CURRICULUM VITAE

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

PERSONAL DATA

Born: April 14, 1950, Staunton, VA

POSITION AND AFFILIATION

Director (2009-Present)
National Institutes of Health
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EDUCATION

1962-1966	Robert E. Lee High School, Staunton, VA
1966-1970	University of Virginia, Charlottesville, VA B.S., Chemistry with Highest Honors
1970-1974	Yale University, New Haven, CT M.Phil., Ph.D., Physical Chemistry
1973-1977	University of North Carolina School of Medicine, Chapel Hill, NC M.D. with Honors

POSTGRADUATE TRAINING

1977-1978	Intern in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1978-1980	Asst. Resident in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1980-1981	Chief Resident in Medicine, North Carolina Memorial Hospital, Chapel Hill, NC
1981-1984	Fellow in Human Genetics and Pediatrics, Yale University School of Medicine, New Haven, CT

ACADEMIC AND GOVERNMENT APPOINTMENTS

1984-1988	Asst. Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor
1987-1988	Asst. Investigator, Howard Hughes Medical Institute, Ann Arbor, MI
1987-1991	Chief, Div. of Medical Genetics, Dept. of Internal Medicine, University of Michigan, Ann Arbor
1988-1991	Assoc. Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor
1988-1991	Assoc. Investigator, Howard Hughes Medical Institute, Ann Arbor, MI
1991-1993	Professor of Internal Medicine and Human Genetics, University of Michigan, Ann Arbor
1991-1993	Investigator, Howard Hughes Medical Institute, Ann Arbor, MI
1993-2003	Professor of Internal Medicine and Human Genetics (on leave), University of Michigan, Ann Arbor
1993-2008	Director, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD
1993-2008	Senior Investigator, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD
2008-2009	Special Volunteer, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD
2009-Present	Senior Investigator, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD
2009-Present	Director, National Institutes of Health, Bethesda, MD

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, 1970-1973
Morehead Foundation Fellow, 1973-1977
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, 1976-1977)
Intern of the Year Award, 1978
Henry C. Fordham Senior Resident Award, 1980
Charles E. Culpepper Foundation Fellow, 1983-1984
Cooley's Anemia Foundation Fellow, 1983-1984
Anthony Renda Research Grant, 1984-1985
Hartford Foundation Fellow, 1985-1987
Jerome Conn Research Award, 1986
Paul di Sant'Agnese 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, National Neurofibromatosis Foundation, 1990
Michiganian 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
E. Mead Johnson Award for Research in Pediatrics, 1992
Honorary Doctor of Science, Yale University, 1992
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
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
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
Jean-Pierre Lecocq Prize, Transgene, S.A., 1994
Lila Gruber Cancer Research Award, American Academy of Dermatology, 1995
Jeffrey Modell Foundation Lifetime Achievement Award, 1995
American Association for Clinical Chemistry National Lectureship Award, 1995
Honorary Doctor of Science, George Washington University, 1996
9th Annual Donald Ware Waddell Lectureship, Arizona Cancer Center, 1997
American Cancer Society/The Society of Surgical Oncology, Basic Science Lecture Annual Award, 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. Dept. 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
Association of Molecular Pathology Award for Excellence in Molecular Diagnostics, 1998
Medical Student Award for Teaching Excellence in Component 1, University of Michigan, 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
George M. Kober Lecture Award, Association of American Physicians, 2000
Honorary Doctor of Science, Brown University, 2000
School of Medicine Commencement Address, University of California, San Diego, 2000
Scientist of the Year, National Disease Research Interchange, 2000
Sheen Award, New Jersey Chapter, American College of Surgeons, 2000
Charles B. Smith Visiting Research Professor, Memorial Sloan-Kettering Cancer Center, 2000
President's Award for Outstanding Recent Contributions in the Field of Public Administration, The American Society for Public Administration, National Capital Area Chapter, 2001
Virginia's Outstanding Scientist, 2001
Commencement Address, The University of Virginia, 2001
Commencement Address and Boucek Award, Loma Linda University School of Medicine, 2001
Victor and Clara Award Lecture, XVII World Congress of Neurology, United Kingdom, 2001
Third Annual Biotechnology Award, Biotechnology Industry Organization and Chemical Heritage Foundation, 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
Commencement Address, Mayo Medical School and Mayo Graduate School, 2002
Physician-in-Chief Pro Tempore, Brigham and Women's Hospital and Harvard Medical School, 2002
Presidential Award, Zeta Beta Sorority, 2002
Lifetime Achievement Award, Virginia Biotechnology Association, 2002
Gairdner Foundation International Award of Merit, 2002
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 Dept. of Energy Gold Award, 2003
Colonel Sanders Lifetime Achievement Award, March of Dimes, 2004
Honorary Doctor of Science, Baylor College of Medicine, 2004

Commencement Address, Baylor College of Medicine, 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 Honorary Degree, 2005
William Allan Award, American Society of Human Genetics, 2005
ASCO Science of Oncology Award, 2006
Commencement Address, Randolph-Macon University, 2006
Commencement Address, University of Connecticut School of Medicine, 2006
Antoine Marfan Award of the National Marfan Association, 2006
Honorary Doctor of Science, University of Miami School of Medicine, 2007
Jiminez Diaz Memorial Lecture and Award, Madrid, Spain, 2007
Presidential Medal of Freedom, 2007
Will Rogers Prize, 2007
Honorary Doctor of Science, University of Michigan, 2007
Honorary Doctor of Science, University of Maryland, Baltimore, 2008
Andrus Award, American Association of Retired Persons, 2008
Inamori Ethics Prize, 2008
Michael J. Scotti Award, National Coalition for Health Professional Education in Genetics, 2008
Nevada Medal Award, The Desert Research Institute, 2009
Honorary Doctor of Humane Letters, Virginia Commonwealth University, 2009
Appointed as an Ordinary Member of the Pontifical Academy of Sciences, 2009
H. Richard Nesson Award, Biomedical Science Careers Student Conference, 2009
Philip Hauge Abelson Award, American Association for the Advancement of Science, 2009
National Medal of Science, 2009
Co-recipient, Albany Medical Center Prize, 2010
Pro Bono Humanum Award, Prix Galien, 2012
Honorary Doctor of Science, Princeton University, 2013
Wallop Howard Leadership Award, Congressional Awards Foundation, 2013
Ronald and Nancy Reagan Research Award, Alzheimers Association, 2014
Tribeca Film Festival Disruptive Innovation Award, 2014
Productive Lives Award, Brain and Behavior Research Foundation, 2014
Evert Jan Thomassen a Thuessink medal, University of Groningen, 2015
Inductee, The Irish American Hall of Fame, 2015
Pioneering Biomedical Research Award, Society for Women's Health Research, 2015
Lupus Foundation of America's National Advocacy Award Recipient, 2015

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, 2006-2009
American Federation for Clinical Research, 1985-present
American Society for Microbiology, 1985-1998
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
    President-elect, 2008; President, 2009 (resigned to become NIH Director)
    Council, 2001-2009

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
American Academy of Achievement, Board of Directors, 2006-2009
The Hastings Center (Fellow), 2008-2009
The Trinity Forum (Senior Fellow), 2008-2009

TEACHING ACTIVITIES (UNIVERSITY OF MICHIGAN)

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

COMMITTEE AND ADMINISTRATIVE SERVICE

Chairman, House Staff Council, North Carolina Memorial Hospital, 1980-1981
Executive Committee of the Medical Staff, North Carolina Hospital, 1980-1981
Search Committee, Microbiology and Immunology Chairmanship, University of Michigan Medical School, 1985-1986
Graduate Admissions Committee, Dept. of Human Genetics, University of Michigan Medical School, 1985-1986
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, Dept. 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 grant proposals to RFA "Immortalized Cells for Cystic Fibrosis Research," 1988
Scientific Advisory Board, National Neurofibromatosis Foundation, 1988-1993
Co-Chairman, Research Advisory Board, National Neurofibromatosis Foundation, 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, 1991-1993
Director (1990-1991), and Assoc. Director (1991-1993), Executive Committee for "Experimental Models for Gene Therapy" Program Project, University of Michigan Medical School
Director, Executive Committee for "Genomic Technology and Genetic Disease," Human Genome Center, University of Michigan Medical School, 1990-1993
Co-Director, Center for Molecular Genetics, University of Michigan Medical School, 1990-1991
External Advisory Committee, Washington University Human Genome Center, 1991-1993
Chairman, Cystic Fibrosis Foundation Conference on "Gene Therapy for Cystic Fibrosis," 1991
Member, Medical and Scientific Advisory Board of the National Vascular Malformations Foundation, 1991-1993
Member, Medical Advisory Board, HHT Foundation International, 1993
Member, Scientific Advisory Board, National Marfan Foundation, 1993
Chairman, National Advisory Council on Human Genome Research, 1993-2008

Member, Search Committee for Institute Director, NINDS/NIH, 1993-1994
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, 1994-1999
Member, Senior Biomedical Research Service Advisory Committee, NIH, 1995-1998
Member, Cancer Genetics Working Group, National Cancer Institute, 1996-1999
Chair, Board of Governors, Center for Inherited Disease Research, 1996-2008
Co-Chair, Steering Committee, National Coalition for Health Professional Education in Genetics, 1996-2002
Member, Interagency Group on Genetic Testing, Dept. of Health and Human Services, 1997-2000
Chair, NCBI Resources Committee, NIH, 1998-2002
Member, Search Committee for Institute Director, NIDDK/NIH, 1998-1999
Liaison Member, Secretary's Advisory Committee on Genetic Testing, 1999-2002
Member, Association of American Academy of Physicians Council, 2001-present
Co-Chair, Search Committee for Institute Director, NIMH/NIH, 2001-2002
Chair, Board of Directors, National Coalition for Health Professional Education in Genetics, 2002-2008
Member, NIH Administrative Restructuring Advisory Committee, 2003
Co-Chair, Search Committee for Institute Director, NHLBI/NIH, 2003-2004
Member, NIH Steering Committee, 2003-2006
Liaison Member, Secretary's Advisory Committee on Genetics, Health and Society, 2003-2008
Co-chair, NIH Intramural Research Working Group, 2003-2006
Co-chair, NIH Roadmap Implementation Group: Building Blocks, Pathways, and Networks, 2003-2008
Co-chair, NIH Roadmap Implementation Group: Molecular Libraries and Imaging, 2003-present
NIH Liaison to Dept. of Energy, 2004-2007
Member, Committee to Structure the Office of Planning and Strategic Initiatives (OPASI), 2005-2008
Member, NIH Morale Committee, 2005-2008
Chair, Genetic Association Information Network (GAIN) Steering Committee, 2006-2008
Vice-Chair, NCBI/NIH Resource Committee, 2006- 2008
Co-chair, Search Committee, NIDDK/NIH, 2006
Member, Veterans Administration Genomics Committee, 2006-2008
Co-chair, Senior Oversight Committee for the NIH Genome-Wide Association Study Data Sharing Policy, 2007-2008
Member, Obama Transition Team for Dept. of Health and Human Services, 2008-2009
Member, HHS American Recovery and Reinvestment Act Implementation Team, 2009-2010
Member, HHS Healthy Weight Task Force, 2009-2010
Co-chair, National Science and Technology Council Committee on Science (NSTC-CoS), 2009-present
Liaison Member, Foundation for NIH, 2009-present
Member, Reagan-Udall Foundation Board, 2009-present
Member, Heads of International Research Organizations (HIROs), 2009-present
    Chair, 2013-present
Member, U.S. Global Health Initiative Strategic Council, 2010-2012
    Chair, Research Committee
Co-chair, Joint NIH-FDA Leadership Council, 2010-present
Member, Board of Governors, Patient Centered Outcomes Research Institute, 2010-present
Co-Chair, Target Validation Consortium Steering Committee, 2012 – 2013
Co-chair, Accelerating Medicines Partnership, 2013-present

BIBLIOGRAPHY

PUBLICATIONS IN SCIENTIFIC JOURNALS

Peer Reviewed (includes science policy articles for which review was conducted by journal editor)

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**, 3732-3737 (1972).

3. Collins FS, Preston RK, Cross RJ. Vibrationally inelastic scattering of H+ + H2. *Chem Phys Lett*. **25**, 608-610 (1974).

4. Collins FS, Cross RJ. Vibrationally inelastic scattering at high energies. H+ + H2. *J Chem Phys*. **65**, 644-652 (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**, 456-463 (1978).

7. Collins FS, Summer GK, Schwartz RP, Parke JC. Neonatal argininosuccinic aciduria-survival after early diagnosis and dietary management. *J Pediatr*. **96**, 429-431 (1980).

8. Collins FS, Orringer EP. Pulmonary hypertension and cor pulmonale in the sickle hemoglobinopathies. *Am J Med*. **73**, 814-821 (1982).

9. Collins FS, Mahoney MJ. Hydrocephalus and abnormal digits after failed first-trimester prostaglandin abortion attempt. *J Pediatr*. **102**, 620-621 (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**, 4469-4479 (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**, 4894-4898 (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**, 1292-1296 (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**, 529-538 (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 Sci USA*. **81**, 6812-6816 (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**, 325-326 (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**, 551-554 (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**, 2551-2557 (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**, 1046-1049 (1987).

20. Treisman J, Collins FS. Adult Turner syndrome associated with chylous ascites and vascular anomalies. *Clin Genet*. **31**, 218-223 (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**, 461-489 (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, Iannazzi JA, Roy JC… Martuza RL, Breakefield XO, Gusella JF. Genetic linkage of von Recklinghausen neurofibromatosis to the nerve growth factor receptor gene. *Cell*. **49**, 589-594 (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**, 540-544 (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**, 532-534 (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**, 1797-1803 (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**, 353-357 (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**, 361-363 (1987).

30. Engelke DR, Hoener PA, Collins FS. Direct sequencing of enzymatically amplified human genomic DNA. *Proc Natl Acad Sci USA*. **85**, 544-548 (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**, 1305-1317 (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**, 149-154 (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**, 884-888 (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**, 1744-1747 (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**, 346-354 (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**, 6437-6441 (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**, 167-172 (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. *Ann Rev Respir Dis*. **138**, 965-968 (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**, 5310-5322 (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**, 13-19 (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**, 33-37 (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**, 51-57 (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**, 1665-1677 (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**, 1085-1087 (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).

50. Collins FS, O'Connell P, Ponder BA, Seizinger BR. Progress towards identifying the neurofibromatosis (NF1) gene. *Trends Genet*. **5**, 217-221 (1989).

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