

**Medical University of South Carolina
Curriculum Vitae**

Name: G. Shashidhar Pai

Office address:

Department of Pediatrics
Medical University of South Carolina
Charleston, SC, 29425

Education:

Years	Location	Degree
1963-64	Sri Bhuvanendra College, Karkal, India	None
1964-70	Mysore Medical College, Mysore, India	M.B.;B.S.

Internship:

1970-71	Krishnarajendra Hospitals, Mysore India
1971-72	Kasturba Medical College, Manipal, India
1972-73	Intern, Memorial Hospital, Worcester, MA

Residency and Fellowship:

1973-75	Resident-Pediatrics, Henry Ford Hospital, Detroit
1975-76	Chief Resident, Henry Ford Hospital, Detroit
1977-79	Post-doctoral Fellow in Pediatrics (Genetics), Johns Hopkins University

Board Certification:

1977	Certified, American Board of Pediatrics
1982	Certified, American Board of Medical Genetics (Clinical Genetics and Cytogenetics)

Faculty Appointments:

1979-84	Medical University of SC Assistant Professor
1984-94	Associate Professor
1994-	Professor
1999-	Professor of Obstetrics and Gynecology, University of SC School of Medicine

Administrative Appointments:

1979-1994	Director, Division of Genetics, Department of Pediatrics
1979-84	Scientific Director, Cytogenetics Laboratory, Department of Pathology
7/90-10/90	Acting Director. Cytogenetic Laboratory, Department of Pathology and Laboratory Medicine
5/1/1994-	Director, Division of Genetics and Child Development, Department of Pediatrics
7/97 –10/98	Acting Director, Cytogenetics Laboratory
1995 –	Medical Director, Masters Program in Genetic Counseling, USC School of Medicine

Hospital Appointments:

1979	Active	Medical University Hospital
1980	Consulting	Roper Hospital
1980	Consulting	Bon Secours -St. Francis Xavier Hospital

Awards/Honors:

1986-87	Excellence in Teaching Award
1987-88	Excellence in Teaching Award
	Nominated for Golden Apple (Teaching) Award
1996-97	Faculty Excellence Award - College of Medicine Student Council

Membership in Professional Societies:

National and International Societies

- American Society of Human Genetics
- Southern Society for Pediatric Research
- American Medical Association
- Indian Society of Human Genetics
- Founding Member, AAP Section on Genetics
- Founding Fellow, American College of Medical Genetics
- Association of Professors of Human and Medical Genetics

Local Societies

- Charleston County Medical Society
- South Carolina Medical Association

Service on National and Regional Boards, Panels and Committees:

Grant reviewer for National Science Foundation	1981
Member, Scientific Discipline Committee on Cytogenetics, Pediatric Oncology Group	1982-83
Member, Metabolic and Genetic Diseases Advisory Committee, South Carolina Department of Health	1984-87
Grant reviewer for March of Dimes - Birth Defects Foundation, Biomedical and Behavioral Sciences Research Grants Program	1985
Member, South Eastern Regional Genetics Group (SERGG) Newborn Screening for Sickle Cell Advisory Committee USPHS Region IV	1985-88
Member, Grant review panel, Genetics Division, DHHS Bureau of Health Care Delivery and Assistance	1987
Consultant: South Carolina Department of Health and Environmental Control, Statewide Newborn Screening for Hemoglobinopathies	1987-91
Member, South Carolina Department of Health and Environmental Control, Commissioner's Task Force on Genetic Services	1991-92
Program Committee, 17th Annual Meeting of the Southern Genetics Group, Litchfield, SC 7/7-7/9	1994
Organizer, Workshop on Caring for Children with Special Needs: Focus on sickle cell and genetic disorders Charleston, SC April 27-28,	1995
Member, AAP Committee on Developmental Pediatrics	1995

Editorial Service

Journal of the American Academy of Dermatology	1989-94, 1996-98
American Journal of Hematology	1991
American Journal of Human Genetics	1991
New England Journal of Medicine	1991
American Journal of Medical Genetics	1992
Southern Medical Journal	1993
Editorial Reviewer, Journal of Medical Genetics	1997
Editorial Reviewer, American Journal of Medical Genetics	1997
Guest Editor, Indian Journal of Pediatrics	2000
Journal of Child Neurology	2005
European Journal of Human Genetics	2005
The American Journal of Medical Sciences	2006

Elective Office Held

Executive Committee Member, South Eastern Regional Genetics Group (SERGG)	1993-94
Board of Directors, South Eastern Regional Genetics Group	1994-99

Invited Lectureships at National and International Meetings

Invited Speaker and Seminar Leader, Genetics for the Clinician: A CME Course sponsored by the Medical Council of India and the All India Institute of Medical Sciences, New Delhi, January 5-7, 1991.

Pediatric Grand Rounds at the Memorial Sloan-Kettering Cancer Center, New York, "Dyskeratosis Congenita: Clinical and Genetic Aspects of a Cancer Predisposing Syndrome", April 11, 1991.

Seminar on "Dyskeratosis Congenita", Department of Human Genetics, College of Health Sciences, Guadalajara, Mexico, April 15, 1991.

Seminar: "On the Tracks of a Cancer Predisposing Gene". Department of Medical Genetics, Oregon Health Sciences University, Portland, Oregon, September 24, 1991.

Invited Speaker: The Joseph Johnston Pediatric Society Meeting, Detroit, Michigan, October 29-30, 1993.

Visiting Professor: University of Tennessee Health Sciences Center, Division of Genetics, September 29-30, 1994.

Invited Speaker - Symposium on Genetics for the Neonatologist - 37th Annual Meeting of the Indian Academy of Pediatrics- Cochin, India, January 10-13, 1998.

Invited Speaker - Update in Genetics - Third Postgraduate Course in Genetics Kasturba Medical College, Manipal, India, January 14-15, 1998.

Invited Member, Committee to Plan an International Multicenter Clinical Trial of Lovastatin Therapy for X-linked Adrenoleukodystrophy. Baltimore, Maryland, January 22-23, 1999.

Invited Participant, United Leukodystrophy Foundation Annual Meeting, DeKalb, IL, July, 1999.

Extramural Support for Academic Activities:

Completed Projects

Development of a Statewide Referral Center for genetic diagnosis, counseling and prenatal diagnosis. Supported by March of Dimes Birth - Defects Foundation Center Grant C-330, 1979-1984, \$145,000

Neonatal screening for hemoglobinopathies: A pilot study. Supported by the South Carolina Department of Health & Environmental Control, 07/01/1984 - 06/30/1986 \$ 42,000

Impact of neonatal screening and early intervention on the mortality and morbidity due to hemoglobinopathies. A SPRANS Demonstration Project, USPHS, Maternal and Child Health Bureau, Grant # MCJ-451003, 10/1/1985 - 9/30/1988 \$ 184,000

Cancer and Birth Defects Surveillance System for Communities Around the Savannah River Site: Supplement to Parent Grant DE-FG09-91SR18217. October 1, 1994 - September, 30, 1996, \$ 125,000/year 10% effort. (P.I. Dr. John P. Dunbar)

Intramural Grants:

Cytogenetic and molecular analysis of dyskeratosis congenita. Funded by the University Research Committee, 11/1/1990 - 10/31/92, \$ 17,060, P.I. 20% effort

Cytogenetic and molecular analysis of dyskeratosis congenita. Children's Hospital Fund 12/1/90 - 10/31/1992, \$ 36,067, P.I. 10% effort

Current Projects:

Genetic Services Contract with the South Carolina, DHEC 1993-94, \$78,000 per year. P.I. 25% effort

The South Carolina Neural Tube Defect Prevention Initiative. P.I. for Region III, 1993-1997, \$ 59,000 per year. 10% effort, Funded by the USPHS, CDC Grant # U85/CCU408774-01 Subcontract with Greenwood Genetic center (P.I. Roger E. Stevenson, M.D.)

Familial Adenomatous Polyposis (FAP) Registry in South Carolina. - Healthy South Carolina Initiative - 7/1/97- 6/30/2000. Co-Investigator, (Brenda Hoffman, P.I.) \$152,805, Year 1.

Lovastatin Therapy for X-linked Adrenoleukodystrophy –

Principal Investigator. Supported by:

University Research Committee, 10/1/97 - 9/30/98, \$ 15,000

Health Sciences Foundation - Children's Hospital Fund - 4/01/98 - 10/31/99, \$45,000.

Merck Co. 11/1/99-6/30/2000, \$ 25,000

Multicenter International Placebo Controlled Trial of Lovastatin Therapy for Adrenoleukodystrophy - Hugo Moser, M.D. P.I. (submitted)

Committee Activities (excluding teaching related):

Member, Ad-hoc Review Committee, University Research Committee	1990,1991
Chairman, Ad-hoc Review Committee, University Research Committee	1991
Basic Sciences Course Directors Committee	1987-1996
Ambulatory Care Quality Improvement Committee	1994-1996
Pediatric Ambulatory Care- Interdisciplinary Team	1997- 1999
MUH Credentialing Subcommitte,	2005 –

Major Clinical Responsibilities:

Pediatric Genetics Clinic, 2 days per week
 Rural Area Outreach Clinics, Monthly in Florence, Beaufort and Georgetown
 Pediatric House staff supervision and in-patient consultation service, daily
 Supervision of Genetic Counselors, Prenatal Wellness Center, as needed

Major Administrative Responsibilities:

Division Director, providing leadership for 9 full-time and 1 part time faculty.

Major Current Teaching Responsibilities:

Course Director, Basic and Clinical Genetics - 3 credit hr. Course for freshman medical students (taught since 1979).

Individual Process Assessment for Parallel Curriculum students (Problem-based learning)

Bedside and clinical teaching and didactic sessions for clinical year students, residents Post-doctoral Fellows

Clinical teaching/supervision of clinical rotation for students - Masters in Genetic Counseling Program – USC School of Medicine.

BIBLIOGRAPHY

A. Original Articles in Peer Reviewed Journals ([†]Student; [‡]Resident; #Post-doctoral Fellow)

1. Pai GS[†], Lipsitz DJ[†]: Clonidine Poisoning. Pediatrics 58:749-50, 1975.
2. VanDyke DL, Logan M, Weiss L, Pai GS[†]: The origin and behavior of two isodicentric bisatellited chromosomes. American Journal of Human Genetics 29: 294-300, 1977.
3. Pai GS[†], Leach DC, Weiss L, Wolff C, VanDyke DL: Thyroid abnormalities in twenty children with Turner syndrome. Journal of Pediatrics 91: 267-69, 1977.
4. Pai GS[#], Valle D, Thomas GH, Rosenbaum K: Cluster of trisomy 13 live births. Lancet I: 613, 1978.
5. Pai GS[#], Thomas GH, Leonard CO, Ward JC, Valle DL, Pyeritz RE: Syndromes due to chromosomal abnormalities: partial trisomy 22, interstitial deletion 13q and trisomy 8. Johns Hopkins Medical Journal 145: 162-169, 1979.
6. Valle D, Pai GS[#], Thomas GH, Pyeritz RE: Homocystinuria due to cystathionine beta-synthase deficiency: clinical manifestations and therapy. Johns Hopkins Medical Journal 146: 110-117, 1980.
7. Pai GS[#], Thomas GH: A new R-banding technique in clinical cytogenetics. Human Genetics 54: 41-45, 1980.
8. Pai GS[#], Sprengel JA, Do TT, Marenzi C, Migeon BR: Localisation of loci for hypoxanthine phosphoribosyl transferase and glucose-6-phosphate dehydrogenase and biochemical evidence of nonrandom X chromosome expression from studies of a human X-autosome translocation. Proceedings of the National Academy of Sciences (USA) 77: 2810-2813, 1980.
9. Pai GS[#], Thomas GH, Mahoney W, Migeon BR: Complex chromosome rearrangements: report of a new case and literature review. Clinical Genetics 18: 436-444, 1980.
10. Tsang KY, Pai GS, Fudenberg HH: Characterization of a newly established human osteosarcoma cell line LM-1. In Vitro 17: 308-314, 1981.
11. Pai GS, Thomas GH, Benke P: Absence of constitutive heterochromatin in a partially identified supernumerary marker chromosome. Journal of Medical Genetics 18: 392-394, 1981.
12. Miller JM, McCarter L, Pai GS, Horger EO III: Hygroma cervicis: antepartum ultrasonic findings. Journal of Reproductive Medicine 26: 567-570, 1981.
13. Pai GS, Grush OC, Shuman C: Hematological abnormalities in triploidy. American Journal of Diseases of Children 136: 367-369, 1982.
14. Pai GS: Cardiac and ophthalmic malformations and in-utero exposure to dilantin. Pediatrics 70: 327, 1982.
15. Pai GS, Rogers JF, Sommer A: Identical MR/MCA syndrome due to del(2)(q32) in two sisters with intrachromosomal insertional translocation in their father. American Journal of Medical Genetics 14: 189-198, 1983.

16. Tsang KY, Fudenberg HH, Sun DC, Pai GS, Bishop LR, Sager S: Cultivation of human osteosarcoma cell lines in serum-free hormone-supplemented medium. In Vitro 19: 515-521, 1983.
17. Horger EO III, Pai GS: Ultrasound in the diagnosis of fetal malformations: implications for obstetric management. American Journal of Obstetrics and Gynecology 147: 163-167, 1983.
18. Powers JM, Moser AB, Upshur JK, Bradford BF, Pai GS, Kohn PH, Frias J, Tiffany C: Fetal cerebro-hepato-renal (Zellweger) syndrome: dysmorphic, biochemical and pathological findings in four affected fetuses. Human Pathology 16: 610-620, 1985.
19. Pai GS, Levkoff AH, Leithiser RE Jr.: Median cleft of the upper lip associated with lipomas of the central nervous system and cutaneous polyps. American Journal of Medical Genetics 26: 921-924, 1987.
20. Pai GS, Shields SM, Houser PM^{*}: Segregation of inverted chromosome 13 in families ascertained through liveborn recombinant offspring. American Journal of Medical Genetics 27: 127-133, 1987.
21. Fakh MH[#], Williamson HO, Seymour EQ, Pai GS: Concurrence of the Holt-Oram syndrome and the Rokitansky-Kuster-Hauser syndrome: A case report. Journal of Reproductive Medicine 32: 549-550, 1987.
22. Pai GS, Macpherson RI: Idiopathic multicentric osteolysis: Report of 2 new cases and literature review. American Journal of Medical Genetics 29: 929-936, 1988.
23. Stanley WS, Pai GS, Horger EO III, Yan Y, McNeal K: Incidental detection of premature centromere separation (PCS) in amniocytes associated with a mild form of Roberts syndrome. Prenatal Diagnosis 8: 565-569, 1988.
24. Pai GS: Minimal expression of Beckwith-Wiedemann syndrome and bilateral Wilms tumor. American Journal of Diseases of Children 142: 586, 1988.
25. Pai GS, Morgan S, Whetsell C⁺: Etiologic heterogeneity in dyskeratosis congenita. American Journal of Medical Genetics 32: 63-66, 1989.
26. Pai GS, Yan Y, DeBauche DM[#], Stanley WS, Paul SR: Bleomycin hypersensitivity in dyskeratosis congenita fibroblasts, lymphocytes and transformed lymphoblasts. Cytogenetics and Cell Genetics 52: 186-189, 1989.
27. Macpherson RI, Pai GS: Hajdu-Cheney syndrome: Case report and review of the literature. Dysmorphology and Clinical Genetics 3: 70-78, 1989.
28. Pai GS, Shapiro SD, Macpherson RI: Exclusion of allelism and close linkage between neurofibromatosis and multiple cartilaginous exostoses. American Journal of Medical Genetics 32: 145, 1989.
29. DeBauche DM[#], Pai GS, Stanley WS: Enhanced G₂ chromatid radio-sensitivity in dyskeratosis congenita fibroblasts. American Journal of Human Genetics 46: 350-357, 1990.
30. Sahn EE, Garen PD, Pai GS, Levkoff AH, Hagerty RC, Maize JC: Multiple rhabdomyomatous mesenchymal hamartomas of skin. American Journal of Dermatopathology 12: 485-491, 1990.
31. Salinas CF, Pai GS, Vera CL, Milutinovic J, Hagerty R, Cooper JD, Cagna DR: Variability of

- expression of the oro-facio-digital syndrome, Type I in black females: six cases. American Journal of Medical Genetics 38: 574-582, 1991.
32. McCall S^{*}, Ramzy MI[#], Cure' JK, Pai GS: Encephalocranio-cutaneous lipomatosis and the Proteus syndrome: distinct entities with overlapping manifestations. American Journal of Medical Genetics 43: 662-668, 1992.
 33. Ning Y[#], Yan Y, Pai GS, Gross AJ: Heterozygote detection through bleomycin induced G₂ chromatid breakage in families with dyskeratosis congenita. Cancer Genetics and Cytogenetics 60: 31-34, 1992.
 34. Cantu' ES, Khan TA, Pai GS: Fluorescence in situ hybridization (FISH) of a whole-arm translocation involving chromosomes 18 and 20 with alpha-satellite DNA probes: Detection of a centromeric DNA break? American Journal of Medical Genetics 44: 340-344, 1992.
 35. Nicholls RD, Pai GS, Cantu' ES, Gottlieb W: Paternal uni-parental disomy of chromosome 15 in a child with Angelman syndrome. Ann. Neurology 32: 512-518, 1992.
 36. Sahn EE, Cook J, Gross RH, Garen PD, Pai GS: Musculoapo-neurotic fibromatosis (extra-abdominal desmoid tumor) in a child with idiopathic multicentric osteolysis. Pediatric Dermatology 10:49-53, 1993.
 37. Rodgers BL^{*}, Vanner LV, Pai GS, Sens MA: Walker-Warburg syndrome: Report of three affected sibs. American Journal of Medical Genetics 49:198-201, 1994.
 38. Parsons J^{*}, Sahn EE, Holden KR, Pai GS: Neurological disease in a child with hepatoerythropoietic porphyria. Pediatric Dermatology 11: 216-211, 1994.
 39. Batista DAS[#], Pai GS, Stetten G: Molecular analysis of a complex chromosomal rearrangement and a review of familial cases American Journal of Medical Genetics 53: 255-263,1994.
 40. Dyer DS⁺, Small KW, Wilson ME, Pai GS: Alstrom syndrome: A case misdiagnosed as Bardet-Biedl. Journal of Pediatric Ophthalmology and Strabismus 31: 272-274,1994.
 41. Metzenberg AB, Pan Y, Das S, Pai GS, Gitschier J: Molecular evidence that the p55 gene is not responsible for either of two Xq28-linked disorders: Emery-Dreifuss muscular dystrophy and dyskeratosis congenita. American Journal of Human Genetics 54: 920-922, 1994.
 42. Das S, Metzenberg A, Pai GS, Gitschier J: Mutational analysis of the biglycan gene excludes it as a candidate gene for X-linked dominant chondrodysplasia punctata, dyskeratosis congenita and incontinentia pigmenti. American Journal of Human Genetics 54: 922-925, 1994.
 43. Jani M, Torchia BS, Pai GS, Migeon BR: Molecular characteriza-tion of tiny ring X chromosomes from females with functional X chromosome disomy and lack of cis X inactivation. Genomics 27: 182-188, 1995.
 44. Cantu' ES, Jacobs DF, Pai GS: An atypical Turner syndrome patient with ring X chromosome mosaicism. Annals of Clinical and Laboratory Science 25: 60-65, 1995.
 45. Porter AD^{*}, Saylor CF, Pai GS: Psychological adjustment of children and adolescents neurofibromatosis Children's Health Care 24: 223-234, 1995.
 46. Joseph M[#], Pai GS, Holden KR, Herman G: X-linked myotubular myopathy: Clinical observations in ten additional cases. American Journal of Medical Genetics 59:168-173, 1995.

47. Cantu' ES, Eicher DJ, Pai GS, Donahue CJ, Harley RA: Mosaic vs. non-mosaic trisomy 9: report of a liveborn infant evaluated by fluorescence in situ hybridization and review of the literature. American Journal of Medical Genetics 62:330-335,1996.
48. Buebel MS, Salinas CF, Pai GS, Macpherson RI, Greer MK, Perez-Comas A: A Seckel-like new syndrome of primordial dwarfism. American Journal of Medical Genetics 64:447-52,1996.
49. Joseph M[#], Cantu' ES, Pai GS, Willi SM, Papenhausen PR, Weiss L: Xp pseudoautosomal gene haploinsufficiency and linear growth deficiency in three girls with chromosome Xp22-Yq11 translocation. Journal of Medical Genetics 33: 906-911, 1996.
50. Singh I, Kremser K, Ghosh B, Singh AK, Pai GS: Abnormality in translational regulation of catalase expression in disorders of peroxisomal biogenesis. Journal of Neurochemistry 67:2373-2378, 1996.
51. Hammond LS, Macias MM, Tarleton JC, Pai GS: Fragile X syndrome due to deletions in the FMR1 gene: a new case and review of the literature. Am J Med Genet 72:430-434, 1997.
52. Pai GS, Hane B, Joseph M[#], Nelson R, Hammond LS, Arena JF, Lubs HA, Stevenson RE, Schwartz CE: A new X-linked recessive syndrome of mental retardation and mild dysmorphism maps to Xq28. J Med Genet 34:529-534, 1997.
53. Kulling D, Green J, Pai GS, Baron PL, Lewin D, Hoffman BJ: Familial adenomatous polyposis registry in South Carolina. J. South Carolina Med Asso . 93: 405-409,1997.
54. Greer MK, Brown FR, III, Pai GS, Chowdhry S, Klein A: Cognitive, adaptive and linguistic skills of children with Williams syndrome. Am J Med Genet 74:521-5,1997.
55. Southgate WM, Wagner CL, Shields SM, Cantu ES, Pai GS: Mosaic trisomy 8: A cautionary note regarding missed antenatal diagnosis. J. Perinatology 18: 78-80,1998.
56. Chahal A, Khan M, Pai GS, Barbosa E, Singh I: Restoration of phytanic acid oxidation in Refsum disease fibroblasts from patients with mutations in the phytanoyl-CoA hydroxylase gene. FEBS Letts. 429: 119-122, 1998.
57. Milewicz DM, Chen H, Park E-S, Petty EM, Zaghi H, Pai GS, Wiling M, Patel V: Reduced penetrance and variable expressivity of familial thoracic aortic aneurysms/dissections. Am J Cardiol 82:474-479,1998.
58. Singh I, Khan M, Key LL, Pai GS: Lovastatin for X- adrenoleukodystrophy (Letter). N Engl J Med 339 (10): 702-3,1998.
59. Meyer-Cohen J, Pai GS, Conradi S: Lethal multiple pterygium syndrome in four male fetuses in a family: Evidence for an X-linked recessive form? (Letter) Am J Med Genet 82:97-99,1999.
60. Knight SW, Heiss N, Vulliamy TJ, Greschner S, Stavrides G, Pai GS, Lestringant G, Varma N, Mason PJ, Dokal I, Poustka A. X-linked dyskeratosis congenita is predominantly caused by missense mutations in the DKC1 gene. Am J Hum Genet. 65:50-58,1999.
61. Pai GS, Khan M, Barbosa E, Key LL, Cure' JK, Betros R and Singh I. Lovastatin therapy for X-linked adrenoleukodystrophy: Clinical and biochemical observations on twelve patients. Molecular Genetics and Metabolism 69:312-322,2000.
62. Leonardi M, Pai GS and Lebel R: The Ritscher-Schinzel (3C) syndrome: Report of four new

cases and review. Am J Med Genet (in press)

63. Stevenson RE, Allen WP, Pai GS, Best R, Seaver LH, Dean J, Thompson S. Decline in prevalence of neural tube defects in a high-risk region of the United States. *Pediatrics* 106(4): 677-683,2000.
64. Pai GS and Gadewar S: Diagnostic approach to children with birth defects. *Indian J. Pediatrics* 67(11): 819-823, 2000.
65. Macpherson RI, Pai GS: Evaluation of newborns with skeletal dysplasias. *Indian J. Pediatrics* 67(12) 907-913,2000.
66. Spranger J, Self S, Clarkson KB, **Pai GS**: Ischiopspinal dysostosis with rib gaps and nephroblastomatosis [Review]. *Clin. Dysmorph* 10:19-23,2001.
67. Currier SC, Lee CK, Chang BS, Bodell AL, **Pai GS, et al**: Mutations in POMT1 are found in a minority of patients with the Walker-Warburg syndrome. *AJMG* 133: 53-57, 2005.
68. Christiansen LR, Lage JM, Wolff DJ, **Pai GS**, Harley RA: Mosaic duplication 1(q11q14) in an infant with nephroblastomatosis and mineralization of extraplacental membranes. *Pediatric Dev Pathol.* 8: 115-123, 2005.
69. Schule, B.; Oviedo, A.; Johnston, K.; **Pai, S, Francke, U**. Inactivating mutations in ESCO2 cause SC phocomelia and Roberts syndrome: no phenotype-genotype correlation. *Am. J. Hum. Genet.* 77: 1117-1128, 2005. PubMed ID : [16380922](#)
70. Suriano G, Yew S, Ferriera P, Senz J, Kaurah P. Rao C, Sears D, **Pai GS**, et al. Characterization of a recurrent germ line mutation of the E-cadherin gene: implications for genetic testing and clinical management. *Clin. Cancer Res.* 11: 5401-5409,2005.
71. Friez MJ, Jones JR, Clarkson K, Lubs H, ABuelo D, Pai GS, Simensen R, Schwartz C, Stevenson RE. Recurrent infections, Hypotonia, and mental retardation caused by duplication of MECP2 and adjacent region in Xq28. *Pediatrics* 118: 1687-1695, 2006.
72. Kmetz E, Burgess G, **Pai GS**: Incontinentia pigmenti with foreshortened hand : Evidence for the significance of NFκβ receptor in human malformation. *Pediatric Dermatology*
73. [Stuart SW](#), [King CH](#), [Pai GS](#). Autism spectrum disorder, Klinefelter syndrome, and chromosome 3p21.31 duplication: a case report. [MedGenMed.](#) 2007 Dec 18;9(4):60.

B. Book Chapters, Invited Review Articles, etc.

1. Pai GS: Ano-rectal malformations: Genetic aspects. Dialogues in Pediatric Urology, 7: 4, 1984.
2. Pai GS: Genetic counseling in pediatric practice. Indian Journal of Pediatrics 53: 471-476, 1986.
3. Pai GS, Houser PM*: Neonatal screening for sickling hemoglobinopathies in South Carolina: Can the promise be fulfilled? J. South Carolina Medical Association 83: 243-245, 1987.
4. Pai GS (1990) Heart-Hand syndrome. In: Mary Louise Buyse (ed) Birth Defects Encyclopedia, Blackwell Scientific Publications, Cambridge, pp 850-851.
5. Carpenter NJ, Pai GS (1990): Dyskeratosis congenita. In: Mary Louise Buyse (ed) Birth Defects Encyclopedia, Blackwell Scientific Publications, Cambridge, pp 567-568.

6. Pai GS: Diagnostic approach to a child with mental retardation. Indian Pediatrics 31: 879-890, 1994.
7. Pai GS, Lewandowski R and Borgaonkar D. (ed); Handbook of chromosomal syndromes Wiley and Sons, New York Publishers, 2003.

C. Abstracts Presented at National or International Scientific Meetings (*Student; *Resident; #Post-doctoral Fellow)

- 1a. Pai GS, Leach DC, Weiss L, et al: Thyroid abnormalities in 20 children with Turner syndrome. Presented at the Annual Meeting of the Society for Pediatric Research, Pediatric Research 11: 429, 1977.
- 2a. Pai GS, Thomas GH: Evaluation of a high resolution R-banding technique. Presented at the Annual Birth Defects Conference, Chicago, Proceedings of the Annual Birth Defects Conf, Chicago 1979.
- 3a. Pai GS, Migeon BR, Thomas GH, et al: Nonrandom inactivation of the X-chromosome in a child with a complex 4 chromosome rearrangement including two X-autosome translocations. Presented at the Annual Meeting of the American Society of Human Genetics. American Journal of Human Genetics 31(6): 106A, 1979.
- 4a. Pai GS, Salinas CF: Cytological abnormalities in Roberts syndrome. Presented at the Annual Meeting of the American Society of Human Genetics, New York, 1980, American Journal of Human Genetics 32(6): 82A, 1980.
- 5a. Rogers JF, Floyd CM, Pai GS: Monosomy 2q32 secondary to an intrachromosomal shift and meiotic crossover in a paternal chromosome 2. Presented at the 32nd Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 33(6): 118A, 1981.
- 6a. Rogers JF, Pai GS: Partial trisomy 13pter->q22: due to malsegregation of a complex maternal translocation (7:8:13) phenotype-karyotype correlation from an analysis of 12 cases. Presented at the 33rd Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 34(6): 140A, 1982.
- 7a. Pai GS, Shields SM, Houser P*: Segregation of inv(13) chromosomes in families ascertained through offspring with rec(13)(dup q) syndrome. Presented at the 35th Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 36: 108S, 1984.
- 8a. Houser P*, Pai GS: A prospective study of low frequency X-aneuploidy in women with multiple reproductive mishaps. Presented at the 35th Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 36(4): 96S, 1984.
- 9a. Stanley WS, Morgan S, Grush O, Khan T, Pai GS: XY/XX chimerism in a girl with acute lymphoblastic leukemia. Presented at the Annual Meeting of the Southern Society for Pediatric Research, Clinical Research 32: 886A, 1984.
- 10a. Powers JM, Moser HW, Upshur JK, Bradford BF, Pai GS, et al.: Fetal cerebro-hepato-renal (Zellweger) syndrome: dysmorphic, biochemical, radiological and pathological findings in an affected fetus. Neurology 274, 1984.
- 11a. Haines JH*, Clarke MC*, Pai GS, Seigle ML, Pilia PA#, Williams AV, Ainsworth SK: Clinical, biochemical and pathological studies on a large black kindred with Fabry's disease. Presented at

- the 37th Annual Meeting of the American Society of Human Genetics, American J. Human Genetics 41: 66A, 1987.
- 12a. Stanley WS, Pai GS, Horger EO III, Yan Y, McNeal KS: Prenatal detection of premature centromere separation (PCS) in the absence of a family history of Roberts syndrome. Presented at the 37th Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 41: 286A, 1987.
- 13a. Whetsell C⁺, Pai GS, Morgan S, Tennyson R: Etiologic heterogeneity in dyskeratosis congenita. Presented at the Annual Meeting of the Southern Society for Pediatric Research, Clinical Research 36: 60A, 1988.
- 14a. DeBauche DM[#], Pai GS, Stanley WS: Hypersensitivity to X-irradiation-induced G₂ chromatid damage in dyskeratosis congenita. Presented at the 38th Annual Meeting of the American Society of Human Genetics, American Journal of Human Genetics 43: A22 [suppl], 1988.
- 15a. Sahn E, Maize J, Garen P, Pai GS, Hagerty R: Multiple rhabdomyomatous mesenchymal hamartomas of the skin. American Journal of Dermatopathology 12: 316A, 1990.
- 16a. Khan TA, Cantu' ES, Pai GS: First report of monosomy 18p and trisomy 20p karyotypic imbalance. Presented at Association of Cytogenetic Technologists Meeting, Vancouver, Canada, 1991.
- 17a. Pai GS, Salinas CF, Morgan SK, Sahn E: Clinical and cytogenetic observations on 8 families with Dyskeratosis congenita (DC) and estimation of proportions due to X-linked and autosomal inheritance. Presented at the 8th International Congress of Human Genetics, Washington, DC, 1991. American Journal of Human Genetics 49(4): 154, 1991.
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