrome in 48 well-defined new individuals. American Journal of Medical Genetics, 132A:265-272, 2005.

156. White S.M., Graham, Jr. J.M., Kerr B., Gripp K., Weksburg R., Cytrynbaum C., Reeder J.L., Stewart F.J., Edwards M., Wilson M., Bankier A.: The adult phenotype in Costello syndrome. American Journal of Medical Genetics, 136A:128-135, 2005.

157. Graham, J.M., Jr.,: Marshall J. Edwards: discoverer of maternal hyperthermia as a human teratogen. Birth Defects Research (Part A), 73:857-864, 2005.

158. Graham, Jr., J.M. Shaw G.M.: Gene/environment interactions in rare diseases that include common birth defects. Birth Defects Research (Part A), 73:865-867, 2005.

159. Lalani, S.R., Safiullah, A.A., Fernbach S.D., Harutyunyan K.G., Thaller, C., Peterson L.E., McPherson J.D., Gibbs R.A., White L.D., Heffner M., Davenport S.L.H., Graham, Jr., J.M., Bacino C.A., Glass N.L., Towbin J.A., Craigen W.J., Neish S.R., Lin A.E., Belmont J.W.: Spectrum of CHD7 mutations in 110 individuals with CHARGE syndrome and genotype-phenotype correlation. American Journal of Human Genetics, 78:303-314, 2006.

160. Graham, J.M., Jr., Lee J.: Bosma arhinia microphthalmia syndrome. American Journal of Medical Genetics, 140A:189-193, 2006.

161. Graham J.M., Jr.: Tummy time is important. Clinical Pediatrics, 45:119-221, 2006.

162. Doswell, B,H., Visootsak J., Brady A.N., Graham, Jr., J.M.: Turner syndrome: an update and review for the primary pediatrician. Clinical Pediatrics, 45:301-313, 2006.

163. Graham J.M., Jr., Kumar A.: Diagnosis and management of extensive vertex birth molding. Clinical Pediatrics, 45:672-678, 2006.

164. Wang R., Jones J.R., Chen S., Rogers R.C., Friez M.J., Schwartz C.E., Graham Jr., J.M.: A previously unreported mutation in a Currarino syndrome kindred. American Journal of Medical Genetics, 140A: 1923-1930, 2006.

165. Adam, M.P., Schelley S., Gallagher R., Brady A.N., Barr K., Blumberg B., Shieh J.T.C., Graham, Jr., J.M., Slavotinek A., Martin M., Keppler-Noreuil K., Storm A.L., Hudgins L: Clinical features and management issues in Mowat-Wilson syndrome. American Journal of Medical Genetics, 140A: 2730-2741, 2006 (winner of John Opitz award for best paper, 2006).

166. Conway R, Danielpour M, Graham Jr., J.M.: Surgical management of cerebellar tonsillar herniation in three patients with macrocephaly-cutis marmorata telangiectatica congenita. Journal of Neurosurgery (4 Suppl Pediatrics), 106:296-301, Editorial and Response 292-295, 2007.

167. Risheg H., Graham, Jr., J.M., Clark R.D., Rodgers R.C., Opitz J.M., Moeschler J.B., Pfeiffer A.P., May M., Joseph S.M., Jones J.R., Stevenson R.E., Schwartz C.E., Friez M.J.: A recurrent mutation in *MED12* leading to R961W causes Opitz-Kaveggia syndrome. Nature Genetics, 39:451-453, 2007, published online March 4, 2007.

168. Chen C, Visootsak J, Dills S, Graham Jr., JM: Prader-Willi Syndrome: An Update and Review for the Primary Pediatrician. Clinical Pediatrics, 46:580-591, 2007.

169. Ballif BC, Hornor SA, Jenkins E, Madan-Khetarpah S, Surti U, Jackson K, Asamoah A, Farnsworth PL, Gordon C. Gowans GC, Conway RL, Graham, Jr. JM, Medne L, Zackai EH, Tamim H. Shaikh TH, Peggy S. Eis PS, Bassem A. Bejjani BA, Shaffer LG: Discovery of a previously unrecognized microdeletion syndrome of 16p11.2-p12.2. Nature Genetics, 39:1071-1073, 2007, published online August 19, 2007.

170. Visootsak J, Rosner B., Dykens E., Tartaglia N., Graham, Jr., J.M.: Behavioral phenotype of sex chromosome aneuploidies: 48,XXYY, 48,XXXY, and 49,XXXXY. American Journal of Medical Genetics, 143A:1198-1203, 2007.

171. Borozdin W, Graham, Jr. JM, Bohm D, Bamshed MJ, Spranger S, Burke L, Olney AH, Leipoldt M, Kohlhase J: Multigene deletions on chromosome 20q13.13-q13.2 including SALL4 result in an extended phenotype of Okihiro syndrome plus developmental delay. Human Mutation, 28:830, published online August 2007.

172. Vatanavicharn N., Graham Jr., J.M., Curry C.J., Pepkowitz S., Lachman R.S., Rimoin D.L., Wilcox, W.R.: Diaphanospondylodysostosis: six new cases and exclusion of the candidate genes, *Pax1* and *Meox1*. American Journal of Medical Genetics, 143A:2292-2302 2007.

173. Conway R.L., Pressman B., Dobyns B., Danielpouir M., Lee, J., Butler M.G., Zachai E., Close L., Saitta S.C., Clericuzio C., Milunsky J., Hoyme G., Shieh J., Moeschler J.B., Crandall B., Lauzon J.L., Graham Jr., J.M.: Neuroimaging findings in macrocephaly-capillary malformation syndrome: a longitudinal study of 17 patients. American Journal of Medical Genetics, 143A:2981-3008, 2007.

174. Sanchez-Lara, P.A., Graham, Jr., J.M., Lee J., Hing A.V., Cunningham M.: The morphogenesis of wormian bones: a study of craniosynostosis and purposeful cranial deformation. American Journal of Medical Genetics, 143A:3243-3251, 2007.

175. Laury A., Sanchez-Lara P.A., Pepkowitz S., Graham, Jr., J.M.: A study of 534 fetal pathology cases from prenatal diagnosis referrals analyzed from 1989 through 2000, American Journal of Medical Genetics, 143A:3107-3120, 2007.

176. Graham, Jr., J.M., Visootsak, J., Huddleson L., Dykens, E., Clark, R.D., Jones, K.L., Moeschler, J.B., Opitz J.M., Morford J., Simensen R., Rogers, R.C., Lyons M.J., Schwartz,, C.E., Friez, M.J., Stevenson, R.E.: Behavioral features in 10 patients with FG syndrome (Opitz-Kaveggia syndrome) and the p.R961W mutation in the MED12 gene. American Journal of Medical Genetics, 146A:3011-3017, 2008.

177. Lyons, M.J., Graham, Jr., J.M., Neri, G., Hunter, A.G.D., Clark, R.D., Rogers, R.C., Moscarda, M., Simensen, R., Dodd, J., Robertson S., Dupont, B.R., Friez, M.J., Schwartz C.E., Stevenson, R.E.: Clinical experience in the evaluation of 30 patients with a prior diagnosis of FG syndrome. Journal of Medical Genetics, 46:9-13, 2009.

178. Adam, M.P., Hudgins L., Carey, J.C., Hall, B.D., Coleman K., Gripp K.W., Perez-Aytes A., Graham, Jr., J.M.: Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Birth Defects Research Part A; Clinical and Molecular Teratology, 85:13-19, 2009.

179. Hall, B.D., Graham, Jr., J.M., Cassidy, S.B., Opitz, J.M.: Elements of morphology: standard terminology for the periorbital region. American Journal of Medical Genetics, 149A:29-39, 2009.

180. Dai, L., Bellugi, U., Chen, X.-N., Pulst-Korenberg, A.M., Jarvinen-Pasley, A., Tirosh-Wagner, T., Eis P.S., Graham, Jr., J.M., Mills, D., Searcy, Y., Korenberg J.R.: Is it Williams syndrome? GTF2IRD1 implicated in visual-spatial construction and GTF2I in sociability revealed by high resolution arrays. American Journal of Medical Genetics, 149A:302-314, 2009.

181. Graham J.M., Jr., Kramer, N., Bejjani, B., Thiel, C.T., Carta, C., Neri, G, Tartaglia M., Zenker, M.: Genomic duplication of *PTPN11* is an uncommon cause of Noonan syndrome. American Journal of Medical Genetics, 149A:2122-2128, 2009.

182. Pariani M.J., Spencer A., Graham, Jr., J.M., Rimoin D.L.: A 785 kb deletion of 3p14.1p13, including the FOXP1 gene, associated with speech delay, contractures, hypertonia and blepharophimosis. European Journal of Medical Genetics, 52:123-127, 2009.

183. Clark, R.D. Graham, Jr., J.M., Friez M.J., Hoo, J.J., Jones, K.L., McKeown C., Moeschler, J.B., Raymond F.L., Rogers, R.C., Schwartz, C.E., Battaglia A., Lyons M.J., Stevenson, R.E.: The clinical phenotype of FG (Opitz-Kaveggia): an algorithm for diagnostic testing. Genetics in Medicine, 11:769-775, 2009.

184. Visootsak J. and Graham, Jr. J.M.: Social function in multiple X and Y chromosome disorders: XXY, XYY, XXYY, XXXY. Developmental Disabilities Research Reviews, 15:328-332, 2009.

185. El-Gharbawy A.H., Peeden J.N., Lachman R.S., Graham, Jr., J.M., Moore S.R., Rimoin D.L.: Severe cleidocranial dysplasia and hypophosphatasia in a child with microdeletion of the C-terminal region of *RUNX2*. American journal of Medical Genetics, 152A:169-174, 2010.

186. Sanchez-Lara P.A., Carmichael S.L., Graham, Jr., J.M., Lammer E.J., Shaw G. M., Ma C., Rasmussen S.A., National Birth Defects Prevention Study: Fetal constraint as a potential risk factor for craniosynostosis. American Journal of Medical Genetics, 152A:394-400, 2010.

187. Graham J.M., Jr., Spencer A.H., Grinberg I., Niesen C., Platt L.D., Maya M., Namavar Y., Baas F., Dobyns W.: Molecular and neuroimaging findings in pontocerebellar hypoplasia type 2 (PCH2): Is prenatal diagnosis possible? American Journal of Medical Genetics, 152A:2268-2276, 2010.

188. Graham Jr., J.M., Clark R.D., Moeschler J.B., Rogers R.C.: Behavioral features in young adults with FG syndrome (Opitz-Kaveggia syndrome). American Journal of Medical Genetics, 154C:477-485, 2010.

189. Johnston JJ, Sapp JC, Turner JT, Amor D, Artimos S, Aleck KA, Bocian J, Bodurtha JN, Cox G, Curry C, Day R, Donnai D, Field M, Fujiwara I, Gabbet M, Gal M, Graham Jr. JM, Hedera P, Hennekam RCM, Hirsch J., Hopkin R., Jabs EW, Kayserili H, Kidd AMJ, Kimonis V, Lin AE, Lynch SA, Maisenbacher M, Mansour S, McGaughran J, Mehta L, Murphy H, Ragada M, Robin NH, Rope AF, Rosenbaum KN, Schaefer GB, Shealy A, Smith W, Soller M, Sommer AM, Stalker HJ, Steiner B, Stefan M, Tilstra D, Tomkins S., Trapane P, Tsai A, Van Allen MI, Vasudevan PC, Zabel B, Zunich J, Black GC, Biesecker LB: Molecular analysis expands the spectrum of phenotypes associated with *GLI3* mutations. Human Mutation, 10:1142-1154, 2010.

190. Namavar Y, Barth PG, Kasher PR, Brockman K, Bernerr G, Writzl K, Ventura K, Neufeld-Kaiser W, Ferrierro DM, Basel-Vanagaite L, Eggens V, Krageloh-Mann I, De Meirleir L, King M, Graham, Jr. JM, von Moers A, Knoers N, Sztriha L, Korinthenberg R, Dobyns WB, Baas F, Poll-The BT: Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 134:143-156, 2011.

191. Busse T., Graham, Jr., J.M., Feldman G., Perin J., Catherwood A., Knowlton R., Rappaport E.F., Emanuel B., Driscol D.A., Saitta S.C.: Hign resolution genomic arrays identify CNVs that phenocopy the chromosome 22q11.2 deletion syndrome, Human Mutation, 32:91-97, 2011.

192. Bern D., Yoshimura S-I, Nunes-Bastos R., Bond F.F., Kurlan M.A., Rahman F., Handley M.T.W., Hadzhiev Y., Masood I., Straatman-Iwanowska A.A., Cullinane A.R., McNeill A., Pasha S.S., Kirby G.A., Foster K., Ahmed Z., Morton J.E., Williams D., Graham, Jr. J.M., Dobyns W.B., Burglen L., Ainsworth J.R., Gissen P., Muller F, Maher E.R., Barr F.A., Aligianis I.A.: Loss-of-function mutations in RAB18 cause Warburg Micro syndrome. American Journal of Human Genetics 88:499-507, 2011.

193. Nemec U., Nemec S.F., Krakow D., Brugger P.C., Malinger G., Graham J.M., Jr., Rimoin D.L., Prayer D.: The skeleton and musculature on foetal MRI. Insights Imaging, 2:309-318, 2011.

194. Burkardt D.D., Rosenfeld J.A., Helgeson M.L. Angle B., Banks V., Smith W.E., Gripp K.W., Moline J., Moran R.T., Niyazov D.M., Stevens C.A., Zackai E., Lebel R.R., Ashley D.G., Kramer N., Lachman R.S., Graham, J.M. Jr. Distinctive phenotype in 9 patients with deletion of chromosome 1q24-q25. American Journal of Medical Genetics, 155A:1336-1351, 2011.

195. Nemec S., Kasprian G., Brugger P., Bettelheim D., Amann G., Nemec U., Rotmensch S., Graham, J.M., Jr, Rimoin D.L, Lachman R., Prayer D.: Abnormalities of the upper extremity on fetal MRI. Ultrasound in Obstetrics and Gynecology, 38:559-567, 2011.

196. Nemec S.F., Ursula U., Weber M., Kasprian G., Brugger P. C., Krestan C. R., Rotmensch S., Rimoin D. L., Graham J. M., Jr, Prayer D.: Male sexual development *in utero*: testicular descent on prenatal MRI. Ultrasound in Obstetrics and Gynecology, 38:688-694, 2011.

197. Nemec S., Nemec U., Weber M., Rotmensch S., Kasprian G., Brugger P., Krestan C.R., Rimoin D.L, Graham, J.M., Jr, Prayer D.: Female external female genitalia on fetal MRI. Ultrasound in Obstetrics and Gynecology, 38:695-700, 2011.

198. Nemec S.F., Höftberger R., Nemec U., Bettelheim D., Brugger P. C., Kasprian G., Amann G., Rotmensch S., Graham J. M. Jr., Rimoin D. L., Daniela Prayer D. Fetal akinesia and associated abnormalities on prenatal MRI. Prenatal Diagnosis, 31:484-490, 2011.

199. Nemec S.F., Kasprian G., Brugger P. C., Bettelheim D., Nemec U., Krestan C. R., Rotmensch S., Rimoin D. L., Graham J. M. Jr., Prayer D. Abnormalities of the penis in utero – hypospadius on fetal MRI. J Perinatal Med. 39:451-456, 2011.

200. Nemec S. F., Horcher E., Kasprian G., Brugger P.C., Bettelheim D., Amann G., Nemec U., Rotmensch S., Rimoin D.L., Graham J.M., Jr., Prayer D.: Tumor disease and associated congenital abnormalities on prenatal MRI. European Journal of Radiology, 81:e115-e122, 2012.

201. Nemec S. F., Nemec U., Weber M., Brugger P.C., Bettelheim D., Krestan C. R., Rimoin D. L., Graham J. M. Jr., Prayer D.: Penile biometry on prenatal MR imaging. Ultrasound in Obstetrics and Gynecology, 39:330-335, 2012.

202. Nemec S., Nemec U., Bettelheim D., Brugger P.C., Horcher E., Schopf V., Graham J.M. Jr., Rimoin D.L., Prayer D.: Ovarian cysts on prenatal MRI. European Journal of Radiology, 81:1934-1944, 2012.

203. Nemec S.F., Brugger P.C., Nemec U., Bettelheim D., Kasprian G., Amann G., RimoinD.L., Graham J.M. Jr., Prayer D.: Situs anomalies on prenatal MRI. European Journal of Radiology, 81:e495-e501, 2012.

204. Klaassens M., Reinstein E., Hilhorst-Hofstee Y., Schrander J.J.P., Malfait F., Staal H., ten Have L.C., Blaauw J., Roggeveen H.C.J., Krakow D., De Paepe A., van Steensel M.A.M., Pals G., Graham, Jr., J.M., Schrander-Stumpel C.T.R.M.: Ehlers-Danlos syndrome arthrochalasia type (VIIA-B) – expanding the phenotype: from prenatal life through adulthood. Clinical Genetics, 82:121-130, 2012.

205. Noh G. J., Graham J. M., Jr.: 2q23.1 microdeletion of the *MBD5* gene in a female with seizures, developmental delay and distinct dysmorphic features, European Journal of Medical Genetics, 55: 354-357, 2012.

206. Graham J. M., Jr., GLUT1 deficiency syndrome as a cause of encephalopathy that includes cognitive disability, treatment-resistant infantile epilepsy and a complex movement disorder. European Journal of Medical Genetics 55(5):332-334, 2012.

207. Noh G.J., Tavyev Asher Y. J., Graham, J. M., Jr.: Clinical review of genetic epileptic encephalopathies. European Journal of Medical Genetics, 55(5):281-298, 2012.

208. O'Leary R.E., Shih J.C., Hyland K., Kramer N., Tavyev Asher Y.J., Graham J.M., Jr.: De novo microdeletion of Xp11.3 exclusively encompassing the monoamine oxidase A and B genes in a male infant with episodic hypotonia: a genomics approach to personalized medicine. European Journal of Medical Genetics, 55(5):349-353, 2012.

209. Nemec S.F., Nemec U., Brugger P.C., Bettelheim D., Rotmensch S., Graham J. M. Jr., Rimoin D. L., Daniela Prayer D.: MR imaging of the fetal musculskeletal system. Prenatal Diagnosis, 32:205-213, 2012.

210. Rimoin L., Graham J.M., Jr.: Blistering skin disorders in the neonate. Clinical Pediatrics, 51:685-688, 2012.

211. Rimoin L., Graham J.M., Jr.: Ichthyotic skin disorders in the neonate. Clinical Pediatrics, 51:791-795, 2012.

212. Mirzaa G.M., Conway R.L., Gripp K.W., Lerman-Sagie T., Siegel D.H., deVries L.S., Lev D., Kramer N., Hopkins E., Graham, J.M. Jr., Dobbyns W.B. Megalencephaly-Capillary Malformation (MCAP) and Megalencephaly- Polydactyly- Polymicrogyria-Hydrocephalus (MPPH) syndromes: two closely-related disorders of brain overgrowth and abnormal brain and body morphogenesis. American Journal of Medical Genetics, Part A, 158A: 269-291, 2012 (Winner of 2013 John M. Opitz Young Investigator Award from Wiley-Liss for the best 2012 paper by a fellow in the American Journal of Medical Genetics).

213. Lee H., Graham, Jr., J.M., Rimoin D.L., Lachman R.S., Krejci P., Nelson S.F., Krakow D., Cohn D.H.: Exome sequencing identifies mutations in *PDE4D* in acrodysostosis. American Journal of Human Genetics, 90:746-751, 2012.

214. Nemec U., Nemec S.F., Brugger P.C., Bettelheim D., Weber M., Graham J.M. Jr., Rimoin D.L., Prayer D.: Male genital abnormalities in intrauterine growth restriction, Prenatal Diagnosis, 32:4 27-431, 2012.

215. Nemec U., Nemec S.F., Weber M., Brugger P.C., Bettelheim D., Wadhawan I., Kolb A., Graham J.M. Jr., Rimoin D.L., Prayer D.: Clubfeet and associated abnormalities on fetal magnetic resonance imaging. Prenatal Diagnosis, 32:822-828, 2012. 2012.

216. Moh W., Graham Jr. J.M., Isha Wadhawan I, Sanchez-Lara Pa: Extrinsic factors influencing fetal deformations and intrauterine growth restriction. Journal of Pregnancy, 2012:750485. Epub 2012 Jul 19.

217. Riviere J-B, Mirzaa GM, O'Roak BJ, Beddaoui M, Alcanrtara D., Conway RJ, St-Onge J, Schwartzentruber, JA, Gripp, KW, Nikkel, SM, Worthylake T, Sullivan CT, Ward TR, Butler HE, Kramer NA, Albrecht B, Armour CM, Armstrong L, Caluferiu O, Cytrynbaum C, Drolet BA, Innes AM, Lauzon JL, Lin AE, Mancini GMS, Meschino WS, Reggin JD, Saggar AK, Lerman-Sagie T, Uyanik G, Weksberg R, Zirin B, Beaulieu CL, FORGE Canada Consortium, Majewski J, Bulman DE, O'Driscoll M, Shendure J, Graham Jr. JM, Boycott KM, Dobyns WB: *De novo* germline and postzygotic mutations in *AKT3*, *PIK3R2* and *PIK3CA* cause a spectrum of related megalencephaly syndromes, Nature Genetics, 44(8):934-40. 2012.

218. Shah H., Bens S., Caliebe A., Graham Jr. J.M., Girisha K.M.: Growth retardation, intellectual disability, facial dysmorphism, cataract, thoracic hypoplasia and skeletal abnormalities: a novel phenotype. American Journal of Medical Genetics, Am J Med Genet Part A. 158A: 2941-2945, 2012

219. Reinstein E, Frentz S, Morgan T, Garcıa-Minaur S, Leventer RJ, McGillivray G, Pariani1 M, van der Steen A, Pope M, Holder-Espinasse M, Scott R, Thompson EM, Robertson T, Coppin B, Siegel R, Zurita MB, Rodriguez JI, Morales1 C, Rodrigues Y, Arcas J, Saggar A, Horton M, Zackai E, Graham, Jr., JM, Rimoin DL and Robertson SP: Vascular and connective tissue anomalies associated with X-linked periventricular heterotopia due to mutations in *FLNA*. European Journal of Human Genetics, 21:494-502, 2013.

220. McDonell L.M., Mirzaa G.M, Alcantara D., Schwartzentruber J., Carter M., Clericuzio C.L., Graham Jr., J.M., Rosendahl D., Polster, Acsadi T.G., Townshend, S., Williams, S., Isidor B., Smyser C., Paciokowski A., Willing M., Beaulieu C.L., Marcadier J., Geraghty M.T., Majewski J., Bulman D.E., Dobyns W.B., O'Driscoll M., Boycott, K.M. on behalf of the FORGE Canada Consortium. Microcephaly-Capillary Malformation Syndrome: A Novel RASopathy Caused by Mutations in the Deubiquitinating Enzyme *STAMBP*. Nature Genetics 45: 556-562, 2013.

221. Alkalay AL, Graham, Jr. JM, Kotton RH, Forbis SA, Fuscaldo MC, Simmons CF, Klein AH: Very low birth weight infant outcome stratified by gestational age and birth weight. Neonatal Intensive Care 24:18-23, 2013.

222. Russell B., Graham Jr., J.M.: Expanding our knowledge of conditions associated with the *ASXL* gene family. Genome Medicine, 5:16, 2013.

223. Mirzah G., Conway R., Graham Jr., J.M., Dobyns W.B.: PIK3CA-Related Segmental Overgrowth. In: Pagon RA, Adam MP, Bird TD, et al., editors. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2013. Available from: http://www.ncbi.nlm.nih.gov/books/NBK153722/ Aug. 15, 2013.

224. Graham, Jr. JM, and Schwartz CE: *MED12*-related disorders. American Journal of Medical Genetics Part A, 161A: 2734-2740, 2013.

225. Handley MT, Morris-Rosendahl DJ, Brown S, Macdonald F, Hardy C, Bem D, Carpanini SM, Borck G, Martorell L, Izzi C, Faravelli F, Accorsi P, Pinelli L, Lina Basel-Vanagaite LB, Peretz G, Abdel-Salam GMH, Zaki MS, Jansen A, Mowat D, Glass I, Stewart H, Mancini G, Lederer D, Roscioli T, Giuliano F, Plomp AS, Rolfs A, Graham Jr JM, Seemanova E, Poo P, Garcıa-Cazorla A, Edery P, Jackson IJ, Eamonn R. Maher ER, Aligianis IA: Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype–phenotype correlations in Warburg Micro syndrome and Martsolf syndrome. Human Mutatation 34:686–696, 2013.

226. Santen GWE, Aten E, Vulto-van Silfhout AT, Pottinger C, van Bon BW, Van Minderhout IJ, Snowdowne R, van der Lane CA, Boogaard M, Linssen MM, Vijhuizen L, van der Wielen MJR, Vollebregt MJE, Vollebregt MJ; the Coffin-Siris consortium Breuning MH, Kriek M, van Haeringen A, den Dunnen JT, Hoischen A, Clayton-Smith J, de Vries BB, Hennekam RC, van Belzen MJ, Almureikhi M, Baban A, Barbosa M, Ben-Omran T, Berry K, Bigoni S, Boute O, Brueton L, van der Burgt I, Canham N, Chandler KE, Chrzanowska K, Collins AL, de Toni T, Dean J, den Hollander NS, Flore LA, Fryer A, Gardham A, Graham JM Jr, Harrison V, Horn D, Jongmans MC, Josifova D, Kant SG, Kapoor S, Kingston H, Kini U, Kleefstra T, Krajewska-Walasek M, Kramer N, Maas SM, Maciel P, Mancini GM, Maystadt I, McKee S, Milunsky JM, Nampoothiri S, Newbury-Ecob R, Nikkel SM, Parker MJ, Pérez-Jurado LA, Robertson SP, Rooryck C, Shears D, Silengo M, Singh A, Smigiel R, Soares G, Splitt M, Stewart H, Sweeney E, Tassabehji M, Tuysuz B, van Eerde AM, Vincent-Delorme C, Wilson LC, Yesil G: Coffin-Siris syndrome and the BAF complex: genetype-phenotype study in 63 patients. Human Mutation 34:1519-1528, 2013.

227. Ezgu F.S., Krejci P., Li S., deSousa C, Graham, Jr. J.M., Hansmann I., He W., Porpora K., Wand D., Wertelecki W., Schneider A., Wilcox W.R.: Phenotype-genotype correlations in patients with the Marinesco-Sjogren syndrome. Clinical Genetics, 86:74-84, 2014.

228. Burkhardt D.D., Graham, Jr. JM, Short S.S., Frykman P.: Advances in Hirschsprung disease genetics and treatment strategies: an update for the primary care pediatrician. Clinical Pediatrics, 53:71-81.2014.

229. Au PY, Racher HE Graham, Jr. JM, Kramer N., Lowery RB, Parboosingh JS, Innes AM, FORGE Canada Consortium: De novo exon 1 missense mutations of *SKI* and Shprintzen-Goldberg syndrome: two new cases and a clinical review. American Journal of Medical Genetics Part A, 164: 346-352, 2014.

230. Kuo AA, Tritasavit S, Graham Jr., J.M.: Congenital muscular torticollis and positional plagiocephaly. Pediatrics in Review 35:79-87; 2014.

231. Girisha KM, Abdollahpour H, Shah H, Bhavani SLG, Graham JM Jr., Boggula VR, Phadke SR, Kutsche K: A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers and scoliosis, American Journal of Medical Genetics, Patr A, 164:1035-1040, 2014.

232. Babkina N, Graham Jr, JM: New genetic testing in prenatal diagnosis. Seminars in Fetal and Neonatal Medicine, 19: 214- 219, 2014.

233. Keppler-Noreuil KM, Sapp JC, Lindhurst MJ, Parker VER, Blumhorst C, Darling T, Tosi LL, Huson SM, Whitehouse RW, Jakkula E, Grant I, Balasubramanian M, Chandler KE, Fraser JL, Gucev Z, Crow YJ, Manace LM, Clark RD, Sellars EA, Pena LDM, Krishnamurth V, Shuen A, Braverman N, Cunningham ML, Sutton VR, Tasic V, Graham Jr JM, Geer Jr J, A. Henderson A, Semple RK, Biesecker LG: Clinical delineation and natural history of *PI3KCA* –related overgrowth spectrum. American Journal of Medical Genetics, Part A 164:1713-33., 2014.

234. Hunter AG, Graham JM, Neri G, Rogers RC, Stevenson RE, Turner G, Friez MJ. The intellectual disabilities evaluation and advice system (IDEAS): Outcome of the first 55 cases. Am J Med Genet Part A 164A: 1102–1117, 2014.

235. Dhamija R, Graham, Jr, JM, Smaoui, N, Erik Thorland E, Kirmani S: Novel de novo *SPOCK1* mutation in a proband with developmental delay, microcephaly and agenesis of corpus callosum. European Journal of Medical Genetics 57: 181e-184, 2014.

236. McMillin MJ, Beck AE, Chong JX, Shively KM, Buckingham KJ, Gildersleeve HI, Aracena MI, Aylsworth AS, Bitoun P, Carey J, Clericuzio CL, Crow YJ, Curry CJ, Devriendt K, Everman DB, Fryer A, Gibson K, Giovannuci-Uzielli ML, Graham Jr. JM, Hall JG, Hecht JT, Heidenreich RA, Hurst JA, Irani S, Krapels IPC, Leroy JG, Mowat D, Plant G, Robertson SP, Schorry EK, Stumpel C, Scott RH, Seaver LH, Sherr E, Splitt M, Stewart H, Temel S, Weaver DW, Whiteford M, Williams MS, Tabor HK, Smith JD, Shendure J, Nickerson DA, University of Washington Center for Mendelian Genomics, Bamshad MJ: Mutations in P*IEZO2* Cause Gordon Syndrome, Marden-Walker Syndrome and Distal Arthrogryposis Type 5. The American Journal of Human Genetics, 94: 734-744, 2014.

237. Shih EM, Graham Jr. JM: Review of genetic and environmental factors leading to hypospadias. European Journal of Medical Genetics, 57: 453-463, 2014.

238. Graham Jr. JM, Hennekam RC: Genetics of common malformations. European Journal of Medical Genetics, 57:353-354, 2014.

239. Castillo A, Kramer N, Schwartz CE, Miles JH, DuPont BR, Rosenfeld JA, Graham Jr, JM: 19q13.32 microdeletion syndrome; three new cases. European Journal of Medical Genetics, 57:654-658, 2014.

240. Russell B, Johnston JJ, Biesecker LG, Kramer N, Pickart A, Rhead W, Tan W-H, Brownstein CA, Clarkson LK, Dobson A, Rosenberg AZ, Graham Jr., JM: Clinical management of patients with *ASXL1* mutations and Bohring-Opitz Syndrome, emphasizing the need for Wilms tumor surveillance. American Journal of Medical Genetics, in press, April 2015.

241. Barber JCK, Rosenfeld JA, Graham, Jr. JM, Kramer N., Lachlan K, Bateman MS, Collinson MN, Stadheim BF, Turner CLS, Gauthier JN, Reimschisell TE, Queshi AM, Dabir TA, Humphries MW, Marble M, Huang T, Beal SJ, Massiah J, Taylor EJ, Wyn S: Inside the 8p23.1 duplication syndrome; eight microduplications of likely or uncertain clinical significance. American Journal of Medical Genetics, in press, April 2015.

242. Wentzensen IM, Johnston JJ, Patton JH, Graham JM Jr., Sapp JC, Biesecker LG: Exome sequencing identifies a mutation in *OFD1* in a male with Joubert Syndrome, orofaciodigital spectrum anomalies, and complex polydactyly. Submitted to Human Genome Variation, April 2015.

243. Babkina N, Deignan JL, Lee H, Vilain E, Sanjar R, Giurgea I, Mowat D, Graham Jr. JM: Early infantile epileptic encephalopathy with a *de novo* variant in *ZEB2* identified by exome sequencing. Submitted to European Journal of Medical Genetics, April 2015.

244. Tenney J, Graham Jr, JM, Dobyns WB, Gleeson JG: A male with preaxial polydactyly, Joubert syndrome and *OFD1* mutation discovered by exome sequencing. In preparation, 2015.

245. Wyszynski D.F., Carmen W.J., Cantor A.B., Graham, Jr., J.M., Kunz L.H., Slavotinek A.M., Kirby R.S. Seeger J.: Pregnancy and birth outcomes among women with idiopathic thrombocytopenic purpura. In preparation, 2015.

246. Mirzaa G.M., Conway R.L., Kramer N., Graham, Jr. J.M., Dobyns W.B.: The megalencephaly-capillary malformation (MCAP) syndrome without megalencephaly: expanding the spectrum of a unique overgrowth disorder. In preparation, 2015.

247. Mencias I, Vatanavicharn N, Borozdin W, Kohlhase J, Graham, Jr., J.M.: Wildervanck syndrome: an asymmetrical phenotype with discordant expression in monozygous twins. In preparation, 2015.

248. Reinstein E., Graham, Jr, J.M., Falk R, Schreck R: Double autosomal/sex chromosome aneuploidy revisited: mechanisms reviewed. In preparation, 2015.

249. Farington-Rock C., Graham, Jr., J.M., Taylor K.D., Kirilova V., Cohn D., Roberts S.P., Krakow D.: Germline and somatic mosaicism for filamin B mutations in Larsen syndrome. In revision for Journal of Medical Genetics, 2015.

250. Beck A.E., Hudgins L., Carey J.C., Graham, Jr., J.M., Grix A.W., Dent K.M., Chen E., Abdul-Rahman O., Kimonos V., Lazzeroni L.C., Bamshad M., Hoyme H.E., Francke U: Phenotypic analysis of familial oculo-auriculo-vertebral spectrum, In preparation, 2015.

INVITED RESEARCH PUBLICATIONS

1. Graham J.M. Jr.: Craniostenosis: A new approach to management. Pediatric Annals, 10:258-264, 1981.

2. Walzer S., Graham J.M. Jr., Bashir A.S. and Silbert A.R.: Preliminary observations on language and learning in XXY boys. Birth Defects Original Article Series, 18(4):185-192, 1982.

3. Koskinen-Moffett L.K., Moffett B.C. and Graham J.M. Jr.: Cranial synostosis and intrauterine compression: a developmental study of human sutures, In: Factors and Mechanisms Influencing Bone Growth, New York, AR Liss, Inc., pp. 365-378, 1982

4. Graham J.M. Jr. Alterations in head shape as a consequence of fetal head constraint. Seminars in Perinatology, 7:257-269, 1983.

5. Graham J.M. Jr., Limb anomalies as a consequence of spatially-restricting uterine environments. In: Fallon JF and Caplan AI (Eds.) Limb Development and Regeneration: Part A, New York, AR Liss, Inc., pp. 413-422, 1983.

6. Graham J.M. Jr., Harris M., Frank J.E., Little G.A. and Klein R.Z., Congenital hypothalamic hamartoblastoma syndrome: Natural history and genetic implications., In: Bartsocas CS (Ed.) Endocrine Genetics and Genetics of Growth, New York, AR Liss, Inc., pp 163-174, 1985.

7. Graham J.M. Jr., The effects of alcohol consumption during pregnancy. Prevention of Physical and Mental Defects, Part C: Basic and Medical Science, Education, and Future Strategies, New York, AR Liss, Inc., pp 335-339, 1985.

8. Graham J.M. Jr., The association between limb anomalies and spatially-restricting uterine environments. Prevention of Physical and Mental Defects, Part C: Basic and Medical Science, Education, and Future Strategies, New York, A.R. Liss, Inc., pp. 99-103, 1985.

9. Walzer S., Bashir A.S., Graham J.M. Jr., Silbert A.R., Lange N.T., Denapoli M.F. and Richmond J.B.: The behavioral development of boys with X chromosome aneuploidy: Impact of reactive style on the educational intervention for learning deficits. Birth Defects Original Article Series 22(3):1-21, 1986.

10. Graham J.M. Jr. and Edwards M.J., Teratogenic effects of maternal hyperthermia. In: Proceedings of the 9th Environmental Medicine Symposium: Environmental Factors and Developing Brain, Supplement of Annals of the Research Institute of Environmental Medicine, pp. 365-374, Nagoya University, Japan, 1989.

11. Graham J.M. Jr., Clinical approach to human central nervous system defects. In: Proceedings of the 9th Environmental Medicine Symposium: Environmental Factors and Developing Brain, Supplement of Annals of the Research Institute of Environmental Medicine, pp. 375-387, Nagoya University, Japan, 1989.Graham JM Jr., Meill E.A. Cardiac defects in CHARGE Association, (parent support organization newsletter) CHARGE Accounts, 2(1):2-3, 1991.

12. Sharony R., Pepkowitz S., Hixon H., Machin, G.A., Graham J.M. Jr.: Diprosopus - A pregastrulation defect involving the head, neural tube, heart and diaphragm. Birth Defects: Original Article Series, 29:211-225, 1993.

13. Sharony R., Hixon H., Pepkowitz S., Carlson D., Platt L., Graham J.M. Jr.: Experience with a fetal dysmorphology/pathology service in an academic medical center. Birth Defects: Original Article Series, 29:195-203, 1993.

14. Graham J.M. Jr.: Plagiocephaly-torticollis deformation sequence. Gene Scene, 3(1):3-4, 1998.

15. Lawand C., Prasad, C., Graham, Jr. J.M., Blake, K.D.: CHARGE association - looking ahead. Canadian Paediatric Surveillance Program Resource Binder. 1999-2002.

16. Blake K., Graham, Jr., J.M., Prasad C., Smith I.S.: Quarterly Report. Incidence and Prevalence of CHARGE Association/Syndrome. Published by the Canadian Pediatric Surveillance Program (CPSP) 2002 Results: 20-24. http://www.cps.ca/english/CPSP/index.htm

17. Lawand C, Graham JM, Jr, Prasad C, Blake KD (2003). CHARGE association / syndrome: Looking ahead. Published by the Canadian Pediatric Surveillance Program (CPSP) 2003 Resources. http://www.cps.ca/english/CPSP/index.htm

18. Carey J.C., Curry C.J.R., Grix A.W., Golabi M., Graham, Jr., J.M., Buehler B.A.: A tribute to Bryan Hall: Festschrift 2003. American Journal of Medical Genetics, 123A:1-4, 2003.

19. Blake K., Graham, Jr., J.M., Prasad C., Smith I.S.: CHARGE Association/Syndrome (September 2001 through August 2004). Published by the Canadian Pediatric Surveillance Program (CPSP) 2003 Results: 20-24. http://www.cps.ca/english/CPSP/index.htm

20. Blake K., Graham, Jr., J.M., Prasad C., Smith I.S.: CHARGE Association/Syndrome (September 2001 through August 2004) Final Report. Published by the Canadian Pediatric Surveillance Program (CPSP) 2004 Results: 27-31. http://www.cps.ca/english/CPSP/index.htm

BOOKS AND BOOK CHAPTERS

1. Graham J.M. Jr., Phillips E.L.R., Herman C.S. and Little R.E.: Manual for the Assessment of Fetal Alcohol Effects, Seattle, University of Washington Press, 1982.

2. Graham J.M. Jr., Chapter 20: Congenital anomalies, In: Levine, M.D., Carey, W.B., Crocker, A.C. and Gross, R.T. (Eds.) Developmental Behavioral Pediatrics, Philadelphia, WB Saunders Co., pp. 363-389, 1983.

3. Graham J.M. Jr., Bashir A.S. and Stark R.E., Chapter 40: Communicative disorders, In: Levine, M.D., Carey, W.B., Crocker, A.C. and Gross, R.T. (Eds.) Developmental Behavioral Pediatrics, Philadelphia, WB Saunders Co., pp. 847-864, 1983.

4. Graham J.M. Jr. and Edwards M.J., Chapter 11.3: Teratogenic effects of maternal hyperthermia, In: Gericke GS and Op't Hof J (Eds.) Genetic Perspectives in Fetal and Neonatal Medicine, Pretoria, Republic of South Africa, Haum Publishers, pp 44-56, 1983.

5. Graham J.M. Jr., Chapter 11.4: Adverse effects from uterine restrictive influences: Fetal deformation, In: Gericke GS and Op't Hof J. (Eds.) Genetic Perspectives in Fetal and Neonatal Medicine, Pretoria, Republic of South Africa, Haum Publishers, pp 56-93, 1983.

6. Graham J.M. Jr. and Edwards S.T., Chapter 17: Congenital malformations of the alimentary tract. In: Walker WA and Watkins J (Eds.) Nutrition in Pediatrics Basic Sciences and Clinical Application, Boston, Little Brown and Co., pp 675-705, 1985.

7. Graham J.M. Jr., Current issues in alcohol teratogenesis. In: Shenker JG and Weinstein D (Eds) The Intrauterine Life-Management and Therapy, Amsterdam, Elsevier Science Publishers, pp 383-388, 1986.

8. Graham J.M. Jr., Smith's Recognizable Patterns of Human Deformation, 2nd Edition, Philadelphia, W.B. Saunders Co., 1988.

9. Rimoin D.L. and Graham J.M. Jr., Chapter 16: Short Stature. In: Rimoin D.L. and Emery A.E.H. (Eds.) Principles and Practice of Medical Genetics, Edinburgh, U.K., Churchill Livingstone, Vol 1, pp 225-234, 1990.

10. Cooley, W.C., Graham, E.S., Moeschler, J.B. and Graham, J.M. Jr.: Reactions of mothers and medical professionals to a film of parents of persons with Down syndrome, Chigier E (Ed.) Looking Up At Down Syndrome, London, England, Freund Publishing House Ltd, pp 109-117, 1990.

11. Charman CE, and Graham J.M. Jr., Hypothalamic hamartoblastoma syndrome, congenital, Buyse, M.L. (Ed.), Birth Defects Encyclopedia, Cambridge MA, Blackwell Scientific Publications, pp 932-934, 1990.

12. Graham J.M. Jr., Rawsley B.E., Nordgren R.E., and Ionasescu I., Emery-Dreifuss syndrome, Buyse, M.L., (Ed.), Birth Defects Encyclopedia, Cambridge MA, Blackwell Scientific Publications, pp 613-614, 1990.

13. Halal F., Moeschler J.B. and Graham J.M. Jr., Radial-renal-ocular syndrome, Buyse ML (Ed.), Birth Defects Encyclopedia, Cambridge MA, Blackwell Scientific Publications, pp 1455-1456, 1990.

14. Graham J.M. Jr., Craniofacial deformation, Gorlin R.J., Cohen M.M., Jr., and Levin L.S. (Eds), Syndromes of the Head and Neck, 3rd edition, New York, Oxford University Press, pp 1-4, 1990.

15. Gibbs B., Moeschler J.B., Graham J.M. Jr., and Andrew C. Readings and Resources: A Guide for Educational and Medical Professionals Caring for Children with Genetic and Prenatally Determined Conditions. Hanover, NH, Clinical Genetics and Child Development Center, pp 1-144, 1991.

16. Graham J.M. Jr., Chapter 25: Congenital anomalies, In: Levine, M.D., Carey, W.B., Crocker, A.C. (Eds.) Developmental Behavioral Pediatrics, 2nd Edition, Philadelphia, WB Saunders Co., pp 229-243, 1992.

17. Bashir, A.S, Stark, R.E, Graham J.M. Jr., Chapter 64: Communication disorders, In: Levine, M.D., Carey, W.B., Crocker, A.C. (Eds.) Developmental Behavioral Pediatrics, 2nd Edition, Philadelphia, WB Saunders Co., pp 557-568, 1992.

18. Graham J.M. Jr., Donahue K.C., Hall J.G.: Human anomalies and cultural practices. In: Human Malformations and Related Anomalies, (Eds): Stevenson R. and Hall J., New York, Oxford University Press, Vol 1:169-181, 1993.

19. Graham J.M., Jr.: Chapter 3: When is it best to be born? A morphological perspective: craniofacial deformations. In: Amiel-Tison, C. and Stewart, A., The Newborn Infant: One Brain for Life, Paris, INSERM Press, pp 23-38, 1994.

20. Graham J.M. Jr. and Rimoin D.L.: Chapter 39: Abnormal body size and proportion. In: Rimoin D.L., Connor J.M., Pyeritz R.E., Emery A.E.H. (Eds.), Principles and Practice of Medical Genetics (3rd Edition), New York NY, Churchill Livingstone, pp 737-752, 1996.

21. Graham J.M. Jr.: Craniofacial deformation. Balliere's Clinical Paediatrics, 6:293-315, 1998.

22. Ming J.E., Stiehm E.R., Graham J.M. Jr. Chapter 9: Syndromes associated with immunodeficiency, Advances in Pediatrics, 46:271-351, 1999.

23. Graham, Jr., J.M.: Deformaciones craneofaciales. In Sola, A., Rogido, M. Cuidados Especiales del Feto y el ReciÈn Nacido. Buenos Aires, CientÌfica Interamericana, p184-188, 2000.

24. Graham, Jr., J.M, Rimoin, D.L.: Alteraciones del Crecimiento. In Sola, A., Rogido, M. Cuidados Especiales del Feto y el ReciÈn Nacido. Buenos Aires, CientÌfica Interamericana,. p 189-196, 2000.

25. Cooley, W.C., Graham, Jr., J.M.: Down Syndrome. In: Stockman, J.A., Lohr, J.A., (Eds.), Essence of Office Pediatrics. Philadelphia PA, W.B. Saunders Co., p. 117, 2001.

26. Graham J.M. Jr. and Rimoin D.L.: Chapter: Abnormal body size and proportion. In: Rimoin D.L., Connor J.M., Pyeritz R.E., Emery A.E.H. (Eds.), Principles and Practice of Medical Genetics (4th Edition), New York NY, Churchill Livingstone, pp 1066-1082, 2002.

27. Graham J.M. Jr., Chapter: Mixoploidy. In: Creighton T.E. (Ed.), Wiley Encyclopedia of Molecular Medicine, New York NY, John Wiley & Sons, pp. 2100-2103, 2002.

28. Graham J.M. Jr. FG Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, pp 193-194, 2003.

29. Graham J.M. Jr. Hallermann-Streiff Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, p 202, 2003.

30. Graham J.M. Jr. CHARGE Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, pp 167-168, 2003.

31. Visootsak J, Graham, Jr., JM; Klinefelter syndrome and its variants. Orphanet encyclopedia, March 2003: http://www.orpha.net/data/patho/GB/uk-KS.html

32. Ming J.E., Graham Jr., J.M.: Chapter 24: Genetic disorders, including syndromic immunodeficiencies. In: Stiehm E.R., Ochs H.D., Winkelstein J.A. (Eds.), Immunologic Disorders in Infants and Children. Fifth Edition, W. B. Saunders Co., pp 785-819, 2004.

33. Simpson J.L., Graham Jr., J.M., Samango-Sprouse C., Swerdloff R.: Chapter 28, Klinefelter Syndrome. In: Cassidy S.B., Allanson J.E. (Eds.), Management of Genetic Syndromes. 2nd Edition, Wiley-Liss, Hoboken NJ, pp 323-333, 2005.

34. Graham J.M. Jr.: Chapter 7: Skull. In: Human Malformations and Related Anomalies, (Eds): Stevenson R.E. and Hall J.G, 2nd Edition, New York, Oxford University Press, pp 221-265, 2005.

35. Visootsak J, Graham, Jr., JM; Klinefelter syndrome and its variants. Orphanet encyclopedia, September 2006: http://www.orpha.net/data/patho/GB/uk-KS.html

36. Graham J.M. Jr. and Rimoin D.L.: Chapter 41: Abnormal body size and proportion. In: Rimoin D.L., Connor J.M., Pyeritz R.E., Korf B.R. (Eds.), Emery and Rimoin's Principles and Practice of Medical Genetics (5th Edition), New York NY, Churchill Livingstone, pp 948-963, 2007.

37. Mortier G., Graham J.M. Jr. and Rimoin D.L.: Chapter 17: Short Stature Syndromes. In: Kelnar C. J. H., Savage M.O., Saenger P., and Cowell C.T. (Eds.), Growth Disorders (2nd Edition), Hodder Arnold Press, London, UK, pp 261-282, 2007.

38. Graham J.M. Jr., Smith's Recognizable Patterns of Human Deformation, 3rd Edition, Philadelphia, Elsevier-W.B. Saunders Co., Philadelphia, PA, 2007.

39. Graham, JM Jr..: Chapter 1. Deformations and Disruptions. In: Hennekam RCM, Allanson J, Krantz I. (Eds) Gorlin's Syndromes of the Head and Neck. (5th Edition), Oxford University Press, New York NY, pp 3-24, 2010.

40. Visootsak J., Graham Jr., J.M., Samango-Sprouse C., Swerdloff R. Simpson J.L.: Chapter 33, Klinefelter Syndrome. In: Cassidy S.B., Allanson J.E. (Eds.), Management of Genetic Syndromes. 3rd Edition, Wiley-Liss, Hoboken NJ, pp 479-494, 2010.

41. Graham JM Jr.: B Hale, A. Scialli, MS Tassinari (Eds). Chapter 6. How do genes affect the risk of having a child with a birth defect? Teratology Primer 2nd Edition, Teratology Society, pp17-19. 2010.

42. Nemec S.F., Brugger P.C., Kasprian G., Nemec U., Graham J.M., Jr., Prayer D.: The skeleton and musculature. In Prayer D. (Ed.). Medical Radiology Diagnostic Imaging: Fetal MRI, 1st Edition, Springer,-Verlag, Berlin Heidelberg Germany, pp 235-246, 2011.

43. Sanchez-Lara, P.A., Graham Jr., J.M.: Congenital Anomalies of the Skull. In: Swaiman K., Ashwal S., Ferriero D., Schor N. (Eds.), Swaiman's Pediatric Neurology: Priniciples and Practice 5th Edition, Elsevier, New York NY, pp 247-262, 2012.

44. Graham J.M. Jr. Burkardt D.D. and Rimoin D.L.: Chapter 38: Abnormal body size and proportion. In: Rimoin D.L., Pyeritz R.E., Korf B.R. (Eds.), Emery and Rimoin's Principles and Practice of Medical Genetics, 6th Edition, New York NY, Elsevier, pp 1022-1045, 2013.

45. Graham J.M. Jr. CHARGE Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, 2013.

46. Graham J.M. Jr. Hallermann-Streiff Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, 2013.

47. Graham J.M. Jr. Larsen Syndrome. In: The NORD Guide to Rare Disorders. Philadelphia, PA. Lippincott Williams & Wilkins, 2013.

48. Ming J.E., Graham Jr., J.M.: Chapter 12: Genetic syndromes with evidence of immune deficiency. In: Sullivan K.E. and Stiehm E.R.(Eds.), Stiehm's Immune Deficiencies. 6th Edition, New York, NY, Elsevier Inc, pp 281-324, 2014.

49. Graham J.M. Jr., Sanchez-Lara, P.A.: Chapter 5: Skull. In: Human Malformations and Related Anomalies, Stevenson R.E., Hall J.G., Everman D. and Solomon B., 3rd Edition, New York, Oxford University Press, in press, 2016.

50. Graham J.M. Jr., Sanchez-Lara P.A.: Smith's Recognizable Patterns of Human Deformation, 4th Edition, Philadelphia, Elsevier-W.B. Saunders Co., Philadelphia, PA, in press 2016.

51. Sanchez-Lara, P.A., Graham Jr., J.M.: Chapter 31: Congenital Anomalies of the Skull. In: Swaiman K., Ashwal S., Ferriero D., Schor N., Finkel R., Gropman A., Pearl P., Shevell M. (Eds.), Swaiman's Pediatric Neurology: Priniciples and Practice 6th Edition, Elsevier, New York NY, in press 2016.

LETTERS TO THE EDITOR

1. Smith D.W., and Graham J.M. Jr.: Fetal alcohol syndrome in child whose parents had stopped drinking. Lancet 2(8141):527, 1979.

2. Graham J.M. Jr., Wertelecki W, O'Conner J.E. and Cohen M.M. Choanal atresia with Down syndrome. Journal of Pediatrics, 98:664, 1981.

3. Graham J.M. Jr., and Smith D.W. Dominantly-inherited pterygium colli. Journal of Pediatrics, 98:664-665, 1981.

4. Frank D.A., Graham J.M. Jr. and Smith D.W. Adoptive children in a dysmorphology clinic: Implications for evaluation of children before adoption. Pediatrics, 68:744-745, 1981.

5. Graham J.M. Jr., Neurologic findings with maternal hyperthermia - Reply. Pediatrics, 69:128-129, 1982.

6. Graham J.M. Jr., and Frank D.A. Dysmorphology and Adoptions - Reply. Pediatrics, 69:665, 1982.

7. Pagon R.A., Zonana J, and Graham J.M. Jr. CHARGE Association. Pediatrics, 70:827-828, 1982.

8. Graham J.M. Jr., Rawnsley E.F., Simmons G.M. Crow H.C. Confirmation of xeroradiography as an important postmortem fetal diagnostic technique. American Journal of Obstetrics and Gynecology, 153:347, 1985.

9. Cooley W. C., Graham J. M. Jr., Letter to the editor: Clinical note concerning smaller caliber of subglottic region in children with Down syndrome. Clinical Pediatrics, 30(11):646, 1991.

10. Graham J.M. Jr., Letter to the editor: Life before death. Clinical Pediatrics, 14(1):87, 1994.

11. Biesecker L.G., Topf K., Graham J.M. Jr., Letter to the editor: Familial Pallister-Hall syndrome, Journal of Medical Genetics, 31(9):740, 1994.

12. Bacino C.A., Graham J.M. Jr., Letter to the editor: Clinical and molecular studies in full trisomy 22: Further delineation of the phenotype and review of the literature. Reply to Dr. Hirschhorn. American Journal of Medical Genetics, 62:99, 1995.

13. Bacino C.A., Graham J.M. Jr., Letter to the editor: Clinical and molecular studies in full trisomy 22: Further delineation of the phenotype and review of the literature. Reply to Dr. Robinson and Dr. Kalousek. American Journal of Medical Genetics, 62:101, 1995.

14. Wang M., Matthews K.R., Imaizumi K., Beiraghi S., Blumberg B., Scheuner M., Graham, Jr. J.M., and Godfrey M., Correspondence: P1148A in fibrillin-1 is not a mutation anymore. Nature Genetics 15:12, 1997.

15. Allanson J., Hunter A., Cassidy S., Curry C., Donnai D., Fraser C., Gorlin R., Graham, Jr. J.M., Hall B., Hall J., Jones K.L., Stevenson R., and Winter R., Letter to the Editor: Photographic documentation of syndrome diagnosis. American Journal of Medical Genetics 68:487, 1997.

16. Aylsworth A., Graham, Jr. J.M., Hall J.G., Hoyme H.E., Jones K.L., Stevenson R.E., Correspondence: Morphogenesis: Re: Clinical, natural history, and imaging information on patients included in reports. American Journal of Medical Genetics 119A:93, 2003.

17. Blake K., Graham, Jr., J.M., Prasad C.: Reply to Letter to the Editor by Lowry et al.: An epidemiological analysis of CHARGE syndrome: preliminary results from a Canadian study (Issekutz et al., 2005). American Journal of Medical Genetics, 139A:170-171, 2005.

18. Conway R, Danielpour M, Graham Jr., J.M.: Editorial and Response: Macrocephaly-cutis marmorata telangiectatica congenita. Journal of Neurosurgery (4 Suppl Pediatrics), 106:293-295, 2007.

DOCTORAL THESIS

Graham J.M. Jr. Communicative skills in XXY boys. Division of Communicative Disorders, Department of Health Services Administration, School of Hygiene and Public Health, Johns Hopkins University, Baltimore, Maryland, 1981.

DOCTORAL THESIS ADVISEES

Weng E.Y. Review of Beckwith-Wiedemann syndrome: A prototypical fetal overgrowth disorder. Doctoral Thesis, UCLA Medical School, Los Angeles, CA, 1995.

REVIEWS

1. Little R.E., Graham J.M. Jr. and Samson H.H.: Fetal alcohol effects in humans and animals. Advances in Alcohol and Substance Abuse, 1:103-125, 1982.

2. Graham J.M. Jr., Causes of limb-reduction defects: The contribution of fetal constraint and/or vascular disruption, Clinics in Perinatology, 13:575-591, 1986.

3. Graham J.M. Jr., Rotter J.I., Riccardi V.M. et al: Report of the task force on teaching human genetics in North American Medical Schools. American Journal of Human Genetics, 44:161-165, 1989.

4. Charman C.E., Graham J.M. Jr.: Report of the First Information and Education Committee Human Genetics Teaching Forum. American Journal of Human Genetics 44:605-607, 1989.

5. Rimoin D.L., and Graham J.M. Jr.: Syndromes associated with growth deficiency. Acta Paediatr. Scan. (Suppl) 349:3-10, 1989.

6. Graham J.M. Jr. (Guest Editor), Fetal Dysmorphology, Clinics in Perinatology, 17(3), September, 1990.

7. Graham J.M. Jr. (Guest Editor), Fetal Clinical Genetics, Clinics in Perinatology, 17(4), December, 1990.

8. Graham J.M. Jr., Otto, C., Clinical approach to prenatal detection of human structural defects, Clinics in Perinatology, 17(3):513-546, September, 1990.

9. Edwards M.J., Graham J.M. Jr., Posterior nuchal cystic hygroma, Clinics in Perinatology, 17(3):611-640, September, 1990.

10. Graham JM Jr., Clinical approach to human structural defects, Seminars in Perinatology, 15(1)Suppl 1:2-15, 1991.

11. Sharony R., Graham J.M. Jr. Identification of fetal problems associated with anticonvulsant usage and maternal epilepsy. Obstetrics and Gynecology Clinics of North America, 18(4):933-951, 1991.

12. Graham J.M. Jr., Curry C.J.R., Hoyme H.E., Stevenson R., Hall J.G. Fellowship and career development in dysmorphology and clinical genetics. Pediatric Clinics of North America, 39(2), 349-362, 1992.

13. Graham J.M. Jr. Introduction: Papers on Brachmann-deLange syndrome presented at the 12th Annual David W. Smith Workshop on Morphogenesis and Malformations. American Journal of Medical Genetics, 47:939, 1993.

14. Adams J, Buttar H.S., Chambers C., Collins T.F.X., Graham, Jr., J.M., Jones K.L., Kweder S., Lammer E., Mitala J., Polifka J.E., and Schwetz B.A.: Statement of the Public Affairs Committee of the Teratology Society on Thalidomide. Teratology 62:172-173, 2000.

15. Adams J, Bittner P., Buttar H.S., Chambers C.D., Collins T.F.X., Daston G.P., Filkins K., Flynn T.J., Graham, Jr., J.M., Jones K.L., Kimmel C., Lammer E., Librizzi R., Mitala J., and Polifka J.E.: Statement of the Public Affairs Committee of the Teratology Society on the Fetal Alcohol Syndrome. Teratology, 66:344-347, 2002.

16. Graham J.M. Jr., Kathleen K. Sulik (Guest Editors) Introduction: Twelth Robert J. Gorlin Conference on Dysmorphology, American Journal of Medical Genetics 122A:281-282, 2003.

17. Carey J.C., Curry C.J.R., Grix A.W., Golabi M., Graham J.M. Jr., Buehler B.A.: A tribute to Bryan D. Hall: Festscrift 2003. American Journal of Medical Genetics, 123A:1-4, 2003.

18. Graham J.M. Jr., Hennekam R.C.M.: Preface to Special Issue on Epilepsy and Genetics, European Journal of Medical Genetics, 55(5): 279-280, 2012.

ABSTRACTS AND PRESENTATIONS

1. Graham J.M. Jr.: The effect of d-amphetamine sulfate on susceptibility to audiogenic seizures in DBA/2J mice. Texas Reports on Biology and Medicine, 30:207, 1972.

2. Graham J.M. Jr.: Schreiber R.A. and Zemp J.W. The effect of d-amphetamine sulfate on susceptibility to audiogenic seizures in DBA/2J mice. Fifth International Congress of Pharmacology, San Francisco CA, July 23-28, 1972.

3. Graham J.M. Jr., Schreiber R.A. and Zemp J.W.: The effects of d-amphetamine sulfate on susceptibility to audiogenic seizures in DBA/2J and C57BL/6J mice. Texas Reports on Biology and Medicine, 31:570-571, 1973.

4. Graham J.M. Jr., Schreiber R.A. and Zemp J.U.: D-amphetamine reduces the severity of sound-induced seizures in DBA/2J and C57BL/6J mice. Third Annual Meeting of the Society for Neuroscience, San Diego CA, November 7-10, 1973.

5. Wertelecki W, Graham J.M. Jr., Sergovich F and Granoff D.: The importance of clinical recognition of triploidy. Proceedings of the Fourteenth International Congress of Pediatrics, pp. 85-87, 1974.

6. Graham J.M. and Smith D.W.: The neonate with an engaging head: The soft-headed baby. Clinical Research 27:118A, 1979.

7. Graham J.M. Jr. and Smith D.W.: Dominantly inherited pterygium colli: Cervical lymphectasia malformation complex. Clinical Research 27:101A, 1979.

8. Graham J.M. Jr., DeSaxe M and Smith D.W.: Sagittal craniostenosis: Fetal head constraint as one possible cause. Pediatric Research, 13:485, 1979.

9. Graham J.M. Jr., and Smith D.W.: Parietal cranioteobes in the neonate: Its origin and relevance. Pediatric Research, 13:485, 1979.

10. Graham J.M. Jr., DeSaxe M and Smith D.W.: Sagittal craniostenosis: Fetal head constraint as one possible cause. Teratology, 19:27-28A, 1979.

11. Graham J.M. Jr., De Saxe M, Badura R.J. and Smith D.W.: Fetal head constraint as one possible cause for craniostenosis. 1979 Birth Defects Conference, Chicago IL, June 24-27, 1979.

12. Graham J.M. Jr., Koskinen-Moffett L.K., Moffett B.C. and Smith D.W.: Fetal head constraint: A common cause for craniostenosis. Clinical Research, 28:116A, 1980.

13. Graham J.M. Jr., Miller M.E., Stephan J.M. and Smith D.W.: Limb reduction anomalies and early in-utero limb compression. Clinical Research 28:116A, 1980.

14. Pagon R.A., Graham J.M. Jr., Sybert V.P.: The CHARGE association. Clinical Research, 28:522, 1980.

15. Graham J.M. Jr., Hoehn H.W., Lin M.S. and Smith D.W.: Diploid-triploidy mosaicism: Clinical and cytogenetic features. Pediatric Research, 14:522, 1980.

16. Graham J.M. Jr., Miller M.E., Stephan M.J. and Smith D.W.: Limb reduction anomalies and early in-utero limb compression. Pediatric Research, 14:585, 1980.

17. Graham J.M. Jr., and Smith D.W.: Pterygium colli as a consequence of jugular lymphatic obstruction malformation sequence: Pathogenesis and report of dominant inheritance. Pediatric Research, 14:585, 1980.

18. Miller M.E., Graham J.M. Jr., Higginbottom M.C. and Smith D.R.: Conditions of limb reduction and body wall closure defects: Features in the spectrum of early amnion rupture malformation sequence. Pediatric Research 14:586, 1980.

19. Pleet H.B., Graham J.M. Jr., Harvey M.A. and Smith D.W.: Patterns of malformations resulting from the teratogenic effects of first trimester hyperthermia. Pediatric Research, 14:587, 1980.

20. Pagon R.A., Graham J.M., Jr., Yong S and Kalina R.E.: Coloboma and multiple anomalies - ? an autosomal dominant disorder. Association for Research in Vision and Ophthalmology Meeting, Orlando FL, May 5-9, 1980.

21. Graham J.M. Jr., Miller M.E., Higginbottom M.C., Stephen M.J. and Smith D.W.: Early in-utero compression as a cause for limb deficiency. 1980 Birth Defects Conference, New York City, June 8-11, 1980.

22. Graham J.M. Jr., Smith D.W., Koskinen-Moffett L., Stuntz J.T., Loeser J.D., Clarren S.K., and Moffett B.C.: Craniostenosis caused by fetal head constraint: Correlation of etiology with sutural histology and response to surgery. 1980 Birth Defects Conference, New York City, June 8-11, 1980.

23. Miller M.E., Graham J.M. Jr., Higginbottom M.C. and Smith D.W.: Compressionrelated defects from early amnion rupture: Evidence for mechanical teratogenesis. 1980 Birth Defects Conference, New York City, June 8-11, 1980.

24. Jung J.H., Graham J.M. Jr., Schultz N and Smith D.W.: Monozygotic twinning: The problem of congenital hydranencephaly/porencephaly. 1980 Birth Defects Conference, New York City, June 8-11, 1980.

25. Graham J.M. Jr., Miller M.E., Stephan M.J. and Smith D.W.: Limb reduction anomalies and early in-utero limb compression. Teratology 21:40A, 1980.

26. Graham J.M. Jr., Higginbottom M.C. and Smith D.: Pre-axial polydactyly of the foot associated with early amnion rupture: Evidence for mechanical teratogenesis? Conference on Malformations and Morphogenesis, San Diego CA, September 4-6, 1980.

27. Graham J.M. Jr., Hoehn H., Lin M.S. and Smith D.W.: Diploid-triploid mixoploidy: Clinical and cytogenetic features. Conference on Malformations and Morphogenesis, San Deigo CA, September 4-6, 1980.

28. Smith D.W., Harvey M.A.S., Bunn B.S. and Graham J.M. Jr.: Specific diagnoses and prognoses in SGA infants. Conference on Malformations and Morphogenesis, San Diego CA, September 4-6, 1980.

29. Pagon R.A., Graham J.M. Jr., Zonana J. and Yong S.L.: The CHARGE association, American Society of Human Genetics Meeting, New York City, September 24-27, 1980.

30. Carey J.C., Smith D.W., Graham J.M. Jr., Thain W.S., Haas J.E. and Hall B.D.: New syndrome of developmental/growth retardation, coarse facies, and unusual hands. Clinical Research, 29:130A, 1981.

31. VanAllen M.I., Smith D.W. and Graham J.M. Jr.: Twin reversed arterial perfusion sequence. Clinical Research, 29:133A, 1981.

32. Smith D.W., Harvey M.A.S., Bunn B.S. and Graham J.M. Jr.: Specific diagnoses and prognoses in SGA infants. Clinical Research, 29:133A, 1981.

33. Jung J.H., Graham J.M. Jr., Schultz N. and Smith D.W.: Vascular disruption associated with monozygotic twinning: A possible cause of congenital hydranencephaly porencephaly. Clinical Research 29:132A, 1981.

34. Graham J.M. Jr., Bashir A.S., Walzer S., Start R.E. and Gerald P.S.: Communication skills among unselected XXY boys. Pediatric Research, 15:562, 1981.

35. Graham J.M. Jr., Stephen T.D., Siebert Jr. and Smith D.W.: Determinants in the morphogenesis of muscle tendon insertions. Pediatric Research 15:643, 1981.

36. Graham J.M. Jr., Darby B.L., Barr H.M., Smlth D.W. and Streissguth A.P.: Longterm effects of moderate alcohol consuption during pregnancy. Pediatric Research, 15:643, 1981.

37. Graham J.M. Jr., Clarren S.K., Koskinen-Moffett L., Stuntz J.T., Loeser J.D. and Smith D.W.: Craniostenosis: Correlation of etiology with sutural histology and response to surgery. Pediatric Research, 15:662, 1981.

38. Smith D.W., Harvey M.A.S., Bunn B.S. and Graham J.M. Jr.: Specific diagnosis and prognoses in SGA infants. Pediatric Research, 15:646, 1981.

39. Van Allen M.I., Smith D.W. and Graham J.M. Jr.: Twin reversed arterial perfusion sequence. Teratology, 23:67A, 1981.

40. Graham J.M. Jr., Crow H.C., Rawnsley E.F. and Hoefnagel D.: Use of xeroradiography in the study of abortuses. Teratology, 23:37A, 1981.

41. Graham J.M. Jr., Darby B.L., Barr H.M., Smith D.W. and Streissguth A.P.: Longterm effects of alcohol consumption during pregnancy. Teratology, 23:38A, 1981.

42. Graham J.M. Jr., Clarren S.K., Koskinen-Moffett J.T., Loeser J.D. and Smith D.W.: Craniostenosis: Correlation of etiology with sutural histology and response to surgery. Teratology 23:38A, 1981.

43. Graham J.M. Jr., Stephens T.D., Siebert Jr. and Smith D.W.: Determinants in the morphogenesis of muscle tendon insertions. Teratology, 23:37A, 1981.

44. Smith D.W., Harvey M.A.S., Bunn B.S. and Graham J.M. Jr.: Specific diagnoses and prognoses in SGA infants. Teratology 23:63A, 1981.

45. Graham J.M. Jr., Marin-Padilla M and Hoefnagel D.: Disruptive vascular defects associated with Catergot ingestion during pregnancy. 1981 Birth Defects Conference, San Diego CA, June 14-17, 1981.

46. Graham J.M. Jr., Bashir A.S., Walzer S, Stark R.E. and Gerald P.S.: Communications skills and reading and spelling abilities in unselected XXY boys. 1981 Birth Defects Conference, San Diego CA, June 14-17, 1981.

47. Graham J.M. Jr. and Hoefnagel D.: Bilateral thumb polydcatyly in a girl whose mother shows the Fromont anomaly. Conference on Malformations and Morphogenesis. Hanover, NH, August 23-25, 1981.

48. Marin-Padilla M., Graham J.M. Jr. and Simmons G.H.: Extrinsic vascular disruptions and unilateral limb malformations. Conference on Malformations and Morphogenesis. Hanover, NH, Hampshire, August 23-25, 1981.

49. Graham J.M. Jr., Stephens T.D., Siebert Jr. and Smith D.W.: Determinants in the morphogenesis of muscle tendon insertions. David W. Smith Festscrift. Lake Wilderness Continuing Educational Center, Maple Valley WA, September 18-20, 1981.

50. Koskinen-Moffett LK, Moffett BC and Graham J.M. Jr.: Cranial synostosis and intrauterine compression: A developmental study of human sutures. International Conference on Factors and Mechanisms Influencing Bone Growth. UCLA Center for Health Sciences, Los Angeles CA, January 5-7. 1981.

51. Graham J.M. Jr., Bashir A.S., Walzer S., Stark R.E., Tallal P. and Gerald P.S.: Auditory processing abilities in unselected XXY boys. Clinical Research, 30:118A, 1982.

52. Graham J.M. Jr. and Hoefnagel D.: Thumb polydactyly as part of the range of genetic expression for preaxial limb deficiency. Clinical Research, 30:133A, 1982.

53. Graham J.M. Jr., Crow H.C., Rawnsley E.F. and Hoefnagel D.: Enhanced visualization of soft tissues in the study of aborted fetuses through the use of xeroradiography. Teratology 24:45AA, 1982.

54. Graham J.M. Jr.: Limb anomalies as a consequence of spatially-restricting uterine environments. Third International Conference on Limb Development and Regeneration. Storrs, CT, June 27-July 2, 1982.

55. Graham J.M. Jr.: Limb anomalies as a consequence of spatially- restricting uterine environments. International Conference on Prevention of Mental and Physical Congenital Anomalies, Strasbourg, France, October 11-16, 1982.

56. Graham J.M. Jr.: Effects of alcohol consumption during pregnancy. International Conference on Prevention of Mental and Physical Congenital Anomalies. Strasbourg, France, October 11-16, 1982.

57. Graham J.M. Jr., Ferm R., Ferm V.H.: Maternal binge drinking as a possible cause for neural tube defects. Clinical Research, 31:128A, 1983.

58. Gang D., Wright J., Edwards W., Graham J.M. Jr.: Extensive vertex cutis aplasia associated with atresia of the gall bladder and vascular anomalies. Clinical Research, 31:127A, 1983.

59. Graham J.M. Jr., Perl D., O'Keefe T., Rawnsley E., Little G.A.: Apparent familial recurrence of hypothalamic hamartoblastoma syndrome. Clinical Research 31:128A, 1983.

60. Graham J.M. Jr., Crow H.C., Rawnsley E.F., Simmons G.M., Hoefnagel D.: Enhanced visualization of soft tissues in the study of aborted fetuses through the use of xeroradiography. Clinical Research 31:128A, 1983.

61. Graham J.M. Jr., Ferm R., Ferm V.H.: Maternal binge drinking as a possible cause for neural tube defects. Pediatric Research, 17:299A, 1983.

62. Graham J.M. Jr., Crow H.C., Rawnsley E.F., Simmons G.M., Hoefnagel D.: Enhanced visualization of soft tissues in the study of aborted fetuses through the use of xeroradiography. Pediatric Research, 17:299A, 1983.

63. Graham J.M. Jr., Ferm R., Ferm V.H.: Maternal binge drinking as a possible cause for neural tube defects. Proceedings of the Greenwood Genetic Center, 2:98, 1983.

64. Wright J., Gang D., Edwards W. and Graham J.M. Jr.: Extensive vertex cutis aplasia associated with atresia of the gall bladder and vascular anomalies. Proceedings of the Greenwood Genetic Center, 2:103-104, 1983.

65. Graham J.M. Jr., Perl D., O'Keefe T., Rawnsley E. and Little G.A.: Apparent familial recurrence of hypothalamic hamartoblastoma syndrome. Proceedings of the Greenwood Genetic Center, 2:117-118, 1983.

66. Wertelecki W., Graham J.M. Jr., Sergovich F.R.: Trisomy 22 mosaicism: Turner stigmata and asymmetry. Proceedings of the Greenwood Genetic Center, 2:136-137, 1983.

67. Graham J.M. Jr., Crow H.C. Rawnsley E.F., Simmons G.M. and Hoefnagel D.: Enhanced visualization of soft tissues in the study of aborted fetuses through the use of xeroradiography. Proceedings of the Greenwood Genetic Center, 2:133, 1983.

68. Graham J.M. Jr., Bashir A.S., Walzer S., Stark R.E.: Left hemisphere deficits in unselected XXY Boys. Behavioral Genetics, 13:536, 1983.

69. Graham J.M. Jr., Ferm R., Ferm V.H.: Neural tube defects induced by alcohol and/or heat in golden hamster fetuses. Teratology, 27:45A-46A, 1983.

70. Graham J.M. Jr., Ferm R. and Ferm V.H.: Folate supplementation does not prevent alcohol-or heat-induced neural tube defects. Proceedings of the Greenwood Genetic Center 3:116-117, 1984.

71. Graham J.M. Jr., Boyle W., Troxell J., Cullity G.J. and Beckwith J.B. Bilateral neoplastic kidney disease, pulmonary cystic disease, and fetal macrosomia: A spectrum of developmental abnormalities. Pediatric Research 18:305A, 1984.

72. Graham J.M. Jr., Ferm V.H. and Layton W.: Folate supplementation does not prevent neural tube defects induced by alcohol or heat. Pediatric Research 18:305A, 1984.

73. Graham J.M. Jr., Rawnsley E.F. and Sulik K.K. Vitamin A teratogenesis in a sensitive genetic background. Pediatric Research 18:305A, 1984.

74. Graham J.M. Jr., Troxell J., Boyle W. and Beckwith J.B.: Bilateral neoplastic kidney disease and pulmonary cystic disease: A spectrum of developmental abnormnalities. Teratology 29:32A, 1984.

75. Graham J.M. Jr., Struckmeyer C.L. and Hallowell C.: Dominantly inherited unilateral ectrodactyly: Case report and review of the literature. American Journal of Human Genetics, 36(Supple):53, 1984.

76. Graham J.M. Jr., Harris J., Frank J.E., Little G.A. and Klein R.Z.: Syndromes associated with polydactyly and hypopituitarism. Pediatric Research 19:325A, 1985.

77. Graham J.M. Jr., Meill E., Pagon R.A., Siebert J.R. and Kaplan L.: Conotruncal defects in CHARGE Association: Evidence for neural crest involvement. Pediatric Research 19:325A, 1985.

78. Graham J.M. Jr., Frank J.E., Harris M., Klein R.Z., Saunders R.L., Binder M. and Fratkin J.D.: Congenital hypothalamic hamartoblastoma syndrome: Natural history and etiologic considerations. Tetratology 31:65A, 1985.

79. Graham J.M. Jr., Meill E., Pagon R.A., Siebert J.R. and Kaplan L.C.: Cardiac features of the CHARGE association: Support for involvement of the neural crest. Proceedings of the Greenwood Genetic Center 4:82-83, 1985.

80. Graham J.M. Jr., Harris M., Frank J.E., Little G.E. and Klein R.Z.: Congenital hypothalamic hamartoblastoma syndrome: Natural history and genetic implications. Fourth International Clinical Genetics Seminar, Athens, Greece, May 22-25, 1985.

81. Graham J.M. Jr., Darby B.L., Barr H.M., Streissguth A.P. and Hanson J.M.: Fetal alcohol effects on human morphogenesis: Discriminating features in a prospective study. The 2nd International Symposium: The Fetus as a Patient, Diagnosis and Therapy. Jerusalem, Isreal, May 26-31, 1985.

82. Graham J.M. Jr. and Ferm V.H.: Failure of folate supplementation to protect from neural tube defects induced experimentally by heat or alcohol. The 2nd International Symposium: The fetus as a Patient, Diagnosis and Therapy. Jerusalem, Isreal, May 26-31, 1985.

83. Graham J.M. Jr., Brown F.E. and Hall B.D.: Thumb polydactyly as part of the range of genetic expression for thenar hypoplasia. American Journal of Human Genetics 37:A132, 1985.

84. Rawnsley E.R., Simmons G.M., Graham J.M. Jr. and Crow H.C.: Prenatal diagnosis of X-linked hydrocephalus. American Journal of Human Genetics, 37:A135, 1985.

85. Edwards W.H., Graham, J.M. Jr., Horton W.A., Lachman R.S. and Rimoin D.L.: Otopalatal-digital syndrome, type 2: Severe manifestations in a female infant. American Journal of Human Genetics, 37:A52, 1985.

86. Graham J.M. Jr., Edwards W.H., Horton W.A., Lachman R.S. and Rimoin D.L.: Otopalatal-digital syndrome, type 2, versus atelosteogenesis: severe manifestations in a female infant. Proceedings of the Greenwood Genetic Center 5:112-113, 1986.

87. Graham J.M. Jr., Smith M., Fratkin J., Marin-Padilla M., Nordgren R., Saunders R., Cohen M.M., Klein R.Z.: Congenital hypothalamic hamartomas: A spectrum of developmental abnormality. Pediatric Research 20:338A, 1986.

88. Graham J.M. Jr., Rawnsley E.S., Nordgren R. and Fratkin J.: Autosomal dominant limb-girdle nuscular dystrophy with progressive cardiomyopathy: Report of a large family and delineation of natural history. Pediatric Research 20:337A, 1986.

89. Graham J.M. Jr., Charman C., Klein R.Z., Moeschler J.B.: New aspects of hyperthermia teratogenesis. Teratology 33:80C, 1986.

90. Park J.P., Graham J.M. Jr., Wurster-Hill D.H., Andrews P.A., Colley C., and Dossu J.R.: Partial trisomy 21 associated with microcephaly and parental double NOR. Karyogram 12:88, 1986.

91. Graham J.M. Jr., Charman C., Hartman J., Moeschler J.B.: Malformation syndromes due to maternal hyperthermia. 7th International Congress of Human Genetics. Berlin, West Germany, September 22-27, 1986.

92. Graham J.M. Jr., Charman C., Hartman J., Rawnsley E. and Moeschler, J.B.: Malformation syndromes due to maternal hyperthermia. American Journal of Human Genetics, 39:A255, 1986.

93. Park J.P., Wurster-Hill D.H., Moeschler J.B., Graham J.M. Jr., and Charman C.E.: A de novo interstitial 4p deletion (pl6.1 p 15.2) associated with Wolf syndrome. American Journal of Human Genetics, 39:A128, 1986.

94. Moeschler J.B., Graham J.M. Jr., Rawnsley B.E., Park J.P. Wurster-Hill D.H., and Marin-Padilla M.: Neural tube defects in 13q- syndrome, American Journal of Human Genetics, 39:A72, 1986.

95. Rawnsley B.E., Charman C.E., Crow H. and Graham J.M. Jr.: Prenatal diagnosis of Jeune syndrome. American Journal of Human Genet. 39:A263, 1986.

96. Graham J.M. Jr., Rawnsley E.R., Nordgren R., Fratkin J.: Autosomal dominant limbgirdle muscular dystrophy with progressive cardiomyopathy: report of a large family and delineation of natural history, American Journal of Medical Genetics 25:720, 1986.

97. Graham J.M. Jr., Charman C.E., Moeschler J.B.: Neurogenic club feet associated with maternal hyperthermia. American Journal of Medical Genetics 25:739, 1986.

98. Graham J.M. Jr., Charman C.E., Hartman J., Rawnsley B.E. and Moeschler J.B.: Malformation syndromes due to maternal hyperthermia. Proceedings of the Greenwood Genetics Center, 6:115-116, 1987.

99. Moeschler J.B., Graham J.M. Jr., Rawnsley B.E., Park J.P., Wurster-Hill D.H. and Marin-Padilla M.: Neural tube defects in 13q- syndrome. Proceedings of the Greenwood Genetics Center, 6:140, 1987.

100. Volpe, M.V., Graham J.M. Jr., Miller F.S. III, Riskahl J.S., King R.A. and deMartinville B.: Linkage analysis in X-linked ocular albinism, Pediatric Research 21:295A, 1987.

101. Graham J.M. Jr., Charman C.E. and Chaisson R.: Postnatal Plagiocephaly: pathogenesis and therapy in 40 cases (1981-86), Pediatric Research 21:227A, 1987.

102. Moeschler J.B. Charman C.E., Berg S.Z., Graham J.M. Jr.: Rett syndrome: additional findings from 7 cases. Pediatric Research 21:229A, 1987.

103. Thomas IT, Cantu E.S., Frias J.L. Lafer C., Flannery D.B., Graham J.M. Jr.: Pigmentary abnormalities and chromosomal and genetic mosaicism and chimerism, Pediatric Research 21:294A, 1987.

104. Graham J.M. Jr.: Cranial asymmetry: 78 cases in 6 years (1981-1986), Teratology 35:50A, 1987.

105. Park J.P. and Graham J.M. Jr., Wurster-Hill D.H.: Ring chromosome 12, Karyogram, 13(6):101, 1987.

106. Graham J.M. Jr., deMartinville B., Volpe M.A., Risdahl J.S., Rich S.S., Miller F.S. III and King R.A.: Clinical findings and linkage analysis in X-linked ocular albinism. American Journal of Human Genetics, 41:A64, 1987.

107. Iafolla A.K., Fratkin J.D., Spiegel P.K., Cohen M.M., Moeschler J.B. and Graham J.M. Jr.: Further delineation of the congenital hypothalamic hamartoblastoma syndrome. American Journal of Human Genetics, 41:A277, 1987.

108. Park J.P., Moeschler J.B., Graham J.M. Jr. and Wurster-Hill D.H.: Clinical and cytogenetic findings in 18q deletions: comparison of extreme distal del(l8)(pter q22.3) with more proximal deletions. American Journal of Human Genetics, 41:A137, 1987.

109. Charman C.E., Chaisson R., Donahue K.C. and Graham J.M. Jr.: Pathogenesis and treatment of plagiocephaly (1981-86). American Journal of Human Genetics, 41:A51, 1987.

110. Berg S.Z., Moeschler J.B., Charman C.E. and Graham J.M. Jr.: Genetics counseling issues for Rett syndrome families. American Journal of Human Genetics, 41:A193, 1987.

111. deMartinville B., Graham J.M. Jr., Volpe M.A., Risdahl J.S., Rich S.S., Miller F.S. III and King R.A.: Genetic analysis in X-linked ocular albinism. Presented at the Ninth International Workshop on Human Gene Mapping, Paris France, Sept. 611, 1987.

112. Donahue K.C., Charman C.E., Chaisson R., and Graham J.M. Jr.: Postnatal head deformation: Anthropological observations and applications to the treatment of postnatal plagiocephaly. Neural Crest and Craniofacial Disorders: (Eds. Opitz J.M., Gorlin R.J., Reynolds J.F., and Spano L.M.) A.R. Liss Inc., New York, 1988.

113. Iafolla A.K., Fratkin J.D., Spiegel P.K., Cohen M.M., Moeschler J.B. and Graham J.M. Jr.: Further delineation of the congenital hypothalamic hamartoblastoma syndrome. Proceedings of the Greenwood Genetic Center, 7:188-189, 1988.

114. Graham J.M. Jr., deMartinville B., Volpe M.A., Risdahl J.S., Rich S.S., Pearson P.L., Miller F.S. III and King R.A.: Clinical findings and linkage analysis in X-linked ocular albinism. Proceedings of the Greenwood Genetic Center, 7:222-223, 1988.

115. Graham J.M. Jr., Charman C.E., Chaisson R. and Donahue K.C.: Postnatal head deformation: anthropological observations and applications to the treatment of postnatal plagiocephaly. Proceedings of the Greenwood Genetic Center, 7:156-159, 1988.

116. Moeschler J.B., Marin-Padilla M., Rawnsley E. and Graham J.M. Jr.: Pathogenesis of X-linked hydrocephalus: lessons from fetal neuropathology. Proceedings of the Greenwood Genetic Center 7:159-160, 1988.

117. Graham J.M. Jr., Charman C.E., Chaisson R. and Donahue K.C.: Pathogenesis and treatment of non-synostotic plagiocephaly. Clinical Research 36:218A, 1988.

118. Graham J.M. Jr., deMartinville B., Volpe M.A., Risdahl J.S., Rish S.S., Pearson P.L., Miller F.S. and King R.A.: X-lined ocular albinism: clinical delineation and linkage analysis. Clinical Research 36:207A, 1988.

119. Moeschler J.B., Rawnsley E., Graham J.M. Jr. and Marin-Padilla M.: Pathogenesis of X-linked hydrocephalus: aqueductal stenosis is secondary to compression. Pediatric Research 23:269A, 1988.

120. Graham J.M. Jr., deMartinville B., Volpe M.A., Risdahl J.S., Rich S.S., Miller F.S. and King R.A.: Delineation of X-linked ocular albinism. Pediatric Research 23:267A, 1988.

121. Graham J.M. Jr., Edwards M.J., Lipson A., Uebster W. and Edwards M.J.: Gestational hyperthermia may cause Moebius syndrome. Pediatric Research 23:267A, 1988.

122. Graham J.M. Jr., Iafolla A.K., Fratkin J.D., Spiegel P.K. and Cohen M.M. Jr.: Delineation of congenital hypothalamic hamartoblastoma syndrome. Teratology 37:462, 1988. Presented at the Teratology Society Meeting, Palm Beach, Florida, June 10-15, 1988.

123. Graham J.M. Jr., Edwards M.J., Lipson A.H., Webster W.S. and Edwards M.J.: Gestational hyperthermia as a cause for Moebius syndrome. Teratology 37:461-442, 1988. Presented at the Teratology Society Meeting, Palm Beach, Florida, June 1015, 1988.

124. Graham J.M. Jr., Charman C.E., Chaisson R. and Donahue K.C.: Deformation as a cause and treatment for non-synostotic plagiocephaly. Teratology: The International Journal of Abnormal Development, 38(5~:521, 1988. Presented at the International Federation of Teratology Societies Meeting, Kyoto, Japan, July 1416, 1988.

125. Graham J.M. Jr., Edwards M.J., Lipson A.H., Webster W.S. and Edwards M.J.: Moebius syndrome caused by gestational hyperthermia. Teratology: The International Journal of Abnormal Development, 38t5):520, 1988. Presented at the International Federation of Teratology Societies Meeting, Kyoto, Japan, July 1416, 1988.

126. Graham J.M. Jr., Iafolla A.K., Fratkin J.D., Spiegel P.K. and Cohen M.M. Jr.: Congenital hypothalamic hamartoblastoma syndrome. Teratology: The International Journal of Abnormal Development, 38(5):520, 1988. Presented at the International Federation of Teratology Societies Meeting, Kyoto, Japan, July 14-16, 1988.

127. Berg S.Z., Furey P., and Graham J.M. Jr.: Paralysis of vertical gaze and hepatosplenomegaly as a phenotypic marker for Niemann-Pick disease, type C. American Journal of Human Genetics, 43:A39, 1988. Presented at the 1988 American Society of Human Genetics Meeting, New Orleans LA, Oct. 12-15, 1988.

128. Graham J.M. Jr., Morse R.P., Rockenmacher S., Lin A., Hall B.D., MacLeod P.J. and Pyeritz R.E.: Infantile Marfan Syndrome. American Journal of Human Genetics, 43:A51, 1988.

129. Moeschler J.B., Shear T.R., Gibbs E.D., and Graham J.M. Jr.: Psychoeducational Aspects of Aarskog Syndrome. American Journal of Human Genetics, 43:A62, 1988. Presented at the 1988 American Society of Human Genetics Meeting, New Orleans, LA, Oct. 12-15, 1988.

130. Park J.P., Wurster-Hill D.H., Edwards M.J., and Graham J.M. Jr.: Diploid/polyploid mixoploidy in young adolescents confirmed by cytogenetic analysis of fibroblasts. American Journal of Human Genetics, 43:A63, 1988.

131. Edwards M.J., Moeschler J.B. and Graham J.M. Jr.: Clinical correlations with collagen biochemistry in osteogenesis imperfecta. American Journal of Human Genetics, 43:A82, 1988.

132. Neidich J.A., Nussbaum R.L., Packer R., Graham J.M. Jr., Donnenfeld A.E., Emanuel B.S. and Puck J.M.: Heterogeneity in clinical severity and molecular lesions in Aicardi syndrome. American Journal of Human Genetics, 43:A91, 1988. Presented at the 1988 American Society of Human Genetics Meeting, New Orleans, LA, October 1215, 1988.

133. Rawnsley E., Edwards M.J. and Graham J.M. Jr.: Cystic hygroma and lethal multiple pterygia. American Journal of Human Genetics, 43:A245, 1988.

134. Moeschler J.B., Edwards M.J., Graham J.M. Jr.: Lower extremity equivalent of the Poland anomaly. Pediatric Research, 78A, 1989. Presented at the Society for Pediatric Research Meeting, Washington D.C., May 3, 1989.

135. Graham J.M. Jr., Iafolla A.K., Lipson A.H., Webster W.S. and Edwards M.J.: Gestational hyperthermia as a possible cause for Moebius syndrome. Pediatric Research 25:76A 1989. Presented at the Society for Pediatric Research Meeting, Washington D.C., May 3, 1989.

136. Edwards M.J., Park J., Wurster-Hill D.H., Graham J.M. Jr.: Clinical features of diploid/polyploid mixoploidy in older individuals. Pediatric Research 25:76A, 1989. Presented at the Society for Pediatric Research Meeting, Washington D.C., May 3, 1989.

137. Borenstein T.C., Korenberg J.R., Platt L.D., Davos I., Gans S., Schreck R. and Graham J.M. Jr.: Congenital gastric teratoma in Beckwith-Wiedemann syndrome. Pediatric Research 25:75A, 1989. Presented at the Society for Pediatric Research Meeting, Washington D.C., May 3, 1989.

138. Graham J.M. Jr, Reynolds J.F. and Kelley R.I.: Cardio-Facio-Cutaneous (CFC) syndrome: Studies of peroxisomal and fatty acid metabolism. Proceedings of the Greenwood Genetic Center, 8:142-145, 1989. Presented at the IX David W. Smith Morphogenesis and Malformations Meeting, Oakland CA, Aug. 3-6, 1988.

139. Graham J.M. Jr, Morse R.P., Rochenmacher S., Lin A., Hall B.D., MacLeod P.J. and Pyeritz R.E.: Infantile Marfan syndrome. Proceedings of the Greenwood Genetic Center, 8:213-214, 1989.

140. Graham J.M. Jr, Edwards M.J., Lipson A.H., Webster W.S., Edwards M.J.: Gestational hyperthermia as a possible cause for Moebius syndrome. Proceedings of the Greenwood Genetic Center, 8:213, 1989.

141. Graham J.M. Jr., Furrey P. and Berg S.Z.: Paralysis of vertical gaze and hepatosplenomegaly as a phenotypic marker for Niemann-Pick Disease, Type C. Proceedings of the Greenwood Genetic Center, 8:213, 1989.

142. Edwards M.J., Rawnsley E and Graham J.M. Jr.: Posterior nuchal systic hygroma and lethal multiple pterygia. Proceedings of the Greenwood Genetic Center, 8:156,1989. Presented at the IX David W. Smith Morphogenesis and Malformations Meeting, Oakland CA, Aug. 3-6, 1988.

143. Moeschler J.B., and Graham J.M. Jr.: Radial-renal-ocular syndromes: a phenotypic community. Proceedings of the Greenwood Genetic Center, 8:158-159, 1989. Presented at the IX David W. Smith Morphogenesis and Malformations Meeting, Oakland CA, Aug. 3-6, 1988.

144. Edwards M.J., Moeschler J.B. and Graham J.M. Jr.: Clinical and biochemical correlation in osteogenesis imperfecta. Proceedings of the Greenwood Genetic Center, 8:177, 1989. Presented at the IX David W. Smith Morphogenesis and Malformations Meeting, Oakland CA, Aug. 3-6, 1988.

145. Lin A.E., Morter G. and Graham J.M. Jr.: CNS malformations in the CHARGE association. Proceedings of the Greenwood Genetic Center, 8:202, 1989. Presented at the IX David W. Smith Morphogenesis and Malformations Meeting, Oakland CA, Aug. 3-6, 1988.

146. Graham J.M. Jr.: New syndrome of hypoplastic 1st and 2nd digital rays, microcephaly, and diabetes in siblings born to consanguineous parents. Proceedings of the Greenwood Genetics Center, in press, 1990. Presented at the X David W. Smith Morphogenesis and Malformations Meeting, Madrid, Spain, May 23-27, 1989.

147. Korenberg J.R., Barker D., Fain P., Graham J.M. Jr., Pribyl T., Pulst S.M.: Achondroplasia is not tightly linked to the locus for neurofibromatosis 1. Presented at the Tenth International Workshop on Human Gene Mapping. New Haven, CT, June 10-17, 1989.

148. Graham J.M. Jr.: New syndrome of hypoplastic 1st and 2nd digital rays, microcephaly, and diabetes in siblings born to consanguineous parents. Presented at the March of Dimes Clinical Genetics Conference, Boston MA, July 9-12, 1989.

149. Neidich J.A., Emanuel B.S., Graham J.M. Jr., Nussbaum R.L., Packer R.J. and Puck J.M.: X inactivation and molecular mapping in Aicardi Syndrome. Presented at the March of Dimes Clinical Genetics Conference, Boston MA, July 9-12, 1989.

150. Cooley W.C., Graham E.S., Moeschler J.B., Graham J.M. Jr.: Reactions of mothers and professionals to a film of parents of persons with Down syndrome. Developmental Medicine and Child Neurology, Presented at the American Academy of Cerebral Palsy and Developmental Medicine Meeting, San Francisco, CA, October 26, 1989.

151. Graham J.M. Jr.: New syndrome of type A2 brachydactyly, microcephaly, and diabetes in siblings born to consanguineous parents. American Journal of Human Genetics, 45(4):A76, 1989. Presented at the American Society of Human Genetics Meeting, Baltimore MD, November 11-15, 1989.

152. Cantrell M.A., Simpson E., Bicknell J.N., Pagon R.A., Walker D.C., Thuline H.C., Graham J.M. Jr.: Absence of H-Y antigen expression in a patient with deletion of the long arm of the Y chromosome and isolation of new DNA probes for region 6 of the Y chromosome. American Journal of Human Genetics, 45(4):A134, 1989. Presented at the American Society of Human Genetics Meeting, Baltimore MD, November 11-15, 1989.

153. Francomano C.A., Finkelstein J.E., Yamada Y., Pyeritz R.E., Graham J.M. Jr.: Analysis of the chondroitin sulfate proteoglycan core protein (CSPGCP) gene in achondroplasia and pseudoachondroplasia. American Journal of Human Genetics, 45(4):A140, 1989. Presented at the American Society of Human Genetics Meeting, Baltimore MD, November 11-15, 1989.

154. Pribyl T., Korenberg J.R., Barker D., Fain P., Ren M., Graham J.M. Jr., Pulst S.M.: The gene for achondroplasia is not tightly linked to the locus for neurofibromatosis. American Journal of Human Genetics, 45(4):A157, 1989. Presented at the American Society of Human Genetics Meeting, Baltimore MD, November 11-15, 1989.

155. Richardson, M., Christian C.L., Platt L.D., Santulli T., DePond W., Peckowitz S.H., Graham J.M. Jr.: Lethal cardiac anomalies in fetal Turner syndrome. Clinical Research, 38(1):188A, 1990. Presented at the Western Society of Pediatric Research Meeting, Carmel, CA, February 6-9, 1990.

156. Edwards M.J., Moeschler J.B., Fahy M.J., Hall J.G., Graham J.M. Jr.: History of gestational hyperthermia in two patients with amyoplasia. Clinical Research, 38(1):186A, 1990.

157. Edwards M.J., Rawnsley E., Graham J.M. Jr.: Cystic hygroma and lethal multiple pterygia in a fetus. Clinical Research, 38(1):186A, 1990. Presented at the Western Society of Pediatric Research Meeting, Carmel, CA, February 6-9, 1990.

158. Graham J.M. Jr., Moeschler J.B., Cooley W.C.: Reactions of mothers and professionals to a film of parents of persons with Down syndrome. Clinical Research, 38(1):186A, 1990.

159. Graham J.M. Jr.: New syndrome of type A2 brachydactyly, microcephaly and diabetes in siblings born to consanguineous parents. Clinical Research, 38(1):186A, 1990.

160. Smith A.C.M, Graham J.M. Jr., Hall J.G.: Congenital contractures, brain structural anomalies, cardiac anomalies and renal anomalies. Clinical Research, 38(1):188A, 1990. Presented at the Western Society of Pediatric Research Meeting, Carmel, CA, February 6-9, 1990.

161. Cooley W.C., Graham E.S., Moeschler J.B., Graham J.M. Jr.: Reactions of mothers and medical professionals to a film of parents of persons with Down syndrome. Presented at the Ambulatory Pediatric Society, Region I Annual Meeting, Farmington, CT, February 9, 1990.

162. Edwards M.J., Moeschler J.B., Fahy M.J., Hall J.G., Graham J.M. Jr.: History of gestational hyperthermia in two patients with amyoplasia. Pediatric Research, 27(4):68A, 1990. Presented at the Society for Pediatric Research Meeting, Anaheim, CA, May 7, 1990.

163. Christian C.L., Richardson M., Platt L.D., Santulli T., DePond W., Peckowitz S.H., Graham J.M. Jr.: Turner syndrome and hypoplastic left heart. Pediatric Research, 27(4):67A, 1990.

164. Falik-Borenstein T.C., Graham J.M. Jr., Schreck R.R.: Chimerism versus mosaicism in CVS. Pediatric Research, 27(4):68A, 1990. Presented at the Society for Pediatric Research Meeting, Anaheim, CA, May 7, 1990.

165. Moeschler, J.B., Edwards, M.J., and Graham, J.M. Jr.: Lower extremity equivalent of the Poland anomaly. Teratology 41(5):579, 1990. Presented at the Teratology Society Meeting, Victoria, B.C., June 6, 1990.

166. Edwards, M.J., Moeschler, J.B., Fahy, M., Hall, J.G., and Graham, J.M. Jr.: History of gestational hyperthermia in two patients with amyoplasia. Teratology 41(5):552, 1990. Presented at the Teratology Society Meeting, Victoria, B.C., June 6, 1990.

167. Tick, D.B., Greenberg, F. and Graham, J.M. Jr.: The pattern and form of human somatic structural ectopy. Teratology 41(5):596, 1990. Presented at the Teratology Society Meeting, Victoria, B.C., June 6, 1990.

168. Graham, J.M. Jr.: New Syndrome of hypoplastic 1st and 2nd digital rays, microcephaly, and diabetes in siblings born to consanguinous parents. Proceedings of the Greenwood Genetics Center 9:1103-104, 1990. Presented at the X David W. Smith Workshop on Malformations and Morphogenesis, Madrid, Spain, May 23-29, 1989.

169. Graham J.M. Jr., Edwards Matthew J., Iafolla, A.K., Moeschler J.B., Edwards Marshall J.: Sporadic patterns of anomalies due to maternal hyperthermia: Moebius syndrome and amyoplasia. American Journal of Human Genetics, 47(3):458, 1990

170. Christian C.L., Nagel R.A., and Graham J.M. Jr.: Marinesco-Sjogren syndrome associated with decreased growth hormone levels. American Journal of Human Genetics, 47(3):A51, 1990.

171. Klein J., Hixon H.E.C., Graham J.M. Jr.: Multidisciplinary involvement in the evaluation of aborted fetuses and stillborns: case reports and literature review. American Journal of Human Genetics, 47(3):A279, 1990.

172. Toriello H.V., Moeschler J.M., Graham J.M. Jr., Higgins J.V.: Syndromes with renal, radial and ocular defects: A phenotypic community. American Journal of Human Genetics, 47(3):A80, 1990.

173. Graham, J.M. Jr., Rimoin, D.L., Tolo, V., Jones, M.C. and Marsh, J.L.,: Surgical treatments for congenital anomalies. American Journal of Human Genetics, 47(3):21, 1990. Presented at the American Society of Human Genetics Meeting, Cincinnatti OH, October 16-21, 1990.

174. Graham, J.M. Jr. Edwards M.J., Iafolla, A.K., and Moeschler, J.B.: Sporadic patterns of anomalies due to maternal hyperthermia: Moebius syndrome and amyoplasia. Proceedings of the Greenwood Genetics Center, 10:106, 1991. Presented at the XI David U. Smith Workshop on Malformations and Morphogenesis, Lexington, KY, August 5-8, 1990.

175. Christian C.L., Lachman R.S., Aylesworth A.S., Fujimoto A., Gorlin R.J., Lipson M.H. and Graham J.M. Jr.: Radiological findings in Hallermann-Streiff syndrome: report of five cases and a review of the literature. Proceedings of the Greenwood Genetic Center, 10:123. XII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

176. Graham, J.M. Jr., Edwards, M.J., Iafolla, A.K., Moeschler, J.B.: Sporadic patterns of anomalies due to maternal hyperthermia: Moebius syndrome and amyoplasia. Presented at the 4th Manchester Birth Defects Conference, Manchester, England, October 2-5, 1990

177. Sharony R., Garber A., Schreck R., Platt L., Viskochil D., Graham J.M. Jr.: Radial ray reduction defects as part of the valproic acid embryopathy spectrum. Clinical Research, 39(1):65A, 1991. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 6-9, 1991.

178. Christian C.L., Fleisher D.R., Feldman E.J., Pepkowitz S.H., Iafolla A.K., DiLiberti J.J., Graham J.M. Jr.: Lipid storage myopathy associated with Ruvalcaba-Myhre-Smith syndrome: Treatment with carnitine. Clinical Research, 39(1):64A, 1991. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 6-9, 1991.

179. Curry C.J.R., Micek M., Yu C., Golbus M., Graham J.M. Jr.: Prenatal diagnosis of trisomy 12 mosaicism. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 6-9, 1991. Clinical Research, 39(1):65A, 1991.

180. Klein J., Platt L., Tick D., Carlson D., Pepkowitz S., Graham J.M. Jr.: Fraser syndrome: Prenatal and postnatal diagnosis. Clinical Research, 39(1):134A, 1991.

181. Falik-Borenstein T.C., Graham J.M. Jr., Klein J., Schreck R.R.: Trisomy 8 mosaicism in CVS: postnatal confirmation studies and pregnancy outcome. Pediatric Research, 29(4):69A, 1991.

182. Sharony R., Garber A., Schreck R., Platt L.D., Buehler B.A., Graham J.M. Jr.: Valproate exposure during pregnancy causing radial ray reduction defects. Pediatric Research, 29(4):71A, 1991.

183. Christian C.L, Fleischer D.R., Feldman E.J., Pepkowitz S.H., Iafolla A.K., DiLiberti J.H., Graham J.M. Jr.: Carnitine deficiency and lipid storage myopathy associated with Ruvalcaba-Myhre-Smith syndrome. Pediatric Research, 29(4):68A, 1991. Presented at the The Society for Pediatric Research Meeting, New Orleans, LA, April 29-May 2, 1991.

184. Graham J.M. Jr., Sharony R., Garber A., Schreck R., Platt L.D., Buehler B.A.: Preaxial ray reduction defects valproate exposure during pregnancy. Teratology, 43(5):441, 1991. Presented at the Teratology Society Meeting, Boca Raton, FL, June 25, 1991.

185. Christian C.L., Pressman B.D., Schreck R., Roulston D., Graham J.M. Jr.: Progression of cranial imaging findings in the acrocallosal syndrome. Presented at the March of Dimes Clinical Genetics Conference, Vancouver, B.C. Canada, July 7-10, 1991.

186. Graham J.M. Jr.: Familial progressive cerebral calcification, microcephaly, spastic quadraparesis and failure of developmental progression. Presented at the March of Dimes Clinical Genetics Conference, Vancouver, B.C. Canada, July 7-10, 1991 .

187. Sharony R., Pepkowitz S., Hixon H., Graham J.M. Jr.: Diprosopus - A pregastrulation defect involving the head, neural tube, heart and diaphragm. Proceedings of the Greenwood Genetic Center, 11:80-81, 1992. Presented at the XII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

188. Perez-Aytes A., Carey J.C., Hoyme E.H., Graham J.M. Jr., Aleck K.: Urethral obstruction sequence, lower limb reduction defects, and the vascular disruption hypothesis. Proceedings of the Greenwood Genetic Center, 11:71-72, 1992. Presented at the XII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

189. Moeschler J.B., Graham J.M. Jr.: "Mild" Brachmann-deLange syndrome: phenotypic and developmental characteristics of mildly affected individuals. Proceedings of the Greenwood Genetic Center, 11:113-114, 1992. Presented at the XII David R. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

190. Braddock S., Lachman R., Charman C., Carey J.C., Ireland M., Graham J.M. Jr.: Radiological features in Brachmann-deLange syndrome. Proceedings of the Greenwood Genetic Center, 11:130, 1992. Presented at the XII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

191. Christian C.L., Schreck R., Neufeld N., Graham J.M. Jr.: Mosaic trisomy 13: phenotypic presentations and clinical implications for prenatal diagnosis. Proceedings of the Greenwood Genetics Center, 11:163-164, 1992. Presented at the XII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 27-October 1, 1991.

192. Sharony R., Pepkowitz S., Hixon H., Graham J.M. Jr.: Diprosopus - A pregastrulation defect involving the head, neural tube, heart and diaphragm. Presented at the Second International Workshop on Fetal Genetic Pathology, Big Sky, Montana, October 12-16, 1991.

193. Sharony R., Hixon H., Pepkowitz S., Carlson D., Platt L., Graham J.M. Jr.: Experience with a fetal dysmorphology/pathology service in an academic medical center. Presented at the Second International Workshop on Fetal Genetic Pathology, Big Sky, Montana, October 12-16, 1991.

194. Wagstaff J., Knoll, J.H.M., Fleming J., Kirkness E.F., Martin-Gallardo A., Greenberg F., Graham J.M. Jr., Menninger J., Ward D., Venter J.C., Lalande M. Localization of the GABAA receptor B3 subunit gene (GABRB3) to the Angelman/Prader-Willi region of human chromosome 15. American Journal of Human Genetics, 49(4):422, 1991. Presented at 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

195. Knoll J.H.M., Wagstaff J., Glatt K., Graham J.M. Jr., Greenberg F., Horsthemke B., Lalande M. Molecular analysis of distal 15qllql3: A difference between Angelman and Prader-Willi syndromes. American Journal of Human Genetics, 49(4):18, 1991. Presented at 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

196. Carlson D., Graham J.M. Jr., Platt L.D., Pepkowitz S.H., Klein J. Early Prenatal Diagnosis of Fraser Syndrome. American Journal of Human Genetics, 49(4):129, 1991. Presented at 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

197. Garber A., Klein J., Carlson D., Platt L., Graham J.M. Jr.: Natural history of recurrent, non-syndromal posterior nuchal cystic hygroma. American Journal of Human Genetics, 49(4):138, 1991. Presented at 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

198. Falik-Borenstein T., Graham J.M. Jr., Platt L., Schreck R.: Chromosomal mosaicism in cvs: fetal development and pregnancy outcome, a two years follow-up. American Journal of Human Genetics, 49(4):173, 1991. Presented at 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

199. Bohlman M.C., Edwards M.J., Graham J.M. Jr., Fischel-Ghodsian N.: Deletion in blood mitochondrial DNA in Kearns-Sayre syndrome. American Journal of Human Genetics, 49(4):187, 1991. Presented at the 8th International Congress of Human Genetics, Washington, D.C. October 6-11, 1991.

200. Klein J., Schreck R., Platt L.D., Borenstein T., Graham J.M. Jr.: CVS mosaicism for trisomy 8 and genetic counseling. American Journal of Human Genetics, 49(4):221, 1991. Presented at the 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

201. Christian C.L., Schreck R., Neufeld N., Graham J.M. Jr.: Mosaic trisomy 13: phenotypic presentations. American Journal of Human Genetics, 49(4):259, 1991. Presented at the 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

202. Coons G., Klein J., Schreck R., Pepkowitz S., Graham J.M. Jr.: Flow cytometric diagnosis of clinically suspected triploidy. American Journal of Human Genetics, 49(4):259, 1991. Presented at the 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

203. Roulston D., Altherr M., Wasmuth J.J., Christian C., Graham J.M. Jr., Schreck R.R.: Confirmation of a suspected deletion of 4pl6 by fluorescent in situ hybridization (FISH) with a cosmid probe. American Journal of Human Genetics, 49(4):274, 1991. Presented at the 8th International Congress of Human Genetics, Washington, DC. October 6-11, 1991.

204. Braddock S.R., Lachman R.S., Graham J.M. Jr.: Radiology of mild DeLange syndrome. Clinical Research 40(1):57A, 1992. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 5-8, 1992.

205. Bacino C.A, Platt L.D., Carlson D., Pepkowitz S., Garber A., Graham J.M. Jr.: Prenatal detection and natural history of the Neu-Laxova syndrome. Pediatric Research, 31(4):68A, 1992. Presented at the Society for Pediatric Research Meeting, Baltimore, MD, May 4-7, 1992.

206. Braddock S.R., Charman C.E., Clarren S.K., Graham J.M. Jr.: Distinguishing characteristics and long term outcomes associated with malformational vs. deformational craniosynostosis. Teratology, 45:462, 1992. Presented at the Teratology Society Meeting, Boca Raton, FL, June 28-July 2, 1992.

207. Garber A.P., Carlson D.E., Klein J., Schreck R., Braddock S.R., [Baham J.M. Jr.: Clinical findings in mosaic trisomy 16. Teratology, 45:508, 1992. Presented at the Teratology Society Meeting, Boca Raton, FL, June 28-July 2, 1992.

208. Hsu W.T., Carlson D.E., Klein J., Fischel-Ghodsian N., Graham J.M. Jr., Schreck R.: Prenatal detection of mosaic trisomy 16 associated with neonatal lethal phenotype. Presented at March of Dimes Clinical Genetics Conference, Stanford University, Palo Alto, CA, July 12-15, 1992.

209. Weng E.Y., Moeschler J.B., Graham J.M. Jr.: Longitudinal follow-up in 14 probands with Beckwith-Wiedemann syndrome. Proceedings of the Greenwood Genetics 12:59-60, 1993. Presented at the XIII David W. Smith Workshop on Malformations and Morphogenesis, Wake Forest University, Winston-Salem, NC, August 5-9, 1992.

210. Wilcox W.R., Graham J.M. Jr.: The acrocallosal syndrome: Report of 4 cases, review of the literature, and proposed diagnostic criteria. Proceedings of the Greenwood Genetics 12:56, 1993. Presented at the XIII David W. Smith Workshop on Malformations and Morphogenesis, Wake Forest University, Winston-Salem, NC, August 5-9, 1992.

211. Braddock S.R., Graham J.M. Jr.: Ventruto syndrome: Report of a second family. Proceedings of the Greenwood Genetics 12:106, 1993. Presented at the XIII David W. Smith Workshop on Malformations and Morphogenesis, Wake Forest University, Winston-Salem, NC, August 5-9, 1992.

212. Graham J.M. Jr., Braddock S.R.: Distinguishing characteristics and long term outcomes associated with malformational vs. deformational craniosynostosis. Presented at the 5th Manchester Birth Defects Conference, Manchester, United Kingdom, October 13-16, 1992.

213. Weng E.Y., Moeschler J.B., Graham J.M. Jr.: Longitudinal follow-up in 14 probands with Beckwith-Wiedemann syndrome. American Journal of Human Genetics, 51(4):A311, 1992.

214. Sharony R., Carlson D.E., Graham J.M. Jr., Platt L.D.: Prenatal diagnosis of conjoined twins: associated sonographic anomalies. American Journal of Human Genetics, 51(4):A414, 1992.

215. Weng E.Y., Moeschler J.B., Graham J.M. Jr.: Longitudinal follow-up in 14 probands with Beckwith-Wiedemann syndrome. Clinical Research, 41(1):70A, 1993. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 17-20, 1993.

216. Rabinowitz YS, Wilcox W, Shorr N, Enzer Y, and Graham JM Jr.: Fraser cryptopthalmos syndrome (FCS) with visual potential following eyelid and forniceal reconstruction. Investigative Opthalmology and Visual Science, 1993. Presented at the Association for Research and Vision in Ophthalmology, Sarasota, FL, May 27, 1993.

217. Rabinowitz YS, Wilcox W, Shorr N, Enzer Y, and Graham JM Jr.: Fraser cryptopthalmos syndrome (FCS) with visual potential following eyelid and forniceal reconstruction. Presented at the International Conference on Cornea and Eye Banking, Jerusalem, Israel, June 20-24, 1993.

218. Garber, A., Schreck R., Oeztas S., Carlson D.E., Fischel-Ghodsian N., Hsu W-T, Graham J.M. Jr.: Uniparental disomy for chromosome 16 in a fetus. Presented at the Seventh International Clinical Genetics Seminar on Dysmorphology and Genetics of Cardiovascular Disorders, Pythagorion, Samos, Greece, May 27-June 1, 1993.

219. Graham J.M. Jr., Edwards M.J.: Diploid polyploid mixoploidy in young adults. Teratology 47(5):56, 1993. Presented at the Teratology Society Meeting, Tucson, AZ, June 26-July 1, 1993.

220. Garber A., Schreck R., Oeztas S., Carlson D., Fischel-Ghodsian N., Hsu W., Graham J.M. Jr.: Analysis of prenatally-detected mosaic trisomy 16. Teratology 47(5):57, 1993. Presented at the Teratology Society Meeting, Tucson, AZ, June 26July 1, 1993.

221. Graham J.M. Jr., Skovby F., Cohen M.M.: Spinal cord compromise in Proteus syndrome. Proceedings of the Greenwood Genetics 13:130, 1994. Presented at the XIV David W. Smith Workshop on Malformations and Morphogenesis, Mont-Tremblant, Quebec, Canada, August 12-17, 1993.

222. Bacino C.A., Schreck R., Pepkowitz S., Graham J.M. Jr.: Non-mosaic trisomy 22 surviving through late gestation: delineation of the clinical phenotype. Proceedings of the Greenwood Genetics 13:119, 1994. Presented at the XIV David W. Smith Workshop on Malformations and Morphogenesis, Mont-Tremblant, Quebec, Canada, August 12-17, 1993.

223. Graham J.M. Jr., Prowse M., Hixon H., Johnson C.: Urorectal septal malformation sequence and extrophy of the cloaca malformation sequence: a spectrum of problems in cloacal septation. Proceedings of the Greenwood Genetics 13:74, 1994. Presented at the XIV David W. Smith Workshop on Malformations and Morphogenesis, Mont-Tremblant, Quebec, Canada, August 12-17, 1993.

224. Hsu W-T., Garber A., Carlson D., Fischel-Ghodsian N., Graham J.M. Jr., Linn S., Wheeler M., Oztas, Schreck R.: Prenatal detected trisomy 16 in two phenotypically abnormal newborns. American Journal of Human Genetics, 53(3):A1419, 1993. Presented at the 43rd American Society of Human Genetics Meeting, New Orleans, LA, October 5-9, 1993.

225. Graham J.M. Jr., Skovby F., Cohen M.M.: Spinal cord compromise in Proteus syndrome. American Journal of Human Genetics, 53(3):A443, 1993. Presented at the 43rd American Society of Human Genetics Meeting, New Orleans, LA, October 5-9, 1993.

226. Bacino C.A., Graham J.M. Jr., Fischel-Ghodsian N., Hsu W.T., Schreck, R. Nonmosaic trisomy 22: a lethal trisomy surviving to late gestation. American Journal of Human Genetics, 53(3):A1380, 1993. Presented at the 43rd American Society of Human Genetics Meeting, New Orleans, LA, October 5-9, 1993.

227. Rabinowitz YS, Wilcox W, Shorr N, Enzer Y, and Graham JM Jr.: Fraser cryptopthalmos syndrome (FCS) with visual potential following eyelid and forniceal reconstruction. Presented at the American Academy of Ophthalmology, Chicago, IL, November 8-15, 1993.

228. Ming JE, Stiehm ER, Graham JM Jr.: Immunodeficiency as a component of genetic syndromes. Pediatric Research, 35(4):92A, 1994. Presented at the American Pediatric Society-Society for Pediatric Research Meeting, Seattle, WA, May 2-5, 1994.

229. Ming JE, Stiehm ER, Graham JM Jr.: Immunodeficiency is a component of genetic syndromes. Presented at the Workshop on Primary and Acquired Immunodeficiencies, Orvieto, Italy, June 18-21, 1994.

230. Ming JE, Stiehm ER, Graham JM Jr.: Immunodeficiency as a component of genetic syndromes. Teratology 49(5):405, 1994. Presented at the Teratology Society Meeting, Las Croabas, Puerto, June 25-30, 1994.

231. Mortier, GR, Wilkin DJ, Stern H, De Paepe A, Falk RE, Shohat M, Schwartz CE, Johnson CL, Jones MC, Jones KL, Graham JM Jr, Rimoin DL, Cohn DH.: Clinical features in Stickler syndrome families linked and unlinked to the type II collagen gene. Proceedings of the Greenwood Genetics Center,14:48, 1995. Presented at the XV David W. Smith Workshop on Malformations and Morphogenesis, Tampa, Florida, August 4-9, 1994.

232. Moeschler JB, Guttmacher AE, Graham JM Jr.: Blepharophimosis-cleft palatehypogonadism-growth and mental retardation: a new multiple congenital anomaly/mental retardation syndrome in brothers. Proceedings of the Greenwood Genetics Center, 14:85, 1995. Presented at the XV David W. Smith Workshop on Malformations and Morphogenesis, Tampa, Florida, August 4-9, 1994.

233. Bhatt S, Wilcox WR, Dobyns WB, Pressman BD, Graham JM Jr.: Recurrent frontal cortical dysplasia - A new neuronal migration disorder. Proceedings of the Greenwood Genetics Center, 14:70, 1995. Presented at the XV David W. Smith Workshop on Malformations and Morphogenesis, Tampa, Florida, August 4-9, 1994.

234. Graham JM Jr., Hixon H, Bacino CA, Daack-Hirsch S, Stadler S, Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome: description of a family and report of a sporadic case with de novo 4pl6; 8q24.11 translocation. Proceedings of the Greenwood Genetics Center, 14:41, 1995. Presented at the XV David W. Smith Workshop on Malformations and Morphogenesis, Tampa, Florida, August 4-9, 1994.

235. Bhatt S, Dobyns WB, Pressman BD, Graham JM Jr: Apparent X-linked pachygyria/lissencephaly with agenesis of the corpus callosum. American Journal of Human Genetics 55(3):A78, 1994. Presented at the American Society of Human Genetics Meeting, Montreal, Quebec, October 18-22, 1994.

236. Ming JE, Stiehm ER, Graham JM Jr: Immunodeficiency as a component of recognizable syndromes. American Journal of Human Genetics 55(3):A88, 1994. Presented at the American Society of Human Genetics Meeting, Montreal, Quebec, October 18-22, 1994.

237. Graham JM Jr., Hixon H, Bacino CA, Daack-Hirsch S, Stadler S, Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome: Description of a family and report of a sporadic case with a de novo 4pl6; 8q24.11 translocation. American Journal of Human Genetics 55(3):A107, 1994. Presented at the American Society of Human Genetics Meeting, Montreal, Quebec, October 18-22, 1994.

238. Graham JM Jr., Biesecker LG: Linking, lumping and splitting Pallister-Hall syndrome. Presented at the National Biomedical Research Day, Cedars-Sinai Medical Center, Los Angeles, California, October 21, 1994.

239. Graham JM Jr., Hixon H., Bacino CA, Daack-Hirsch S., Stadler S., Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome with linkage to the branchio-oto-renal syndrome locus at 8ql3.3. Presented at the National Biomedical Research Day, Cedars-Sinai Medical Center, Los Angeles, California, October 21, 1994.

240. Graham JM Jr., Hixon H., Bacino CA, Daack-Hirsch S., Stadler S., Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome with linkage to the branchio-oto-renal syndrome locus at 8ql3.3. Presented at the 6th Manchester Birth Defects Conference, Manchester, United Kingdom, November 1-4, 1994.

241. Graham JM Jr., Biesecker LG: Lumping and splitting clinical entities within the Pallister-Hall syndrome. Presented at the 6th Manchester Birth Defects Conference, Manchester, United Kingdom, November 1-4, 1994.

242. Graham JM Jr., Hixon H., Bacino CA, Daack-Hirsch S., Stadler S., Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome with linkage to the branchio-oto-renal syndrome locus at 8ql3.3. Clinical Research, 43:137A,1995. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 9-11, 1995.

243. Graham JM Jr., Biesecker LG: Lumping and splitting clinical entities within the Pallister-Hall syndrome. Clinical Research, 43:136A, 1995. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 9-11, 1995.

244. Bhatt S, Wilcox WR, Dobyns WB, Pressman BD, Graham JM Jr.: Recurrent frontal cortical dysplasia - A new neuronal migration disorder. Clinical Research, 43:152A, 1995. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 9-11, 1995.

245. Graham JM Jr., Biesecker LG: Genetics of Pallister-Hall syndrome. Presented at the 26th Annual March of Dimes Clinical Genetics Conference and the American College of Medical Genetics 2nd Annual Meeting, Los Angeles, CA, March 6-9, 1995.

246. Kim J, Hall BD, Goodfellow PJ, Graham JM Jr: Report of two families with X-linked cleft palate/ankyloglossia: description of clinical findings. Presented at the 26th Annual March of Dimes Clinical Genetics Conference and the American College of Medical Genetics 2nd Annual Meeting, Los Angeles, CA, March 6-9, 1995.

247. Hixon HH, Krakow D, Pepkowitz SH, Graham JM Jr: Intrauterine fetal demise evaluated through a Fetal Dysmorphology/Pathology Service. Presented at the 26th Annual March of Dimes Clinical Genetics Conference and the American College of Medical Genetics 2nd Annual Meeting, Los Angeles, CA, March 6-9,1995.

248. Graham JM Jr., Biesecker LG: Lumping and splitting within the Pallister-Hall syndrome - A lesson in human pathology. Invited Presentation at the 6th Biennial Congress of the Human Genetics Society, Club Mykonos, Langebaan, South Africa, March 26-29, 1995.

249. Graham JM Jr., Hixon H, Bacino CA, Daack-Hirsch S, Stadler S, Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome with linkage to the branchio-oto-renal syndrome locus at 8ql3.3. Presented at the 6th Biennial Congress of the Human Genetics Society, Club Mykonos, Langebaan, South Africa, March 26-29, 1995.

250. Bhatt S, Wilcox WR, Dobyns WB, Pressman BD, Graham JM Jr.: Recurrent frontal cortical dysplasia - A new neuronal migration disorder. Presented at the 6th Biennial Congress of the South African Human Genetics Society, Club Mykonos, Langebaan, South Africa, March 26-29, 1995.

251. Graham JM Jr., Biesecker LG: Linking, lumping and splitting Pallister-Hall syndrome. Presented at the IV International Fetal Genetic Pathology Workshop, Kruger National Park, South Africa, March 31-April 2, 1995.

252. Graham JM Jr.: Status of the human gene map for craniofacial malformation syndromes. Invited Presentation at the IV International Fetal Genetic Pathology Workshop, Kruger National Park, South Africa, March 31-April 2, 1995.

253. Graham JM Jr., Biesecker LG: The Genetics of Pallister-Hall syndrome. Pediatric Research 37:82A, 1995. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, San Diego, CA, May 7-11, 1995.

254. Graham JM Jr., Hixon H, Bacino CA, Daack-Hirsch S, Stadler S, Murray JC: Autosomal dominant transmission of a Goldenhar-like syndrome with linkage to the branchio-oto-renal syndrome locus at 8ql3.3. Pediatric Research 37:83A, 1995. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, San Diego, CA, May 7-11, 1995.

255. Kim J, Hall BD, Goodfellow PJ, Graham JM Jr: Report of two families with X-linked cleft palate/ankylogossia: description of clinical findings. Pediatric Research 37:83A, 1995. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, San Diego, CA, May 7-11, 1995.

256. Wilcox WR, Graham JM Jr, Reinker K, Hsia YE, Rimoin DL: Marinesco-Sjögren syndrome is a lysosomal lipid storage disorder and is not corrected in vitro by co-culture with control cells. Pediatric Research 37:155A, 1995. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, San Diego, CA, May 7-11, 1995.

257. Graham JM Jr, Biesecker LG: The genetics of Pallister-Hall syndrome. Teratology, 1995. Presented at the Teratology Society Meeting, Newport Beach, CA., June 24-29, 1995.

258. Biesecker LG, Kang S, Abbott M, Allen J, Olney A, Grebe T, Clericuzio C, Graham JM Jr: Linkage studies in Pallister-Hall syndrome. Proceedings of the Greenwood Genetics Center, 15:161, 1996. Presented at the XVI David W. Smith Workshop on Malformations and Morphogenesis, Big Sky, Montana, July 29-August 3, 1995.

259. Graham JM Jr, Lewis RA, Dobyns WB: Cerebro-Oculo-Facial-Skeletal (COFS) syndrome: one spectrum of variable severity, or multiple separate syndromes. Proceedings of the Greenwood Genetics Center, 15:179, 1996. Presented at the XVI David W. Smith Workshop on Malformations and Morphogenesis, Big Sky, Montana, July 29-August 3, 1995.

260. Krakow D., Biesecker L., Van Dop C., Graham J.M. Jr.: Acrodysostosis: is it a G protein abnormality? Proceedings of the Greenwood Genetics Center, 15:165, 1996. Presented at the XVI David W. Smith Workshop on Malformations and Morphogenesis, Big Sky, Montana, July 29-August 3, 1995.

261. Kang S., Graham J.M. Jr., Abbott M., Biesecker L.G.: Linkage analysis of Pallister-Hall syndrome. American Journal of Human Genetics 57:A324, 1995.

262. Graham J.M. Jr.: Fibroblast growth factors and receptors: their role in morphogenesis. American Journal of Human Genetics 57:31, 1995. Presented at the American Society of Human Genetics Meeting, Minneapolis, MN, October 24-28, 1995.

263. Graham JM Jr., Zackai EH, Lewis RA, Dobyns WB: Delineation of cerebro-oculo-facial-skeletal (COFS) syndrome. Journal of Investigative Medicine 44:130A, 1996. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 15-18, 1996.

264. Krakow D, Biesecker L, Van Dop C, Lachman R, Rimoin DL, Graham JM Jr: Acrodysostosis: Is it a G protein abnormality? Journal of Investigative Medicine 44:131A, 1996. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 15-18, 1996.

265. Biesecker LG, Kang S, Allen JC, Grebe TA, Clericuzio CL, Haskins-Olney AH, Graham JM Jr.: Clinical and genetic analysis of Pallister-Hall syndrome. Pediatric Research 39:470, 1996. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, Washington, DC, May 6-10, 1996.

266. Graham JM Jr., Zackai EH, Lewis RA, Dobyns WB: Delineation of cerebro-oculo-facial-skeletal (COFS) syndrome. Pediatric Research 39:478, 1996. Presented at the American Pediatric Society and Society for Pediatric Research Annual Meeting, Washington, DC, May 6-10, 1996.

267. Graham JM Jr., Zackai EH, Lewis RA, Dobyns WB: Lumping and splitting within the cerebro-oculo-facial skeletal (COFS) spectrum of disorders. Teratology 53:18, 1996. Presented at the Teratology Society Meeting, Keystone, CO, June 22-27, 1996.

268. Kang S, Graham JM Jr., Abbott M, Schäffer A, Green ED, Rosenberg M, Allen J, Clericuzio C, Grebe T, Haskins-Olney A, Biesecker LG: Autosomal dominant Pallister-Hall syndrome maps to 7p13. Proceedings of the Greenwood Genetics Center, 16:126, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

269. Krantz D, Rand EB, Genin A, Jones M, Bhatt S, Graham JM Jr, Piccoli DA, Spinner NB: Frequency and consequences of 20p12 deletions in Alagille syndrome. Proceedings of the Greenwood Genetics Center, 16:197-198, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

270. Dibbern K, Rabinowitz Y, Graham JM Jr.: Natural history and surgical treatment of cryptophthalmos in Fraser syndrome. Proceedings of the Greenwood Genetics Center, 16:137-138, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

271. Thomas JA, Pepkowitz S, Graham JM Jr.: Microtia, renal agenesis, and DiGeorge sequence: A new syndrome or diabetic embryopathy? Proceedings of the Greenwood Genetics Center, 16:148-149, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

272. Lucas BC, Cooney DF, Graham JM Jr.: Treatment of plagiocephaly-torticollis deformation sequence. Proceedings of the Greenwood Genetics Center, 16:170-171, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

273. Kerner B, Flaum E, Mathews H, Carlson DE, Pepkowitz SH, Hixon H, Graham JM Jr.: Cervical teratoma: Prenatal diagnosis and long term follow-up. Proceedings of the Greenwood Genetics Center, 16:222-223, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

274. Sampson PD, Streissguth AP, Bookstein FL, Little RE, Clarren SK, DeHaene P, Hanson JW, Graham JM Jr: The incidence of fetal alcohol syndrome and the prevalence of alcohol-related neurodevelopmental disorder. Proceedings of the Greenwood Genetics Center,16:242, 1997. Presented at the XVII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead, CA, September 26-October 1, 1996.

275. Kang S, Graham JM Jr, Abbott M, Schaffer A, Green ED, Rosenberg M, Allen J, Cleriuzio C, Grebe T, Haskins-Olney A, Biesecker LG. Autosomal dominant Pallister-Hall syndrome maps to 7p13. American Journal of Human Genetics, 59:A17, 1996. Presented at the American Society of Human Genetics Meeting, San Francisco, CA, October 29-November 2, 1996.

276. Godfrey M, Wang M, Belleh S, Mathews K, Wahl M, Cederbaum SD, Graham JM Jr, and Clericuzio CL. Clinical and molecular comparisons between severe forms of the Marfan syndrome and congenital contractural arachnodactyly (CCA). American Journal of Human Genetics, 59:A26, 1996. Presented at the American Society of Human Genetics Meeting, San Francisco, CA, October 29-November 2, 1996.

277. Kerner B, Hixon H, Crandall B, and Graham JM Jr. Microcephalic lissencephaly, cleft palate, congenital heart defect, and contractures with absence of the corticospinal tract: a new syndrome? American Journal of Human Genetics, 59:A96, 1996. Presented at the American Society of Human Genetics Meeting, San Francisco, CA, October 29-November 2, 1996.

278. Dibbern K and Graham JM Jr. Two patients with FG syndrome and unusual additional physical fingings. Journal of Investigative Medicine, 45:128A, 1997. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 6-8, 1997.

279. Graham J.M. Jr.: Exposure to electromagnetic fields from a 60Hz power line source and occurrence of bilateral nephroblastomatosis and Wilms tumor. Proceedings of the Thirty-Second Annual Meeting, in press, 1997. Presented at 33rd Annual Meeting of National Council on Radiation Protection and Measurements, Arlington VA, April 2-3, 1997.

280. Graham J.M. Jr.: Dysmorphology and Teratology Club Symposium: Human developmental genes and phenotypes. Presented at the Pediatric Academic Societies' 1997 Annual Meeting, Washington DC, May 4, 1997.

281. Graham J.M. Jr.: Comparison of velo-cardio-facial syndrome with CHARGE association. Invited Presentation at the Seventh Biennial Southern African Society of Human Genetics Congress, Pilanesberg National Park, South Africa, May 18-21, 1997.

282. Graham J.M. Jr. and Biesecker L.G.: Autosomal dominant Pallister-Hall syndrome maps to GLI3 on 7p13. Presented at the Seventh Biennial Southern African Society of Human Genetics Congress, Pilanesberg National Park, South Africa, May 18-21, 1997.

283. Graham J.M. Jr. and Wang E.W.: Craniosynostosis syndromes and FGFR3 mutations without problems in long bone development. Presented at the Third International Skeletal Dysplasia Meeting, Marina Del Rey CA, August 7-9, 1997.

284. Graham J.M. Jr. and Wang E.W.: Craniosynostosis syndromes and FGFR3 mutations without problems in long bone development. Proceedings of the Greenwood Genetics Center, 17:70, 1998. Presented at the XVIII David W. Smith Workshop on Malformations and Morphogenesis, Pawleys Island SC, August 13-17, 1997.

285. Krantz I.D., Li L., Collins C.C., Costa T., Pierpont M.E.M., Piccoli D.A., Hood L., Graham J.M. Jr., and Spinner N.B. Alagille syndrome is caused by mutations in human Jagged1, a ligand for Notch1. Proceedings of the Greenwood Genetics Center, 17:130-131, 1998. Presented at the XVIII David W. Smith Workshop on Malformations and Morphogenesis, Pawleys Island SC, August 13-17, 1997.

286. Graham J.M. Jr. and Jabs E.W.: Use of helmets postoperatively in craniosynostosis syndromes. Journal of Craniofacial Genetics and Developmental Biology, 18:8, 1998. Presented at the 1997 Annual Meeting of the Society of Craniofacial Genetics, Baltimore MD, October 28, 1997.

287. Graham J.M. Jr. and Biesecker L.G.: The Inaugural Tony Lipson Memorial Lecture: Autosomal dominant Pallister-Hall syndrome maps to GLI3 on 7p13. Invited presentation at the Fifth International Federation of Teratology Societies Conference, Sydney, N.S.W., Australia, November 16-19, 1997.

288. Graham J.M. Jr.: Comparison of velo-cardio-facial syndrome with CHARGE association. Invited presentation at the Fifth International Federation of Teratology Societies Conference, Sydney, N.S.W., Australia, November 16-19, 1997.

289. Schweitzer D.N., Przylepa K.A., Graham J.M. Jr., Lachman R.S., and Jabs E.W.: Subtle radiographic findings in Crouzon syndrome with acanthosis nigracans. Journal of Investigative Medicine, 46:120A, 1998. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 5-7, 1998.

290. Graham J.M. Jr.: Ears you ought to know. Invited presentation at International Conference on Ear Reconstruction '98: Choices for the Future, Chateau Lake Louise Banff National Park, Alberta, Canada, March 4-6, 1998.

291. Graham J.M. Jr.: Lipomas and spinal lesions in Proteus syndrome. Invited presentation at NIH Proteus Syndrome Workshop. National Human Genome Research Institute. National Institutes of Health, Bethesda MD, March 19-20, 1998.

292. Shin S., Biesecker L.G., Graham J.M. Jr.: GLI3 transcription factor mutations, subcellular localization, and repressor/activator functions correlate with the genesis of three distinct human limb malformation syndromes. Presented at the 6th International Limb Development and Regeneration Conference. Teratology, 57:111, 1998. Sun Valley ID, May 17-21, 1998.

293. Graham J.M. Jr., Greenberg C.R., Busch D.: UV sensitivity in COFS, MICRO, and Cockayne syndromes - a spectrum of disorders. Teratology, 57:196, 1998. Presented at the Teratology Society Meeting, San Diego CA, June 21-25, 1998.

294. Graham J.M. Jr, Superneau D., Rogers R.C., Corning K., Schwartz C.E., Dykens E.M.: Behavioral and personality characteristics in FG syndrome. Proceedings of the Greenwood Genetics Center, 18:152, 1999. Presented at the XIX David W. Smith Workshop on Malformations and Morphogenesis, Whistler B.C., August 6-10, 1998.

295. Shah B., O'Connor M., Cronin P., Graham J.: Psychopathology in children with varying levels of prenatal alcohol exposure. Scientific Proceedings 14:129. Presented at the 45th Annual Meeting of the American Academy of Child and Adolescent Psychiatry, Anaheim CA, October 26-November 1, 1998.

296. Graham J.M. Jr., Lucas B.: Confirmation of Bosma arhinia microphthalmia syndrome. Presented at the 1998 Annual Meeting of the Society of Craniofacial Genetics, Denver CO, October 27, 1998.

297. Graham J.M. Jr., Greenberg C.R., Anyane-Yeboa K., Jaspers N.G.J., Raams, A., Coleman D.M., Ziffer, D.W., Busch D.: COFS syndrome caused by defective nucleotide excision repair. American Journal of Human Genetics, 63:A33, 1998. Presented at the American Society of Human Genetics Meeting, Denver CO, October 27-31, 1998.

298. Lev-Lehman E., Nawaz Z., Benton C., Dicks D., Graham J.M. Jr., Beaudet A.L.: Analysis of mutations causing Angelman syndrome: mRNA stability and UBE3A activity. American Journal of Human Genetics, 63:A332, 1998. Presented at the American Society of Human Genetics Meeting, Denver CO, October 27-31, 1998.

299. Graham J.M. Jr., Greenberg C.R., Anyane-Yeboa K., Jaspers N.G.J., Raams, A., Coleman D.M., Ziffer, D.W., Busch D.: COFS syndrome may be allelic with Cockayne syndrome and both disorders result from defective nucleotide excision repair. Presented at the 8th Manchester Birth Defects Conference, Manchester UK, November 10-13, 1998.

300. Graham J.M. Jr.: Cole-Hughes macrocephaly-mental metardation syndrome. Presented at the 8th Manchester Birth Defects Conference, Manchester UK, November 10-13, 1998.

301. Graham J.M. Jr, Superneau D., Rogers R.C., Corning K., Schwartz C.E., Dykens E.M.: Behavioral and personality characteristics in FG syndrome. Presented at the 5th International Symposium for the Study of Behavioral Phenotypes, Baltimore MD, November 18-21, 1998.

302. Naqvi S.S., Graham J.M. Jr.: Cole-Hughes macrocephaly-mental metardation syndrome. Presented at the 5th International Symposium for the Study of Behavioral Phenotypes, Baltimore MD, November 18-21, 1998.

303. Graham J.M. Jr.: Cole-Hughes macrocephaly-mental metardation syndrome and associated autistic features. Journal of Investigative Medicine 47:55A, 1999. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 27-30, 1999.

304. Graham J.M. Jr.: Cole-Hughes macrocephaly-mental metardation syndrome and associated autistic features. Pediatric Research, 45:74A, 1999. Presented at the Pediatric Academic Societies Annual Meeting, San Francisco CA, May 1-4, 1999.

305. Graham J.M. Jr.: Bosma arhinia microphthalmia syndrome. Pediatric Research, 45:74A, 1999. Presented at the Pediatric Academic Societies Annual Meeting, San Francisco CA, May 1-4, 1999.

306. Graham J.M. Jr.: Bosma arhinia microphthalmia syndrome. Teratology, 59:389, 1999. Presented at the Teratology Society Meeting, Keystone CO, June 28-July 3, 1999.

307. Graham J.M. Jr., Smith A.K., Lachman R.S., Tolo V.T.: Acrodysostosis associated with spinal stenosis. Proceedings of the Greenwood Genetics Center, 19:184, 2000. Presented at the 4th International Skeletal Dysplasia Meeting, Baden-Baden Germany, July 29-August 1, 1999.

308. Salazar D., Graham J.M. Jr., Lachman R.S., Rimoin D.L., Wilcox W.R.: Spondylometaphyseal dysplasia, giant cell type. Proceedings of the Greenwood Genetics Center, 19:188, 2000. Presented at the 4th International Skeletal Dysplasia Meeting, Baden-Baden Germany, July 29-August 1, 1999.

309. Graham J.M. Jr.: Bosma arhinia microphthalmia syndrome. Proceedings of the Greenwood Genetics Center, 19:76, 2000. Presented at the XX David W. Smith Workshop on Malformations and Morphogenesis, Schlangenbad Germany., August 4-8, 1999.

310. Kerner, B., Graham, J.M., Jr., Golden, J.A., Pepkowitz, S.H., Dobyns, W.B.: Familial lissencephaly with cleft palate and severe cerebellar hypoplasia. Proceedings of the Greenwood Genetics Center, 19:68-69, 2000. Presented at the XX David W. Smith Workshop on Malformations and Morphogenesis, Schlangenbad Germany., August 4-8, 1999.

311. Lachman R.S., Graham J.M. Jr., Smith A.K., Tolo V.T.: Acrodysostosis associated with spinal stenosis. Proceedings of the Greenwood Genetics Center, 19:91-92, 2000. Presented at the XX David W. Smith Workshop on Malformations and Morphogenesis, Schlangenbad Germany., August 4-8, 1999.

312. Biesecker L.G., Graham J.M. Jr., Grebe T., Neri G., Pagon R., Bonneman C., Partington M., Kiloran C.: Mutational spectrum of Pallister-Hall syndrome. American Journal of Human Genetics, 65:A284, 1999. Presented at the American Society of Human Genetics Meeting, San Francisco CA, October 19-23, 1999.

313. Ludecke H.-J., Graham J.M. Jr., Lindow S., Horsthemke B.: Is there a "Golderhar " gene in 4p15.3? American Journal of Human Genetics, 65:A229, 1999. Presented at the American Society of Human Genetics Meeting, San Francisco CA, October 19-23, 1999.

314. Everman D.B., Polinkovsky A., Francomano C.A., Graham J.M. Jr., Goodman F.R., Neri G., Morrison S., Robin N.H., Warman M.L.: Additional CDMP1 mutations suggest that brachydactyly type C is locus homogeneous and incompletely penetrant. American Journal of Human Genetics, 65:A294, 1999. Presented at the American Society of Human Genetics Meeting, San Francisco CA, October 19-23, 1999.

315. Graham J.M. Jr., Meira L.B., Greenberg C.R., Jaspers N.G.J., Busch D., Coleman D.M., Ziffer D.W., Friedberg E.C.: Original COFS syndrome Manitoba aboriginal kindred has a mutation in the Cockayne syndrome group B (CSB) gene. American Journal of Human Genetics, 65:A299, 1999. Presented at the American Society of Human Genetics Meeting, San Francisco CA, October 19-23, 1999.

316. Schweitzer D.N., Lachman R.S., Pressman B.D., Graham J.M. Jr.: Further delineation of the Van Den Ende-Gupta syndrome of blepharophimosis, joint contractures, arachnodactyly and normal development. American Journal of Human Genetics, 65:A343, 1999. Presented at the American Society of Human Genetics Meeting, San Francisco CA, October 19-23, 1999.

317. Graham J.M. Jr., Meira L.B., Greenberg C.R., Busch D., Ziffer D.W., Coleman D.M., Friedberg E.C.: Original COFS syndrome kindred from Manitoba has a mutation in the Cockayne syndome group B (CSB) gene. Journal of Investigative Medicine 48:48A, 2000. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 9-12, 2000.

318. Gordon O.K., Earl D.L., Kreutzman J.T., Hwang J.M., Graham J.M. Jr.: Vertex birth molding: longterm residual effects and suggested management of cone-headed neonates. Journal of Investigative Medicine 48:48A, 2000. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 9-12, 2000.

319. Schweitzer D.N., Earl D.L., Graham J.M., Jr.: Marden-Walker syndrome: case report and review. Genetics in Medicine 2:87, 2000. Presented at Presented at the Annual Clinical Genetics Meeting, Palm Springs, CA, March 9-12, 2000.

320. Graham J.M. Jr., Meira L.B., Greenberg C.R., Busch D.B., Doughty A.T.B., Ziffer D.W., Coleman D.M., Savre-Train I., Friedberg E.C.: Original COFS syndrome kindred from Manitoba has a mutation in the Cockayne syndome group B (CSB) gene. Pediatric Research 47:81A, 2000. Presented at the Pediatric Academic Societies Meeting, Boston MA, May 12-16, 2000.

321. Naqvi S.S.A., Graham J.M. Jr.: Autosomal dominant macrocephaly and autism. Presented at the American Psychiatric Association Annual Meeting, Chicago IL, May 13-15, 2000.

322. Graham J.M. Jr., Krakow D., Lachman R.S.: Radiographic findings and Gs-alpha bioactivity studies and mutation screening in acrodysostosis indicate a different etiology from pseudohypoparathyroidism. Developmental Dynamics, 219:444, 2000. Presented at 7th International Conference on Limb Development and Regeneration, Aussois France, May 21-24, 2000.

323. Graham J.M. Jr., Anyane-Yeboa K., Jaspers N.G.J., Coleman D., Ziffer D.W., Busch D.B.: Genotype-phenotype correlations in COFS syndrome. European Journal of Human Genetics 8:51, 2000. Presented at the European Human Human Genetics Conference 2000, Amsterdam, The Netherlands, May 27-30, 2000.

323. Graham, J.M., Jr.: Management of fragile X syndrome. Presented at 7th International Fragile X Conference. Los Angeles CA., July 19-23, 2000 (Invited Talk).

324. Schwartz C.E., May M., Neri G., Gillessen-Kaesbach G., Graham J.M. Jr., Scott C., Stevenson R.E.: The Aaarskog-Scott syndrome most frequently results from mutations giving rise to a truncated protein. Proceedings of the Greenwood Genetics Center, 20:162-163, 2001. Presented at the XXI David W. Smith Workshop on Malformations and Morphogenesis, San Diego CA, August 1-6, 2000.

325. Wang R., Earl D.L., Ruder R.O., Graham J.M. Jr.: Syndromic ear anomalies and renal ultrasounds. Proceedings of the Greenwood Genetics Center, 20:170, 2001. Presented at the XXI David W. Smith Workshop on Malformations and Morphogenesis, San Diego CA, August 1-6, 2000.

326. Graham, J.M., Jr.: Trigonocephaly and the Opitz C syndrome. Proceedings of the Greenwood Genetics Center, 20:68-69, 2001. Presented at the XXI David W. Smith Workshop on Malformations and Morphogenesis, San Diego CA, August 1-6, 2000.

327. Schweitzer D.N., Yano S., Earl D.L., Graham J.M., Jr.: Johnson-McMillin syndrome, a neuroectodermal syndrome with conductive hearing loss and microtia: report of a new case. American Journal of Human Genetics, 67:118, 2000. Presented at the American Society of Human Genetics Meeting, Philadelphia PA, October 2-6, 2000.

328. Graham, J.M., Jr.: Phenotype and dosage effect in polysomy X and polysomy Y (48,XXXY; 48,XXYY; 49,XXXXY): Clinical consequence and deductions. Presented at XXY/Klinefelter syndrome in 2000: expanding the phenotype and identifying new research directions. National Institutes of Health, Bethesda MD, August 28-29, 2000 (Invited Talk).

329. Graham, J.M., Jr., Schweitzer, D.N., Earl, D.L., Lachman, R.S.: Marden-Walker syndrome and Van Den Ende-Gupta syndrome: clinical delineation and review of differentiating features. Presented at the 9th Manchester Birth Defects Conference, Manchester UK, November 7-10, 2000.

330. Wang R., Earl D.L., Ruder R.O., Graham J.M. Jr.: Syndromic ear anomalies and renal ultrasounds. Journal of Investigative Medicine 49(1):52A, 2001. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 7-10, 2001.

331. Elliott, A.M., Graham, J.M., Jr., Curry, C., Pal, T., Rimoim, D.L., Lachmam, R.S. : Dolichospondylic Dysplasia and the 3-M Syndrome: A Spectrum or Distinct Entities? Genetics in Medicine 3:65, 2001. Presented at Presented at the Annual Clinical Genetics Meeting, Miami FL, March 1-4, 2001.

332. Wang, R., Graham, J.M.Jr.: Inhibition of neural crest cell migration by hyperglycemia: a possible mechanism of teratogenesis in infants of diabetic mothers? Teratology 63:286, 2001. Presented at the Teratology Society Meeting, Montreal , Quebec CA June 23-28, 2001.

333. White SM, Goodman F, Graham JM Jr., Everman D., Delatycki MB, Lachman RS, Rimoin DL, Warman ML, Savarirayan R,: Broad phenotypic spectrum caused by an identical heterozygous CDMP1 mutation in three unrelated families. Presented at the 5th International Skeletal Dysplasia Meeting, Oxford, UK, September 6-8, 2001.

334. Wang R., Danielpour M., Gruskin D., Graham J.M., Jr.: Dandy Walker malformation and tethered spinal cord: a new association? Proceedings of the Greenwood Genetics Center, 21:82-83, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

335. Dawson K., Gruskin D., Kreutzmann J., Pepkowitz S.H., Graham J.M., Jr.: A rapidly progressive myopathy which presented as torticollis and tight hips. Proceedings of the Greenwood Genetics Center, 21:132-133, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

336. Wheeler P. Graham J.M., Jr., Cox T., Lin A., Hall B., Schwartz C.: Two brothers with coloboma, distinctive facial features, short stature and mental retardation: CHARGE association, X-linked mental retardation, FG syndrome or unique condition? Proceedings of the Greenwood Genetics Center, 21:140-141, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

337. Visootsak J., Graham J.M., Jr., Hoyme H.E., Szeftel R., Connor M., Schelley S., Schwartz C., Dykens E.: A new family with Borjeson-Forssman-Lehmann syndrome (BFLS): clinical and behavioral features. Proceedings of the Greenwood Genetics Center, 21:141-142, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

338. Graham J.M., Jr., Earl D.L., Danielpour M., Jabs E.W.: A patient with Boston craniosynostosis. Proceedings of the Greenwood Genetics Center, 21:144, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

339. White S.M., Graham, J.M., Jr., Goodman F.R., Everman D., Delatycki M.B., Lachman R.S., Rimoin D.L., Warman M.L.., Savarirayan R.: Broad phenotypic spectrum caused by an identical heterozygous CDMP1 mutation in three unrelated families. Proceedings of the Greenwood Genetics Center, 21:165-166, 2002. Presented at the XXII David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, September 7-11, 2001.

340. Graham J.M., Jr., Wheeler P., Lin A.E., Hall B.D., Cox T.C., Schwartz C.E.: A new X-linked syndrome with similarity to CHARGE association, Toriello-Carey syndrome, and FG syndrome. Genetic Counseling in press, 2001. Presented at the 10th International Workshop on X-linked Mental Retardation. Frascati Italy, September 19-22, 2001.

341. Graham J.M., Jr., Earl D.L., Danielpour M., Jabs E.W.: A patient with Boston craniosynostosis. Presented at the Society for Craniofacial Genetics Meeting, San Diego CA, October 12, 2001.

342. Graham J.M., Jr., Wheeler P., Lin A.E., Hall B.D., Cox T.C., Schwartz C.E.: A new X-linked syndrome with similarity to CHARGE association, Toriello-Carey syndrome, and FG syndrome. American Journal of Human Genetics 69:310, 2001. Presented at the American Society of Human Genetics Meeting, San Diego CA, October 12-16, 2001.

343. Villard L., Nguyen K., Cardoso C., Lese C., Weiss A., Sifry-Platt, Grix A.W., Graham, J.M. Jr., Winter R.M., Leventer R.J., Dobyns W.B.: X-linked bilateral perisylvian polymicrogyria maps to Xq. American Journal of Human Genetics 69:500, 2001. Presented at the American Society of Human Genetics Meeting, San Diego CA, October 12-16, 2001.

344. De Mollerat X.J., Everman D., Clarkson K., Rogers C., Aylsworth A., Graham J.M., Jr., Stevenson R.E., Schwartz C.E.: p63 mutations are not a major cause of non-syndromic SHSF. American Journal of Human Genetics 69:629, 2001. Presented at the American Society of Human Genetics Meeting, San Diego CA, October 12-16, 2001.

345. Wang R, Danielpour M, Gruskin D, Graham JM, Jr.: Dandy Walker malformation and tethered spinal cord: a new association? Presented at the 2001 American Association of Neurologic Surgeons, Section on Pediatric Neurological Surgery Meeting, New York, NY, Nov. 28 - Dec. 1, 2001.

346. Visootsak J., Graham J.M., Jr., Hoyme H.E., Szeftel R., Connor M., Schelley S., Schwartz C., Dykens E.: A new family with Borjeson-Forssman-Lehmann syndrome (BFLS): clinical and behavioral features. Journal of Investigative Medicine 50(1):54A, 2002. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 6-9, 2002.

347. Donahue K., Graham, J.M., Jr.: Deformation of the human infant cranium. Abstract Book, page A99. Presented at American Association for the Advancement of Science Annual Meeting. Boston MA, February 14-19, 2002.

348. Graham, J.M., Jr.: An academic practice model for clinical genetics services. Abstract Book, page 100. Presented at the Annual Clinical Genetics Meeting, New Orleans, LA, March 14-17, 2002.

349. Sanchez P.A., Graham J.M., Jr., Relan A.: Digital dysmorphology cases: development of interactive, on-line genetics cases. Abstract Book, page 84. Presented at the Annual Clinical Genetics Meeting, New Orleans, LA, March 14-17, 2002.

350. Graham J.M., Jr.: Abnormal head shapes. Invited presentation. 10th Annual Conference on Advanced Practice in Neonatal Care, Atlanta Georgia, April 11-12, 2002.

351. Graham J.M., Jr.: Ear anomalies and facial clefting. Invited presentation. 10th Annual Conference on Advanced Practice in Neonatal Care, Atlanta Georgia, April 11-12, 2002.

352. Visootsak J., Dykens E., Schwartz C., Graham J.M., Jr.: Clinical and behavioral features of Borjeson-Forssman-Lehmann Syndrome (BFLS), compared with Prader-Willi Syndrome (PWS) and Klinefelter Syndrome (KS) variants. Pediatric Research, 51:24A, 2002, Presented at the Pediatric Academic Societies Meeting, Baltimore, MD, May 4-7, 2002.

353. Graham J.M., Jr., Gomez M., Earl D., Kreutzman J., Guo X.: Management and outcomes in 308 cases of torticollis-plagiocephaly deformation sequence. Pediatric Research, 51:150A 2002, Presented at the Pediatric Academic Societies Meeting, Baltimore, MD, May 4-7, 2002.

354. Graham J.M., Jr.: Behavioral and clinical features of FG syndrome. Invited presentation. FG Family Support Group Meeting, Salt Lake City UT, June 20-21, 2002.

355. Graham J.M., Jr.: Diagnosis and management of environmentally determined head shape abnormalities. Teratology 65:301, 2002. Presented at the Teratology Society Meeting, Scottsdale AZ, June 22-27, 2002.

356. Visootsak J., Dykens E., Graham J.M., Jr.: Clinical and behavioral features of Klinefelter Syndrome (KS) variants: 48,XXYY compared to 48,XXXY and 49,XXXXY. Presented at the Society for Developmental and Behavioral Pediatrics Annual Scientific Meeting and Workshops, Seattle WA, September 26-30, 2002.

357. De Mollerat X.J., Everman D.B., Morgan C.T., Clarkson K.B., Rogers R.C., Colby R.S., Aylsworth A.S., Graham J.M., Jr., Stevenson R.E., Schwartz C.E.: p63 mutations are not a major cause of non-syndromic split hand/foot malformation. Proceedings of the Greenwood Genetics Center, 22:145, 2003. Presented at the XXIII David W. Smith Workshop on Malformations and Morphogenesis, Greenville SC, August 7-11, 2002.

358. Graham, J.M., Jr., Earl D.L., Krahn K.N., Murray J: Brachydactyly D with cleft lip and scoliosis, a new broadly variable autosomal dominant syndrome. Proceedings of the Greenwood Genetics Center, 22:101-102, 2003. Presented at the XXIII David W. Smith Workshop on Malformations and Morphogenesis, Greenville SC, August 7-11, 2002.

359. Graham J.M., Jr., Visootsak J., Schwartz C., Rosner B., Dykens E.: Behavioral features of Borjeson-Forssman-Lehmann Syndrome (BFLS), compared with Prader-Willi Syndrome (PWS) and Klinefelter Syndrome (KS) variants. Genetic Counseling, 13:in press, 2002. Presented at the XIIIth European Meeting on Dysmorphology, Strasbourg FRANCE, September 4-7, 2002.

360. Graham, J.M., Jr., Earl D.L., Krahn K.N., Murray J: Brachydactyly D with cleft lip and scoliosis, a new broadly variable autosomal dominant syndrome. Genetic Counseling, 13:in press, 2002. Presented at the XIIIth European Meeting on Dysmorphology, Strasbourg FRANCE, September 4-7, 2002.

361. Lalani S.R., Stockton D.W., Bacino C., Molinari L.M., Glass N.L., Fernbach S.D., Towbin J.A., Craigen W.J., Graham, J.M., Jr., Hefner M.A., Lin A.E., McBride K.L.., Davenport S.L., Martin D. Belmont J.W.: Toward a genetic etiology of CHARGE syndrome. American Journal of Human Genetics 71:281, 2002. Presented at the American Society of Human Genetics Meeting, Baltimore MD, October15-19, 2002.

362. Dawson, K.G., Sebald E., Graham J.M., Jr., Krakow D.: Mutation detection in *NOGGIN* in individuals with multiple synostosis syndrome. American Journal of Human Genetics 71:255, 2002. Presented at the American Society of Human Genetics Meeting, Baltimore MD, October15-19, 2002.

363. Graham J.M., Jr., Visootsak J., Rosner B., Dykens E.: Behavioral features of FG Syndrome. Presented at the Society for the Study of Behavioural Phenotypes Meeting, Whistler BC, November 1-4, 2002.

364. Visootsak J., Dykens E., Schwartz C., Rosner B., Graham J.M., Jr.: Behavioral features of Borjeson-Forssman-Lehmann Syndrome (BFLS), compared with Prader-Willi Syndrome (PWS) and Klinefelter Syndrome (KS) variants. Presented at the Society for the Study of Behavioural Phenotypes Meeting, Whistler BC, November 1-4, 2002.

365. Graham J.M., Jr.: How scientific is physical measurement in behavioral phenotypes? A consideration of the proportional effects of extra X chromosomes on dysmorphic features, language and developmental-behavioral outcomes in Klinefelter syndrome variants. Invited Presentation for the Society for the Study of Behavioural Phenotypes Meeting, Whistler BC, November 1-4, 2002.

366. Graham J.M., Jr., Visootsak J., Schwartz C., Rosner B., Dykens E.: Behavioral features of FG Syndrome. Presented at the 10th Manchester Birth Defects Conference, Manchester UK, November 19-22, 2002.

367. Visootsak J., Dykens E., Rosner B., Graham J.M., Jr.: Clinical and behavioral features of Klinefelter syndrome (KS) variants: 48,XXYY compared to 48,XXXY and 49,XXXXY. Journal of Investigative Medicine 51:S150, 2003. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 29-February 1, 2003.

368. Graham J.M., Jr., Visootsak J., Schwartz C., Rosner B., Dykens E.: Behavioral features of FG Syndrome. Journal of Investigative Medicine 51: S149, 2003. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 29-February 1, 2003.

369. Visootsak J., Dykens E., Rosner B., Graham J.M., Jr.: Clinical and behavioral features of Klinefelter syndrome (KS) variants: 48,XXYY compared to 48,XXXY and 49,XXXXY. Pediatric Research 53: 81A. Presented at the Pediatric Academic Societies Meeting, Seattle, WA, May 3-6, 2003.

370. Graham J.M., Jr., Visootsak J., White S.H., Rosner B., Dykens E., Schwartz C.: Clinical and behavioral features of Borjeson-Forssman-Lehmann Syndrome (BFLS) patients with mutations in PHF6. Pediatric Research 53: 81A. Presented at the Pediatric Academic Societies Meeting, Seattle, WA, May 3-6, 2003.

371. Graham, J.M. Jr., Wheeler P., Tackels-Horne D., Lin A.E., Hall B.D., May M., Short K.M., Schwartz C.E., Cox T.C.: A new X-linked syndrome with agenesis of the corpus callosum, mental retardation, coloboma, micrognathia, and a mutation in the Alpha 4 gene at Xq13, Presented at the Festscrift of Dr. Bryan Hall, Lake Arrowhead CA, March 12, 2003.

372. Lawand CMD, Blake KD, Prasad C, Graham JM Jr. The Cranial Nerve Anomalies of CHARGE association/syndrome (A/S). Paediatrics and Child Health. 8 (Supp B abstract 38): 26B, 2003. Presented at the 80th Annual Meeting of the Canadian Paediartic Society, June 2003.

373. Issekutz KA, Smith IM, Prasad C, Graham JM Jr. Blake KD. The Incidence and Prevalence of CHARGE Association/Syndrome in Canada. Paediatrics and Child Health. 8 (Supp B abstract 38): 26B, 2003. Presented at the 80th Annual Meeting of the Canadian Paediartic Society, June 2003.

374. Graham, J.M. Jr., Wheeler P., Tackels-Horne D., Lin A.E., Hall B.D., May M., Short K.M., Schwartz C.E., Cox T.C.: A new X-linked syndrome with agenesis of the corpus callosum, mental retardation, coloboma, micrognathia, and a mutation in the Alpha 4 gene at Xq13. Birth Defects Research: Part A Clinical and Molecular Teratology, 67:321, 2003. Presented at the Teratology Society Meeting, Philadelphia PA, June 21-26, 2003.

375. Graham, J.M., Jr.: CHARGE syndrome: clinical and behavioral features. International CHARGE Syndrome Family Support Group Meeing, Invited Presentation, Cleveland OH, July 24-27, 2003.

376. Graham J.M., Jr., Kreutzman J., Earl D., Halberg A., Samayoa C., Guo X.: Redefinition of current infant head shape parameters and management of positional brachycephaly. Proceedings of the Greenwood Genetics Center, 23:75, 2004. Presented at the XXIV David W. Smith Workshop on Malformations and Morphogenesis, Vancouver BC, August 7-12, 2003.

377. Wang R., Visootsak J., Graham J.M., Jr.: Midline defects in FG syndrome: does tethered spinal cord contribute to the phenotype? Proceedings of the Greenwood Genetics Center, 23:177-178, 2004. Presented at the XXIV David W. Smith Workshop on Malformations and Morphogenesis, Vancouver BC, August 7-12, 2003.

378. Earl D., Kreutzman J., Field F., Krakow D., Wilcox W., Rimoin D., Lachman R., Graham J.M., Jr.: Larsen syndrome clinical and radiographic delineation. Journal of Investigative Medicine 52:S100, 2004. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 28-31, 2004.

379. Chu C., Rabinowitz Y., Bergwerk K., Scambler P., Graham J.M., Jr.: Genotype-phenotype correlations of mutations in *FRAS1* with Fraser syndrome patients. Journal of Investigative Medicine 52:S133, 2004. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 28-31, 2004.

380. Herman K., Honrubia D., Graham J.M., Jr.: Opitz C trigonocephaly syndrome: a report of three affected siblings from one family. Journal of Investigative Medicine 52:S133, 2004. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 28-31, 2004.

381. Graham, J.M., Jr.: Marshall Edwards Invited Lecture: Cranial deformation in human infants. Birth Defects Research: Part A Clinical and Molecular Teratology, in press, 2004. Presented at the Australian Birth Defects Society Annual Meeting. Brisbane, Australia, February 20-21, 2004.

382. Graham, J.M., Jr.: Invited Lecture: New patterns of malformation in diabetic embryopathy. Birth Defects Research: Part A Clinical and Molecular Teratology, in press, 2004. Presented at the Australian Birth Defects Society Annual Meeting. Brisbane, Australia, February 20-21, 2004.

383. Graham, J.M., Jr.: Invited Lecture: Genetic counseling for oculo-auricular-vertebral sequence. Birth Defects Research: Part A Clinical and Molecular Teratology, in press, 2004. Presented at the Australian Birth Defects Society Annual Meeting. Brisbane, Australia, February 20-21, 2004.

384. Visootsak J., Rosner B., Dykens, E., Graham, J.M., Jr.: Behavioral features of CHARGE syndrome. Pediatric Research 55:272A, 2004. Presented at the Pediatric Academic Societies Meeting, San Francisco, CA, May 1-4, 2004.

385. Graham, J.M. Jr., Earl D., Kreutzman J., Lachman R., Rimoin D.L., Cohn D., Krakow D.: Clinical features in Larsen syndrome patients with FLNB mutations. Proceedings of the Greenwood Genetics Center, 24: 94, 2005. Presented at the XXV David W. Smith Workshop on Malformations and Morphogenesis, Wasatch Mountains, Utah, August 18-21, 2004.

386. Krakow D., Graham Jr., J.M., Robertson S.P., Sebald E. T., Morgan T., King L., Earl D., Kreutzman J., Lachman R.L., Rimoin D.L., Cohn D.H.: Mutations in the gene encoding filamin B disrupt vertebral segmentation, joint formation and skeletogenesis. Proceedings of the Greenwood Genetics Center, 24: 93, 2005. Presented at the XXV David W. Smith Workshop on Malformations and Morphogenesis, Wasatch Mountains, Utah, August 18-21, 2004.

387. Graham, J.M. Jr., Lachman R., Krakow D.: Larsen syndrome: suggested diagnostic criteria for *FLNB* mutation analysis. Presented at the 11th Manchester Birth Defects Conference, Manchester England UK, November 9-12, 2004.

388. Aligianis, I., Johnson, C., Gissen, P., Chen, D., Morgan N.V., Hofman, K., Maina, E., Tee, L., Morton, J., Ainsworth, J.R., Stoodley, M., Pilz, D., Rosser, E., Cole T., Stolte-Dijktra, I., Fieggen, K., Clayton-Smith, J., Shields, J., Newbury-Ecob, R., Horn, D., Warburg M., Megabane, A., Dobyns, W., Graham, Jr., J.M., Bond, J., Trembath, R., Harris, L., Takai, Y., Mundlos, S., Tannahill, D., Woods, C.G., Maher E.R.: Molecular genetics of MICRO syndrome and genotype-phenotype correlations. Presented at the 11th Manchester Birth Defects Conference, Manchester England UK, November 9-12, 2004.

389. Conway R., Graham J.M., Jr., Falk R.: The female phenotype of oto-palatal-digital syndrome type 2: a review and report of possible new features. Journal of Investigative Medicine 53:S99, 2005. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 2-5, 2005.

390. Graham, Jr., J.M.: Bosma arhinia microphthalmia syndrome. Presented at Judy Hall's Festscrift at the Western Society for Pediatric Research Meeting, Carmel, CA, February 2-5, 2005.

391. Graham, Jr., J.M., Robertson S.P., Lachman R.S., Krakow D.: Genotype-phenotype correlations in Larsen syndrome. European Journal of Human Genetics 13 (Supplement 1):131, 2005. Presented at European Society for Human Genetics Meeting, Prague CZ, May 7-10, 2005.

392. Casas K.A., Lee J., Hermam K., Graham J.M., Jr., Li, S.: Pathogenesis of ring chromosome 14 syndrome. Presented at American College of Medical Genetics Meeting, Dallas TX , March 17-20, 2005.

393. Visootsak J., Schwenk K, Dykens E., Phelan MC, Graham, J.M., Jr.: Adaptive and maladaptive behavior in 22q13 deletion syndrome compared to 5p- syndrome. Pediatric Research, 57:2301, 2005. Presented at the Pediatric Academic Societies Meeting, Washington, DC, May 14-17, 2005.

394. Graham, Jr. J.M. Symposium: Gene/environment interactions in rare diseases that include common birth defects. Presented at the Teratology Society Meeting, St. Pete Beach, FL, June 25-30, 2005.

395. Graham J.M., Jr. and Visootsak J.: What's new in FG Syndrome? FG Family Support Group Meeting. Boston MA. June 6-9, 2005.

396. Graham, Jr. J.M., Robertson S.P., Kramer N., Lachman R., Rock C., Krakow D: Joint dislocation disorders caused by mutations in *FLNB*. Invited Plenary Session Talk. Presented at Human Genetics Society of Australasia Annual Meeting, Newcastle NSW Australia. July 27-29, 2005.

397. Graham, Jr. J.M.: Differential diagnosis of disorders resulting in microcephaly, cataracts and microcornea. Invited Symposium Talk. Presented at Human Genetics Society of Australasia Annual Meeting, Newcastle NSW Australia. July 27-29, 2005.

398. Graham, Jr. J.M.: Diagnosis and management of CHARGE syndrome. Invited Dysmorphology Club Talk. Presented at Human Genetics Society of Australasia Annual Meeting, Newcastle NSW Australia. July 27-29, 2005.

399. Martinez J.A., Graham, Jr., J.M.: Klippel-Trenaunay syndrome: role of angiogenic factors in vascular malformations and overgrowth syndromes. Proceedings of the Greenwood Genetics Center, 25:104-105, 2006. Presented at the XXVI David W. Smith Workshop on Malformations and Morphogenesis, Iowa City, IA, August 1-5, 2005.

400. Conway R., Danielpour M., Graham, Jr., J.M.: Macrocephaly cutis marmorata telangiectatica congenita: an overgrowth syndrome with cutaneous vascular anomalies. Proceedings of the Greenwood Genetics Center, 25:91, 2006. Presented at the XXVI David W. Smith Workshop on Malformations and Morphogenesis, Iowa City, IA, August 1-5, 2005.

401. Vatanavicharn N., Graham, Jr., J.M.: Microduplication of genes in the PWS/AS region (15q11-13) can cause different clinical phenotypes. Proceedings of the Greenwood Genetics Center, 25:102, 2006. Presented at the XXVI David W. Smith Workshop on Malformations and Morphogenesis, Iowa City, IA, August 1-5, 2005.

402. Adam, M.P., Schelley S., Gallagher R., Brady N., Barr K., Blumberg B., Shieh J.T.C., Graham, Jr., J.M., Hudgins L., Mowat-Wilson syndrome: an under-recognized cause of severely impaired or absent speech. Proceedings of the Greenwood Genetics Center, 25,:84-85, 2006. Presented at the XXVI David W. Smith Workshop on Malformations and Morphogenesis, Iowa City, IA, August 1-5, 2005.

403. Graham, Jr. J.M., Robertson S.P., Kramer N., Lachman R., Krakow D.: Genotype-phenotype correlations in Larsen syndrome. Proceedings of the Greenwood Genetics Center, 25,66, 2006. Presented at the XXVI David W. Smith Workshop on Malformations and Morphogenesis, Iowa City, IA, August 1-5, 2005.

404. Graham, Jr. J.M., Robertson S.P., Kramer N., Lachman R., Rock C., Krakow D.: Genotype-phenotype correlations in Larsen syndrome. Presented at 16th European Meeting on Dysmorphology, Strasbourg France, September 7-10, 2005.

405. Lalani, S.R., Safiullah, A.A., Fernbach S.D., Molinari L.M., Bacino C.A., Davenport S.L., Heffner M.A., Graham, Jr., J.M., Belmont J.W.: Spectrum on CHD7 mutations in 113 individuals with CHARGE syndrome. American Journal of Human Genetics Abstract 10:19, Presented at the American Society of Human Genetics Meeting, Salt Lake City UT, October 25-29, 2005.

406. Lin A., Bird L., Gillessen-Kaessbach J.G., Grossfeld P., Hamilton R., Hicks D., Innes M., Kerr B., Moog U., Rebolledo M. Vaux K., Wieczorek D., Graham, Jr., J.M., Gripp K.: The spectrum of cardiovascular anomalies in Costello syndrome includes arteriopathy. American Journal of Human Genetics Abstract 624:132, Presented at the American Society of Human Genetics Meeting, Salt Lake City UT, October 25-29, 2005.

407. Wang R., Jones J.R., Chen S., Rogers R.C., Friez M.J., Schwartz C.E., Graham Jr., J.M.: Extreme phenotypic variability and a new mutation in HLXB9 in a Currarino Syndrome kindred. American Journal of Human Genetics Abstract 703:145, Presented at the American Society of Human Genetics Meeting, Salt Lake City UT, October 25-29, 2005.

408. Graham, Jr., J.M.: Invited Speaker and Discussant: Descriptive terminology for the periocular region. 1st NIH Consensus Conference on Dysmorphology Nomenclature, Bethesda MD, December 8-10, 2005.

409. Vatanavicharn N., Graham Jr., J. M., Dawson K., Kohlhase J.: Discordant monozygotic twins with Wildervanck syndrome: a proposed mode of inheritance. Journal of Investigative Medicine 54:S100, 2006. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, February 1-4, 2006.

410. Visootsak J., Rosner B., Dykens E, Tartaglia N, Graham J.M., Jr.: Adaptive and Maladaptive Behavior of Males with Sex Chromosome Aneuploidy. Journal of Investigative Medicine 54(1)S280, 2006. Presented at the Southern Society for Pediatric Research Meeting. Atlanta, GA, March 3-5, 2006. *(SSPR Clinical Science Young Investigator Award)*

411. Conway R.L., Zachai E., Hoyme H.E., Milunsky J.M., Shieh J., Butler M.G., Crandall B., Zinn A., Dorosthar P.C., Graham Jr., J.M.: Macrocephaly-cutis marmorata telangiectatica congenita: a review of 13 patients with attention to clinical features and management. European Journal of Human Genetics14 (Supplement 1): 139, 2006. Presented at European Society for Human Genetics Meeting, Amsterdam NH, May 6-9, 2006.

412. Graham, Jr., J.M.: Invited Presentation: Urethral obstruction malformation sequence and other causes for prune belly syndrome. International Prune Belly Syndrome Support Group Meeting, Torrance CA, July 21, 2006.

413. Graham, Jr., J.M.: Invited Presentation: Differential diagnosis for cerebral overgrowth syndromes. International Sotos Syndrome Support Group Meeting, Orange CA, July 22, 2006.

414. Graham, Jr., J.M.: The morphogenesis of wormian bones. Proceedings of the Greenwood Genetics Center, 26:80-81, 2007. Presented at the 27th David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, Sept 8-12, 2006.

415. Conway R.L., Danielpour M., Pressman B., Butler M.G., Zachai E., Close L., Clericuzio C., Graham Jr., J.M.: Longitudinal analysis of neuroimaging abnormalities in macrocephaly-cutis marmorata telangiectatica congenita. Proceedings of the Greenwood Genetics Center, 26:71-72, 2007. Presented at the 27th David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, Sept 8-12, 2006.

416. Pober B.R., Al-Gazi L., Lacombe D., Chassaing N., Bieth E., Donahoe P.K., McPherson E., Graham Jr., J.M., Hill R.S., Walsh C., Kartarci S.: Locus mapping in Donnai-Barrow syndrome: approach to identify a gene important for congenital diaphragmatic hernia. Proceedings of the Greenwood Genetics Center, 26:53, 2007. Presented at the 27th David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, Sept 8-12, 2006.

417. Vatanavicharn N., Wilcox, Jr., W.R., Graham Jr., J.M., Curry C.J., Pepkowitz S., Lachman R.S., Rimoin D.L.: Diaphanospondylodysostosis (DSD): three new cases and similarities of DSD to the Pax1 knockout mouse. Proceedings of the Greenwood Genetics Center, 26:119, 2007. Presented at the 27th David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, Sept 8-12, 2006.

418. Martinez J.A., Graham, Jr., J.M.: Signaling pathways in nail development: brachydactyly with nail aplasia. Proceedings of the Greenwood Genetics Center, 26:100-101, 2007. Presented at the 27th David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead CA, Sept 8-12, 2006.

419. Risheg H., Friez M.J., Graham. Jr. J.M., Moeschler J.B., Rogers R.C., Opitz J.M., Stevenson R.E., Schwartz C.E.: A novel missense mutation, p.R808W, in the HOPA gene is present in 10% of a cohort of FG syndrome families. American Journal of Human Genetics Abstract Book, Presented at the American Society of Human Genetics Meeting, New Orleans LA, October 9-13 2006.

420. Graham, Jr., J.M.: Diabetic embryopathy. UCSD Course in Human Teratology, La Jolla CA, October 28, 2006.

421. Graham, Jr., J.M., Rock C., Robertson S., Krakow D.: Allelic disorders associated with mutations in filamin B (*FLNB*). Presented at the 12th Manchester Birth Defects Conference, Manchester UK, November 21-24, 2006.

422. Risheg H., Friez M.J., Tarpey P., Raymond L., Turner G., Gecz J., Porteous M., Graham, Jr. J.M., Opitz J.M., Rogers R.C., Lubs H.A., Stevenson R.E., Schwartz C.E.: Lessons from Opitz FG syndrome and Lujan syndrome. Presented at the 12th Manchester Birth Defects Conference, Manchester UK, November 21-24, 2006.

423. Borozdin W., Graham, Jr. J.M., Bamshad M.J., Leipolldt J., Kohlhase J.: Characteristics of three overlapping microdeletions including *SALL4* renders 5 neighboring genes responsible for severe developmental delay in a patient with Okihiro syndrome. Presented at the 12th Manchester Birth Defects Conference, Manchester UK, November 21-24, 2006.

424. Graham, Jr. J.M.: Maternal diabetes and/or obesity during pregnancy as risk factors for birth defects during pregnancy. Invited Lecture for Research Institute, Ospedale Bambino Gesu, Rome Italy, November 28, 2006.

425. Graham, Jr., J.M.: CHARGE syndrome management: clinical and behavioral features. Invited Lecture for Research Institute, Ospedale Bambino Gesu, Rome Italy, November 28, 2006.

426. Graham, Jr., J.M.: Invited Speaker and Discussant: Descriptive terminology for the periocular region. NIH Consensus Conference on Dysmorphology Nomenclature, Rome IT, November 28-30, 2006.

427. Ballif BC, Hornor SA, Sulpizio SG, Lloyd RM, Minier SL, Rorem EA, Theisen A, Jenkin E, Madan-Khetarpal S, Surti U, Medne SU, Zackai E, Asamoah A, Farnsworth P, Gowans G, Conway RC, Graham Jr JM, Bejjani BA, Shaffer LG; Discovery of a novel microdeletion syndrome on 16p11.2p12.2 and identification of other clinically relevant pericentromeric imbalances by array CGH. Platform presentation at the American College of Medical Genetics Meeting, Nashville TN, March 21-24, 2007.

428. Conway R.L., Pressman B., Dobyns B., Danielpouir M., Lee, J., Butler M.G., Zachai E., Close L., Saitta S.C., Clericuzio C., Milunsky J., Hoyme G., Shieh J., Moeschler J.B., Crandall B., Lauzon J.L., Graham Jr., J.M.: Neuroimaging findings in macrocephaly-cutis marmorata telangiectatica congenital: a longitudinal study of 15 patients. Invited Talk. Festschrift for M Michael Cohen. Salt Lake City UT, March 31, 2007.

429. Graham, Jr., J.M., Sanchez-Lara, P.A., Lee J., Hing A.V., Cunningham M.: The morphogenesis of wormian bones. Invited Talk. Festschrift for M Michael Cohen. Salt Lake City UT, March 31, 2007.

430. Ballif BC, Hornor SA, Jenkins E, Madan-Khetarpal S, Surti U, Jackson K, Asamoah A, Farnsworth P, Gowans G, Conway RL, Graham JM Jr, Medne L, Zackai E, Shaikh TH, Geoghegan J, Selzer R, Eis P, Bejjani BA, Shaffer LG: Discovery of clinically relevant pericentromeric imbalances using array-based comparative genomic hybridization. MC-GARD Molecular Profiling of the Genome Conference. Amsterdam, Netherlands. May 2-5, 2007.

431. Schwartz C.E., Tarpey P.S., Raymond L., Risheg H., Lubs H.A., Opitz J.M., Clark R.D., May M.M., Briault S., Graham, Jr. J.M., Fryns J.P., Piluso G., Chelly J., Verloes A., Skinner C., Rogers R.C., Moeschler J.B., Joseph S.M., Jones J., Gecz J., Raymond F.L., Stratton M., Friez M.J., Stevenson R.E.: Opitz-Kaveggia (FG) and Lujan syndromes are allelic having mutations in the MED12 gene. European Journal of Human Genetics, 15:Supplement 1, 267. Presented at the European Society of Human Genetics Meeting, Nice France, June 16-19, 2007.

432. Graham, JM Jr., Risheh H., Rogers R.C., Clark R.D., Jones K.L., Moeschler J.B., May M., Joseph S.M., Jones J.R., Schwartz C.E., Friez M.J., Stevenson R.E.: Clinical features in patients Clinical features in patients with Opitz-Kaveggia (FG) syndrome and a recurrent mutation, p.R961W, in the MED12 gene. European Journal of Human Genetics, 15:Supplement 1, 59. Presented at the European Society of Human Genetics Meeting, Nice France, June 16-19, 2007.

433. Lyons, M.J., Clark, R.D., Graham, J.M. Jr., Hunter, A.G.D., Neri, G., Rogers, R.C., Stevenson, R.E.: Clinical experience in the evaluation of 30 patients with a prior diagnosis of FG syndrome. Proceedings of the Greenwood Genetics Center, 27:102-103, 2007. Presented at the 28th David W. Smith Workshop on Malformations and Morphogenesis, Williamsburg VA, August 8-12, 2007.

434. Graham, J.M. Jr., Visootsak, J., Dykens, E., Clark, R.D., Jones, K.L., Moeschler, J.B., Rogers, R.C., Schwartz,, C.E., Friez, M.J., Stevenson, R.E.: Behavioral features in patients with Opitz-Kaveggia (FG) syndrome and a recurrent mutation, p.R961W, in the MED12 gene. Proceedings of the Greenwood Genetics Center, 27:101-102, 2007. Presented at the 28th David W. Smith Workshop on Malformations and Morphogenesis, Williamsburg VA, August 8-12, 2007.

435. Pober, B.R., Al-Gazali, L., Hill, R.S., Donnai, D., Black, G.C.M., Bieth, E., Chassaing, N., Lacombe, D., Devriendt, K., Teebi, A., Lacassie, Y., Graham, J.M. Jr., McPherson, E., Toriello, H., Loscertales, M., Robson, C., MacLaughlin, D.T., Noonan, K.M., Russell, M.K., Walsh, C.A., Donahoe, P.K., and Kantarci, S.: Mutations in megalin cause Donnai-Barrow Facio-Oculo-Acoustico-Renal syndrome. Proceedings of the Greenwood Genetics Center, 27:67-68, 2007. Presented at the 28th David W. Smith Workshop on Malformations and Morphogenesis, Williamsburg VA, August 8-12, 2007.

436. Sanchez-Lara, P.A., Graham, J.M. Jr., Lee, J., Hing, A.V., Cunningham, M.: Wormian bones in non-syndromic craniosynostosis. Proceedings of the Greenwood Genetics Center, 27:128, 2007. Presented at the 28th David W. Smith Workshop on Malformations and Morphogenesis, Williamsburg VA, August 8-12, 2007.

437. Clark, R.D., Graham, J.M. Jr., Stevenson, R.E., Rogers, R.C., Jones, K.L., Moeschler, J.B., Friez, M.J., Schwartz, C.E.: Opitz-Kaveggia (FG) syndrome revisited: the clinical phenotype in 10 affected males with the *MED12* mutation R961W. Proceedings of the Greenwood Genetics Center, 27:100-101, 2007. Presented at the 28th David W. Smith Workshop on Malformations and Morphogenesis, Williamsburg VA, August 8-12, 2007.

438. Graham, J.M. Jr., Clark, R.D., Visootsak, J., Dykens, E., Jones, K.L., Moeschler, J.B., Rogers, R.C., Simenson, R, Schwartz,, C.E., Friez, M.J., Stevenson, R.E.: FG syndrome (Opitz-Kaveggia syndrome): clinical and behavioral phenotype in males with *MED12* mutation, p.R961W. Genetic Counseling, in press, Presented at the 18th European Meeting on Dysmorphology, Strasbourg, FR, September 5-7, 2007.

439. Graham, J.M., Jr.: Invited presentation: Differential diagnosis: generalized overgrowth disorders. 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

440. Graham, J.M., Jr.: Invited presentation: Differential diagnosis: generalized overgrowth disorders. 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

441. Graham, J.M., Jr.: Invited presentation: Dysmorphic features of the skull and face, 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

442. Graham, J.M., Jr.: Invited presentation: Dysmorphic features of the trunk and limbs, 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

443. Graham, J.M., Jr.: Invited presentation: Deformations and deformation patterns, 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

444. Graham, J.M., Jr.: Invited presentation: Disruptions and disruptive patterns, 1st Course in Clinical Dysmorphology, European School of Genetic Medicine, Bologna IT, September 9-12, 2007.

445. Graham, J.M. Jr., Clark, R.D., Visootsak, J., Dykens, E., Jones, K.L., Moeschler, J.B., Rogers, R.C., Simenson, R, Schwartz,, C.E., Friez, M.J., Stevenson, R.E.: FG syndrome (Opitz-Kaveggia syndrome): clinical and behavioral phenotype in males with *MED12* mutation, p.R961W. Genetic Counseling, in press, Presented at the 13th International Meeting on Fragile X and Mental Retardation, Venice, IT, October 3-6, 2007.

446. Graham, J.M., Jr.; Invited Talk: Pierre Robin Sequence. Children's Hospital of Orange County Craniofacial Symposium. Orange CA, October 13, 2007.

447. Graham, J.M. Jr., Visootsak, J., Dykens, E., Clark, R.D., Jones, K.L., Moeschler, J.B., Rogers, R.C., Schwartz,, C.E., Friez, M.J., Stevenson, R.E.: Behavioral features in patients with FG (Opitz-Kaveggia) syndrome and a recurrent mutation, p.R961W, in the *MED12* gene. American Society of Human Genetics Abstract Book p156, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

448. Sanchez-Lara, P.A., Graham, J.M. Jr., Lee, J., Hing, A.V., Cunningham, M.: The morphogenesis of wormian bones: a study of craniosynostosis and purposeful cranial deformation. American Society of Human Genetics Abstract Book p149, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

449. Conway R.L., Pressman B., Dobyns B., Butler M.G., Zachai E., Saitta S.C., Campbell, L., Clericuzio C., Milunsky J., Hoyme G., Shieh J., Moeschler J.B., Crandall B., Lauzon J.L., Visckochil D., Harding B., Graham Jr., J.M.: Neuroimaging findings in macrocephaly-cutis marmorata telangiectatica congenital. American Society of Human Genetics Abstract Book p138, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

450. Carr C.W., Zhang J., Carron J.D., Lachman R.S., Graham, J.M., Jr., Kramer N.A., Abdul-Rahman O.A.: Van Den Ende Gupta syndrome: expansion of the phenotype and confirmation of autosomal recessive inheritance. American Society of Human Genetics Abstract Book p147, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

451. Clark R.D., Graham J.M., Jr., Stevenson R.E., Rogers R.C., Jones K.L., Moeschler J.B. Friez M.J., Schwartz C.E.: Opitz-Kaveggia (FG) syndrome revisited: the clinical phenotype in 10 affected males with *MED12* mutation R961W. American Society of Human Genetics Abstract Book p158, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

452. Bernstein J.A., Alkuraya, F.S., Armstrong L., Chen K.C., Clericuzio C., Graham J.M., Jr., Stoler J., Saal H.M., Stevens C.A., Cherry A.M., Hoyme H.E.: Duplication 22q11.2: clinically heterogeneous new syndrome or genetic polymorphism? American Society of Human Genetics Abstract Book p170, Presented at the American Society of Human Genetics Meeting, San Diego CA, October 23-27, 2007.

453. Graham J.M., Jr. Invited Lecture: Craniofacial malformations, deformations disruptions and dysplasias. Indian Academy of Pediatrics Course on Genetics for the Practicing Pediatrician. New Delhi, India. December 16-16, 2007.

454. Graham J.M., Jr. Invited Lecture: Common malformation syndromes in pediatric practice. Indian Academy of Pediatrics Course on Genetics for the Practicing Pediatrician. New Delhi, India. December 16-16, 2007.

455. Graham J.M., Jr. Invited Lecture: Congenital overgrowth syndromes. Indian Academy of Pediatrics Course on Genetics for the Practicing Pediatrician. New Delhi, India. December 16-16, 2007.

456. Graham J.M., Jr., Visootsak J., Dykens E., Clark R.D., Jones, K.L., Moeschler J.B., Rogers R.C., Schwartz C.E.,Friez M.J., Stevenson R.E.: Clinical and behavioral features in patients with FG (Opitz-Kaveggia) syndrome and a recurrent mutation, p.R961W, in the MED12 gene. American College of Medical Genetics Annual Meeting, Phoenix AZ, March 12-16, 2008.

457. Graham J.M., Jr. Invited Lecture: Larsen Syndrome. Second European Course in Clinical Dysmorphology. Rome, Italy. March 28-29, 2008.

458. Adam, M.P., Hudgins L., Carey, J.C., Hall, B.D., Coleman K., Gripp K.W., Perez-Aytes A., Graham, Jr., J.M.: Invited Presentation. Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Lewis B. Holmes Festschrift, Boston MA, May 10, 2008.

459. Graham J.M., Jr. Course Organizer, Invited Presentation: Dysmorphic features of the face and skull. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

460. Graham J.M., Jr. Course Organizer, Invited Presentation: Teratogenic effects of maternal diabetes and/or obesity. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

461. Graham J.M., Jr. Course Organizer, Invited Presentation: Fetal alcohol syndrome. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

462. Graham J.M., Jr. Course Organizer, Invited Presentation: Effects of antidepressant drugs and cigarettes during pregnancy. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

463. Graham J.M., Jr. Course Organizer, Invited Presentation: Disruptions and disruptive effects. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

464. Graham J.M., Jr. Course Organizer, Invited Presentation: Teratogenicity of retinoids and immunosuppressants. Second European School of Medical Genetics Course in Clinical Dysmorphology. Bertinoro, Italy May 12-15, 2008.

465. Graham J.M., Jr. Invited Presentation: Teratogenic effects of maternal diabetes and/or obesity. Perinatal Advisory Committee of Los Angeles County (PAC-LAC) Annual Conference. Los Angeles CA, May 29, 2008.

466. Graham J.M., Jr. Invited Presentation: Review of array comparative genomic hybridization in the evaluation of autism. Signature Scientific Microarray Conference. Spokane WA, June 20-21, 2008.

467. Graham J.M., Jr., Kramer, N., Bejjani, B., Thiel, C.T., Carta, C., Neri, G,Tartagli,a M., Zenker, M: Duplication of *PTPN11* in a boy with Noonan syndrome. Signature Scientific Microarray Conference. Spokane WA, June 20-21, 2008.

468. Graham, Jr., J.M., Adam, M.P., Hudgins L., Carey, J.C., Hall, B.D., Coleman K., Gripp K.W., Perez-Aytes A.: Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Presented at the Teratology Society Meeting, Monterey CA, June 29-July 2, 2008.

469. Sanchez-Lara, PA, Carmichael, S., Graham, jM. Jr. Lammer, E, Shaw, G, Rasmussen, S.A. and the National Birth Defects Prevention Study: Fetal constraint as a potential risk factor for craniosynostosis: Presented at the Teratology Society Meeting, Monterey CA, June 29-July 2, 2008.

470. Graham J.M., Jr., Kramer, N., Bejjani, B., Thiel, C.T., Carta, C., Neri, G, Tartaglia M., Zenker, M: Duplication of *PTPN11* in a boy with Noonan syndrome. Presented at the 29th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblanc, Quebec, Canada, August 8-12, 2008.

471. Adam, M.P., Hudgins L., Carey, J.C., Hall, B.D., Coleman K., Gripp K.W., Perez-Aytes A., Graham, Jr., J.M.: Preaxial hallucal polydactyly as a marker for diabetic embryopathy. Presented at the 29th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblanc, Quebec, Canada, August 8-12, 2008.

472. Sanchez-Lara, PA, Carmichael, S., Graham, jM. Jr. Lammer, E, Shaw, G, Rasmussen, S.A. and the National Birth Defects Prevention Study: Fetal constraint as a potential risk factor for craniosynostosis. Presented at the 29th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblanc, Quebec, Canada, August 8-12, 2008.

473. Spencer, A., Lara- Sanchez, P. Dobyns, W., Golden, J., Schwartz, C., Bannykh, S, Krakow, D. Graham, J.M., Jr.: An unusual case of lissencephaly with distinctive neuropathology: a new syndrome compared with other known lissencephaly syndromes. Presented at the 29th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblanc, Quebec, Canada, August 8-12, 2008.

474. Graham J.M., Jr., Kramer, N., Bejjani, B., Thiel, C.T., Carta, C., Neri, G, Tartaglia M., Zenker, M: Duplication of *PTPN11* in a boy with Noonan syndrome. Presented at the 13th Manchester Dysmorphology Conference, Manchester England, October 28-61, 2008.

475. Sanchez-Lara, PA, Carmichael, S., Graham, jM. Jr. Lammer, E, Shaw, G, Rasmussen, S.A. and the National Birth Defects Prevention Study: Fetal constraint as a potential risk factor for craniosynostosis. Presented at the American Society of Human Genetics Meeting, Philadelphia, PA, November 11-15, 2008.

476. Graham J.M., Jr. Invited Presenation: Teratogenic impact of maternal gestational diabetes. 15th Annual Conference: California Association of Neonatologists: Current Topics and Controversies in Perinatal and Neonatal Medicine. Coronado CA, March 6-8, 2009.

477. Spencer A, Pariani M, Graham J.M., Jr., Rimoin D: Deletion of FOXP1 is associated with speech delay, contractures, hypertonia and blepharophimosis. Presented at the American College of Medical Genetics Meeting. Abstract 232; page 155. Tampa FL, March 25-28, 2009.

478. Graham J.M., Jr. Invited Presentation: Uterine and Placental Factors. Massachusetts General Hospital Postgraduate Course in Human Teratogens. Boston MA, April 26-28, 2009.

479. Graham, J.M., Jr.: Invited Presentation: Medical genetics evaluation in children with disabilities: why does it matter? Spring Medical Surgical / Behavioral Science Conference, Bad Kissingen, Germany, May 17-21, 2009.

480. Graham, J.M., Jr.: Invited Presentation: Diagnosis-based management of children (and families) with genetic syndromes. Spring Medical Surgical / Behavioral Science Conference, Bad Kissingen, Germany, May 17-21, 2009.

481. Graham J.M., Jr., Merrill A., Krakow D.: Is use of the term autosomal recessive Larsen syndrome justified for autosomal recessive sulfation disorders? Presented at the 30th David W. Smith Workshop on Malformations and Morphogenesis, Children's Hospital of Philadelphia, Philadelphia PA, August 5-9, 2009.

482. Mencias I., Kohlhase J., Borozdin W., Graham J.M., Jr.: Wildervanck syndrome: an asymmetrical phenotype with discordant expression in monozygous twins. Presented at the 30th David W. Smith Workshop on Malformations and Morphogenesis, Children's Hospital of Philadelphia, Philadelphia PA, August 5-9, 2009.

483. Spencer A., Grinberg I., van Ruissen F., Namavar Y., Baas F., Plat L., Dobyns W., Graham J.M., Jr.: Pontocerebellar hypoplasia type II in twins caused by a homozygous mutayion in TSEN54: Is prenatal diagnosis possible? Presented at the 30th David W. Smith Workshop on Malformations and Morphogenesis, Children's Hospital of Philadelphia, Philadelphia PA, August 5-9, 2009.

484. Clark, R.D., Graham, Jr., J.M., Friez M.J., Hoo, J.J., Jones, K.L., McKeown C., Moeschler, J.B., Raymond F.L., Rogers, R.C., Schwartz, C.E., Battaglia A., Lyons M.J., Stevenson, R.E.: The clinical phenotype of FG (Opitz-Kaveggia): an algorithm for diagnostic testing. Presented at the British Society of Human Genetics Conference, University of Warwick, Coventry UK, August 31-September 2, 2009.

485. Graham, J.M., Jr.: Invited Presentation: Common syndromic prenatal-onset growth disorders. Landstuhl Regional Medical Center Grand Rounds, Heidelberg, Germany, September 1, 2009.

486. Graham J.M., Jr., Spencer A., Grinberg I., Platt L., Maya M., van Ruissen F., Namavar Y., Baas F., Platt L., Dobyns W.: Molecular and neuroimaging findings in pontocerebellar hypoplasia type II. Platform presention at the 20th European Meeting on Dysmorphology, Strasbourg France, September 4-5, 2009.

487. Clark, R.D., Graham, Jr., J.M., Friez M.J., Hoo, J.J., Jones, K.L., McKeown C., Moeschler, J.B., Raymond F.L., Rogers, R.C., Schwartz, C.E., Battaglia A., Lyons M.J., Stevenson, R.E.: The clinical phenotype of FG syndrome: an algorithm for diagnostic testing. Platform presentation at the American Society of Human Genetics Meeting, Abstract 228, page 87, Honolulu, HI, October 20-24, 2009.

488. Burkardt D., Rosenfeld J., Angle B., Banks V., Gripp K.W., Helgeson M., Kramer N., Moline J., Moran R., Niyazov D.M., Smith W., Stevens C., Zackai E., Lachman R.S., Graham, J.M. Jr.: Patients with deletion 1q24-q25 have a recognizable syndrome. Platform presention at the American College of Medical Genetics Meeting, Albuquerque NM, March 24-28, 2010.

489. Burkardt D., Rosenfeld J., Angle B., Banks V., Gripp K.W., Helgeson M., Kramer N., Moline J., Moran R., Niyazov D.M., Smith W., Stevens C., Zackai E., Lachman R.S., Graham, J.M. Jr.: Distinctive phenotype in 8 patients with deletion of chromosome 1q24-q25. Platform presention at the Pediatric Academic Societies Meeting, Vancouver BC, May 1-4, 2010.

490. Sun A, Petrin AL, May M, Chaubrey A, Murray JC, Smith RJH, Schwartz CE, Kramer N, Graham, Jr JM: A new gene for Branchio-Oto-Renal syndrome in an extended pedigree: AHI1. Presented at the 31th David W. Smith Workshop on Malformations and Morphogenesis, Alderbrrook Resort, Union WA, August 27-September 1, 2010.

491. Graham, J.M. Jr., Burkardt D., Rosenfeld J., Helgeson M., Angle B., Banks V., Smith W., Gripp K.W., Moline J., Moran R., Niyazov D.M., Stevens C., Zackai E., Lebel R.R., Ashley D., Kramer N., Lachman R.S.: Distinctive phenotype in 9 patients with deletion of chromosome 1q24-q25. Presented at the 31th David W. Smith Workshop on Malformations and Morphogenesis, Alderbrrook Resort, Union WA, August 27-September 1, 2010.

492. Graham, J.M. Jr.' Burkardt D., Rosenfeld J., Helgeson M., Angle B., Banks V., Smith W., Gripp K.W., Moline J., Moran R., Niyazov D.M., Stevens C., Zackai E., Lebel R.R., Ashley D., Kramer N., Lachman R.S.: Distinctive phenotype in 9 patients with deletion of chromosome 1q24-q25. Presented at the 14th Manchester Dysmorphology Conference, Manchester England, October 11-14, 2010.

493. Noh GY, Graham, J.M. Jr.: Elucidating the complexity of epilepsy: clues from 2q23.1 deletion. Journal of Investigative Medicine 59;111, 2011. Presented at the Western Society for Pediatric Research Meeting, Carmel, CA, January 26-29, 2011.

494. Graham, J.M. Jr.: Invited presentation: Common limb malformation syndromes, First Indo-US Symposium on Skeletal Dysplasia, Lucknow, India, Febuary 12-13, 2011.

495. Graham, J.M. Jr.: Invited presentation: Overgrowth syndromes, First Indo-US Symposium on Skeletal Dysplasia, Lucknow, India, Febuary 12-13, 2011.

496. Graham, J.M. Jr.: Cardinal features and characteristicbehaviors in FG syndrome. Invited Presentation, ACMG Program Guide and Abstracts p.277, Presented at the American College of Medical Genetics Annual Meeting, Vancouver BC, March 16-20, 2011.

497. Graham, J.M. Jr., Kramer N., Funari V., Klein O., Seidel K., Kantaputra P., Taylor K.D.: Autosomal dominant natal teeth with selective tooth agenesis. ACMG Program Guide and Abstracts p.228, Presented at the American College of Medical Genetics Annual Meeting, Vancouver BC, March 16-20, 2011.

498. Noh GY, Graham, J.M. Jr.: Elucidating the complexity of epilepsy: clues from 2q23.1 deletion. ACMG Program Guide and Abstracts p.197, Presented at the American College of Medical Genetics Annual Meeting, Vancouver BC, March 16-20, 2011.

499. Graham, J.M. Jr., Kramer N., Funari V., Klein O., Seidel K., Kantaputra P., Taylor K.D.: Autosomal dominant natal teeth with selective tooth agenesis. Presented at European Society for Human Genetics Annual Meeting, Amsterdam, Netherlands May 28-31, 2011.

500. Klaassens M., Reinstein E., Hilhorst-Hofstee Y., Schrander J.J.P., Malfait F., Staal H., Speth L., ten Have L.C., Blaauw J., Roggeveen H.C.J., De Paepe A., van Steensel M.A.M., Pals G., Graham, Jr., J.M., Schrander-Stumpel C.T.R.M.: Ehlers-Danlos syndrome arthrochalasia type (VIIA-B) – expanding the phenotype: from prenatal life through adulthood. Presented at European Society for Human Genetics Annual Meeting, Amsterdam, Netherlands May 28-31, 2011.

501. Graham, J.M. Jr., Kramer N., Funari V., Klein O., Seidel K., Kantaputra P., Taylor K.D.: Autosomal dominant natal teeth with selective tooth agenesis. Presented at the 32nd David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead UCLA Conference Center, Lake Arrowhead CA, September 9-14, 2011.

502. Dobyns W.B., Mirzaa G.M., Graham, J.M. Jr.: Insights on overgrowth from the macrocephaly-capillary malformation syndrome. Presented at the 32nd David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead UCLA Conference Center, Lake Arrowhead CA, September 9-14, 2011.

503. Sun A., Taylor K., Kramer N., Graham, J. M., Jr.: Novel locus identified for Branchio-Oto-Renal syndrome. Presented at the 32nd David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead UCLA Conference Center, Lake Arrowhead CA, September 9-14, 2011.

504. Reinstein E., Graham J.M. Jr., Rimoin D.L.: Ehlers-Danlos syndrome with periventricular nodular heterotopia (EDS-PNH): expanding the phenotype. Presented at the 32nd David W. Smith Workshop on Malformations and Morphogenesis, Lake Arrowhead UCLA Conference Center, Lake Arrowhead CA, September 9-14, 2011.

505. Graham, J.M. Jr., Kramer N., Funari V., Klein O., Seidel K., Kantaputra P., Taylor K.D.: Autosomal dominant natal teeth with selective tooth agenesis. Presented at 34th Annual Society of Craniofacial Genetics and Developmental Biology Meeting, Montreal, October 11, 2011, American Journal of Medical Genetics, Part A, p4, 2012.

506. Graham, J.M. Jr., Kramer N., Funari V., Klein O., Seidel K., Kantaputra P., Taylor K.D.: Autosomal dominant natal teeth with selective tooth agenesis. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

507. Sun A., Taylor K., Kramer N., Graham, J.M. Jr.: Novel locus identified for Branchio-Oto-Renal syndrome. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

508. Lee H., Graham J.M. Jr., Rimoin D.L., Lachman R.S., Nelson S.F., Krakow D., Cohn D.H.: Acrodysostosis: exome sequencing identifies mutations in PDE4D encoding phosphodiesterase 4D. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

509. Stevens C.G., Yagnik G. Qi L. Cherkez E., Sanchez-Lara P.A., Kimonis V., Stoler J., Cunningham M., Graham J.M. Jr., Boyadiev S.A.: Clinical and epidemiolocal analysis of nonsyndromic craniosynostosis. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

510. Noh GY, Graham, J.M. Jr.: Elucidating the complexity of epilepsy: clues from 2q23.1 deletion. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

511. Probst F.J., Corrigan R.R., Zabriskie R.C., Murdock D.R., Hamid R., Tiller G.E., Phillips J.A., Kramer N., Graham J.M. Jr., Bainbridge M.N., jin W., Wang L.L., Gibbs R.A., Plon S.E.: Linkage analysis and whole-exome sequencing on families with multiple lipomatosis. Presented at 12th International Congress of Human Genetics, Montreal, October 11-15, 2011.

512. O'Leary R., Shih J.C., Hyland K., Kawamata N., Tavyev-Asher Y. J., Graham J. M., Jr.: De novo microdeletion of Xp11.3 targeting the monoamine oxidase A and B genes in a male infant with episodic hypotonia: a genomics approach to personalized medicine. Presented at Western Society for Pediatric Research, Carmel CA, January 26-28, 2012 (Winner WSPR Genetics Subspecialty Award).

513. Kramer N.A., Falk R.E., Graham J.M., Jr.: Evidence of a population specific mutation in *USH1G* that leads to Usher syndrome in the Filipino population. Presented at American College of Medical Genetics Annual Meeting, Charlotte NC, March 27-31, 2012.

514. Graham J.M., Jr., O'Leary R., Shih J.C., Hyland K., Kawamata N., Tavyev-Asher Y. J.: De novo microdeletion of Xp11.3 targeting the monoamine oxidase A and B genes in a male infant with episodic hypotonia. Presented at the 33rd David W. Smith Workshop on Malformations and Morphogenesis, Legacy Lodge Conference Center, Atlanta GA, August 8-12, 2012.

515. Carter M.T., Mirzaa G.M., McDonell L.M., Clericuzio C., Aesadi G., Graham, J.M., Jr., Dobyns W.B., Boycott K.M.: Microcephaly-Capillary Malformation Syndrome (MIC-CAP): further clinical delineation of four new patients. Presented at the 33rd David W. Smith Workshop on Malformations and Morphogenesis, Legacy Lodge Conference Center, Atlanta GA, August 8-12, 2012.

516. Graham J.M., Jr.: Invited Talk: Genetics of neonatal seizures and early infantile epileptic encephalopathy. Presented at Indo-US Symposium on Disorders in the developing Brain. October 27-28, 2012, Kasturba Medical College, Manipal India.

517. Graham J.M., Jr.: Evaluation of epilepsy syndromes in Genetics clinic. Presented at Indo-US Symposium on Disorders in the developing Brain. October 27-28, 2012, Kasturba Medical College, Manipal India.

518. Basel-Vanagaite L., Kasrlinsky L., Wolf L., Shohat M., Skinner C., Rogers C., Stevenson R., Schwartz C.E., Graham J.M., Jr.,: Computer-aided facial recognition of individuals with FG (Opitz-Kaveggia) syndrome caused by p.Arg961Trp mutation in *MED12*, Presented at American Society of Human Genetics Meeting, November 6-10, 2012.

519. Russell B, Nasiak M, Kramer N, Johnston JJ, Biesecker LG, Graham JM Jr.: Diagnosis and management of Bohring-Opitz Syndrome caused by *de novo ASXL1* mutations. Presented at Western Society for Pediatric Research, Carmel CA, January 24-26, 2013.

520. Bale S., Graham Jr., J.M., Cohen J., Kramer N.: Laboratory and Clinical Perspectives on the Technology and Applications of Whole Exome Sequencing. National Society of Genetic Counselors Webinar, March 28, 2013.

521. Graham Jr. JM, Russell B, Johnston JJ, Biesecker LG: Diagnosis and management of Bohring-Opitz Syndrome with or without *ASXL1* mutations. Presented at the European Human Conference, Paris FR, June 8-11, 2013.

522. Graham Jr, JM. Pediatric issues in Marfan syndrome. National Marfan Foundation Annual Family Conference, Los Angeles CA, August 3, 2013.

523. Mirzaa G.H., Adams C, Kramer N, Conway R.L., Graham, Jr. J.M., Dobyns W.B.: Molecular insights into mosaic megalencephaly disorders. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

524. Graham, Jr., J.M., Russell B., Kramer N., Johnston J.J., Biesecker L.G.: Diagnosis and management of Bohring-Opitz syndrome. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

525. Hunter AGW, Graham, Jr., JM, Neri G, Rogers RC, Stevenson RE, Turner G, Friez MJ: The Intellectual Disabilities Evaluation and Advice System (IDEAS): outcome of the first 55 cases. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

526. Castillo A., Kramer N., Lausch E., Zachai, EH, Hakonsrson H, Saita S, Graham Jr. J.M.: Infantile systemic hyalinosis versus infantile myofibromatosis. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

527. Mirzaa GM, Adams C, Kramer N, Conway R, Graham, Jr. JM, Dobyns WB: Molecular insights into mosaic megalencephaly disorders. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

528. McDonnell LM, Mirzza GH, Alcantara D, Carter Melissa T, Graham, J. JM, Dobyns WB, O'Driscoll M, Boycott KM: Mutations in *STAMBB* cause microcephaly capillary malformation syndrome. Presented at the 34th David W. Smith Workshop on Malformations and Morphogenesis, Mont Tremblant, Quebec, CA, August 9-14, 2013.

529. McMillan MJ, Beck AE, Chong JX, Shively KM, Buckingham KJ, Gildersleeve HJ, Splitt M, Aylesworth AS, Krapels IPC, Curry CJ, Alvarez MIA, Hecht JT, Hurst J, Scott R, Graham, Jr JM, Smith JD, Tabor HK, Shendure J, Nickerson DA, Banshad MJ: Mutations in *PIEZO2* cause Gordon syndrome, Marden Walker syndrome and distal arthrogryposis type 5. Presented at the American Society of Human Genetics Meeting, Boston MA, October 22-26, 2013.

530. Dhamija R, Graham, Jr, JM, Thorland E, Kirmani S: Novel *denovo SPOCK1* mutation in a proband with developmental delay, microcephaly and agenesis of corpus callosum. Presented at the American Society of Human Genetics Meeting, Boston MA, October 22-26, 2013.

531. Graham, Jr. JM: Lessons from the exome. Pediatric Grand Rounds, Dartmouth-Hitchock Medical Center, Lebanon NH, October 2, 2013.

532. Shih EM, Graham, Jr. JM, Vitazka P, Pitukcheewanont P: Duplication of 17p13.3 involving *SERPINF1* associated with an unclassified type of metaphyseal dysplasia. Presented at Western Society for Pediatric Research, Carmel CA, January 24, 2014, Journal of Investigative Medicine, 62:232, 2014.

533. Graham, Jr. JM: Lessons from the exome. Pediatric Grand Rounds, Harbor-UCLA Medical Center, Torrance CA, February 13, 2014.

534. Graham Jr. JM: The many faces of hemimegalencephaly. Invited Presentation. Scientific Workshop on Brain Plasticity, Hemisheric Specialization, and Neurorehabilitation After Cerebral Hemisherectomy, Paradise Pier Hotel, Anaheim CA, July 9-12, 2014.

535. Graham Jr., JM, McMillin MJ, Chong J, Beck A, Bamshad M: Long-term follow-up of a patient with Msarden-Walker syndrome and a c.8056C>T *PIEZO2* mutation and comparison with Gordon syndrome and c.8057G>A mutations in *PIEZO2*. Presented at the 35th David W. Smith Workshop on Malformations and Morphogenesis, Madison WI, July 25-30, 2014.

536. Tenney J, Graham Jr, JM, Dobyns WB, Gleeson JG: A male with preaxial polydactyly, Joubert syndrome and *OFD1* mutation discovered by exome sequencing. Presented at the 35th David W. Smith Workshop on Malformations and Morphogenesis, Madison WI, July 25-30, 2014.

537. Babkina N, Giurgea I, Mowat D, Graham Jr. JM: Early infantile epileptic encephalopathy with a de novo variant in *ZEB2* discovered by exome sequencing. Presented at the 35th David W. Smith Workshop on Malformations and Morphogenesis, Madison WI, July 25-30, 2014.

538. Russell B, Johnston JJ, Biesecker LG, Kramer N, Pickart A, Rhead W, Tan W-H, Brownstein CA, Clarkson LK, Dobson A, Rosenberg AZ, Graham Jr., JM: Clinical management of patients with *ASXL1* mutations and Bohring-Opitz Syndrome, emphasizing the need for Wilms tumor surveillance. Presented at the 64th Annual American Society for Human Genetics Meeting, October 18-22, 2014, San Diego CA.

539. Graham Jr, JM: Everything you needed to know about plagiocephaly and craniosynostosis but were afraid to ask. (Invited Presentation). AAP Chapter 2 Town Hall Meeting, October 22, 2014, Woodland Hills CA.

540. Graham Jr, JM: Congenital overgrowth syndromes. (Invited Presentation). Presented at the INDO-US Symposium on Genomic Insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology & First Annual Meeting of the Indian Academy of Medical Genetics, November 7-9, 2014, Hyderabad, India.

541. Graham Jr, JM: MED-12 Related Disorders. (Invited Presentation). Presented at the INDO-US Symposium on Genomic Insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology & First Annual Meeting of the Indian Academy of Medical Genetics, November 7-9, 2014, Hyderabad, India.

542. Graham Jr., JM: Syndromes of Primordial Short Stature. (Invited Presentation). Presented at the INDO-US Symposium on Genomic Insights into Human Morphogenesis: Prenatal, Postnatal and Molecular Dysmorphology & First Annual Meeting of the Indian Academy of Medical Genetics, November 7-9, 2014, Hyderabad, India.

543. Graham Jr, JM: Infant head shape abnormalities (2015). (Invited Presentation). CSMC Pediatric Grand Rounds, January 15, 2015, Los Angeles, CA.

544. Graham JM, Jr, Zadeh N, Sagi-Dain L, Shalev SA: *KCNK9* imprinting syndrome – a treatable disorder? Presented at the 36th David W. Smith Workshop on Malformations and Morphogenesis, St Michaels MD, August 14-19, 2015.

545. Pierson TM, Rajaraman R, Delgado M, Srour M, Graham JM Jr, Venkateswaran S: GATAD2B-associated neurodevelopmental disorder (GAND); clinical and molecular insights. Presented at the 4th Annual Child Neurology Society Meeting, Washington DC, October 7-10, 2015.