

## CURRICULUM VITAE

Clair Ann Francomano, M.D.

April 18, 2005

*Current Title:* Chief, Human Genetics and Integrative Medicine Section  
Laboratory of Genetics  
National Institute on Aging

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Baltimore, Maryland 21210

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*Place of Birth:* Mt. Vernon, New York

*Spouse:* John L.S. Thorpe

*Children:* Emily Catherine Thorpe, born 8/1/89  
Charles Eric Thorpe, born 8/9/91

### *Education:*

1976 B.A., Yale University, New Haven, CT, magna cum laude

1980 M.D., Johns Hopkins University School of Medicine, Baltimore, MD

### *Postgraduate Training and Fellowship Appointments:*

1980-81 Internship, Department of Medicine, Johns Hopkins Hospital

1981-82 Residency, Department of Medicine, Johns Hopkins Hospital

1982-83 Clinical Fellowship, Medical and Pediatric Genetics, Johns Hopkins University  
School of Medicine

1983-84      Research Fellowship, Medical and Pediatric Genetics, Johns Hopkins University School of Medicine

*Faculty Appointments:*

1984-85      Assistant, Medical and Pediatric Genetics, Johns Hopkins University School of Medicine

1985-92      Assistant Professor, Medicine and Pediatrics, Johns Hopkins University School of Medicine

1990-94      Director, Greenberg Center for Skeletal Dysplasias, Johns Hopkins University School of Medicine

1992-        Associate Professor, Medicine and Pediatrics, Center for Medical Genetics, Johns Hopkins University School of Medicine

1996-        Associate Professor (Adjunct), Department of Health Policy and Management, Johns Hopkins University School of Hygiene and Public Health

*Hospital and University Administrative Appointments:*

1981-82      Medical Board of the Johns Hopkins Hospital

1981-82      Executive Committee, Medical Board of the Johns Hopkins Hospital

1981-84      Committee on House staff and Postdoctoral Programs, Johns Hopkins University School of Medicine

1983-84      Medical School Council, Johns Hopkins University School of Medicine

1985-94      Internship Selection Committee, Department of Medicine, Johns Hopkins University School of Medicine

1988-90      Committee on Admissions, Johns Hopkins University School of Medicine

1988-90      Provost's Committee on the Status of Women at Johns Hopkins University

1991-94      Pediatric Clinical Research Unit Advisory Board

1991-94      General Clinical Research Centers Advisory Committee

1991-94      Committee on Women and Minorities, Johns Hopkins University School of Medicine

1992-94      Task Force on Women's Academic Careers in Medicine, Department of Medicine, Johns Hopkins University School of Medicine

1992-94      Planning Committee, Physician and Society Course

1992-94      Coordinator, Physician and Research Unit, Physician and Society Course, Johns Hopkins University School of Medicine

1992-94      Student Awards Committee, Johns Hopkins University School of Medicine

1992-94      Associate Secretary/Treasurer, Johns Hopkins Medical and Surgical Association Council

1994-        Councilor, Johns Hopkins Medical and Surgical Association Council

*NIH Administrative Appointments:*

1994 -99      Chief, Medical Genetics Branch, National Human Genome Research Institute

1995 - 98      Director, Metropolitan Washington Medical Genetics Fellowship Program

1995 - 96      Search Committee for Director, Center for Medical Genetics, Georgetown University Medical Center

1995 - 96 National Human Genome Research Institute Reinvention Committee  
 1996 -2001 Medical Executive Committee, National Institutes of Health Clinical Center  
 1996 - Senior Biomedical Research Service (SBRS) Policy Board, NIH  
 1996 -2001 Clinical Director, National Human Genome Research Institute  
 1996 - 98 NIH Committee on Inclusion of Children in Research  
 1996 - 97 NIH Committee on Recruitment and Career Development of NIH Clinical  
 Researchers. Subcommittee Deputy Chair, Trends in Clinical Research Activities.  
 1996 - Board of Tutors, Clinical Research Training Program  
 1997 - 99 Clinical Research Revitalization Committee, NIH  
 1997 - 99 Managed Care Workgroup, NIH  
 1998 -2000 Central Tenure Committee, NIH  
 2001 - Chief, Human Genetics and Integrative Medicine Section, Laboratory of  
 Genetics, National Institute on Aging  
 2003 - Promotion and Tenure Committee, National Institute on Aging

*Specialty Certification:*

1983 American Board of Internal Medicine  
 1984 American Board of Medical Genetics, Clinical Genetics  
 1993 American Board of Medical Genetics, Clinical Molecular Genetics  
 2003 American Board of Medical Genetics, Clinical Molecular Genetics recertification

*Licensure:* M.D., Maryland, #D25334

*Awards and Honors:*

1992 Honorary Life Member, Little People of America  
 1994 Antoine Marfan Award, National Marfan Foundation  
 1994 American Society of Clinical Investigation  
 1995 Fellow, Executive Leadership in Academic Medicine (ELAM) Program  
 1998 Presidential Guest Speaker, American Academy of Orthopedic Surgery  
 1999 CD Christian Distinguished Scientist, Society for Gynecological Investigation  
 2001 Keynote Speaker, North American Spine Society  
 2003 Third Annual Leaman Lectureship, Cincinnati Children's Hospital Medical  
 Center

*Memberships in Academic Societies:*

1982 - 86 Associate, American College of Physicians  
 1984 - American Society of Human Genetics  
 1987 - Medical Advisory Board, Little People of America  
 1988 - American Federation for Clinical Research  
 1988 - American College of Physicians  
 1989 - 91 Maryland State Commission on Hereditary and Congenital Disorders  
 1992 - American Medical Association  
 1992 - American College of Medical Genetics, Founding Fellow  
 1993 - 95 Chairman, Board of Directors, Stetler Research Fund for Women Physicians

1993 - 96 Board of Directors, Human Growth Foundation  
 1993 - Organizing Committee, Bone Dysplasia Society  
 1995 - 96 Joint Rapid Action Team on Clinical Genetics Training, American Society of Human Genetics and American College of Medical Genetics  
 1995 - 96 Process Action Team on a Consolidated Military Genetics/DNA Program  
 1995 - Professional Advisory Board, National Marfan Foundation  
 1996 - ELUM, Alumnae of the Executive Leadership in Academic Medicine Fellowship  
 1996 - Board of Directors, Center for Human Genetics, Bar Harbor, ME  
 1998-2000 Item Writer for Clinical Genetics Board Certification Examination, American Board of Medical Genetics  
 1999- Executive Committee, International Society on Dysplasias of the Skeletal  
 2000 National Advisory Council for Women 2000: Forging a Vision; Flora Stone Mather Alumnae Association  
 2001-2003 President, International Society on Dysplasias of the Skeleton

*Evaluative and Editorial Positions:*

1989 - Editorial Board, Annals of Medicine (Helsinki)  
 1993 - 99 Scientific Director for Clinical Disorders, Online Mendelian Inheritance in Man  
 1996 - Genetics Editor, Stedman's Medical Dictionary  
 1989-90 Ad Hoc Review Committee, National Institute Diabetes, Digestive & Kidney Diseases, National Institutes of Health  
 1992 Ad Hoc Review Committee, National Heart Lung and Blood Institute, National Institutes of Health  
 1992 Ad Hoc member, Mammalian Genetics Study Section, National Institutes of Health  
 1993 - 96 Biology I Study Section, National Institutes of Health  
 2001 - Book Reviewer, Johns Hopkins University Press  
 2001 Guest Editor, American Journal of Medical Genetics Seminars in Medical Genetics: Latest Developments in Skeletal Dysplasias  
 2002 Member, Multidisciplinary Review Panel, Genome Canada  
 2002 Guest Editor, American Journal of Medical Genetics Seminars in Medical Genetics: Medical Genetic Studies in the Amish  
 2004 Member, Multidisciplinary Review Panel, Genome Canada

*Research Support (prior to 1996)*

**Collagen Genes in Heritable Connective Tissue Disorders**

Physician Scientist Award 5 K11 AR01361

Principal Investigator

Percent of Effort: 95

Date: 12/1/84 - 6/30/89

Total Direct Costs: \$307,273

Total Indirect Costs: \$24,582

**Molecular Genetic Studies of Polycystic Kidney Disease**

R01 AM 37065

Principal Investigator

Percent of Effort: 20

Date: 7/1/86 - 6/30/89

Total Direct Costs: \$193,000

Total Indirect Costs: \$123,523

**Molecular Genetic Studies of Hereditary Connective Tissue Disorders**

Clinical Scientist Award (Institutional)

Principal Investigator

Percent of Effort: Supplemental Salary Support to PSA 5 K11 AR01361

Date: 7/1/88 - 6/30/90

Total Direct Costs: \$36,500

Total Indirect Costs: \$0

**Mapping the Chromosomes of Man**

Subproject: Project III: Mapping Selected Skeletal Dysplasias

Program Project 5 PO1 HG00373

Subproject Principal Investigator

Percent of Effort: 20

Date: 9/29/88 - 8/31/93

Total Direct Costs Subproject: \$433,981

Total Indirect Costs Subproject: \$277,271

**A Human Genome Database**

Program Project NIH P41HG00586, DOE DE-FC02-91 ER61230

Subproject Principal Investigator

Percent of Effort: 25

Date: 9/1/91 - 8/31/94

Total Direct Costs: \$15,900,000

Subproject Total Direct Costs: \$340,713

**Molecular Biology of the Marfan Syndrome**

NIH R01AR41135

Principal Investigator

Percent of Effort: 40

Date: 3/15/92 - 1/31/95

Total Direct Costs: \$578,070

Total Indirect Costs: \$374,031

**Metaphyseal Dysplasia Research**

Simon Grant (Gift Account)

Principal Investigator

Percent of Effort: 10 (overlap with Program Project SP01 HG00373)

Date: 9/1/92 - 6/30/95

Total Direct Costs: \$417,279

Total Indirect Costs: \$43,963

**Epidemiology of Oral Clefts**

NIH R01DE10293

Co-Investigator

Percent of Effort: 10

Date: 2/1/93 - 1/31/98

Total Direct Costs: \$1,516,911

Total Indirect Costs: \$924,945

*Protocols of the Intramural Program, National Human Genome Research Institute*

- 1994-2000 Clinical and Molecular Manifestations of Six Categories of Genetic Disorders, 95-HG-010, Principal Investigator
- 1996-2002 Clinical and Molecular Manifestations of Heritable Connective Tissue Disorders, 97-HG-0089, Principal Investigator
- 1996-2000 Issues surrounding Prenatal Genetic Testing for Achondroplasia, 96-HG-0123, Principal Investigator
- 1997-2002 Clinical and Molecular Manifestations of Human Skeletal Dysplasias and Short Stature, 98-HG-0119, Principal Investigator
- 1997-2001 Genetic Studies in the Amish and Mennonites, 97-HG-0192, Principal Investigator
- 1999- 2000 A Pilot Study of Mind-Body Therapy for Chronic Pain In Ehlers-Danlos Syndrome, 00-HG-0054, Principal Investigator

*Protocols of the Intramural Program, National Institute on Aging*

- 2003- Clinical and Molecular Manifestations of Human Skeletal Dysplasias and Short Stature
- 2003- Clinical and Molecular Manifestations of Heritable Connective Tissue Disorders
- 2003- Measuring the Human Energy Field in Patients with Organ Failure and Healthy Subjects
- 2002- Molecular Analysis of the *Klotho* Gene in Diseases of Aging

## PUBLICATIONS

### *Original Peer-reviewed Papers:*

1. Cunningham JR, Renie WA, **Francomano CA**, Maumenee IH, and Pyeritz RE. Pseudoxanthoma elasticum: Treatment of gastrointestinal hemorrhage by arterial embolization and observations on autosomal dominant inheritance. *Johns Hopkins Med J* 147(4):168-173, 1980.
2. Trojak JE, Polmar SH, Winkelstein JA, Hsu S, **Francomano CA**, Pierce GE, Scillian JJ, Gale AN, and McKusick VA. Immunologic studies of cartilage-hair hypoplasia in the Amish. *Johns Hopkins Med J* 148(4):157-164, 1981.
3. Nunez AM, **Francomano CA**, Young MF, Martin GR, and Yamada Y. Isolation and partial characterization of genomic clones coding for a human pro alpha 1(II) collagen chain and demonstration of restriction fragment polymorphism at the 3' end of the gene. *Biochemistry* 24(23):6343-6348, 1985.
4. Hecht JT, **Francomano CA**, Horton WA, and Annegers JF. Mortality in achondroplasia. *Am J Hum Genet* 41(3):454-464, 1987.
5. Killen PD, **Francomano CA**, Yamada Y, Modi WS, and O'Brien SJ. Partial structure of the human alpha 2(IV) collagen chain and chromosomal localization of the gene (COL4A2). *Hum Genet* 77(4):318-24, 1987.
6. **Francomano CA**, Liberfarb RM, Hirose T, Maumenee IH, Streeten EA, Meyers DA, and Pyeritz RE. The Stickler syndrome: Evidence for close linkage to the structural gene for type II collagen. *Genomics* 1(4):293-296, 1987.
7. Bartholomew DW, Batshaw ML, Allen RH, Roe CR, Rosenblatt D, Valle DL, and **Francomano CA**. Therapeutic approaches to cobalamin-C methylmalonic acidemia and homocystinuria. *J Ped* 112(1):32-39, 1988.
8. **Francomano CA**, Streeten EA, Meyers DA, and Pyeritz RE. Marfan syndrome: Exclusion of genetic linkage to three major collagen genes. *Am J Med Genet* 29(2):457-462, 1988.
9. **Francomano CA** and Pyeritz RE. Achondroplasia is not caused by mutation in the gene for type II collagen. *Am J Med Genet* 29(4):955-961, 1988.
10. **Francomano CA**, Liberfarb RM, Hirose T, Maumenee IH, Streeten EA, Meyers DA, and Pyeritz RE. The Stickler syndrome is closely linked to COL2A1, the structural gene for type II collagen. *Pathol Immunopathol Res* 7(1-2):104-106, 1988.

11. Cutting GR, Kazazian HH Jr., Antonarakis SE, Killen PD, Yamada Y and **Francomano CA**. Macrorestriction mapping of COL4A1 and COL4A2 collagen genes on human chromosome 13q34. *Genomics* 3(3):256-263, 1988.
12. Blanton SH, Sarfarazi M, Eiberg H, deGroot J, Farndon PA, Child AH, Pope FM, Peltonen L, **Francomano CA**, Boileau C, and Tsipouras P. An exclusion map of Marfan syndrome. *Am J Med Genet* 27(2):73-77, 1990.
13. Finkelstein JE, **Francomano CA**, Brusilow SW, and Traystman MD. Use of denaturing gradient gel electrophoresis for detection of mutation and prospective diagnosis in late onset ornithine transcarbamylase deficiency. *Genomics* 7(2):167-172, 1990.
14. Aryanpur J, Hurko O, **Francomano CA**, Wang H, and Carson B. Craniocervical decompression for cervicomedullary compression in pediatric patients with achondroplasia. *J Neurosurg* 73(3):375-382, 1990.
15. Dietz HC, Pyeritz RE, Hall BD, Cadle RG, Hamosh A, Schwartz J, Meyers DA, and **Francomano CA**. The Marfan syndrome locus: Confirmation of assignment to chromosome 15 and identification of tightly linked markers at 15q15-q21.3. *Genomics* 9(2):355-361, 1991.
16. Finkelstein JE, Doege K, Yamada Y, Pyeritz RE, Graham JM Jr., Moeschler JB, Pauli RM, Hecht JT, and **Francomano CA**. Analysis of the chondroitin sulfate proteoglycan core protein (CSPGCP) gene in achondroplasia and pseudoachondroplasia. *Am J Hum Genet* 48(1):97-102, 1991.
17. **Francomano CA**, Cutting GR, McCormick MK, Chu ML, Timpl R, Hong HK, and Antonarakis SE. The COL6A1 and COL6A2 genes exist as a gene cluster and detect highly informative DNA polymorphisms in the telomeric region of human chromosome 21q. *Hum Genet* 87(2):162-166, 1991.
18. Puffenberger E and **Francomano CA**. PCR-based detection of polymorphic DdeI and KpnI sites in intron 5 of the adenylate kinase (AK1) gene. *Nucleic Acids Research* 19(5):1161, 1991.
19. Dietz HC, Cutting GR, Pyeritz RE, Maslen CL, Sakai LY, Corson GM, Puffenberger EG, Hamosh A, Nanthakumar EJ, Curristin SM, Stetten G, Meyers DA, and **Francomano CA**. Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene. *Nature* 352(6333)July:337-339, 1991.
20. Kainulainen K, Steinmann B, Collins F, Dietz HC, **Francomano CA**, Child A, Kilpatrick MW, Brock DJH, Keston M, Pyeritz RE, and Peltonen L. Marfan syndrome: No evidence for heterogeneity in different populations, and more precise mapping of the gene. *Am J Hum Genet* 49(3):662-667, 1991.
21. Hecht JT, Blanton SH, Wang Y, Daiger SP, Horton WA, Rhodes C, Yamada Y, and **Francomano CA**. Exclusion of human proteoglycan link protein (CRTL1) and type II collagen (COL2A1) genes in pseudoachondroplasia. *Am J Med Genet* 44(4): 420-424, 1992.

22. Dietz HC, Pyeritz RE, Sakai LY, Corson GM, Kendzior RJ Jr, Puffenberger EG, **Francomano CA**, and Cutting GR. Marfan phenotype variability in a family segregating a missense mutation in the epidermal growth factor-like motif of the fibrillin gene. *J Clin Invest* 89(5):1674-1680, 1992.
23. Hamosh A, McDonald JW, Valle D, **Francomano CA**, Niedermeyer E, and Johnston MV. Dextromethorphan and high-dose benzoate therapy for nonketotic hyperglycinemia in an infant. *J Peds* 121(1):131-135, 1992.
24. Schwindinger WF, **Francomano CA**, and Levine MA. Identification of a mutation in the gene encoding the alpha subunit of the stimulatory G-protein of adenylyl cyclase in McCune-Albright syndrome. *Proc Nat Acad Sci* 89(11):5152-5156, 1992.
25. Dietz HC, Saraiva JM, Pyeritz RE, Cutting GR, and **Francomano CA**. Clustering of fibrillin (FBN1) missense mutations in Marfan syndrome patients at cysteine residues in EGF-like domains. *Human Mutation* 1(5):366-374, 1992.
26. Amelung PJ, Panhuysen CIM, Postma DS, Levitt RC, Koeter GH, **Francomano CA**, Bleeker ER, and Meyers DA. Atopy and bronchial hyperresponsiveness: exclusion of linkage to markers on chromosomes 11q and 6p. *Clin Exper Allergy* 22(12):1077-1084, 1992.
27. Dietz HC, Valle D, **Francomano CA**, Kendzior RJ Jr., Pyeritz RE, and Cutting GR. Skipping of constitutive exons in vivo induced by nonsense mutations. *Science* 259(5095)Jan.:680-683, 1993.
28. Peterson GM, **Francomano CA**, Kinzler K, Vogelstein B, and Nakamura Y. Presymptomatic direct detection of adenomatous polyposis coli (APC) gene mutations in Familial Adenomatous Polyposis. *Human Genetics* 91(4):307-311, 1993.
29. Cook A, Raskind W, Blanton SH, Pauli RM, Gregg RG, **Francomano CA**, Puffenberger E, Conrad EU, Schmale G, Schellenberg G, Wijsman E, Hecht JT, Wells D, and Wagner MJ. Genetic heterogeneity in families with hereditary multiple exostoses. *Am J Hum Genet* 53(1):71-79, 1993.
30. Dietz HC, McIntosh I, Sakai LY, Corson GM, Chalberg SC, Pyeritz RE, and **Francomano CA**. Four novel FBN1 mutations: Significance for mutant transcript level and EGF-like domain calcium binding in the pathogenesis of Marfan syndrome. *Genomics* 17(2):468-475, 1993.
31. Warman ML, Abbott MH, Apte SS, Hefferon TW, McIntosh I, Cohn DH, Hecht JT, Olsen BR, and **Francomano CA**. A type X collagen mutation causes Schmid metaphyseal chondrodysplasia. *Nature Genetics* 5(1):79-82, 1993.
32. Hecht JT, **Francomano CA**, Briggs MD, Deere M, Conner B, Horton WA, Warman M, Cohn DH, and Blanton SM. Linkage of typical pseudoachondroplasia to chromosome 19. *Genomics* 18(3):661-666, 1993.

33. McIntosh I, Abbott MH, Warman ML, Olsen BR, and **Francomano CA**. Additional mutations of type X collagen confirm COL10A1 as the Schmid metaphyseal chondrodysplasia locus. *Hum Mol Genet* 3(2):303-307, 1994.
34. **Francomano CA**, Ortiz DeLuna RI, Hefferon TW, Bellus GA, Turner CE, Taylor E, Meyers DA, Blanton SH, Murray JC, McIntosh I, and Hecht J. Localization of the achondroplasia gene to the distal 2.5 Mb of human chromosome 4p. *Hum Mol Genet* 3(5):787-792, 1994.
35. Piersall LD, Dietz HC, Hall BD, Cadel RG, Pyeritz RE, **Francomano CA**, and McIntosh I. Substitution of a cystine residue in a non-calcium binding, EGF-like domain of fibrillin segregates with the Marfan syndrome in a large kindred. *Hum Mol Genet* 3(6):1013-1014, 1994.
36. Sulisalo T, Klockars J, Mäkitie O, **Francomano CA**, de la Chapelle A, Kaitila I and Sistonen P, M Karayiorgou et al. High resolution linkage disequilibrium mapping of the Cartilage-Hair Hypoplasia gene. *Am J Hum Genet* 55(5):937-945, 1994.
37. Pearson P, **Francomano C**, Foster P, Bocchini C, Li P, McKusick V. The status of Online Mendelian Inheritance in Man (OMIM) medio 1994. *Nuc Acids Res* 22:3470-3473, 1994.
38. Sulisalo T, **Francomano CA**, Sistonen P, Maher JF, McKusick VA, De La Chapelle A, and Kaitila I. High-resolution genetic mapping of the cartilage hair hypoplasia (CHH) gene in Amish and Finnish families. *Genomics* 20(3):347-353, 1994.
39. Karayiorgou M, Kasch L, Lasseter VK, Hwang J, Elango R, Bernardini DJ, Kimberland M, Barb R, **Francomano CA**, Wolyniec PS, Lamacz M, Nestadt G, Meyers D, Ott J, Childs B, Antonarakis S, Kazazian HH, Housman DE, and Pulver AE. Report from the Maryland epidemiology schizophrenia linkage study--no evidence for link between schizophrenia and a number of candidate and other genomic regions using a complex dominant model. *Am J Med Genet* 54(4):345-353, 1994.
40. Bellus GA, Hefferon TW, Ortiz DeLuna RI, Hecht JT, Horton WA, Machado M, Kaitila I, McIntosh I, and **Francomano CA**. Achondroplasia is defined by recurrent G380R mutations of FGFR3. *Am J Hum Genet* 56:368-373, 1995.
41. McIntosh I, Abbott MH, and **Francomano CA**. Concentration of mutations causing Schmid metaphyseal chondrodysplasia in the C-terminal non-collagenous domain of type X collagen. *Hum Mutat* 5(2):121-125, 1995.
42. Hwang SJ, Beaty TH, Panny SR, Joseph MJ, Street NA, Gordon S, McIntosh I, and **Francomano CA**. Association study of transforming growth factor alpha (TGF $\alpha$ ) Taq I polymorphism and oral clefts: Indication of gene-environment interaction in a population based sample of infants with birth defects. *Am J Epidem* 141(7):629-36, 1995.
43. Bellus GA, McIntosh I, Smith EA, Aylesworth AS, Kaitila I, Horton WA, Greenshaw G, Hecht JT, and **Francomano CA**. A recurrent mutation in the tyrosine kinase domain of

- fibroblast growth factor receptor 3 causes hypochondroplasia. *Nature Genetics* 10(3):357-359, 1995.
44. Polymeropoulos MH, Ortiz DeLuna RI, Ide SE, Torres R, Rubenstein J, and **Francomano CA**. The gene for pyknodysostosis maps to human chromosome 1cen-q21. *Nature Genet* 10(2):238-239, 1995.
  45. Greenspan DS, Northrup H, McAlister KA, **Francomano CA**, Werstrup RJ, Marchuk DA, and Kwiatkowski DJ. COL5A1: Fine genetic mapping and exclusion as candidate gene in families with nail-patella syndrome, tuberous sclerosis I, hereditary hemorrhagic telangiectasia, and Ehlers-Danlos syndrome type II. *Genomics* 25(3):737-9, 1995.
  46. Polymeropoulos M, Poush J, Rubenstein JR, and **Francomano CA**. Localization of the gene (SYM1) for proximal symphalangism to human chromosome 17q21-q22. *Genomics* 27(2):225-9, 1995.
  47. Hecht JT, Llerrara CA, Greenhaw GA, **Francomano CA**, Bellus GA, and Blanton SH. Confirmatory linkage of hypochondroplasia to chromosome arm 4p. *J Med Genet* 57(3):505-6, 1995.
  48. Hecht JT, Nelson LD, Crowder E, Wang Y, Elder FFB, Harrison WR, **Francomano CA**, Prange CK, Lennon GG, Deere M, and Lawler J. Mutations in exon 17B of cartilage oligomeric matrix protein (COMP) cause pseudoachondroplasia. *Nature Genet* 10(3):325-329, 1995.
  49. Nijbroek G, Sood S, McIntosh I, **Francomano CA**, Bull E, Pereira L, Ramirez F, Pyeritz RE, and Dietz HC. Fifteen novel FBN1 mutations causing Marfan syndrome detected by heteroduplex analysis of genomic amplicons. *Am J Human Genet* 57(1):8-21, 1995.
  50. Polymeropoulos MH, Ide SE, Wright M, Goodship J, Weissenbach J, Pyeritz RE, DaSilva EO, Ortiz De Luna RI, and **Francomano CA**. The gene for Ellis-van Creveld syndrome is located on 4p16. *Genomics* 35(1):1-5, 1996.
  51. Fried LP, **Francomano CA**, Stokes EJ, MacDonald SM, Wagner EM, Carbone KM, Bias WB, Newman MM and Stobo JD. Career development for women in academic medicine: Multiple interventions in a department of medicine. *JAMA* 276(11):898-905, 1996.
  52. Bellus GA, Gaudenz K, Zackai EH, Clark LA, Szabo J, **Francomano CA**, and Muenke M. Identical mutations in three different fibroblast growth factor receptor genes in autosomal dominant craniosynostosis syndromes. *Nature Genet* 14(2):174-176, 1996.
  53. Johnson MR, Polymeropoulos MH, Vos HL, Ortiz De Luna RI, and **Francomano CA**. A nonsense mutation in the cathepsin K gene observed in a family with pycnodysostosis. *Genome Research* 6(11):1050-1055, 1996.

54. Ide SE, Ortiz De Luna R, **Francomano CA**, and Polymeropoulos MH. Exclusion of the MSX1 homeobox gene as the gene for the Ellis van Creveld syndrome in the Amish. *Hum Genet* 98(5):572-575, 1996.
55. Polymeropoulos MH, Ide SE, and **Francomano CA**. Brachydactyly type C gene maps to human chromosome 12q24. *Genomics* 38(1):45-50, 1996.
56. McIntosh I, Clough MV, Schaffer AA, Puffenberger EG, Horton VK, Peters K, Abbott MH, Roig CM, Cutone S, Ozelius L, Kwiatkowski DJ, Pyeritz RE, Brown LJ, Pauli RM, McCormick MK, and **Francomano CA**. Fine mapping of the nail-patella syndrome locus at 9q34. *Am J Hum Genet* 60(1):133-142, 1997.
57. Moloney DM, Wall SA, Ashworth GJ, Oldridge M, Glass IA, **Francomano CA**, Muenke M, and Wilkie AOM. Prevalence of Pro250Arg mutation of fibroblast growth factor receptor 3 in coronal craniosynostosis. *Lancet* 349(9058):1059-1062, 1997.
58. Muenke M, Gripp KW, McDonald-McGinn DM, Gaudenz K, Whitaker LA, Bartlett SP, Markowitz RI, Robin NH, Nwokoro N, Mulvihill JJ, Losken W, Mulliken JB, Guttmacher AE, Wilroy RS, Clarke LA, Hollway G, Ades L, Haan E, Mulley JC, Cohen MM, Bellus GA, **Francomano CA**, Moloney DM, Wall SA, Wilkie AOM, and Zackai EH. A unique point mutation in the fibroblast growth factor receptor 3 (FGFR3) defines a new craniosynostosis syndrome. *Am J Hum Genet* 60(3):555-564, 1997.
59. McIntosh I, Clough MV, Schaffer AA, **Francomano CA**, McCormick MK. Fine mapping of the nail-patella syndrome locus and integration of new markers into the 9q34 map. *Ann Hum Genet* 61: 216-217, 1997
60. Johnson MR, Wilkin DJ, Vos HL, Ortiz De Luna RI, Dehejia AM, Polymeropoulos MH, and **Francomano CA**. Characterization of human extracellular matrix protein 1 gene within the Pycnodysostosis candidate region on chromosome 1q21. *Matrix Bio* 16(5): 289-292, 1997.
61. Stone DL, Agarwala R, Schaffer AA, Weber JL, Vaske D, Oda T, Chandrasekharappa SC, **Francomano CA**, and Biesecker LB. Genetic and physical mapping of the McKusick-Kaufmann (hydrometrocolpos-polydactyly) syndrome. *Hum Mol Genet* 7(3):475-481, 1998.
62. Mogayzel PJ Jr, Carroll JL, Loughlin GM, Hurko O, **Francomano CA**, and Marcus CL. Sleep-disordered breathing in children with achondroplasia. *J Pediatr* 132(4):667-671, 1998.
63. Agarwala R, Biesecker LG, Hopkins KA, **Francomano CA**, and Schäffer AA. Software for constructing and verifying pedigrees within large genealogies and an application to the Old Order Amish of Lancaster county. *Genome Res* 8(3):211-221, 1998.
64. O'Connell AC, Dent B, Brennan T, and **Francomano CA**. Pycnodysostosis: Oro-Facial manifestations in two pediatric patients. *Pediatr Dent* 20(3):204-207, 1998.

65. Chang S, Rosenberg MJ, Morton H, **Francomano CA**, and Biesecker LG. Identification of a mutation in liver glycogen phosphorylase in glycogen phosphorylase I glycogen storage disease type VI. *Hum Mol Genet* 7(5):865-70, 1998.
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