

CURRICULUM VITAE

Francis Sellers Collins, M.D., Ph.D.

PERSONAL DATA

Born: April 14, 1950, Staunton, VA
Spouse: Diane Lynn Baker, M.S.
Children: Margaret Collins Hill, M.D.
Elizabeth Collins Fraker, M.S.W.

POSITION AND AFFILIATION

Director, 1993-present National Human Genome Research Institute National Institutes of Health 31 Center Drive MSC 2152 Bldg. 31, Room 4B09 Bethesda, MD 20892-2152 Phone: 301-594-7185 Fax: 301-402-0837 Email: fc23a@nih.gov	Laboratory Address: Senior Investigator Genome Technology Branch NIH/NHGRI 50 South Drive MSC 8004 Bldg. 50, Rm. 5314 Bethesda, MD 20892-8004
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EDUCATION

1962-66	Robert E. Lee High School, Staunton, VA
1966-70	University of Virginia, Charlottesville, VA B.S., Chemistry with Highest Honors
1970-74	Yale University, New Haven, CT M.Phil., Ph.D., Physical Chemistry
1973-77	University of North Carolina School of Medicine, Chapel Hill, NC M.D. with Honors

POSTGRADUATE TRAINING

1977-78	Intern in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1978-80	Assistant Resident in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1980-81	Chief Resident in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1981-84	Fellow in Human Genetics and Pediatrics, Yale University School of Medicine, New Haven, CT

ACADEMIC AND GOVERNMENT APPOINTMENTS

1984-88	Assistant Professor of Internal Medicine and Human Genetics, University of Michigan Medical School, Ann Arbor, MI
1987-88	Assistant Investigator, Howard Hughes Medical Institute, Ann Arbor, MI
1987-91	Chief, Division of Medical Genetics, Department of Internal Medicine, University of Michigan, Ann Arbor, MI
1988-91	Associate Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor, MI
1988-91	Associate Investigator, Howard Hughes Medical Institute, Ann Arbor, Michigan
1991-93	Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor, MI
1991-93	Investigator, Howard Hughes Medical Institute, Ann Arbor, MI
1993-2003	Professor of Internal Medicine and Human Genetics (on leave), University of Michigan, Ann Arbor, MI
1993-present	Director, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD

SCIENTIFIC ACTIVITIES

EDITORIAL BOARDS

Associate Editor, *The American Journal of Human Genetics*, 1986-1989
Associate Editor, *Genomics*, 1986-2003
Associate Editor, *Genes, Chromosomes, Cancer*, 1989-present
Associate Editor, *Somatic Cell and Molecular Genetics*, 1989-present
Associate Editor, *Human Molecular Genetics*, 1991-present
Communicating Editor, *Human Mutation*, 1992-present
Associate Editor, *Functional and Integrative Genomics*, 1999-present

CERTIFICATION AND LICENSURE

Board of Medical Examiners, State of North Carolina, 1977 (#21760)
American Board of Internal Medicine, 1980 (#75740)
Board of Medical Examiners, State of Connecticut, 1981 (#022557)
American Board of Medical Genetics, 1984
Board of Medical Examiners, State of Michigan, 1984 (#046960)
Board of Physician Quality Assurance, State of Maryland, 2000 (#D0055707)

HONORS AND AWARDS

Dean's Prize for Academic Excellence, 1970
National Science Foundation Graduate Fellowship, 1970-73
Morehead Foundation Fellow, 1973-77
Alumni Loyalty Merit Award, 1974
Deborah C. Leary Research Award, 1975
First Prize, Student Research Day, 1975
Riggins Scholarship, 1975
Heusner Pupil Award, 1976
W. Reece Berryhill Scholarship, 1976
Medical Faculty Award, 1976
Isaac Hall Manning Award, 1977
Alpha Omega Alpha, elected junior year (president of UNC chapter, 1976-77)
Intern of the Year Award, 1978
Henry C. Fordham Senior Resident Award, 1980
Charles E. Culpepper Foundation Fellow, 1983-84
Cooley's Anemia Foundation Fellow, 1983-84
Anthony Renda Research Grant, 1984-85
Hartford Foundation Fellow, 1985-87
Jerome Conn Research Award, 1986
Paul di Sant'Agnes Award of the Cystic Fibrosis Foundation, 1989
Honorary Doctor of Science, Emory University, 1990
James A. Shannon Lecturer, Massachusetts General Hospital, 1990
Elizabeth Crosby Teaching Award, 1990
Gairdner Foundation International Award, 1990
Von Recklinghausen Award of the National Neurofibromatosis Foundation, 1990
Michigania of the Year Award, *The Detroit News*, 1990
Lynen Medal of the Miami Bio/Technology Symposium, 1991
Young Investigator Award of the American Federation for Clinical Research, 1991
Honorary Doctor of Humane Letters, Mary Baldwin College, 1991
Doris Tulcin Award for Cystic Fibrosis Research, 1991
Distinguished Faculty Achievement Award, University of Michigan, 1991
National Medical Research Award, National Health Council, 1991
Dickson Prize, University of Pittsburgh, 1991
88th Christian A. Herter Lecturer, New York University Medical Center, 1992

E. Mead Johnson Award for Research in Pediatrics, 1992
Honorary Doctor of Science, Yale University, 1992
Joseph Levy Memorial Lecturer, The Queen's University of Belfast, 1992
Priscilla White Lecturer, Joslin Diabetes Center, 1993
Richard and Hinda Rosenthal Award, American College of Physicians, 1993
Jack St. Clair Kilby Award, 1993
Honorary Doctor of Science, The Mount Sinai School of Medicine, 1993
Kaiser Permanente Award for Excellence in Teaching, University of Michigan, 1993
National Medical Research Award (Huntington's Disease Collaborative Research), National Health Council, 1993
Sarstedt Prize for Scientific Research, Dresden, Germany, 1993
CIBA-Geigy/Drew Award in Biomedical Research, Drew University, 1993
John M. Nokes Lecture, University of Virginia, 1994
National Organization for Rare Disorders Scientific Leadership Award, 1994
Honorary Doctor of Science, Commencement Speaker, University of North Carolina, 1994
Nelson Award, University of California, Davis, 1994
Lovelace Institute Award for Excellence in Environmental Research, 1994
George D. Aiken Lecture, University of Vermont, 1994
American Academy of Achievement Golden Plate Award, 1994
Steven C. Beering Award for Outstanding Achievement in Biomedical Science, Indiana University, 1994
Baxter Award for Distinguished Research in Biomedical Sciences, Association of American Medical Colleges, 1994
Willis M. Tate Distinguished Lecture, Southern Methodist University, Dallas, Texas, 1994
Jean-Pierre Lecocq Prize, Transgene, S.A., Strasbourg, France, 1994
Lila Gruber Cancer Research Award, American Academy of Dermatology, 1995
Jeffrey Modell Foundation Lifetime Achievement Award, The Jeffrey Modell Foundation, Inc., 1995
Neuhauser Lecture, The Society for Pediatric Radiology, 1995
Lee Farr Lecture, Yale University, 1995
American Association for Clinical Chemistry National Lectureship Award, 1995
Hatfield Lecture, Oregon Health Sciences University, 1995
Susan G. Komen Breast Cancer Foundation National Award for Scientific Distinction, 1995
John Hickam Lecture, Central Society for Clinical Research, 1995
Medical Research Council Lecture, Society of Toxicology, 1996
Lineberger Lecturer, University of North Carolina, 1996
University of California, Los Angeles, Lectureship Award, 1996
Hollister Lecture, Northwestern University, 1996
Honorary Doctor of Science, George Washington University, 1996
9th Annual Donald Ware Waddell Lectureship, Arizona Cancer Center, 1997
Second Annual James Watson Lecture, The Genome Action Coalition, 1997
James Earle Ash Lecture, Armed Forces Institute of Pathology, 1997
American Cancer Society/The Society of Surgical Oncology, Basic Science Lecture Annual Award, 1997
Department of Pediatrics 75th Annual Guest Lecture, University of Michigan, 1997
Brain Blades Memorial Lecture, George Washington University, 1997
Breath of Life Award, Cystic Fibrosis Foundation, 1997
Klemperer Award Lecture, American College of Rheumatology, 1997
Commissioned Officers Association of the U.S. Public Health Service Health Leader of the Year Award, 1997
Meritorious Executive Award, U.S. Department of Health and Human Services, 1997
Mendel Medal, Villanova University, 1998
Carl W. Gottschalk Award and Lecture, The University of North Carolina, 1998
Honorary Doctor of Science, University of Pennsylvania, 1998
Champions of Pediatric Research Award, Children's National Medical Center, 1998
Ralph Spielman Memorial Lecture, Bucknell University, 1998
Noble Lecture Respondent, Harvard University, 1998
Fifth Annual Maurice Galante Lecture, University of California, San Francisco, 1998
Third Annual James Watson Lecture, The Genome Action Coalition, 1998
Association of Molecular Pathology Award for Excellence in Molecular Diagnostics, 1998
American Heart Association Lewis A. Conner Convocation Lecture, 1998
Medical Student Award for Teaching Excellence in Component 1, University of Michigan, 1999
Shattuck Lecture, Massachusetts Medical Society, 1999

Wilbur Lucius Cross Medal, Yale Graduate School Association, 1999
The Computerworld Smithsonian Institution Award, 1999
Arthur S. Flemming Award, The George Washington University, 1999
New York Academy of Sciences, Genetics in the New Millennium Distinguished Honoree, 2000
Presidential State of the Union Honoree, 2000
Dr. Martin Rodbell Lecture, National Institute of Environmental and Health Sciences, 2000
Association of American Physicians, George M. Kober Lecture Award, 2000
Honorary Doctor of Science, Brown University, 2000
University of California, San Diego, School of Medicine Commencement Address, 2000
University of Michigan Medical School Commencement, Keynote Speaker, 2000
Carter Lecture, British Society for Human Genetics, 2000
Neel Distinguished Research Lecture, American Academy of Otolaryngology, 2000
Scientist of the Year, National Disease Research Interchange, 2000
Tinsley Randolph Harrison Lecture, Vanderbilt University, 2000
Sheen Award, New Jersey Chapter, American College of Surgeons, 2000
Charles B. Smith Visiting Research Professor, Memorial Sloan-Kettering Cancer Center, 2000
Hilldale Lecture on Biological Sciences, University of Wisconsin-Madison, 2001
The American Society for Public Administration, The National Capital Area Chapter, President's Award for Outstanding Recent Contributions in the Field of Public Administration, 2001
Virginia's Outstanding Scientist, 2001
Cosgrove Lecture, The American College of Obstetricians and Gynecologists 50th Anniversary Meeting, 2001
The University of Virginia Commencement Address, 2001
Loma Linda University School of Medicine Commencement Address and Boucek Award, 2001
Victor and Clara Award Lecture, XVII World Congress of Neurology, United Kingdom, 2001
Biotechnology Industry Organization and Chemical Heritage Foundation Third Annual Biotechnology Award, 2001
Daniel Nathans Memorial Lecture, Van Andel Research Institute, 2001
Guthrie Family Humanitarian Award, Huntington's Disease Society of America, 2001
Spain's Prince of Asturias Award for Technical and Scientific Research, 2001
Distinguished Achievement and Leadership Award, American Skin Association, 2001
Scientific Achievement Medal, House of Delegates, American Medical Association, 2001
Warren Triennial Prize Lecture, Massachusetts General Hospital, 2002
Willis M. Tate Distinguished Lecture, Southern Methodist University, 2002
Brennan Lecture, Georgetown University, 2002
20th Annual Spicer-Breckenridge Memorial Lecture, University of North Carolina, 2002
Mayo Medical School and Mayo Graduate School Commencement Address, 2002
Physician-in-Chief Pro Tempore, Brigham and Women's Hospital and Harvard Medical School, 2002
Presidential Award, Zeta Beta Sorority, 2002
Joseph Leiter Lecture, National Library of Medicine and Medical Library Association, 2002
Bernard Sachs Lecturer, Child Neurology Society, 2002
Lifetime Achievement Award, Virginia Biotechnology Association, 2002
Gairdner Foundation International Award of Merit, 2002
Stokes Lecturer, University of Pennsylvania, 2002
William Belden Noble Lecturer, Harvard Memorial Church, 2003
51st National Prayer Breakfast Leadership Luncheon Speaker, 2003
American College of Physicians-American Society of Internal Medicine Award, 2003
Walker Prize, Science Museum of Boston, Massachusetts, 2003
Detroit Science & Technology Leadership Award, 2003
Secretary of the Department of Energy Gold Award, 2003
Colonel Sanders Lifetime Achievement Award, March of Dimes, 2004
Honorary Doctor of Science, Baylor College of Medicine, 2004
Baylor College of Medicine Commencement Address, 2004
Bio-IT World President's Award, 2004
Albert Einstein Award for Outstanding Achievements in the Life Sciences, The Jerusalem Fund, 2004
American Society for Clinical Investigation Award, 2005
Northwestern University Commencement Address, 2005
William Allan Award, American Society of Human Genetics, 2005
ASCO Science of Oncology Award and Lecture, 2006

Randolph-Macon University Commencement Address, 2006
University of Connecticut School of Medicine Commencement Address, 2006
Antonie Marfan Award, 2006
Honorary Doctor of Science, University of Miami School of Medicine, 2007
Jiminez Diaz Memorial Lecture and Award, Madrid, Spain, 2007

MEMBERSHIP AND OFFICES IN PROFESSIONAL SOCIETIES

American Association for the Advancement of Science, 1982-present
American Society of Human Genetics, 1983-present
 Human Genome Committee, 1989-1993
 Ad Hoc Committee on Cystic Fibrosis Screening, 1989-1993
 Board of Directors, 1991-1993
American Scientific Affiliation, 1984-present
 Advisory Council Member 2006-present
American Federation for Clinical Research, 1985-present
American Society for Microbiology, 1985-present
American Society for Hematology, 1988-1998
American Society for Clinical Investigation, 1988-present
Human Genome Organization (HUGO), 1989-present
 Executive Council, 1989-1993
Institute of Medicine, 1991-present
Association of American Physicians, 1992-present
 Council, 2001-present
American Medical Association, 1993-present
American College of Medical Genetics (Founding Fellow), 1993-present
National Academy of Sciences, 1993-present
Molecular Medicine Society (Charter Member), 1994-present
American Academy of Arts and Sciences (Fellow), 1998-present

TEACHING ACTIVITIES (UNIVERSITY OF MICHIGAN)

Course Co-Director with Drs. Thomas Gelehrter and David Ginsburg of Medical Genetics (first year medical student course), 1986-2002
Speaker, Advances in Internal Medicine Course, 1984-1992
Director, Internal Medicine Symposium on "Molecular Genetics and Clinical Medicine: The Emerging Interface," June 1985
Course Director, Genetics Short Course on "Human Gene Mapping," May 1986
Guest Lecturer, Human Genetics 542, 1989-1993

COMMITTEE AND ADMINISTRATIVE SERVICE

Chairman, House Staff Council, North Carolina Memorial Hospital, 1980-81
Executive Committee of the Medical Staff, North Carolina Hospital, 1980-81
Search Committee, Chairmanship of Microbiology and Immunology, University of Michigan Medical School, 1985-86
Graduate Admissions Committee, Department of Human Genetics, University of Michigan Medical School, 1985-86
Chairman, Preliminary Exam Committee on Genetics and Nucleic Acids, Cell and Molecular Biology Program, University of Michigan Medical School, 1986
Nomenclature and Clinical Diseases Committees, International Workshop in Human Gene Mapping (HGM9), 1987
Member, Scientific Advisory Board, Hereditary Disease Foundation, 1987-1993
Director, Neurofibromatosis Center, University of Michigan Medical Center, 1987-1993
Research Advisory Committee, Department of Human Genetics, University of Michigan Medical School, 1987-1993

Co-Chairman, Steering Committee, International Consortium on NF1 Linkage Analysis (sponsored by the National Neurofibromatosis Foundation), 1988

Member, NIH Ad Hoc Program Advisory Committee on Complex Genomes, 1988

Member, NIH Ad Hoc Study Section to review grants submitted in response to RFA "Immortalized Cells for Cystic Fibrosis Research," 1988

Scientific Advisory Board, National Neurofibromatosis Foundation, 1988-1993

Co-Chairman, Research Advisory Board, 1989-1993

Chairman, Ad Hoc Study Section to review proposals on "Gene Therapy for Cystic Fibrosis," Cystic Fibrosis Foundation, 1989

Vice-Chairman, Gordon Conference on Molecular Genetics, 1989

External Advisory Committee, Duke University Program on Neurogenetics, 1989-1993

Co-Chairman, Third Annual North American Conference on Cystic Fibrosis, 1989

Co-Chairman, International Conference on Cystic Fibrosis, 1990

Member, NIH Ad Hoc Study Section to review proposals on an "Index Marker Genetic Map," 1990

Chairman, Gordon Conference on Molecular Genetics, 1991

Chairman, Neurofibromatosis Workshop, International Congress of Human Genetics, 1991

Member, NIH Advisory Council to the National Center for Human Genome Research, 1991-93

Director, Executive Committee for the "Experimental Models for Gene Therapy" Program Project, University of Michigan Medical School, 1990-91

Associate Director, 1991-93

Director, Executive Committee for "Genomic Technology and Genetic Disease," Human Genome Center, University of Michigan Medical School, 1990-93

Co-Director, Center for Molecular Genetics, University of Michigan Medical School, 1990-91

External Advisory Committee, Washington University Human Genome Center, 1991-93

Chairman, Cystic Fibrosis Foundation Conference on "Gene Therapy for Cystic Fibrosis," 1991

Member, Medical and Scientific Advisory Board of the National Vascular Malformations Foundation, 1991-93

Member, Medical Advisory Board, HHT Foundation International, 1993

Member, Scientific Advisory Board, National Marfan Foundation, 1993

Chairman, National Advisory Council on Human Genome Research, 1993-present

Member, Search Committee for Institute Director, NIH/NINDS, 1993-94

Co-Chair, Breakout Panel on "Basic Science," The Secretary's Conference on Breast Cancer, 1993

Co-Chair, Breakout Panel on "Internationalization of Research," Forum on Science and the National Interest, 1994

Co-Chair, Working Group on "Hereditary Susceptibility," National Action Plan for Breast Cancer, 1994-99

Member, Senior Biomedical Research Service Advisory Committee, NIH, 1995-98

Member, Cancer Genetics Working Group, National Cancer Institute, 1996-99

Chair, Board of Governors, Center for Inherited Disease Research, 1996-present

Co-Chair, Steering Committee, National Coalition for Health Professional Education in Genetics, 1996-2002

Member, Interagency Group on Genetic Testing, Department for Health and Human Services, 1997-2000

Chair, NCBI Resources Committee, NIH, 1998-2002

Member, Search Committee for Institute Director, NIH/NIDDK, 1998-99

Liaison Member, Secretary's Advisory Committee on Genetic Testing, 1999-2002

Member, American Academy of Physicians Council, 2001-present

Co-Chair, Search Committee for Institute Director, NIH/NIMH, 2001-02

Chair, Board of Directors, National Coalition for Health Professional Education in Genetics, 2002-present

Member, NIH Administrative Restructuring Advisory Committee, 2003

Co-Chair, Search Committee for Institute Director, NIH/NHLBI, 2003-2004

Member, NIH Steering Committee, 2003-2006

Liaison Member, Secretary's Advisory Committee on Genetics, Health and Society, 2003-present

Co-chair, NIH Intramural Research Working Group, 2003-2006

Co-chair, NIH Roadmap Implementation Group: Building Blocks, Pathways, and Networks, 2003-present

Co-chair, NIH Roadmap Implementation Group: Molecular Libraries and Imaging, 2003-present

NIH Liaison to Department of Energy, 2004-present

Member, Committee to Structure the Office of Planning and Strategic Initiatives (OPASI), 2005-present

Member, NIH Morale Committee, 2005-present

Vice-Chair, NIH/NCBI Resource Committee, 2006- present

Co-chair, Search Committee National Institute of Diabetes and Digestive and Kidney Diseases, 2006

Member, VA Genomics Committee, 2006-present

BIBLIOGRAPHY

PUBLICATIONS IN SCIENTIFIC JOURNALS

Peer Reviewed

1. Trindle CO, Collins FS. Energy-based formalism for mapping analysis of concerted reactions. *Int J Quantum Chem.* **4**, 195-204 (1971).
2. Collins FS, George JK, Trindle CO. Molecular orbital view of the stereochemical behavior in the interaction of bicyclo[2.1.0]pentane and unsaturated molecules. *J Am Chem Soc.* **94**, 3732-37 (1972).
3. Collins FS, Preston RK, Cross RJ. Vibrationally inelastic scattering of $H^+ + H_2$. *Chem Phys Lett.* **25**, 608-10 (1974).
4. Collins FS, Cross RJ. Vibrationally inelastic scattering at high energies. $H^+ + H_2$. *J Chem Phys.* **65**, 644-52 (1976).
5. Collins FS, Ney RL, Hadler NM, McMillan CW, Mangano C. The medical dilemma—professional demands and personal needs. *The Pharos.* **41**, 29-34. (1978).
6. Collins FS, Summer GK. Determination of glutamine and glutamine acid in biological fluids by gas chromatography. *J Chromatogr.* **145**, 456-63 (1978).
7. Collins FS, Summer GK, Schwartz RP, Parke JC. Neonatal argininosuccinic aciduria-survival after early diagnosis and dietary management. *J Pediatr.* **96**, 429-31 (1980).
8. Collins FS, Orringer EP. Pulmonary hypertension and cor pulmonale in the sickle hemoglobinopathies. *Am J Med.* **73**, 814-21 (1982).
9. Collins FS, Mahoney MJ. Hydrocephalus and abnormal digits after failed first-trimester prostaglandin abortion attempt. *J Pediatr.* **102**, 620-1 (1983).
10. Collins FS, Weissman SM. The molecular genetics of human hemoglobin. *Prog Nucl Acids Res Mol Biol.* **31**, 351-458 (1984).
11. Stoeckert CJ, Collins FS, Weissman SM. Human fetal globin DNA sequences suggest novel conversion event. *Nucleic Acids Res.* **12**, 4469-79 (1984).
12. Collins FS, Stoeckert CJ, Serjeant GR, Forget BG, Weissman SM. G gamma beta+ hereditary persistence of fetal hemoglobin: cosmid cloning and identification of a specific mutation 5' to the G gamma gene. *Proc. Natl. Acad. Sci. USA.* **81**, 4894-8 (1984).
13. Collins FS, Boehm CD, Waber PG, Stoeckert CJ, Weissman SM, Forget BG, Kazazian HH. Concordance of a point mutation 5' to the G gamma globin gene with G gamma beta +: Hereditary persistence of fetal hemoglobin in the black population. *Blood.* **64**, 1292-6 (1984).
14. Jennings T, Duray PH, Collins FS, Battaglini J, Enzinger FM. Infantile myofibromatosis: evidence for an autosomal dominant disorder. *Am J Surg Path.* **8**, 529-38 (1984).
15. Collins FS, Weissman SM. Directional cloning of DNA fragments at a large distance from an initial probe: a circularization method. *Proc Natl Acad Sc. USA.* **81**, 6812-6 (1984).
16. Collins FS, Metherall JE, Yamakawa J, Pan J, Weissman SM, Forget BG. A point mutation in the A gamma-globin gene promoter in Greek hereditary persistence of fetal haemoglobin. *Nature.* **313**, 325-6 (1985).

17. Waber PG, Bender MA, Gelinas RE, Kattamis C, Karaklis A, Sofroniadou K, Stamatoyannopoulos G, Collins FS, Forget BG, Kazazian HH. Concordance of a point mutation 5' to the A gamma-globin gene with A gamma beta+ hereditary persistence of fetal hemoglobin in Greeks. *Blood*. **67**, 551-4 (1986).
18. Metherall JE, Collins FS, Pan J, Weissman SM, Forget BG. Beta zero thalassemia caused by a base substitution that creates an alternative splice acceptor site in an intron. *EMBO J*. **5**, 2551-7 (1986).
19. Collins FS, Drumm ML, Cole JL, Lockwood WK, Vande Woude GF, Iannuzzi MC. Construction of a general human chromosome jumping library, with application to cystic fibrosis. *Science*. **235**, 1046-9 (1987).
20. Treisman J, Collins FS. Adult Turner syndrome associated with chylous ascites and vascular anomalies. *Clin Genet*. **31**, 218-23 (1987).
21. Smith CL, Lawrance SK, Gillespie GA, Cantor CR, Weissman SM, Collins FS. Strategies for mapping and cloning macroregions of mammalian genomes. *Methods Enzymol*. **151**, 461-89 (1987).
22. Seizinger BR, Rouleau GA, Ozelius LJ, Lane AH, Faryniarz AG, Chao MV, Huson S, Korf BR, Parry DM, Pericak-Vance MA, Collins FS, Hobbs WJ, Falcone BG, Iannuzzi JA, Roy JC, St. George-Hyslop PS, Tanzi RE, Bothwell MA, Upadhyaya M, Harper P, Goldstein AE, Hoover DL, Bader JL, Spence MA, Mulvihill JJ, Aylsworth AS, Vance JM, Rossenwasser GOD, Gaskell PC, Roses AD, Martuza RL, Breakefield XO, Gusella JF. Genetic linkage of von Recklinghausen neurofibromatosis to the nerve growth factor receptor gene. *Cell*. **49**, 589-94 (1987).
23. Iannuzzi MC, Konkle BA, Ginsburg D, Collins FS. RsaI RFLP in the human von Willebrand factor gene. *Nucleic Acids Res*. **15**, 5909 (1987).
24. Seashore JH, Collins FS, Markowitz RI, Seashore MR. Familial apple peel jejunal atresia: surgical, genetic, and radiographic aspects. *Pediatrics*. **80**, 540-4 (1987).
25. Diehl SR, Boehnke M, Collins FS, Erickson RP, Karolyi IJ, Ploughman LM, Pericak-Vance MA, Aylsworth AS, Roses AD. Linkage analysis of peripheral neurofibromatosis to DNA markers on chromosome 8. *J Med Genet*. **24**, 532-4 (1987).
26. Collins FS, Cole JL, Lockwood WK, Iannuzzi MC. The deletion in both common types of hereditary persistence of fetal hemoglobin is approximately 105 kilobases. *Blood*. **70**, 1797-803 (1987).
27. Konkle BA, Kim S, Iannuzzi MC, Alani R, Collins FS, Ginsburg D. SacI RFLP in the human von Willebrand factor gene. *Nucleic Acids Res*. **15**, 6766 (1987).
28. Stephens K, Riccardi VM, Rising M, Ng S, Green P, Collins FS, Rediker KS, Powers JA, Parker C, Donis-Keller H. Linkage studies with chromosome 17 DNA markers in 45 neurofibromatosis 1 families. *Genomics*. **1**, 353-7 (1987).
29. Diehl SR, Boehnke M, Erickson RP, Baxter AB, Bruce MA, Lieberman JL, Platt DJ, Ploughman LM, Seiler KA, Sweet AM, Collins FS. Linkage analysis of von Recklinghausen neurofibromatosis to DNA markers on chromosome 17. *Genomics*. **1**, 361-3. (1987).
30. Engelke DR, Hoener PA, Collins FS. Direct sequencing of enzymatically amplified human genomic DNA. *Proc Natl Acad Sci USA*. **85**, 544-8 (1988).
31. Kenwrick SJ, Smith TJ, England S, Collins FS, Davies KE. Localisation of the endpoints of deletions in the 5' region of the Duchenne gene using a sequence isolated by chromosome jumping. *Nucleic Acids Res*. **16**, 1305-17 (1988).
32. Butler MG, Fogo AB, Fuchs DA, Collins FS, Dev VG, Phillips JA. Two patients with ring chromosome 15 syndrome. *Am J Med Genet*. **29**, 149-54 (1988).

33. Bloch DB, Bloch KD, Iannuzzi M, Collins FS, Neer EJ, Seidman JG, Morton CC. The gene for the alpha i1 subunit of human guanine nucleotide binding protein maps near the cystic fibrosis locus. *Am J Hum Genet.* **42**, 884-8 (1988).
34. Roth MS, Collins FS, Ginsburg D. Sizing of the human T cell receptor alpha locus and detection of a large deletion in the Molt-4 Cell line. *Blood.* **71**, 1744-7 (1988).
35. Drumm ML, Smith CL, Dean M, Cole JL, Iannuzzi MC, Collins FS. Physical mapping of the cystic fibrosis region by pulsed-field gel electrophoresis. *Genomics.* **2**, 346-54 (1988).
36. Richards JE, Gilliam TC, Cole JL, Drumm ML, Wasmuth JJ, Gusella JF, Collins FS. Chromosome jumping from D4S10 (G8) toward the Huntington disease gene. *Proc Natl Acad Sci USA.* **85**, 6437-41 (1988).
37. Marchuk D, Collins FS. pYAC-RC, a yeast artificial chromosome vector for cloning DNA cut with infrequently cutting restriction endonucleases. *Nucleic Acids Res.* **16**, 7743 (1988).
38. Fountain JW, Lockwood WK, Collins FS. Transfection of primary human skin fibroblasts by electroporation. *Gene.* **68**, 167-72 (1988).
39. Iannuzzi MC, Weber JL, Yankaskas J, Boucher R, Collins FS. The introduction of biologically active foreign genes into human respiratory epithelial cells using electroporation. *Am Rev Respir Dis.* **138**, 965-8 (1988).
40. Gumucio DL, Rood KL, Gray TA, Riordan MF, Sartor CI, Collins FS. Nuclear proteins that bind the human gamma-globin gene promoter: alterations in binding produced by point mutations associated with hereditary persistence of fetal hemoglobin. *Mol Cell Biol.* **8**, 5310-22 (1988).
41. Collins FS, Ponder BA, Seizinger BR, Epstein CJ. The von Recklinghausen neurofibromatosis region on chromosome 17—genetic and physical maps come into focus. *Am J Hum Genet.* **44**, 1-5 (1989).
42. Stephens K, Green P, Riccardi VM, Ng S, Rising M, Barker D, Darby JK, Falls KM, Collins FS, Willard HF, Donis-Keller H. Genetic analysis of eight loci tightly linked to neurofibromatosis I. *Am J Hum Genet.* **44**, 13-9 (1989).
43. Diehl SR, Boehnke M, Erickson RP, Ploughman LM, Seiler KA, Lieberman JL, Clarke HB, Bruce MA, Schorry EK, Pericak-Vance M, O'Connell P, Collins FS. A refined genetic map of the region of chromosome 17 surrounding the von Recklinghausen neurofibromatosis (NF1) gene. *Am J Hum Genet.* **44**, 33-7 (1989).
44. O'Connell P, Leach RJ, Ledbetter DH, Cawthon RM, Culver M, Eldridge JR, Frej AK, Holm TR, Wolff E, Thayer MJ, Schafer AJ, Fountain JW, Wallace MR, Collins FS, Skolnick MH, Rich DC, Fournier REK, Baty BJ, Carey JC, Leppert MF, Lathrop GM, Lalouel JM, White RL. Fine structure DNA mapping studies of the chromosomal region harboring the genetic defect in neurofibromatosis type I. *Am J Hum Genet.* **44**, 51-7 (1989).
45. Fountain JW, Wallace MR, Brereton AM, O'Connell P, White RL, Rich DC, Ledbetter DH, Leach RJ, Fournier RE, Menon AG, Gusella JF, Barker D, Stephens K, Collins FS. Physical mapping of the von Recklinghausen neurofibromatosis region on chromosome 17. *Am J Hum Genet.* **44**, 58-67 (1989).
46. Iannuzzi MC, Dean M, Drumm JL, Hidaka N, Cole JL, Perry A, Stewart C, Gerrard B, Collins FS. Isolation of additional polymorphic clones from the cystic fibrosis region, using chromosome jumping from D7S8. *Am J Hum Genet.* **44**, 695-703 (1989).
47. Wallace MR, Fountain JW, Brereton AM, Collins FS. Direct construction of a chromosome-specific NotI linking library from flow-sorted chromosomes. *Nucleic Acids Res.* **17**, 1665-77 (1989).
48. Fountain JW, Wallace MR, Bruce MA, Seizinger BR, Menon AG, Gusella JF, Michels VV, Schmidt MA, Dewald GW, Collins FS. Physical mapping of a translocation breakpoint in neurofibromatosis. *Science.* **244**, 1085-7 (1989).

49. Boehnke M, Arnheim N, Li H, Collins FS. Fine-structure genetic mapping of human chromosomes using the polymerase chain reaction on single sperm: experimental design considerations. *Am J Hum Genet.* **45**, 21-32 (1989).
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