Committee on Energy and Commerce
U.S. House of Representatives
Witness Disclosure Requirement - "Truth in Testimony"
Required by House Rule XI, Clause 2(g)

1. Your Name: Francis S. Collins, M.D., Ph.D.		
2. Are you testifying on behalf of the Federal, or a State or local government entity?	**Yes** (circled)	**No**
3. Are you testifying on behalf of an entity that is not a government entity?	**Yes**	**No** (circled)
4. Other than yourself, please list which entity or entities you are representing: NIH/HHS		
5. Please list any Federal grants or contracts (including subgrants or subcontracts) that you or the entity you represent have received on or after October 1, 2009:		
6. If your answer to the question in item 3 in this form is "yes," please describe your position or representational capacity with the entity or entities you are representing:		
7. If your answer to the question in item 3 is "yes," do any of the entities disclosed in item 4 have parent organizations, subsidiaries, or partnerships that you are not representing in your testimony?	**Yes**	**No**
8. If the answer to the question in item 3 is "yes," please list any Federal grants or contracts (including subgrants or subcontracts) that were received by the entities listed under the question in item 4 on or after October 1, 2009, that exceed 10 percent of the revenue of the entities in the year received, including the source and amount of each grant or contract to be listed:		
9. Please attach your curriculum vitae to your completed disclosure form.		

Signature: [redacted] **Date:** 19 Jun 2012

Biographical Sketch of Francis S. Collins, M.D., Ph.D.

FRANCIS S. COLLINS, MD, PHD

Francis S. Collins, M.D., Ph.D. is the Director of the National Institutes of Health (NIH). In that role he oversees the work of the largest supporter of biomedical research in the world, spanning the spectrum from basic to clinical research.

Dr. Collins is a physician-geneticist noted for his landmark discoveries of disease genes and his leadership of the international Human Genome Project, which culminated in April 2003 with the completion of a finished sequence of the human DNA instruction book. He served as director of the National Human Genome Research Institute at the NIH from 1993-2008.

Before coming to the NIH, Dr. Collins was a Howard Hughes Medical Institute investigator at the University of Michigan. He is an elected member of the Institute of Medicine and the National Academy of Sciences, was awarded the Presidential Medal of Freedom in November 2007, and received the National Medal of Science in 2009.

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
One Center Drive MSC 0148
Building 1, Room 126
Bethesda, Maryland 20892-0148

Phone: [redacted]
Fax:
E-mail:

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, Genome Technology Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda, MD
2009-Present	Senior Investigator, Genome Technology Branch, 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
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, 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
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
Dept. 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. 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
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
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
Keynote Speaker, University of Michigan Medical School Commencement, 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
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
Cosgrove Lecture, The American College of Obstetricians and Gynecologists 50th Anniversary Meeting, 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
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
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
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 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 and Lecture, 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
Keynote Speaker, AAAS Personalized Medicine: Planning for the Future Colloquium, 2009
Keynote Speaker, mHealth Summit, Foundation for the National Institutes of Health, 2009
H. Richard Nesson Award, Biomedical Science Careers Student Conference, 2009
Keynote Speaker, Society for Neuroscience Annual Meeting, 2009
Keynote Speaker, Cystic Fibrosis Foundation International Conference, 2009
Philip Hauge Abelson Award, American Association for the Advancement of Science, 2009
National Medal of Science, 2009
Keynote Speaker, Annual Meeting of the National Postdoctoral Association, 2010
Co-recipient, Albany Medical Center Prize, 2010

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
Member, National Science and Technology Council (NSTC) Administration Science and Technology Priorities, 2009-2010
Co-chair, National Science and Technology Council Committee on Science (NSTC-CoS), 2009-2010
Member, National Science and Technology Council Intelligence Science Board (NSTC-ISB), 2009-2010
Member, Reagan-Udall Foundation Board, 2009-present
Member, Heads of International Research Organizations (HIRO), 2009-present
Member, Global Health Initiative Strategic Council, 2010-present
Chair, Research Committee
Co-chair, Joint NIH-FDA Leadership Council, 2010-present
Board Member, Patient Centered Outcomes Research Institute, 2010-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).

51. Ledbetter DH, Ledbetter SA, vanTuinen P, Summers KM, Robinson TJ, Nakamura Y, Wolff R, White R, Barker DF, Wallace MR, Collins FS, Dobyns WB. Molecular dissection of a contiguous gene syndrome: frequent submicroscopic deletions, evolutionary conserved sequences, and a hypomethylated "island" in the Miller-Dieker chromosome region. *Proc Natl Acad Sci USA.* **86**, 5136-5140 (1989).

52. Rommens JM, Iannuzzi MC, Kerem B, Drumm JL, Melmer G, Dean M, Rozmahel R, Cole JL, Kennedy D, Hidaka N, Zsiga M, Buchwald M, Riordan JR, Tsui LC, Collins FS. Identification of the cystic fibrosis gene: chromosome walking and jumping. *Science.* **245**, 1059-1065 (1989).

53. Riordan JR, Rommens JM, Kerem B, Alon N, Rozmahel R, Grzelczak Z, Zielenski J, Lok S, Plavic N, Chou JL, Drumm ML, Iannuzzi MC, Collins FS, Tsui LC. Identification of the cystic fibrosis gene: cloning and characterization of complementary DNA. *Science.* **245**, 1066-1073 (1989).

54. Cremers FP, van de Pol DJ, Wieringa B, Collins FS, Sankila EM, Siu VM, Flintoff WF, Brunsmann F, Blonden LA, Ropers HH. Chromosomal jumping from the DXS165 locus allows molecular characterization of four microdeletions and a de novo chromosome X/13 translocation associated with choroideremia. *Proc Natl Acad Sci USA.* **86**, 7510-7514 (1989).

55. Merry DE, Lesko JG, Sosnoski DM, Lewis RA, Lubinsky M, Trask B, van den Engh G, Collins FS, Nussbaum RL. Choroideremia and deafness with stapes fixation: a contiguous gene deletion syndrome in Xq21. *Am J Hum Genet.* **45**, 530-540 (1989).

56. Fain PR, Goldgar DE, Wallace MR, Collins FS, Wright E, Nguyen K, Barker DF. Refined physical and genetic mapping of the NF1 region on chromosome 17. *Am J Hum Genet.* **45**, 721-728 (1989).

57. Nigro JM, Baker SJ, Preisinger AC, Jessup JM, Hostetter R, Cleary K, Bigner SH, Davidson N, Baylin S, Devilee P, Glover TW, Collins FS, Weston A, Modali R, Harris CC, Vogelstein B. Mutations in the p53 gene occur in diverse human tumour types. *Nature*. **342**, 705-708 (1989).

58. Bucan M, Zimmer M, Whaley WL, Poustka A, Youngman S, Allitto BA, Ormondroyd E, Smith B, Pohl TM, MacDonald M, Bates GE, Richards J, Volinia S, Gilliam TC, Sedlacek Z, Collins FS, Wasmuth JJ, Shaw DJ, Gusella JF, Frischauf AM, Lehrrach H. Physical maps of 4p16.3, the area expected to contain the Huntington disease mutation. *Genomics*. **6**, 1-15 (1990).

59. Gumucio DL, Lockwood WK, Weber JL, Saulino AM, Delgrosso K, Surrey S, Schwartz E, Goodman M, Collins FS. The –175T----C mutation increases promoter strength in erythroid cells: correlation with evolutionary conservation of binding sites for two trans-acting factors. *Blood*. **75**, 756-761 (1990).

60. Lemna WK, Feldman GL, Kerem B, Fernbach SD, Zevkovich EP, O'Brien WE, Riordan JR, Collins FS, Tsui LC, Beaudet AL. Mutation analysis for heterozygote detection and the prenatal diagnosis of cystic fibrosis. *N Engl J Med*. **322**, 291-296 (1990).

61. Merry DE, Lesko JG, Siu V, Flintoff WF, Collins FS, Lewis RA, Nussbaum RL. DXS165 detects a translocation breakpoint in a woman with choroideremia and a de novo X; 13 translocation. *Genomics*. **6**, 609-615 (1990).

62. Patel PI, Ledbetter DH, Frances S, Franco B, Wallace MR, Collins FS, Lupski JR. Isolation of a polymorphic DNA sequence (LL101) from the short arm of chromosome 17 [D17S251]. *Nucleic Acids Res*. **18**, 1087 (1990).

63. Donlon TA, Krensky AM, Wallace MR, Collins FS, Lovett M, Clayberger C. Localization of a human T-Cell-specific gene, RANTES (D17S136E), to chromosome 17q11.2-q12. *Genomics*. **6**, 548-553 (1990).

64. Dean M, Drumm ML, Stewart C, Gerrard B, Perry A, Hidaka N, Cole JL, Collins FS, Iannuzzi MC. Approaches to localizing disease genes as applied to cystic fibrosis. *Nucleic Acids Res*. **18**, 345-350 (1990).

65. Ledbetter SA, Wallace MR, Collins FS, Ledbetter DH. Human chromosome 17 NotI linking clones and their use in long-range restriction mapping of the Miller-Dieker chromosome region (MDCR) in 17p13.3. *Genomics*. **7**, 264-269 (1990).

66. Wallace MR, Marchuk DA, Andersen LB, Letcher R, Odeh HM, Saulino AM, Fountain JW, Brereton A, Nicholson J, Mitchell AL, Collins FS. Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients. *Science*. **249**, 181-186 (1990).

67. Lux SE, Tse WT, Menninger JC, John KM, Harris P, Shalev O, Chilcote RR, Marchesi SL, Watkins PC, Bennett V, McIntosh S, Collins FS, Francke U, Ward DC, Forget BG. Hereditary spherocytosis associated with deletion of the human erythrocyte ankyrin gene on chromosome 8. *Nature*. **345**, 736-739 (1990).

68. Walker AP, Collins FS, Siddique T, Yamaoka LH, Herbstreith MH, Pericak-Vance MA, Secore SL, Hung WY, Goate AM, Hardy JA, Patterson D, Roses AD, Bartlett RJ. D21S194, a jump clone from D21S16. *Nucleic Acids Res*. **18**, 1931 (1990).

69. O'Connell P, Viskochil D, Buchberg AM, Fountain J, Cawthon RM, Culver M, Stevens J, Rich DC, Ledbetter DH, Wallace M, Carey JC, Jenkins NA, Copeland NG, Collins FS, White RL. The human homolog of murine Evi-2 lies between two von Recklinghausen neurofibromatosis translocations. *Genomics*. **7**, 547-554 (1990).

70. Gray TA, Gumucio DL, Collins FS. A two-dimensional system for DNA binding proteins. *Technique*. **2**, 147-154 (1990).

71. Drumm ML, Pope HA, Cliff WH, Rommens JM, Marvin SA, Tsui LC, Collins FS, Frizzell RA, Wilson JM. Correction of the cystic fibrosis defect in vitro by retrovirus-mediated gene transfer. *Cell*. **62**, 1227-1233 (1990).

72. Rabbitts P, Bergh J, Douglas J, Collins FS, Waters J. A submicroscopic homozygous deletion at the D3S3 locus in a cell line isolated from a small lung carcinoma. *Genes Chromosomes Cancer*. **2**, 231-238 (1990).

73. Dean M, Amos JA, Lynch J, Romeo G, Devoto M, Ward K, Halley D, Oostra B, Ferrari M, Russo S, Weir BS, Finn PB, Collins FS, Iannuzzi MC. Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. *Hum Genet.* **85**, 275-278 (1990).

74. Beaudet AL, Kazazian HH, Bowman JE, Chakravarti A, Collins FS, Davis JG, Davis PB, Desnick RJ, Elias S, Fost N, Holtzman NA, Kaback M, Klinger K, Reilly PR, Rosenstein B, Rowley PT, Short EM, Smith ACM, Sorenson JR, Tsui LC, Williamson R, Lamontagne N. Statement from the National Institutes of Health workshop on population screening for the cystic fibrosis gene. *N Engl J Med.* **323**, 70-71 (1990).

75. Ballester R, Marchuk D, Boguski M, Saulino A, Letcher R, Wigler M, Collins FS. The NF1 locus encodes a protein functionally related to mammalian GAP and yeast IRA proteins. *Cell.* **63**, 851-859 (1990).

76. Kerem BS, Zielenski J, Markiewicz D, Bozon D, Gazit E, Yahav J, Kennedy D, Riordan JR, Collins FS, Rommens JM, Tsui LC. Identification of mutations in regions corresponding to the two putative nucleotide (ATP)-binding folds of the cystic fibrosis gene. *Proc Natl Acad Sci USA.* **87**, 8447-8451 (1990).

77. Weber JL, Kwitek AE, May PE, Wallace MR, Collins FS, Ledbetter DH. Dinucleotide repeat polymorphisms at the D17S250 and D17S261 loci. *Nucleic Acids Res.* **18**, 4640 (1990).

78. Wallace MR, Andersen LB, Fountain JW, Odeh HM, Viskochil D, Marchuk DA, O'Connell P, White R, Collins FS. A chromosome jump crosses a translocation breakpoint in the von Recklinghausen neurofibromatosis region. *Genes Chromosomes Cancer.* **2**, 271-277 (1990).

79. Upadhyaya M, Cheryson A, Broadhead W. Fryer A, Shaw DJ, Huson S, Wallace MR, Andersen LB, Marchuk DA, Viskochil D, Balck D, O'Connell P, Collins FS, Harper PS. A 90 kb DNA deletion associated with neurofibromatosis type 1. *J Med Genet.* **27**, 738-741 (1990).

80. Andersen LB, Wallace MR, Marchuk DA, Cawthon RM, Odeh HM, Letcher R, White RL, Collins FS. A polymorphic cDNA probe on chromosome 17q11.2 located within the NF1 gene [D17S376]. *Nucleic Acids Res.* **19**, 197 (1991).

81. Iannuzzi MC, Stern RC, Collins FS, Hon CT, Hidaka N, Strong T, Becker L, Drumm ML, White MB, Gerrard B, Dean M. Two frameshift mutations in the cystic fibrosis gene. *Am J Hum Genet.* **48**, 227-231 (1991).

82. Allitto BA, MacDonald ME, Bucan M, Richards J, Romano D, Whaley WL, Falcone B, Ianazzi J, Wexler NS, Wasmuth JJ, Collins FS, Lehrach H, Haines JL, Gusella JF. Increased recombination adjacent to the Huntington disease-linked D4S10 marker. *Genomics.* **9**, 104-112 (1991).

83. Iannuzzi MC, Hidaka N, Boehnke M, Bruck ME, Hanna WT, Collins FS, Ginsburg D. Analysis of the relationship of von Willebrand disease (vWD) and hereditary hemorrhagic telangiectasia and identification of a potential type IIA vWD mutation (IIe865 to Thr). *Am J Hum Genet.* **48**, 757-763 (1991).

84. Cawthon RM, Andersen LB, Buchberg AM, Xu GF, O'Connell P, Viskochil D, Weiss RB, Wallace MR, Marchuk DA, Culver M, Jenkins NA, Copeland NG, Collins FS, White RL. cDNA sequence and genomic structure of EV12B, a gene lying within an intron of the neurofibromatosis type 1 gene. *Genomics.* **9**, 446-460 (1991).

85. Marchuk D, Drumm M, Saulino A, Collins FS. Construction of T-vectors, a rapid and general system for direct cloning of unmodified PCR products. *Nucleic Acids Res.* **19**, 1154 (1991).

86. Andersen LB, Wallace MR, Marchuk DA, Tavakkol R, Mitchell A, Saulino AM, Collins FS. A highly polymorphic cDNA probe in the NF1 gene. *Nucleic Acids Res.* **19**, 3754 (1991).

87. Kainulainen K, Steinmann B, Collins FS, Dietz HC, Francomano CA, Child A, Kilpatrick MW, Brock DJ, Keston M, Pyeritz RE, Peltonen L. Marfan syndrome: no evidence for heterogeneity in different populations, and more precise mapping of the gene. *Am J Hum Genet.* **49**, 662-667 (1991).

88. Gibson AL, Wagner LM, Collins FS, Oxender DL. A bacterial system for investigating transport effects of cystic fibrosis-associated mutations. *Science*. **254**, 109-111 (1991).

89. Shukla H, Gillespie GA, Srivastava R, Collins FS, Chorney MJ. A class I jumping clone places the HLA-G gene approximately 100 kilobases from HLA-H within the HLA-A subregion of the human MHC. *Genomics*. **10**, 905-914 (1991).

90. Gumucio DL, Rood KL, Blanchard-McQuate KL, Gray TA, Saulino A, Collins FS. Interaction of Sp1 with the human gamma globin promoter: binding and transactivation of normal and mutant promoters. *Blood*. **78**, 1853-1863 (1991).

91. Wallace MR, Andersen LB, Saulino AM, Gregory PE, Glover TW, Collins FS. A de novo Alu insertion results in neurofibromatosis type 1. *Nature*. **353**, 864-866 (1991).

92. Gutmann DH, Wood DL, Collins FS. Identification of the neurofibromatosis type 1 gene product. *Proc Natl Acad Sci USA*. **88**, 9658-9662 (1991).

93. Marchuk DA, Saulino AM, Tavakkol R, Swaroop M, Wallace MR, Andersen LB, Mitchell AL, Gutmann DH, Boguski M, Collins FS. cDNA cloning of the type 1 neurofibromatosis gene: complete sequence of the NF1 gene product. *Genomics*. **11**, 931-940 (1991).

94. Goldberg NS, Collins FS. The hunt for the neurofibromatosis gene. *Arch Dermatol*. **127**, 1705-1707 (1991).

95. Strong TV, Smit LS, Turpin SV, Cole JL, Hon Ct, Markiewicz D, Petty TL, Craig MW, Rosenow EC 3rd, Tsui LC, Iannuzzi MC, Knowles MR, Collins FS. Cystic fibrosis gene mutation in two sisters with mild disease and normal sweat electrolyte levels. *N Engl J Med*. **325**, 1630-1634 (1991).

96. Drumm ML, Wilkinson DJ, Smit LS, Worrell RT, Strong TV, Frizzell RA, Dawson DC, Collins FS. Chloride conductance expressed by delta F508 and other mutant CFTRs in Xenopus oocytes. *Science*. **254**, 1797-1799 (1991).

97. Ton CC, Hirvonen H, Miwa H, Weil MM, Monaghan P, Jordan T, van Heyningen V, Hastie ND, Meijers-Heijboer H, Drechsler M, Collins FS, Swaroop A, Strong LC, Saunders GF. Positional cloning and characterization of a paired box- and homeobox-containing gene from the aniridia region. *Cell*. **67**, 1059-1074 (1991).

98. Collins FS. Medical and ethical consequences of the Human Genome Project. *J Clinical Ethics*. **2**, 261-267 (1991).

99. Biesecker L, Bowles-Biesecker B, Collins FS, Kaback M, Wilfond B. General population screening for cystic fibrosis is premature. *Am J Hum Genet*. **50**, 438-439 (1992).

100. Krauss RD, Bubien JK, Drumm ML, Zheng T, Peiper SC, Collins FS, Kirk KL, Frizzell RA, Rado TA. Transfection of wild-type CFTR into cystic fibrosis lymphocytes restores chloride conductance at G1 of the cell cycle. *EMBO J*. **11**, 875-883 (1992).

101. Gusella JF, Altherr MR, McClatchey AI, Doucette-Stamm LA, Tagle D, Plummer S, Groot N, Barnes G, Hummerich H, Collins FS, Housman De, Lehrrach H, Macdonald ME, Bates GP, Wasmuth JJ. Sequence-tagged sites (STSs) spanning 4p16.3 and the Huntington disease candidate region. *Genomics*. **13**, 75-80 (1992).

102. Collins FS. Positional cloning: let's not call it reverse anymore. *Nat Genet*. **1**, 3-6 (1992).

103. Basu TN, Gutmann DH, Fletcher JA, Glover TW, Collins FS, Downward J. Aberrant regulation of ras proteins in malignant tumour cells from type 1 neurofibromatosis patients. *Nature*. **356**, 713-715 (1992).

104. MacDonald ME, Novelletto A, Lin C, Tagle D, Barnes G, Bates G, Taylor S, Allitto B, Altherr M, Myers R, Lehrach H, Collins FS, Wasmuth JJ, Frontali M, Gusella JF. The Huntington's disease candidate region exhibits many different haplotypes. *Nat Genet.* **1**, 99-103 (1992).

105. Collins FS. Cystic fibrosis: molecular biology and therapeutic implications. *Science.* **256**, 774-779 (1992).

106. Bates GP, Valdes J, Hummerich H, Baxendale S, Le Paslier DL, Monaco AP, Tagle D, MacDonald ME, Altherr M, Ross M, Wasmuth JJ, Gusella JF, Cohen D, Collins FS, Lehrach H. Characterization of a yeast artificial chromosome contig spanning the Huntington's disease gene candidate region. *Nat Genet.* **1**, 180-187 (1992).

107. Tagle DA, Collins FS. An optimized Alu-PCR primer pair for human-specific amplification of YACs and somatic cell hybrids. *Hum Mol Genet.* **1**, 121-122 (1992).

108. Gutmann DH, Collins FS. Recent progress toward understanding the molecular biology of von Recklinghausen neurofibromatosis. *Ann Neurol.* **31**, 555-561 (1992).

109. Marchuk DA, Tavakkol R, Wallace MR, Brownstein BH, Taillon-Miller P, Fong CT, Legius E. Andersen LB, Glover TW, Collins FS. A yeast artificial chromosome contig encompassing the type 1 neurofibromatosis gene. *Genomics.* **13**, 672-680 (1992).

110. Tagle DA, Blanchard-McQuate KL, Collins FS. Dinucleotide repeat polymorphism in the Huntington's disease region at the D4S43 locus. *Hum Mol Genet.* **1**, 215 (1992).

111. Legius E, Marchuk DA, Hall BK, Andersen LB, Wallace MR, Collins FS, Glover TW. NF1-related locus on chromosome 15. *Genomics.* **13**, 1316-1318 (1992).

112. Snell RG, Thompson LM, Tagle DA, Holloway TL, Barnes G, Harley HG, Sandkuijl LA, MacDonald ME, Collins FS, Gusella JF, Harper PS, Shaw DJ. A recombination event that redefines the Huntington disease region. *Am J Hum Genet.* **51**, 357-362 (1992).

113. McCombie WR, Martin-Gallardo A, Gocayne JD, FitzGerald M, Dubnick M, Kelley JM, Castilla L, Liu LI, Wallace S, Trapp S, Tagle DA, Whaley WL, Cheng S, Gusella JF, Frischauf AM, Poustka A, Lehrach H, Collins FS, Kerlavage AR, Fields C, Venter JC. Expressed genes, Alu repeats and polymorphisms in cosmids sequenced from chromosome 4p16.3. *Nat Genet.* **1**, 348-353 (1992).

114. Gumucio DL, Heilstedt-Williamson H, Gray TA, Tarle SA, Shelton DA, Tagle DA, Slightom JL, Goodman M, Collins FS. Phylogenetic footprinting reveals a nuclear protein which binds to silencer sequences in the human gamma and epsilon globin genes. *Mol Cell Biol.* **12**, 4919-4929 (1992).

115. Kayes LM, Schroeder WT, Marchuk DA, Collins FS, Riccardi VM, Duvic M, Stephens K. The gene for a novel epidermal antigen maps near the neurofibromatosis 1 gene. *Genomics.* **14**, 369-376 (1992).

116. Kamei T, Fukushima Y, Shibata A, Hayashi Y, Tachibana N, Takeda I, Niikawa N, Collins FS, Takahashi K, Masumura S. DNA deletion in patients with von Recklinghausen neurofibromatosis. *Clin Genet.* **42**, 53-54 (1992).

117. Andersen LB, Ballester R, Marchuk DA, Chang E, Gutmann DH, Saulino AM, Camonis J, Wigler M, Collins FS. A conserved alternative splice in the von Recklinghausen neurofibromatosis (NF1) gene produces two neurofibromin isoforms, both of which have GTPase-activating protein activity. *Mol Cell Biol.* **13**, 487-495 (1993).

118. Andersen LB, Fountain JW, Gutmann DH, Tarle SA, Glover TW, Dracopoli NC, Housman DE, Collins FS. Mutations in the neurofibromatosis 1 gene in sporadic malignant melanoma cell lines. *Nat Genet.* **3**, 118-121 (1993).

119. Legius E, Marchuk DA, Collins FS, Glover TW. Somatic deletion of the neurofibromatosis type 1 gene in a neurofibrosarcoma supports a tumour suppressor gene hypothesis. *Nat Genet.* **3**, 122-126 (1993).

120. Gutmann DH, Boguski M, Marchuk D, Wigler M, Collins FS, Ballester R. Analysis of the neurofibromatosis type 1 (NF1) GAP-related domain by site-directed mutagenesis. *Oncogene.* **8**, 761-769 (1993).

121. Strong TV, Smit LS, Nasr SZ, Wood DL, Cole JL, Iannuzzi MC, Stern R, Collins FS. Characterization of an intron 12 splice donor mutation in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. *Hum Mutat.* **1**, 380-387 (1993).

122. Martin-Gallardo A, Marchuk DA, Kerlavage AR, Venter JG, Collins FS, Wallace MR. Sequencing and analysis of genomic fragments from the NF1 locus. *DNA Sequence.* **3**, 237-243 (1993).

123. Strong TV, Wilkinson DJ, Mansoura MK, Devor DC, Henze K, Yang Y, Wilson JM, Cohn JA, Dawson DC, Frizzell RA, Collins FS. Expression of an abundant alternatively spliced form of the cystic fibrosis transmembrane conductance regulator (CFTR) gene is not associated with a cAMP-activated chloride conductance. *Hum Mol Genet.* **2**, 225-230 (1993).

124. The Huntington's Disease Collaborative Research Group. A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell.* **72**, 971-983 (1993).

125. Gutmann DH, Collins FS. The neurofibromatosis type 1 gene and its protein product, neurofibromin. *Neuron.* **10**, 335-343 (1993).

126. Chandrasekharappa SC, Gross LA, King SE, Collins FS. The human NME2 gene lies within 18kb of NME1 in chromosome 17. *Genes Chromosomes Cancer.* **6**, 245-248 (1993).

127. Biesecker BB, Boehnke M, Calzone K, Markel DS, Garber JE, Collins FS, Weber BL. Genetic counseling for families with inherited susceptibility to breast and ovarian cancer. *JAMA.* **269**, 1970-1974 (1993).

128. Chamberlain JS, Boehnke M, Frank TS, Kiousis S, Xu J, Guo SW, Hauser ER, Norum RA, Helmbold EA, Markel DS, Keshavarzi SM, Jackson CE, Calzone K, Garber J, Collins FS, Weber BL. BRCA1 maps proximal to D17S579 on chromosome 17q21 by genetic analysis. *Am J Hum Genet.* **52**, 792-798 (1993).

129. Tagle DA, Blanchard-McQuate KL, Valdes J, Castilla L, MacDonald ME, Gusella JF, Collins FS. Dinucleotide repeat polymorphism in the Huntington's disease region at the D4S182 locus. *Hum Mol Genet.* **2**, 489 (1993).

130. Smit LS, Nasr SZ, Iannuzzi MC, Collins FS. An African-American cystic fibrosis patient homozygous for a novel frameshift mutation associated with reduced CFTR mRNA levels. *Hum Mutat.* **2**, 148-151 (1993).

131. Valdes JM, Tagle DA, Elmer LW, Collins FS. A simple non-radioactive method for diagnosis of Huntington's disease. *Hum Mol Genet.* **2**, 633-634 (1993).

132. Gutmann DH, Andersen LB, Cole JL, Swaroop M, Collins FS. An alternatively-spliced mRNA in the carboxy terminus of the neurofibromatosis type 1 (NF1) gene is expressed in muscle. *Hum Mol Genet.* **2**, 989-992 (1993).

133. Gregory PE, Gutmann DH, Mitchell A, Park S, Boguski M, Jacks T, Wood DL, Jove R, Collins FS. Neurofibromatosis type 1 gene product (neurofibromin) associates with microtubules. *Somat Cell Mol Genet.* **19**, 265-274 (1993).

134. Tagle DA, Blanchard-McQuate KL, Valdes J, Castilla L, Collins FS. Dinucleotide repeat polymorphisms at the D4S126 and D4S114 loci. *Hum Mol Genet.* **2**, 1077 (1993).

135. Andersen LB, Tarle SA, Marchuk DA, Legius E, Collins FS. A compound nucleotide repeat in the neurofibromatosis (NF1) gene. *Hum Mol Genet.* **2**, 1083 (1993).

136. Liu P, Claxton DF, Marlton P, Hajra A, Siciliano J, Freedman M, Chandrasekharappa SC, Yanagisawa K, Stallings RL, Collins FS, Siciliano MJ. Identification of yeast artificial chromosomes containing the inversion 16 p-arm breakpoint associated with acute myelomonocytic leukemia. *Blood.* **82**, 716-721 (1993).

137. Koh J, Sferra TJ, Collins FS. Characterization of the cystic fibrosis transmembrane conductance regulator promoter region: Chromatin context and tissue-specificity. *J Biol Chem.* **268**, 15912-15921 (1993).

138. Yang Y, Devor DC, Engelhardt JF, Ernst SA, Strong TV, Collins FS, Cohn JA, Frizzell RA, Wilson JM. Molecular basis of defective anion transport in L cells expressing recombinant forms of CFTR. *Hum Mol Genet.* **2**, 1253-1261 (1993).

139. Liu P, Tarle SA, Hajra A, Claxton DF, Marlton P, Freedman M, Siciliano MJ, Collins FS. Fusion between transcription factor CBF beta/PEBP2 beta and a myosin heavy chain in acute myeloid leukemia. *Science.* **261**, 1041-1044 (1993).

140. Gutmann DH, Tennekoon GI, Cole JL, Collins FS, Rutkowski JL. Modulation of the neurofibromatosis type 1 gene product, neurofibromin, during Schwann cell differentiation. *J Neurosci Res.* **36**, 216-223 (1993).

141. Flejter WL, Barcroft CL, Guo SW, Lynch ED, Boehnke M, Chandrasekharappa SC, Hayes S, Collins FS, Weber BL, Glover TW. Multicolor FISH mapping with Alu-PCR-amplified YAC clone DNA determines the order of markers in the BRCA1 region on chromosome 17q12-q21. *Genomics.* **17**, 624-631 (1993).

142. Deng Z, Liu P, Marlton P, Claxton DF, Lane S, Callen DF, Collins FS, Siciliano MJ. Smooth muscle myosin heavy chain locus (MYH11) maps to 16p13.13-p13.12 and establishes a new region of conserved synteny between human 16p and mouse 16. *Genomics.* **18**, 156-159 (1993).

143. Coleman SD, Collins FS, Wallace MR. Characterization of a single base-pair deletion in neurofibromatosis type 1. *Hum Mol Genet.* **2**, 1709-1711 (1993).

144. Abel KJ, Boehnke M, Prahalad M, Ho P, Flejter WL, Watkins M, VanderStoep J, Chandrasekharappa SC, Collins FS, Glover TW, Weber BL. A radiation hybrid map of the BRCA1 region of chromosome 17q12-q21. *Genomics.* **17**, 632-641 (1993).

145. Strong TV, Tagle DA, Valdes JM, Elmer LW, Boehm K, Swaroop M, Kaatz KW, Collins FS, Albin RL. Widespread expression of the human and rat Huntington's disease gene in brain and nonneural tissues. *Nat Genet.* **5**, 259-265 (1993).

146. Gutmann DH, Collins FS. Neurofibromatosis type 1: beyond positional cloning. *Arch Neurol.* **50**, 1185-1193 (1993).

147. Flejter WL, Watkins M, Abel KJ, Chandrasekharappa SC, Weber BL, Collins FS, Glover TW. Isolation and characterization of somatic cell hybrids with breakpoints spanning 17q22-->q24. *Cytogenet Cell Genet.* **64**, 222-223 (1993).

148. Guyer MS, Collins FS. The Human Genome Project and the future of medicine. *Am J Dis Child.* **147**, 1145-1152 (1993).

149. Gutmann DH, Tennekoon GI, Cole JL, Collins FS, Rutkowski JL. Modulation of the neurofibromatosis type 1 gene product, neurofibromin, during Schwann cell differentiation. *J Neurosci Res.* **36**, 216-223 (1993).

150. Chandrasekharappa SC, King SE, Freedman ML, Hayes ST, Bowcock AM, Collins FS. The CA repeat marker D17S791 is located within 40 kb of the WNT3 gene on chromosome 17q. *Genomics.* **18**, 728-729 (1993).

151. Strong TV, Boehm K, Collins FS. Localization of cystic fibrosis transmembrane conductance regulator mRNA in the human gastrointestinal tract by in situ hybridization. *J Clin Invest.* **193**, 347-354 (1993).

152. Cohn JA, Strong TV, Picciotto MR, Nairn AC, Collins FS, Fitz JG. Localization of the cystic fibrosis transmembrane conductance regulator in human bile duct epithelial cells. *Gastroenterology.* **105**, 1857-1864 (1993).

153. Smit LS, Wilkinson DJ, Mansoura MK, Collins FS, Dawson DC. Functional roles of the nucleotide-binding folds in the activation of the cystic fibrosis transmembrane conductance regulator. *Proc Natl Acad Sci USA*. **90**, 9963-9967 (1993).

154. McDonald MT, Papenberg KA, Ghosh S, Glatfelter AA, Biesecker BB, Helmbold EA, Markel DS, Zolotor A, McKinnon WC, Vanderstoep JL, Jackson CE, Iannuzzi M, Collins FS, Boehnke M, Porteous ME, Guttmacher AE, Marchuk DA. A disease locus for hereditary haemorrhagic telangiectasia maps to chromosome 9q33-34. *Nat Genet*. **6**, 197-204 (1994).

155. Horiuchi T, Hatta N, Matsumoto M, Ohtsuka H, Collins FS, Kobayashi Y, Fujita S. Nonsense mutations at Arg-1947 in two cases of familial neurofibromatosis type 1 in Japanese. *Hum Genet*. **93**, 81-83 (1994).

156. National Advisory Council for Human Genome Research. Statement on use of DNA testing for presymptomatic identification of cancer risk. *JAMA*. **271**, 785 (1994).

157. Claxton DF, Liu P, Hsu HB, Marlton P, Hester J, Collins FS, Deisseroth AB, Rowley JD, Siciliano MJ. Detection of fusion transcripts generated by the inversion 16 chromosome in acute myelogenous leukemia. *Blood*. **83**, 1750-1756 (1994).

158. Gutmann DH, Cole JL, Stone WJ, Ponder BA, Collins FS. Loss of neurofibromin in adrenal gland tumors from patients with neurofibromatosis type I. *Genes Chromosomes Cancer*. **10**, 55-58 (1994).

159. Legius E, Hall BK, Wallace MR, Collins FS, Glover TW. Ten base pair duplication in exon 38 of the NF1 gene. *Hum Mol Genet*. **3**, 829-830 (1994).

160. Chandrasekharappa SC, Friedman L, King SE, Lee YH, Welsch P, Bowcock AM, Weber BL, King MC, Collins FS. The gene for pancreatic polypeptide (PPY) and the anonymous marker D17S78 are within 45 kb of each other on chromosome 17q21. *Genomics*. **21**, 458-460 (1994).

161. Nakamura T, Nemoto T, Arai M, Yamazaki Y, Kasuga T, Gutmann DH, Collins FS, Ishikawa T. Specific expression of the neurofibromatosis type 1 gene (NF1) in the hamster Schwann cell. *Am J Pathol*. **144**, 549-555 (1994).

162. Couch FJ, Abel KJ, Brody LC, Boehnke M, Collins FS, Weber BL. Localization of the gene for ATP citrate lyase (ACLY) distal to gastrin (GAS) and proximal to D17S856 on chromosome 17q12-q21. *Genomics*. **21**, 444-446 (1994).

163. Hajra A, Martin-Gallardo A, Tarle SA, Freedman M, Wilson-Gunn S, Bernards A, Collins FS. DNA sequences in the promoter region of the NF1 gene are highly conserved between human and mouse. *Genomics*. **21**, 649-652 (1994).

164. Valdes JM, Tagle DA, Collins FS. Island rescue PCR: a rapid and efficient method for isolating transcribed sequences from yeast artificial chromosomes and cosmids. *Proc Natl Acad Sci USA*. **91**, 5377-5381 (1994).

165. Sartin EA, Doran SE, Riddell MG, Herrera GA, Tennyson GS, D'Andrea G, Whitley RD, Collins FS. Characterization of naturally occurring cutaneous neurofibromatosis in Holstein cattle: a disorder resembling neurofibromatosis type 1 in humans. *Am J Pathol*. **145**, 1168-1174 (1994).

166. Nasr SZ, Strong TV, Collins FS. Identification of a CFTR frameshift mutation (1013 delAA) in trans to delta F508 in a pancreatic sufficient cystic fibrosis patient. *Hum Mol Genet*. **3**, 2063-2064 (1994).

167. Rotman G, Vanagaite L, Collins FS, Shiloh Y. Three dinucleotide repeat polymorphisms at the ataxia-telangiectasia locus. *Hum Mol Genet*. **3**, 2079 (1994).

168. Rotman G, Savitsky K, Ziv Y, Cole CG, Higgins MJ, Bar-Am I, Dunham I, Bar-Shira A, Vanagaite L, Qin S, Nowak NJ, Chandrasekharappa SC, Lehrrach H, Avivi L, Shows TB, Collins FS, Bentley DR, Shiloh Y. A YAC

contig spanning the ataxia-telangiectasia locus (groups A and C) at 11q22-23. *Hum Molec Genet.* **24**, 234-242 (1994).

169. Vanagaite L, Savitsky K, Rotman G, Ziv Y, Gerken SC, White R, Weissenbach J, Gillett G, Benham FJ, Richard CW 3rd, Collins FS, Shiloh Y. Physical localization of microsatellite markers at the ataxia-telangiectasia locus at 11q22-q23. *Genomics.* **22**, 231-233 (1994).

170. Castilla LH, Couch FJ, Erdos MR, Hoskins KF, Calzone K, Garber JE, Boyd J, Lubin MB, Deshano ML, Brody LC, Collins FS, Weber BL. Mutations in the BRCA1 gene in families with early-onset breast and ovarian cancer. *Nat Genet.* **8**, 387-391 (1994).

171. Weber BL, Abel KJ, Brody LC, Flejter WL, Chandrasekharappa SC, Couch FJ, Merajver SD, Collins FS. Familial breast cancer: approaching the isolation of a susceptibility gene. *Cancer.* **74**, 1013-1020 (1994).

172. Couch FJ, Kiousis S, Castilla LH, Xu J, Chandrasekharappa SC, Chamberlain JS, Collins FS, Weber BL. Characterization of 10 new polymorphic dinucleotide repeats and generation of a high-density microsatellite-based physical map of the BRCA1 region of chromosome 17q21. *Genomics.* **24**, 419-424 (1994).

173. Gumucio DL, Shelton DA, Blanchard-McQuate K, Gray T, Tarle S, Heilstedt-Williamson H, Slightom JL, Collins FS, Goodman M. Differential phylogenetic footprinting as a means to identify base changes responsible for recruitment of the anthropoid gamma gene to a fetal expression pattern. *J Biol Chem.* **269**, 15371-15380 (1994).

174. Brody LC, Abel KJ, Castilla LH, Couch FJ, McKinley DR, Yin G, Ho PP, Merajver S, Chandrasekharappa SC, Xu J, Cole JL, Struewing JP, Valdes JM, Collins FS, Weber BL. Construction of a transcription map surrounding the BRCA1 locus of human chromosome 17. *Genomics.* **25**, 238-247 (1995).

175. Malton P, Claxton DF, Liu P, Estey EH, Beran M, LeBeau M, Testa JR, Collins FS, Siciliano MJ. Molecular characterization of 16p deletions associated with inversion 16 defines the critical fusion for leukemogenesis. *Blood.* **85**, 772-779 (1995).

176. Smit LS, Strong TV, Wilkinson DJ, Macek M Jr, Mansoura MK, Wood DL, Cole JL, Cutting GR, Cohn JA, Dawson DC, Collins FS. Missense mutation (G480C) in the CFTR gene associated with protein mislocalization but normal chloride channel activity. *Hum Mol Genet.* **4**, 269-273 (1995).

177. Hajra A, Liu PP, Wang Q, Kelley CA, Stacy T, Adelstein RS, Speck NA, Collins FS. The leukemic core binding factor beta-smooth muscle myosin heavy chain (CBF beta-SMMHC) chimeric protein requires both CBF beta and myosin heavy chain domains for transformation of NIH 3T3 cells. *Proc Natl Acad Sci USA.* **92**, 1926-1930 (1995). Retraction in: Adelstein RS, Collins FS, Hajra A, Kelley CA, Liu PP, Speck NA, Stacy T, Wang Q. *Proc Natl Acad Sci USA.* **93**, 15523 (1996).

178. Collins FS. Positional cloning moves from perditional to traditional. *Nat Genet.* **9**, 347-350 (1995).

179. Merajver SD, Pham TM, Caduff RF, Chen M, Poy EL, Cooney KA, Weber BL, Collins FS, Johnston C, Frank TS. Somatic mutations in the BRCA1 gene in sporadic ovarian tumours. *Nat Genet.* **9**, 439-443 (1995).

180. Wijmenga C, Speck NA, Dracopoli NC, Hofker MH, Liu P, Collins FS. Identification of a new murine runt domain-containing gene, Cbfa3, and localization of the human homolog, CBFA3, to chromosome 1p35-pter. *Genomics.* **26**, 611-614 (1995).

181. Hajra A, Collins FS. Structure of the leukemia-associated human CBFB gene. Genomics 26, 571-579 (1995). Retraction in Hajra A, Collins FS. *Genomics.* **38**, 107 (1996).

182. Merajver SD, Frank TS, Xu J, Pham TM, Calzone KA, Bennett-Baker P, Chamberlain J, Boyd J, Garber JE, Collins FS, Weber BL. Germline BRCA1 mutations and loss of the wild-type allele in tumors from families with early onset breast and ovarian cancer. *Clin Cancer Res.* **1**, 539-544 (1995).

183. Couch FJ, Garber J, Kiousis S, Calzone K, Hauser ER, Merajver SD, Frank TS, Boehnke M, Chamberlain JS, Collins FS, Weber BL. Genetic analysis of eight breast-ovarian cancer families with suspected BRCA1 mutations. *J Natl Cancer Inst Monogr.* **17**, 9-14 (1995).

184. Weber BL, Abel KJ, Couch FJ, Merajver SD, Chandrasekharappa SC, Castilla L, McKinley D, Ho PP, Calzone K, Thomas FS, Xu J, Brody LC, Collins FS. Progress toward isolation of a breast cancer susceptibility gene. BRCA1. *Cold Spring Harbor Symp Quant Biol.* **59**, 531-536 (1995).

185. Liu P, Siedel N, Bodine D, Speck N, Tarle S, Collins FS. Acute myelocytic leukemia with inv(16) produces a chimeric transcription factor with a myosin heavy chain tail. *Cold Spring Harbor Symp Quant Biol.* **59**, 547-553 (1995).

186. Vanagaite L, James MR, Rotman G, Savitsky K, Bar-Shira A, Gilad S, Ziv Y, Uchenik V, Sartiel A, Collins FS, Sheffield VC, Richard CW, Weissenbach J, Shiloh Y. A high-density microsatellite map of the ataxia-telangiectasia locus. *Hum Genet.* **95**, 451-454 (1995).

187. Shurtleff SA, Meyers S, Hiebert SW, Raimondi SC, Head DR, Willman CL, Wolman S, Slovak ML, Carroll AJ, Behm F, Hulshof MG, Motroni TA, Okuda T, Liu P, Collins FS, Downing JR. Heterogeneity of CBF beta/MYH11 fusion messages encoded by the inv(16)(p13q22) and the t(16;16)(p13;q22) in acute myelogenous leukemia. *Blood.* **85**, 3695-3703 (1995).

188. Struewing JP, Brody LC, Erdos MR, Kase RG, Giambarresi TR, Smith SA, Collins FS, Tucker MA. Detection of eight BRCA1 mutations in 10 breast/ovarian cancer families, including one family with male breast cancer. *Am J Hum Genet.* **57**, 1-7 (1995).

189. Liu PP, Hajra A, Wijmenga C, Collins FS. Molecular pathogenesis of the chromosome 16 inversion in the M4Eo subtype of acute myeloid leukemia. *Blood.* **85**, 2289-302 (1995). One paragraph in this review has been withdrawn; see *Blood.* **89**, 1842 (1997).

190. Savitsky K, Bar-Shira A, Gilad S, Rotman G, Ziv Y, Vanagaite L, Tagle DA, Smith S, Uziel T, Sfez S, Ashkenazi M, Pecker I, Frydman M, Harnik R, Patanjali SR, Simmons A, Clines GA, Sartiel A, Gatti RA, Chesa L, Sanal O, Lavin MF, Jaspers NGJ, Taylor AMR, Arlett CF, Miki T, Weissman SM, Lovett M, Collins FS, Shiloh Y. A single ataxia telangiectasia gene with a product similar to PI-3 kinase. *Science.* **268**, 1749-1753 (1995).

191. Russell MW, Dick M 2nd, Campbell RM, Hulse JE, Munroe DJ, Bric E, Housman DE, Collins FS, Brody LC. Localization of Romano-Ward long QT syndrome gene, LQT1, to the interval between tyrosine hydroxylase (TH) and D11S1349. *Am J Hum Genet.* **57**, 503-507 (1995).

192. Asamoah A, North K, Doran S, Wagstaff J, Ogle R, Collins FS, Korf BR. 17q inversion involving the neurofibromatosis type one locus in a family with neurofibromatosis type one. *Am J Med Genet.* **60**, 312-316 (1995).

193. Trent JM, Weber B, Guan XY, Zhang J, Collins FS, Abel K, Diamond A, Meltzer P. Microdissection and microcloning of chromosomal alterations in human breast cancer. *Breast Cancer Res Treat.* **33**, 95-102 (1995).

194. Hajra A, Liu PP, Speck NA, Collins FS. Overexpression of core-binding factor alpha (CBF alpha) reverses cellular transformation by the CBF beta-smooth muscle myosin heavy chain chimeric oncoprotein. *Mol Cell Biol.* **15**, 4980-4989 (1995). Retraction in Hajra A, Liu PP, Speck NA, Collins FS. *Mol Cell Biol.* **16**, 7185 (1996).

195. Weber BL, Abel KJ, Couch FJ, Merajver S, Castilla L, Brody LC, Collins FS. Transcript identification in the BRCA1 candidate region. *Breast Cancer Res Treat.* **33**, 115-124 (1995).

196. Rotman G, Vanagaite L, Collins FS, Shiloh Y. Rapid identification of polymorphic CA-repeats in YAC clones. *Mol Biotechnol.* **3**, 85-92 (1995).

197. Struewing JP, Abeliovich D, Peretz T, Avishai N, Kaback MM, Collins FS, Brody LC. The carrier frequency of the BRCA1 185delAG mutation is approximately 1 percent in Ashkenazi Jewish individuals. *Nat Genet.* **11**, 198-200 (1995).

198. Hudson KL, Rothenberg KH, Andrews LB, Kahn MJ, Collins FS. Genetic discrimination and health insurance: an urgent need for reform. *Science.* **270**, 391-393 (1995).

199. Collins FS. Ahead of schedule and under budget: the Genome Project passes its fifth birthday. *Proc Natl Acad Sci USA.* **92**, 10821-10823 (1995).

200. Guyer MS, Collins FS. How is the Human Genome Project doing, and what have we learned so far? *Proc Natl Acad Sci USA.* **92**, 10841-10848 (1995).

201. Savitsky K, Sfez S, Tagle DA, Ziv Y, Sartiel A, Collins FS, Shiloh Y, Rotman G. The complete sequence of the coding region of the ATM gene reveals similarity to cell cycle regulators in different species. *Hum Mol Genet.* **4**, 2025-2032 (1995).

202. Smith JR, Carpten JD, Brownstein MJ, Ghosh S, Magnuson VL, Gilbert DA, Trent JM, Collins FS. Approach to genotyping errors caused by nontemplated nucleotide addition by Taq DNA polymerase. *Genome Res.* **5**, 312-317 (1995).

203. Ho PP, Couch FJ, Brody LC, Abel KJ, Boehnke M, Shearon TH, Chandrasekharappa SC, Collins FS, Weber BL. Localization of the human homolog of the yeast cell Div. control 27 gene (CDC27) proximal to ITGB3 on human chromosome 17q21.3. *Somat Cell Mol Genet.* **21**, 351-355 (1995).

204. Morrow DM, Tagle DA, Shiloh Y, Collins FS, Hieter P. TEL1, a S. cerevisiae homolog of the human gene mutated in ataxia telangiectasia, is functionally related to the yeast checkpoint gene MEC1. *Cell.* **82**, 831-840 (1995).

205. Collins FS. BRCA1—lots of mutations, lots of dilemmas. *N Engl J Med.* **334**, 186-188 (1996).

206. Bennett-Baker PE, Kiousis S, Chandrasekharappa SC, King SE, Abel KJ, Collins FS, Weber BL, Chamberlain JS. Isolation of tetranucleotide repeat polymorphisms flanking the BRCA1 gene. *Genomics.* **32**, 163-167 (1996).

207. Nasr SZ, Strong TV, Mansoura MK, Dawson DC, Collins FS. Novel missense mutation (G314R) in a cystic fibrosis patient with hepatic failure. *Hum Mutat.* **7**, 151-154 (1996).

208. Rosenfeld MA, Collins FS. Gene therapy for cystic fibrosis. Chest. 109, 241-252 (1996).

209. Wijmenga C, Gregory PE, Hajra A, Schrock E, Ried T, Eils R, Liu PP, Collins FS. Core binding factor beta-smooth muscle myosin heavy chain chimeric protein involved in acute myeloid leukemia forms unusual nuclear rod-like structures in transformed NIH 3T3 cells. *Proc Natl Acad Sci USA.* **93**, 1630-1635 (1996). Partial retraction in *Proc Natl Acad Sci USA.* **93**, 15522 (1996).

210. Liu PP, Wijmenga C, Hajra A, Blake TB, Kelley CA, Adelstein RS, Bagg A, Rector J, Cotelingam J, Willman CL, Collins FS. Identification of the chimeric protein product of the CBFB-MYH11 fusion gene in inv (16) leukemia cells. *Genes Chromosomes Cancer. 16*, 77-87 (1996). Partial retraction in *Genes Chromosomes Cancer.* **18**, 71 (1997).

211. Pecker I, Avraham KB, Gilbert DJ, Savitsky K, Rotman G, Harnik R, Fukao T, Schrock E, Hirotsune S, Tagle DA, Collins FS, Wynshaw-Boris A, Ried T, Copeland NG, Jenkins NA, Shiloh Y, Ziv Y. Identification and chromosomal localization of Atm, the mouse homolog of the ataxia-telangiectasia gene. *Genomics.* **35**, 39-45 (1996).

212. Russell MW, Munroe DJ, Bric E, Housman DE, Dietz-Band J, Riethman HC, Collins FS, Brody LC. A 500-kb physical map and contig from the Harvey ras-1 gene to the 11p telomere. *Genomics.* **35**, 353-360 (1996).

213. Russell MW, Dick M 2nd, Collins FS, Brody LC. KVLQT1 mutations in three families with familial or sporadic long QT syndrome. *Hum Mol Genet.* **5**, 1319-24 (1996).

214. Burke W, Kahn MJE, Garber JE, Collins FS. "First do no harm" also applies to cancer susceptibility testing. *Cancer J Sci Am.* **2**, 250 (1996).

215. Barlow C, Hirotsune S, Paylor R, Liyanage M, Eckhaus M, Collins F, Shiloh Y, Crawly JN, Ried T, Tagle D, Wynshaw-Boris A. Atm-deficient mice: a paradigm of ataxia telangiectasia. *Cell.* **86**, 159-171 (1996).

216. Abel KJ, Brody LC, Valdes JM, Erdos MR, McKinley DR, Castilla LH, Merajver SD, Couch FJ, Friedman LS, Ostermeyer EA, Lynch ED, King MC, Welcsh PL, Osborne-Lawrence S, Spillman M, Bowcock AM, Collins FS, Weber BL. Characterization of EZH1, a human homolog of Drosophila Enhancer of zeste near BRCA1. *Genomics.* **37**, 161-171 (1996).

217. Wilkinson DJ, Mansoura MK, Watson PY, Smit LS, Collins FS, Dawson DC. CFTR: the nucleotide binding folds regulate the accessibility and stability of the activated state. *J Gen Physiol.* **107**, 103-119 (1996).

218. Magnuson VL, Ally DS, Nylund SJ, Karanjawala ZE, Rayman JB, Knapp JI, Lowe AL, Ghosh S, Collins FS. Substrate nucleotide-determined non-templated addition of adenine by Taq DNA polymerase: implications for PCR-based genotyping and cloning. *Biotechniques.* **21**, 700-709 (1996).

219. Lubensky IA, Debelenko LV, Zhuang Z, Emmert-Buck MR, Dong Q, Chandrasekharappa SC, Guru SC, Manickam P, Olufemi SE, Marx SJ, Spiegel AM, Collins FS, Liotta LA. Allelic deletions on chromosome 11q13 in multiple tumors from individual MEN1 patients. *Cancer Res.* **56**, 5272-5278 (1996).

220. Castilla LH, Wijmenga C, Wang Q, Stacy T, Speck NA, Eckhaus M, Marin-Padilla M, Collins FS, Wynshaw-Boris A, Liu PP. Failure of embryonic hematopoiesis and lethal hemorrhages in mouse embryos heterozygous for a knocked-in leukemia gene CBFB-MYH11. *Cell.* **87**, 687-696 (1996).

221. Smith JR, Freije D, Carpten JD, Gronberg H, Xu J, Isaacs SD, Brownstein MJ, Bova GS, Guo H, Bujnovszky P, Nusskern DR, Damber JE, Bergh A, Emanuelsson M, Kallioniemi OP, Walker-Daniels J, Bailey-Wilson JE, Beaty TH, Meyers DA, Walsh PC, Collins FS, Trent JM, Issacs WB. Major susceptibility locus for prostate cancer on chromosome 1 suggested by a genome-wide search. *Science.* **274**, 1371-1374 (1996).

222. Hacia JG, Brody LC, Chee MS, Fodor SP, Collins FS. Detection of heterozygous mutations in BRCA1 using high density oligonucleotide arrays and two-colour fluorescence analysis. *Nat Genet.* **14**, 441-447 (1996).

223. Ghosh S, Collins FS. The geneticist's approach to complex disease. *Annu Rev Med.* **47**, 333-353 (1996).

224. Russell MW, du Manoir S, Munroe DJ, Collins FS, Brody LC. Chromosomal localization of 15 ion channel genes. *Somat Cell Mol Genet.* **22**, 425-431 (1996).

225. Smalley SL, Collins FS. Brief report: genetic, prenatal, and immunologic factors. *J Autism Dev Disord.* **26**, 195-198 (1996).

226. Ghosh S, Karanjawala ZE, Hauser ER, Ally D, Knapp JI, Rayman JB, Musick A, Tannenbaum J, Te C, Shapiro S, Eldridge W, Musick T, Martin C, Smith JR, Carpten JD, Brownstein MJ, Powell JI, Whiten R, Chines P, Nylund SJ, Magnuson VL, Boehnke M, Collins FS. Methods for precise sizing, automated binning of alleles, and reduction of error rates in large-scale genotyping using fluorescently labeled dinucleotide markers. FUSION (Finland-U.S. Investigation of NIDDM Genetics) Study Group. *Genome Res.* **7**, 165-178 (1997).

227. Brown KD, Ziv Y, Sadanandan SN, Chessa L, Collins FS, Shiloh Y, Tagle DA. The ataxia-telangiectasia gene product, a constitutively expressed nuclear protein that is not up-regulated following genome damage. *Proc Natl Acad Sci USA.* **94**, 1840-1845 (1997).

228. Russell MW, du Manoir S, Collins FS, Brody LC. Cloning of the human NADH ubiquinone oxidoreductase subunit B13: localization to chromosome 7q32 and identification of a pseudogene on 11p15. *Mamm Genome.* **8**, 60-61 (1997).

229. Debelenko LV, Emmert-Buck MR, Manickam P, Kester M, Guru SC, DiFranco EM, Olufemi SE, Agarwal S, Lubensky IA, Zhuang Z, Burns AL, Spiegel AM, Liotta LA, Collins FS, Marx SJ, Chandrasekharappa SC. Haplotype analysis defines a minimal interval for the multiple endocrine neoplasia type 1 (MEN1) gene. *Cancer Res.* **57**, 1039-1042 (1997).

230. Highsmith WE Jr, Burch LH, Zhou Z, Olsen JC, Strong TV, Smith T, Friedman KJ, Silverman LM, Boucher RC, Collins FS, Knowles MR. Identification of a splice site mutation (2789 +5 G > A) associated with small amounts of normal CFTR mRNA and mild cystic fibrosis. *Hum Mutat.* **9**, 332-338 (1997).

231. Rothenberg K, Fuller B, Rothstein M, Duster T, Ellis Kahn MJ, Cunningham R, Fine B, Hudson K, King MC, Murphy P, Swergold G, Collins FS. Genetic information and the workplace: legislative approaches and policy changes. *Science.* **275**, 1755-1757 (1997).

232. Chandrasekharappa SC, Guru SC, Manickam P, Olufemi SE, Collins FS, Emmert-Buck MR, Debelenko LV, Zhuang Z, Lubensky IA, Liotta LA, Crabtree JS, Wang Y, Roe BA, Weismann J, Boguski MS, Agarwal SK, Kester MB, Kim YS, Heppner C, Dong Z, Spiegel AM, Burns AL, Marx SJ. Positional cloning of the gene for multiple endocrine neoplasia-type 1. *Science.* **276**, 404-407 (1997).

233. Dong Q, Debelenko LV, Chandrasekharappa SC, Emmert-Buck MR, Zhuang Z, Guru SC, Manickam P, Skarulis M, Lubensky IA, Liotta LA, Collins FS, Marx SJ, Spiegel AM. Loss of heterozygosity at 11q13. analysis of pituitary tumors, lung carcinoids, lipomas, and other uncommon tumors in subjects with familial multiple endocrine neoplasia type 1. *J Clin Endocrinol Metab.* **82**, 1416-1420 (1997).

234. Sood R, Blake T, Aksentijevich I, Wood G, Chen X, Gardner D, Shelton DA, Mangelsdorf M, Orsborn A, Pras E, Balow JE Jr, Centola M, Deng Z, Zaks N, Chen X, Richards N, Fischel-Ghodsian N, Rotter JI, Pras M, Shohat M, Deaven LL, Gumucio DL, Callen DF, Richards RI, Collins FS, Liu PP, Kastner DL, Doggett NA. Construction of a 1-Mb restriction-mapped cosmid contig containing the candidate region for the familial Mediterranean fever locus (MEFV) on chromosome 16p13.3. *Genomics.* **42**, 83-95 (1997).

235. Humphrey JS, Salim A, Erdos MR, Collins FS, Brody LC, Klausner RD. Human BRCA1 inhibits growth in yeast: potential use in diagnostic testing. *Proc Natl Acad Sci USA.* **94**, 5820-5825 (1997).

236. Oda T, Elkahloun AG, Pike BL, Okajima K, Krantz ID, Genin A, Piccoli DA, Meltzer PS, Spinner NB, Collins FS, Chandrasekharappa SC. Mutations in the human Jagged 1 gene are responsible for Alagille syndrome. *Nat Genet.* **16**, 235-242 (1997).

237. Debelenko LV, Zhuang Z, Emmert-Buck MR, Chandrasekharappa SC, Manickam P, Guru SC, Marx SJ, Skarulis MC, Speigel AM, Collins FS, Jensen RT, Liotta LA, Lubensky IA. Allelic deletions on chromosome 11q13 in multiple endocrine neoplasia type 1-associated and sporadic gastrinomas and pancreatic endocrine tumors. *Cancer Res.* **57**, 2238-2243 (1997).

238. Agarwal SK, Kester MB, Debelenko LV, Heppner C, Emmert-Buck MR, Skarulis MC, Doppman JL, Kim YS, Lubensky IA, Zhuang Z, Green JS, Guru SC, Manickam P, Olufemi SE, Liotta LA, Chandrasekharappa SC, Collins FS, Spiegel AM, Burns AL, Marx SJ. Germline mutations of the MEN1 gene in familial multiple endocrine neoplasia type 1 and related states. *Hum Mol Genet.* **6**, 1169-1175 (1997).

239. Guru SC, Olufemi SE, Manickam P, Cummings C, Gieser LM, Pike BL, Bittner ML, Jiang Y, Chinault AC, Nowak NJ, Brzozowska A, Crabtree JS, Wang Y, Roe BA, Weisemann JM, Boguski MS, Agarwal SK, Burns AL, Spiegel AM, Marx SJ, Flejter WL, de Jong PJ, Collins FS, Chandrasekharappa SC. A 2.8-Mb clone contig of the multiple endocrine neoplasia type 1 (MEN1) region at 11q13. *Genomics.* **42**, 436-445 (1997).

240. Wilkinson DJ, Strong TV, Mansoura MK, Wood DL, Smith SS, Collins FS, Dawson DC. CFTR activation: additive effects of stimulatory and inhibitory phosphorylation sites in the R domain. *Am J Physiol.* **273**, L127-133 (1997).

241. Heppner C, Kester MB, Agarwal SK, Debelenko LV, Emmert-Buck MR, Guru SC, Manickam P, Olufemi SE, Skarulis MC, Doppman JL, Alexander RH, Kim YS, Saggar SK, Lubensky IA, Zhuang Z, Liotta LA, Chandrasekharappa SC, Collins FS, Spiegel AM, Burns AL, Marx SJ. Somatic mutation of the MEN1 gene in parathyroid tumours. *Nat Genet.* **16**, 375-378 (1997).

242. The International FMF Consortium. Ancient missense mutations in a new member of the RoRet gene family are likely to cause familial Mediterranean fever. *Cell.* **90**, 797-807 (1997).

243. Guru SC, Agarwal SK, Manickam P, Olufemi SE, Crabtree JS, Weismemann JM, Kester MB, Kim YS, Wang Y, Emmert-Buck MR, Liotta LA, Spiegel AM, Boguski MS, Roe BA, Collins FS, Marx SJ, Burns L, Chandrasekharappa SC. A transcript map for the 2.8-Mb region containing the multiple endocrine neoplasia type 1 locus. *Genome Res.* **7**, 725-735 (1997).

244. Balow JE Jr, Shelton DA, Orsborn A, Mangelsdorf M, Aksentijevich I, Blake T, Sood R, Gardner D, Liu R, Pras E, Levy EN, Centola M, Deng Z, Zaks N, Wood G, Chen X, Richards N, Shohat M, Livneh A, Pras M, Doggett NA, Collins FS, Liu PP, Rotter JI, Fischel-Ghodsian N, Gumucio D, Richards RI, Kastner DL. A high-resolution genetic map of the familial Mediterranean fever candidate region allows identification of haplotype-sharing among ethnic groups. *Genomics.* **44**, 280-291 (1997).

245. Woodage T, Basrai MA, Baxevanis AD, Hieter P, Collins FS. Characterization of the CHD family of proteins. *Proc Natl Acad Sci USA.* **94**, 11472-11477 (1997).

246. Manickam P, Guru SC, Debelenko LV, Agarwal SK, Olufemi SE, Weisemann JM, Boguski MS, Crabtree JS, Wang Y, Roe BA, Lubensky IA, Zhuang Z, Kester MB, Burns AL, Spiegel AM, Marx SJ, Liotta LA, Emmert-Buck MR, Collins FS, Chandrasekharappa SC. Eighteen new polymorphic markers in the multiple endocrine neoplasia type 1 (MEN1) region. *Hum Genet.* **101**, 102-108 (1997).

247. Zhuang Z, Vortmeyer AO, Pack S, Huang S, Pham TA, Wang C, Park WS, Agarwal SK, Debelenko LV, Kester M, Guru SC, Manickam P, Olufemi SE, Yu F, Heppner C,... Collins FS,... Jensen RT, Liotta LA, Lubensky IA. Somatic mutations of the MEN1 tumor suppressor gene in sporadic gastrinomas and insulinomas. *Cancer Res.* **57**, 4682-4686 (1997).

248. Gronberg H, Isaacs SD, Smith JR, Carpten JD, Bova GS, Freije D, Xu J, Meyers DA, Collins FS, Trent JM, Walsh PC, Isaacs WB. Characteristics of prostrate cancer in families potentially linked to the hereditary prostrate cancer 1 (HPC1) locus. *JAMA.* **278**, 1251-1255 (1997).

249. Collins FS, Guyer MS, Chakravarti A. Variations on a theme: cataloging human DNA sequence variation. *Science.* **278**, 1580-1581 (1997).

250. Debelenko LV, Brambilla E, Agarwal SK, Swalwell JI, Kester MB, Lubensky IA, Zhuang Z, Guru SC, Manickam P, Olufemi SE, Chandrasekharappa SC, Crabtree JS, Kim YS, Heppner C, Burns AL,... Collins FS, Travis WD, Emmert-Buck MR. Identification of MEN1 gene mutations in sporadic carcinoid tumors of the lung. *Hum Mol Genet.* **6**, 2285-2290 (1997).

251. Emmert-Buck MR, Lubensky IA, Dong Q, Manickam P, Guru SC, Kester MB, Olufemi SE, Agarwal S, Burns AL, Spiegel AM, Collins FS, Marx SJ, Zhuang Z, Liotta LA, Chandrasekharappa SC, Debelenko LV. Localization of the multiple endocrine neoplasia type I (MEN1) gene based on tumor loss of heterozygosity analysis. *Cancer Res.* **57**, 1855-1858 (1997).

252. Zhuang Z, Ezzat SZ, Vortmeyer AO, Weil R, Oldfield EH, Park WS, Pack S, Huang S, Agarwal SK, Guru SC, Manickam P, Debelenko LV, Kester MB, Olufemi SE, Heppner C,... Collins FS,... Liotta LA, Asa SL, Lubensky IA. Mutations of the MEN1 tumor suppressor gene in pituitary tumors. *Cancer Res.* **57**, 5446-5451 (1997).

253. Hacia JG, Makalowski W, Edgemon K, Erdos MR, Robbins CM, Fodor SP, Brody LC, Collins FS. Evolutionary sequence comparisons using high-density oligonucleotide arrays. *Nat Genet.* **18**, 155-158 (1998).

254. Guru SC, Goldsmith PK, Burns AL, Marx SJ, Spiegel AM, Collins FS, Chandrasekharappa SC. Menin, the product of the MEN1 gene, is a nuclear protein. *Proc Natl Acad Sci USA.* **95**, 1630-1634 (1998).

255. Olufemi SE, Green JS, Manickam P, Guru SC, Agarwal SK, Kester MB, Dong Q, Burns AL, Spiegel AM, Marx SJ, Collins FS, Chandrasekharappa SC. Common ancestral mutation in the MEN1 gene is likely responsible for the prolactinoma variant of MEN1 (MEN1Burin) in four kindreds from Newfoundland. *Hum Mutat.* **11**, 264-269 (1998).

256. Mansoura MK, Smith SS, Choi AD, Richards NW, Strong TV, Drumm ML, Collins FS, Dawson DC. Cystic fibrosis transmembrane conductance regulator (CFTR) anion binding as a probe of the pore. *Biophys J.* **74**, 1320-1332 (1998).

257. Emmert-Buck MR, Debelenko LV, Agarwal S, Kester MB, Manickam P, Zhuang Z, Guru SC, Olufemi SE, Burns AL, Chandrasekharappa SC, Lubensky IA, Liotta LA, Skarulis MC, Spiegel AM, Marx SJ, Collins FS. 11q13 allelotype analysis in 27 northern American MEN1 kindreds identifies two distinct founder chromosomes. *Mol Genet Metab.* **63**, 151-155 (1998).

258. Hacia JG, Brody LC, Collins FS. New approaches to BRCA1 mutation detection. *Breast Disease.* **10**, 45-59 (1998).

259. Valle T, Tuomilehto J, Bergman RN, Ghosh S, Hauser ER, Eriksson J, Nylund SJ, Kohtamaki K, Toivanen L, Vidgren G, Tuomilehto-Wolf E, Ehnholm C, Blaschak J, Langefeld CD, Watanabe RM,... Buchanan TA, Collins F, Boehnke M. Mapping genes for NIDDM. Design of the Finland-United States Investigation of NIDDM Genetics (FUSION) Study. *Diabetes Care.* **21**, 949-958 (1998).

260. Krkljus S, Abernathy CR, Johnson JS, Williams CA, Driscoll DJ, Zori R, Stalker HJ, Rasmussen SA, Collins FS, Kousseff BG, Baumbach L, Wallace MR. Analysis of CpG C-to-T mutations in neurofibromatosis type 1. Mutations in brief no. 129 online. *Hum Mutat.* **11**, 411 (1998).

261. Burke W, Thomson E, Khoury MJ, McDonnell SM, Press N, Adams PC, Barton JC, Beutler E, Brittenham G, Buchanan A, Clayton EW, Cogswell ME, Meslin EM, Motulsky AG, Powell LW, Sigal E, Wilfond BS, Collins FS. Hereditary hemochromatosis: gene discovery and its implications for population-based screening. *JAMA.* **280**, 172-178 (1998).

262. Agarwal SK, Debelenko LV, Kester MB, Guru SC, Manickam P, Olufemi SE, Skarulis MC, Heppner C, Crabtree JS, Lubensky IA, Zhuang Z, Kim YS, Chandrasekharappa SC, Collins FS, Liotta LA, Spiegel AM, Burns AL, Emmert-Buck MR, Marx SJ. Analysis of recurrent germline mutations in the MEN1 gene encountered in apparently unrelated families. *Hum Mutat.* **12**, 75-82 (1998).

263. Guru SC, Manickam P, Crabtree JS, Olufemi SE, Agarwal SK, Debelenko LV, Zhuang Z, Lubensky IA, Kester MB, Kim YS, Heppner C, Weisemann JM, Boguski MS, Wang Y, Roe BA, Burns AL, Liotta LA, Speigel AM, Emmert-Buck MR, Marx SJ, Collins FS, Chandrasekharappa SC. Identification and characterization of the multiple endocrine neoplasia type 1 (MEN1) gene. *J Intern Med.* **243**, 433-439 (1998).

264. Marx SJ, Agarwal SK, Kester MB, Heppner C, Kim YS, Emmert-Buck MR, Debelenko LV, Lubensky IA, Zhuang Z, Guru SC, Manickam P, Olufemi SE, Skarulis MC, Doppman JL, Alexander RH, Liotta LA, Collins FS, Chandrasekharappa SC, Spiegel AM, Burns AL. Germline and somatic mutation of the gene for multiple endocrine neoplasia type 1 (MEN1). *J Intern Med.* **243**, 447-453 (1998).

265. Ghosh S, Hauser ER, Magnuson VL, Valle T, Ally DS, Karanjawala ZE, Rayman JB, Knapp JI, Musick A, Tannenbaum J, Te C, Eldridge W, Shapiro S, Musick T, Martin C,... Tuomilehto J, Collins FS, Boehnke M. A large sample of Finnish diabetic sib-pairs reveals no evidence for a non-insulin-dependent diabetes mellitus susceptibility locus at 2qter. *J Clin Invest.* **102**, 704-709 (1998).

266. Hacia JG, Edgemon K, Sun B, Stern D, Fodor SP, Collins FS. Two color hybridization analysis using high density oligonucleotide arrays and energy transfer dyes. *Nucleic Acids Res.* **26**, 3865-3866 (1998).

267. Mansoura MK, Collins FS. Medical implications of the genetic revolution. *J Health Care Law & Policy.* **1**, 329-352 (1998).

268. Marx S, Spiegel AM, Skarulis MC, Doppman JL, Collins FS, Liotta LA. Multiple endocrine neoplasia type 1: clinical and genetic topics. *Ann Intern Med.* **129**, 484-494 (1998).

269. Xu J, Meyers D, Freije D, Isaacs S, Wiley K, Nusskern D, Ewing C, Wilkens E, Bujnovszky P, Bova GS, Walsh P, Isaacs W, Schleutker J, Matikainen M, Tammela T,... Collins FS, Brownstein M, Trent J. Evidence for a prostate cancer susceptibility locus on the X chromosome. *Nat Genet.* **20**, 175-179 (1998).

270. Karanjawala ZE, Collins FS. Genetics in the context of medical practice. *JAMA.* **280**, 1533-1534 (1998).

271. Hacia JG, Woski SA, Fidanza J, Edgemon K, Hunt N, McGall G, Fodor SP, Collins FS. Enhanced high density oligonucleotide array-based sequence analysis using modified nucleoside triphosphates. *Nucleic Acids Res.* **26**, 4975-4982 (1998).

272. Hacia JG, Brody LC, Collins FS. Applications of DNA chips for genomic analysis. *Mol Psychiatry.* **3**, 483-492 (1998).

273. Collins FS, Patrinos A, Jordan E, Chakravarti A, Gesteland R, Walters L. New goals for the U.S. Human Genome Project. 1998-2003. *Science.* **282**, 682-689 (1998).

274. Collins FS, Brooks LD, Chakravarti A. A DNA polymorphism discovery resource for research on human genetic variation. *Genome Res.* **8**, 1229-1231 (1998).

275. Hacia JG, Sun B, Hunt N, Edgemon K, Mosbrook D, Robbins C, Fodor SP, Tagle DA, Collins FS. Strategies for mutational analysis of the large multiexon ATM gene using high-density oligonucleotide arrays. *Genome Res.* **8**, 1245-1258 (1998).

276. Centola M, Chen X, Sood R, Deng Z, Aksentijevich I, Blake T, Ricke DO, Chen X, Wood G, Zaks N, Richards N, Krizman D, Mansfield E, Apostolou S, Liu J,... Collins FS, Liu PP, Fischel-Ghodsian N, Kastner DL. Construction of an approximately 700-kb transcript map around the familial Mediterranean fever locus on human chromosome 16p13.3. *Genome Res.* **8**, 1172-1191 (1998).

277. Agarwal SK, Guru SC, Heppner C, Erdos MR, Collins RM, Park SY, Saggar S, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ, Burns AL. Menin interacts with the AP1 transcription factor JunD and represses JunD-activated transcription. *Cell.* **96**, 143-152 (1999).

278. Ghosh S, Langefeld CD, Ally D, Watanabe RM, Hauser ER, Magnuson VL, Nylund SJ, Valle T, Eriksson J, Bergman RN, Tuomilehto J, Collins FS, Boehnke M. The W64R variant of the g3-adrenergic receptor is not associated with Type II diabetes or obesity in a large Finnish sample. *Diabetologia.* **42**, 238-244 (1999).

279. Ghosh S, Watanabe RM, Hauser ER, Valle T, Magnuson VL, Erdos MR, Langefeld CD, Balow J Jr, Ally DS, Kohtamaki K, Chines P, Birznieks G, Kaleta HS, Musick A, Te C,... Bergman RN, Collins FS, Boehnke M. Type 2 diabetes: evidence for linkage on chromosome 20 in 716 Finnish affected sib pairs. *Proc Natl Acad Sci USA.* **96**, 2198-2203 (1999).

280. Watanabe RM, Valle T, Hauser ER, Ghosh S, Eriksson J, Kohtamaki K, Ehnholm C, Tuomilehto J, Collins FS, Bergman RN, Boehnke M. Familiality of quantitative metabolic traits in Finnish families with non-insulin-dependent mellitus: Finland-United States Investigation of NIDDM Genetics (FUSION) Study investigators. *Hum Hered.* **49**, 159-168 (1999).

281. Hacia JG, Fan JB, Ryder O, Jin L, Edgemon K, Ghandour G, Mayer RA, Sun B, Hsie L, Robbins CM, Brody LC, Wang D, Lander ES, Lipshutz R, Fodor SP, Collins FS. Determination of ancestral alleles for human single-nucleotide polymorphisms using high-density oligonucleotide arrays. *Nat Genet.* **22**, 164-167 (1999).

282. Guru SC, Crabtree JS, Brown KD, Dunn KJ, Manickam P, Prasad NB, Wangsa D, Burns AL, Spiegel AM, Marx SJ, Pavan WJ, Collins FS, Chandrasekharappa SC. Isolation, genomic organization, and expression analysis of Men1, the murine homolog of the MEN1 gene. *Mamm Genome.* **10**, 592-596 (1999).

283. Marx SJ, Agarwal SK, Heppner C, Kim YS, Kester MB, Goldsmith PK, Skarulis MC, Spiegel AM, Burns AL, Debelenko LV, Zhuang Z, Lubensky IA, Liotta LA, Emmert-Buck MR, Guru SC, Manickam P, Crabtree JS, Collins FS, Chandrasekharappa SC. The gene for multiple endocrine neoplasia type 1: recent findings. *Bone.* **25**, 119-122 (1999).

284. Fuller BP, Kahn MJ, Barr PA, Biesecker L, Crowley E, Garber J, Mansoura MK, Murphy P, Murray J, Phillips J, Rothenberg K, Rothstein M, Stopfer J, Swergold G, Weber B, Collins FK, Hudson KL. Privacy in genetics research. *Science.* **285**, 1359-1361 (1999).

285. Kim YS, Burns AL, Goldsmith PK, Heppner C, Park SY, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ. Stable overexpression of MEN1 suppresses tumorigenicity of RAS. *Oncogene.* **18**, 5936-5942 (1999).

286. Hacia JG, Novotny EA, Mayer RA, Woski SA, Ashlock MA, Collins FS. Design of modified oligodeoxyribonucleotide probes to detect telomere repeat sequences in FISH assays. *Nucleic Acids Res.* **27**, 4034-4039 (1999).

287. Strausberg RL, Feingold EA, Klausner RD, Collins FS. The mammalian gene collection. *Science.* **286**, 455-457 (1999).

288. Hacia JG, Collins FS. Mutational analysis using oligonucleotide microarrays. *J Med Genet.* **36**, 730-736 (1999).

289. Lipkin SM, Wang V, Jacoby R, Banerjee-Basu S, Baxevanis AD, Lynch HT, Elliott RM, Collins FS. MLH3: a DNA mismatch repair gene associated with mammalian microsatellite instability. *Nat Genet.* **24**, 27-35 (2000).

290. Debelenko LV, Swalwell JI, Kelley MJ, Brambilla E, Manickam P, Baibakov G, Agarwal SK, Spiegel AM, Marx SJ, Chandrasekharappa SC, Collins FS, Travis WD, Emmert-Buck MR. MEN1 gene mutation analysis of high-grade neuroendocrine lung carcinoma. *Genes Chromosomes Cancer.* **28**, 58-65 (2000).

291. Manickam P, Vogel AM, Agarwal SK, Oda T, Spiegel AM, Marx SJ, Collins FS, Weinstein BM, Chandrasekharappa SC. Isolation, characterization, expression and functional analysis of the zebrafish ortholog of MEN1. *Mamm Genome.* **11**, 448-454 (2000).

292. Karanjawala ZE, Kaariainen H, Ghosh S, Tannenbaum J, Martin C, Ally D, Tuomilehto J, Valle T, Collins FS. Complete maternal isodisomy of chromosome 8 in an individual with an early-onset ileal carcinoid tumor. *Am J Med Genet.* **93**, 207-210 (2000).

293. Hacia JG, Edgemon K, Fang N, Mayer RA, Sudano D, Hunt N, Collins FS. Oligonucleotide microarray based detection of repetitive sequence changes. *Hum Mutat.* **16**, 354-363 (2000).

294. Ghosh S, Watanabe RM, Valle TT, Hauser ER, Magnuson VL, Langefeld CD, Ally DS, Mohlke KL, Silander K, Kohtamaki K, Chines P, Balow Jr J, Birznieks G, Chang J, Eldridge W,... Bergman RN, Collins FS, Boehnke M. The Finland-United States investigation of non-insulin-dependent diabetes mellitus genetics (FUSION) study. I: An autosomal genome scan for genes that predispose to type 2 diabetes. *Am J Hum Genet.* **67**, 1174-1185 (2000).

295. Watanabe RM, Ghosh S, Langefeld CD, Valle TT, Hauser ER, Magnuson VL, Mohlke KL, Silander K, Ally DS, Chines P, Blaschak-Harvan J, Douglas JA, Duren WL, Epstein MP, Fingerlin TE,... Bergman RN, Collins FS, Boehnke M. The Finland-United States investigation of non-insulin-dependent diabetes mellitus genetics (FUSION) study. II: An autosomal genome scan for diabetes-related quantitative-trait loci. *Am J Hum Genet.* **67**, 1186-1200 (2000).

296. Guru SC, Prasad NB, Shin EJ, Hemavathy K, Lu J, Ip YT, Agarwal SK, Marx SJ, Spiegel AM, Collins FS, Oliver B, Chandrasekharappa SC. Characterization of a MEN1 ortholog from Drosophila melanogaster. *Gene.* **263**, 31-38 (2001).

297. International Human Genome Sequencing Consortium. Initial sequencing and analysis of the human genome. *Nature.* **409**, 860-921 (2001).

298. Crabtree JS, Scacheri PC, Ward JM, Garrett-Beal L, Emmert-Buck MR, Edgemon KA, Lorang D, Libutti SK, Chandrasekharappa SC, Marx SJ, Spiegel AM, Collins FS. A mouse model of multiple endocrine neoplasia, type 1, develops multiple endocrine tumors. *Proc Natl Acad Sci USA.* **98**, 1118-1123 (2001).

299. Knapp JI, Heppner C, Hickman AB, Burns AL, Chandrasekharappa SC, Collins FS, Marx SJ, Spiegel AM, Agarwal SK. Identification and characterization of JunD missense mutants that lack menin binding. *Oncogene.* **19**, 4706-4712 (2000).

300. Royal C, Baffoe-Bonnie A, Kittles R, Powell I, Bennett J, Hoke G, Pettaway C, Weinrich S, Vijayakumar S, Ahaghotu C, Mason T, Johnson E, Obeikwe M, Simpson C, Mejia R,... Berg K, Dunston G, Collins FS. Recruitment experience in the first phase of the African American Hereditary Prostrate Cancer (AAHPC) study. *Ann Epidemiol.* **10**, S68-77 (2000).

301. Rotimi CN, Dunston GM, Berg K, Akinsete O, Amoah A, Owusu S, Acheampong J, Boateng K, Oli J, Okafor G, Onyenekwe B, Osotimehin B, Abbiyesuku F, Johnson T, Fasanmade O,... Adegoke O, Bonney G, Collins FS. In search of susceptibility genes for type 2 diabetes in West Africa: the design and results of the first phase of the AADM study. *Ann Epidemiol.* **11**, 51-58 (2001).

302. Douglas JA, Erdos MR, Watanabe RM, Braun A, Johnston CL, Oeth P, Mohlke KL, Valle TT, Ehnholm C, Buchanan TA, Bergman RN, Collins FS, Boehnke M, Tuomilehto J. The peroxisome proliferator-activated receptor-gamma2 Pro12A1a variant: association with type 2 diabetes and trait differences. *Diabetes.* **50**, 886-890 (2001).

303. Powell IJ, Carpten J, Dunston G, Kittles R, Bennett J, Hoke G, Pettaway C, Weinrich S, Vijayakumar S, Ahaghotu CA, Boykin W, Mason T, Royal C, Baffoe-Bonnie A, Bailey-Wilson J, Berg K, Trent J, Collins FS. African-American heredity prostate cancer study; a model for genetic research. *J Natl Med Assoc.* **93**, 120-123 (2001).

304. Lipkin SM, Wang V, Stoler DL, Anderson GR, Krisch I, Hadley D, Lynch HT, Collins FS. Germline and somatic mutation analyses in the DNA mismatch repair gene MLH3: Evidence for somatic mutation in colorectal cancers. *Hum Mutat.* **17**, 389-396 (2001).

305. Mohlke KL, Lange EM, Valle TT, Ghosh S, Magnuson VL, Silander K, Watanabe RM, Chines PS, Bergman RN, Tuomilehto J, Collins FS, Boehnke M. Linkage disequilibrium between microsatellite markers extends beyond 1 cM on chromosome 20 in Finns. *Genome Res.* **11**, 1221-1226 (2001).

306. Heppner C, Bilimoria KY, Agarwal SK, Kester M, Whitty LJ, Guru SC, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ, Burns AL. The tumor suppressor protein menin interacts with NF-kappaB proteins and inhibits NF-kappaB-mediated transactivation. *Oncogene.* **20**, 4917-4925 (2001).

307. Scacheri PC, Crabtree JS, Novotny EA, Garrett-Beal L, Chen A, Edgemon KA, Marx SJ, Spiegel AM, Chandrasekharappa SC, Collins FS. Bidirectional transcriptional activity of PGK-neomycin and unexpected embryonic lethality in heterozygote chimeric knockout mice. *Genesis.* **30**, 259-263 (2001).

308. Fingerlin TE, Erdos MR, Watanabe RM, Wiles KR, Stringham HM, Mohlke KL, Silander K, Valle TT, Buchanan TA, Tuomilehto J, Bergman RN, Boehnke M, Collins FS. Variation in three single nucleotide polymorphisms in the calpain-10 gene not associated with type 2 diabetes in a large Finnish cohort. *Diabetes.* **51**, 1644-1648 (2002).

309. Patenaude AF, Guttmacher AE, Collins FS. Genetic testing and psychology: New roles, new responsibilities. *Am Psychol.* **57**, 271-282 (2002).

310. Lipkin SM, Moens PB, Wang V, Lenzi M, Shanmugarajah D, Gilgeous A, Thomas J, Cheng J, Touchman JW, Green ED, Schwartzberg P, Collins FS, Cohen PE. Meiotic arrest and aneuploidy in MLH3-deficient mice. *Nat Genet.* **31**, 385-390 (2002).

311. Mykytyn K, Nishimura DY, Searby CC, Shastri M, Yen HJ, Beck JS, Braun T, Streb LM, Cornier AS, Cox GF, Fulton AB, Carmi R, Luleci G, Chandrasekharappa SC, Collins FS, Jacobson SG, Heckenlively JR, Weleber RG, Stone EM, Sheffield VC. Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. *Nat Genet.* **31**, 435-438 (2002).

312. Wolfsberg TG, Wetterstrand KA, Guyer MS, Collins FS, Baxevanis AD. A user's guide to the human genome. *Nat Genet.* **32** S, 1-79 (2002).

313. Guttmacher AE, Collins FS. Genomic medicine—a primer. *N Engl J Med.* **347**, 1512-1520 (2002).

314. Mouse Genome Sequencing Consortium. Initial sequencing and comparative analysis of the mouse genome. *Nature.* **420**, 520-562 (2002).

315. Mammalian Gene Collection Program Team. Generation and initial analysis of more than 15,000 full-length human and mouse cDNA sequences. *Proc Natl Acad Sci USA.* **99**, 16899-16903 (2002).

316. Mohlke KL, Erdos MR, Scott LJ, Fingerlin TE, Jackson AU, Silander K, Hollstein P, Boehnke M, Collins FS. High-throughput screening for evidence of association by using mass spectrometry genotyping on DNA pools. *Proc Natl Acad Sci USA.* **99**, 16928-16933 (2002).

317. Marx SJ, Simonds WF, Agarwal SK, Burns AL, Weinstein LE, Cochran C, Skarulis MC, Spiegel AM, Libutti SK, Alexander HR, Chen CC, Chang R, Chandrasekharappa SC, Collins FS. Hyperparathyroidism in hereditary syndromes: special expressions and special managements. *J Bone Miner Res.* **17**, Suppl 2:N37-43 (2002).

318. Sukhodolets KE, Hickman AB, Agarwal SK, Sukhodolets MV, Obungu VH, Novotny EA, Crabtree JS, Chandrasekharappa SC, Collins FS, Spiegel AM, Burns AL, Marx SJ. The 32-kilodalton subunit of replication protein A interacts with menin, the product of the MEN1 tumor suppressor gene. *Mol Cell Biol.* **23**, 493-509 (2003).

319. Eriksson M, Brown WT, Gordon LB, Glynn MW, Singer J, Scott L, Erdos MR, Robbins CM, Moses TY, Berglund P, Dutra A, Pak E, Durkin S, Csoka AB, Boehnke M, Glover TW, Collins FS. Recurrent de novo point mutations in lamin A cause Hutchinson-Gilford progeria syndrome. *Nature.* **423**, 293-298 (2003).

320. Fang NY, Greiner TC, Weisenburger DD, Chan WC, Vose JM, Smith LM, Armitage JO, Mayer RA, Pike BL, Collins FS, Hacia JG. Oligonucleotide microarrays demonstrate the highest frequency of ATM mutations in the mantle cell subtype of lymphoma. *Proc Natl Acad Sci USA.* **100**, 5372-5377 (2003).

321. Crabtree JS, Scacheri PC, Ward JM, McNally SR, Swain GP, Montagna C, Hager JH, Hanahan D, Edlund H, Magnuson MA, Garrett-Beal L, Burns AL, Ried T, Chandrasekharappa SC, Marx SJ, Spiegel AM, Collins FS. Of mice and MEN1: Insulinomas in a conditional mouse knockout. *Mol Cell Biol.* **23**, 6075-6085 (2003).

322. Agarwal SK, Novotny EA, Crabtree JS, Weitzman JB, Yaniv M, Burns AL, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ. Transcription factor JunD, deprived of menin, switches from growth suppressor to growth promoter. *Proc Natl Acad Sci USA.* **100**, 10770-10775 (2003).

323. Rotimi C, Daniel H, Zhou J, Obisesan A, Chen G, Chen Y, Amoah A, Opoku V, Acheampong J, Agyenim-Boateng K, Eghan BA Jr, Oli J, Okafor G, Ofoegbu E, Osotimehin B,... Aje T, Collins F, Dunston G. Prevalence and determinants of diabetic retinopathy and cataracts in West African type 2 diabetes patients. *Ethn Dis.* **13**, S110-117 (2003).

324. Olopade OI, Fackenthal JD, Dunston G, Tainsky MA, Collins FS, Whitfield-Broome C. Breast cancer genetics in African Americans. *Cancer.* **97**, 236-45 (2003).

325. Libutti SK, Crabtree JS, Lorang D, Burns AL, Mazzanti C, Hewitt SM, O'Connor S, Ward JM, Emmert-Buck MR, Remaley A, Miller M, Turner E, Alexander HR, Arnold A, Marx SJ, Collins FS, Spiegel AM. Parathyroid gland-specific deletion of the mouse Men1 gene results in parathyroid neoplasia and hypercalcemic hyperparathyroidism. *Cancer Res.* **63**, 8022-8028 (2003).

326. The International HapMap Consortium. The International HapMap Project. *Nature.* **426**, 789-796 (2004).

327. Crawford GE, Holt IE, Mulliken JC, Tai D, National Institutes of Health Intramural Sequencing Center, Green ED, Wolfsberg TG, Collins FS. Identifying gene regulatory elements by genome-wide recovery of DNase hypersensitive sites. *Proc Natl Acad Sci USA.* **101**, 992-997 (2004).

328. Silander K, Valle TT, Scott LJ, Mohlke KL, Stringham HM, Wiles KR, Duren WL, Doheny KF, Pugh EW, Chines P, Narisu N, White PP, Fingerlin TE, Jackson AU, Li C,... Tuomilehto J, Collins FS, Boehnke M. A large set of Finnish affected sibling pair families with type II diabetes mellitus suggests susceptibility loci on chromosomes 6, 11, and 14. *Diabetes.* **53**, 821-829 (2004).

329. Scacheri PC, Rozenblatt-Rosen O, Caplen NJ, Wolfsberg TG, Umayam L, Lee JC, Hughes CM, Shanmugam KS, Bhattacharjee A, Meyerson M, Collins FS. Short interfering RNAs can induce unexpected and divergent changes in the levels of untargeted proteins in mammalian cells. *Proc Natl Acad Sci USA.* **101**, 1892-1897 (2004).

330. Rotimi CN, Chen G, Guass D, Zhou J, Doumatey A, Furbert-Harris P, Adegoke O, Amoah A, Acheampong J, Agyenim-Boateng K, Egan BA, Oli J, Okafor G, Ofoegbu E, Osotimehin B,... Akinsete O, Dunston G, Collins FS. A genome-wide search for type 2 diabetes susceptibility genes in West Africans. The Africa America Diabetes Mellitus (AADM) study. *Diabetes.* **53**, 838-841 (2004).

331. Ahaghotu C, Baffoe-Bonney A, Kittles R, Pettaway C, Powell I, Royal C, Vijayakumar S, Bennett J, Hoke G, Mason T, Bailey-Wilson J, Boykin W, Berg K, Carpten J, Weinrich S, Trent J, Dunston G, Collins FS. Clinical characteristics of African-American men with hereditary prostate cancer: the AAHPC study. *Prostate Cancer Prostatic Dis.* **7**, 165-169 (2004).

332. Silander K, Mohlke KL, Scott LJ, Peck EC, Hollstein P, Skol AD, Jackson AU, Deloukas P, Hunt S, Stavrides G, Chines PS, Erdos MR, Narisu N, Conneely KN, Li C,... Watanabe RM, Boehnke M, Collins FS. Genetic variation near the hepatocyte nuclear factor-4α gene predicts susceptibility to type 2 diabetes. *Diabetes.* **53**, 1141-1149 (2004).

333. The International HapMap Consortium. Integrating ethics and science in the International HapMap Project. *Nat Rev Genet.* **5**, 467-475 (2004).

334. Goldman RD, Shumaker DK, Erdos MR, Eriksson M, Goldman AE, Gordon LB, Gruenbaum Y, Khuon S, Mendez M, Varga R, Collins FS. Accumulation of mutant lamin A causes progressive changes in nuclear architecture in Hutchinson-Gilford Progeria Syndrome. *Proc Natl Acad Sci USA.* **101**, 8963-8968 (2004).

335. Lipkin SM, Rozek, LS, Rennert G, Yang W, Chen PC, Hacia J, Hunt N, Shin B, Fodor S, Kokoris M, Greenson JK, Fearon E, Lynch H, Collins FS, Gruber SB. The MLH1 D132H variant is associated with susceptibility to sporadic colorectal cancer. *Nat Genet.* **36**, 694-699 (2004).

336. The Comprehensive Knockout Mouse Project Consortium. The knockout mouse project. *Nat Genet.* **36**, 921-924 (2004).

337. Scacheri PC, Kennedy AL, Chin K, Miller MT, Hodgson JG, Gray JW, Marx SJ, Spiegel AM, Collins FS. Pancreatic insulinomas in multiple endocrine neoplasia, type 1 knockout mice can develop in the absence of chromosome instability or microsatellite instability. *Cancer Res.* **64**, 7039-7044 (2004).

338. The MGC Project Team. The status, quality, and expansion of the NIH full-mammalian cDNA project: the mammalian gene collection (MGC). *Genome Res.* **14**, 2121-2127 (2004).

339. International Human Genome Sequencing Consortium. Finishing the euchromatic sequence of the human genome. *Nature.* **434**, 931-945 (2004).

340. The ENCODE Project Consortium. The ENCODE (ENCyclopedia of DNA Elements) Project. *Science.* **306**, 636-640 (2004).

341. Conneely KN, Silander K, Scott LJ, Mohlke KL, Lazaridis KN, Valle TT, Tuomilehto J, Bergman RN, Watanabe RM, Buchanan TA, Collins FS, Boehnke M. Variation in the resistin gene is associated with obesity and insulin-related phenotypes in Finns. *Diabetologia.* **47**, 1782-1788 (2004).

342. Collins FS. What we do and don't know about 'race', 'ethnicity', genetics and health at the dawn of the genome era. *Nat Genet Sup.* **36**, S13-15 (2004).

343. Scacheri PC, Crabtree JS, Kennedy AL, Swain GP, Ward JM, Marx SJ, Spiegel AM, Collins FS. Homozygous loss of menin is well tolerated in liver, a tissue not affected in MEN1. *Mamm Genome.* **15**, 872-877 (2004).

344. Austin CP, Brady LS, Insel TR, Collins FS. NIH Molecular Libraries Initiative. *Science.* **306**, 1138-1139 (2004).

345. Bonham VL, Warshauer-Baker E, Collins FS. Race and ethnicity in the genome era. *Am Psychol.* **60**, 9-15 (2004).

346. Chen Y, Kittles R, Zhou J, Chen G, Adeyemo A, Panguluri RK, Chen W, Amoah A, Opoku V, Acheampong J, Agyenim-Boateng K, Eghan BA, Nyantaki A, Oli J, Okafor G,... Collins FS, Dunston GM, Rotimi CN. Calpain-10 gene polymorphisms and type 2 diabetes in West Africans: The Africa America Diabetes Mellitus (AADM) study. *Ann Epidemiol.* **15**, 153-159 (2005).

347. Adeyemo AA, Johnson T, Acheampong J, Oli J, Okafor G, Amoah A, Owusu S, Agyenim-Boateng K, Dunston G, Collins F, Rotimi C. A genome wide quantitative trait linkage analysis for serum lipids in type 2 diabetes in an African population. *Atherosclerosis.* **181**, 389-397 (2005).

348. Chen G, Adeyemo AA, Johnson T, Zhou J, Amoah A, Owusu S, Acheampong J, Agyenim-Boateng K, Eghan Jr BA, Oli J, Okafor G, Abbiyesuku F, Dunston GM, Chen Y, Collins FS, Rotimi C. A genome-wide scan for quantitative trait loci linked to obesity phenotypes among West Africans. *Int J Obes.* **29**, 255-259 (2005).

349. Capell BC, Erdos MR, Madigan JP, Fiordalisi, Varga R, Conneely KN, Gordon LB, Der CJ, Cox AD, Collins FS. Inhibiting farnesylation of the progerin protein prevents the characteristic nuclear blebbing of HGPS. *Proc Natl Acad Sci USA.* **102**, 12879-12884 (2005).

350. Kittles RA, Baffoe-Bonnie A, Moses T, Robbins C, Ahaghotu C, Huusko P, Pettaway C, Vijayakumar S, Bennett J, Hoke G, Mason T, Weinrich S, Trent J, Collins F,... Dunston G, Powell I, Carpten JD. A common nonsense mutation in EphB2 is associated with prostate cancer risk in African American men with a positive family history. *J Med Genet.* **43**, 507-511 (2005).

351. Guttmacher AE, Collins FS. Realizing the promise of genomics in biomedical research. *JAMA.* **294**, 1399-1402 (2005).

352. Mohlke KL, Skol AD, Scott LJ, Valle TT, Bergman RN, Tuomilehto J, Boehnke M, Collins FS, on behalf of the FUSION Study Group. Evaluation of SLC2A10 (GLUT10) as a candidate gene for type 2 diabetes and related traits in Finns. *Mol Genet Metab.* **85**, 323-327 (2005).

353. The International HapMap Consortium. A haplotype map of the human genome. *Nature.* **437**, 1299-1320 (2005).

354. Mohlke KL, Jackson AU, Scott LJ, Peck EC, Suh YD, Chines PS, Watanabe RM, Buchanan TA, Conneely KN, Erdos MR, Narisu N, Enloe S, Valle TT, Tuomilehto, J, Bergman RN, Boehnke M, Collins FS. Mitochondrial

polymorphisms and susceptibility to type 2 diabetes-related traits in Finns. *Human Genetics*. **118**, 245-254 (2005).

355. Crawford GE, Holt IE, Whittle J, Webb BD, Tai D, Davis S, Margulies EH, Chen Y, Bernat JA, Ginsburg D, Zhou D, Luo S, Vasicek TJ, Daly MJ, Wolfsberg TG, Collins FS. Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS). *Genome Res.* **16**, 123-131 (2006).

356. Weinrich SP, on behalf of the African American Hereditary Prostate Cancer Study Network. Prostate cancer screening in high-risk men. *Cancer.* **106**, 796-803 (2006).

357. Willer CJ, Scott LJ, Bonnycastle LL, Jackson AU, Chines P, Pruim R, Bark CW, Tsai YY, Pugh EW, Doheny KF, Kinnunen L, Mohlke KL, Valle TT, Bergman RN, Tuomilehto J, Collins FS, Boehnke M. Tag SNP selection for Finnish individuals based on the CEPH Utah HapMap database. *Genetic Epidemiol.* **30**, 180-190 (2006).

358. Varga R, Eriksson M, Erdos MR, Olive M, Harten I, Kolodgie F, Capell BC, Cheng J, Faddah D, Perkins S, Avallone H, San H, Qu X, Ganesh S, Gordon LB, Virmani R, Wight TN, Nabel EG, Collins FS. Progressive vascular smooth muscle cell defects in a mouse model of Hutchinson-Gilford progeria syndrome. *Proc Natl Acad Sci USA*. **103**, 3250-3255 (2006).

359. Scacheri PC, Davis S, Odom DT, Crawford GE, Perkins S, Halawi MJ, Agarwal SK, Marx SJ, Spiegel AM, Meltzer PS, Collins FS. Genome-wide analysis of menin binding provides insights to MEN1 tumorigenesis. *PLoS Genet.* **4**, e51 (2006).

360. Alexander DF, Alving BM, Battey JF, Berg JM, Collins FS, Fauci AS, Gallin JI, Grady PA, Hodes RJ, Hrynkow SH, Insel TR, Jones JF, Katz SI, Landis SC, Li TK,... Straus SE, Tabak LA, Volkow ND. Response to: "Rescuing the NIH before it is too late." *J Clin Invest.* **116**, 1462-1463 (2006).

361. Bernat JA, Crawford GE, Ogurstsov AY, Collins FS, Ginsburg D, Kondrashov AS. Distant conserved sequences flanking endothelial-specific promoters contain tissue-specific DNase-hypersensitive sites and over-represented motifs. *Hum Mol Genet.* **15**, 2098-2105 (2006).

362. Shumaker DK, Dechat T, Kohlmaier A, Adam SA, Bozovsky MR, Erdos MR, Eriksson M, Goldman AE, Khuon S, Collins FS, Jenuwein T, Goldman RD. Mutant nuclear lamin A leads to progressive alterations of epigenetic control in premature aging. *Proc Natl Acad Sci USA*. **103**, 8703-8708 (2006).

363. Crawford GE, Davis S, Scacheri PC, Renaud G, Halawi MJ, Erdos MR, Green R, Meltzer PS, Wolfsberg TG, Collins FS. DNase-chip: a high-resolution method to identify DNase I hypersensitive sites using tiled microarrays. *Nature Methods*. **3**, 503-509 (2006).

364. Lewinski MK, Yamashita M, Emerman M, Ciuffi A, Marshall H, Crawford G, Collins F, Shinn P, Leipzig J, Hannenhalli S, Berry CC, Ecker JR, Bushman FD. Retroviral DNA integration: viral and cellular determinants of target-site selection. *PLoS Pathog*. **2**, e60:611-622 (2006).

365. Bonnycastle LL, Willer CJ, Conneely KN, Jackson AU, Burrill CP, Watanabe RM, Chines PS, Narisu N, Scott LJ, Enloe ST, Swift AJ, Duren WL, Stringham HM, Erdos MR, Riebow NL,... Mohlke KL, Boehnke M, Collins FS. Common variants in MODY contribute to risk of type 2 diabetes in Finns. *Diabetes*. **55**, 2534-2540 (2006).

366. Scott LJ, Bonnycastle LL, Willer CJ, Sprau AG, Jackson AU, Narisu N, Duren WL, Chines PS, Stringham HM, Erdos MR, Valle TT, Tuomilehto J, Bergman RN, Mohlke KL, Collins FS, Boehnke M. Association of transcription factor 7-like 2 (TCF7L2) variants with type 2 diabetes in a Finnish sample. *Diabetes*. **55**, 2649-2653 (2006).

367. Capell BC, Collins FS. Human laminopathies: nuclei gone genetically awry. *Nat Rev Genet.* **12**, 940-952 (2006).

368. Manolio TA, Bailey-Wilson JE, Collins FS. Genes, environment, and the value of prospective cohort studies. *Nat Rev Genet.* **7**, 812-820 (2006).

369. Cerrato A, Parisi M, Santa Anna S, Missirlis F, Guru S, Agarwal S, Sturgill D, Talbot T, Spiegel A, Collins F, Chandrasekharappa S, Marx S and Oliver B. Genetic interactions between Drosophila melanogaster menin and Jun/Fos. *Dev Biol.* **298**, 59-70 (2006).

370. Shtir C, Nagakawa IS, Duren WL, Conneely KN, Scott LJ, Silander K, Valle TT, Tuomilehto J, Buchanan TA, Bergman RN, Collins FS, Boehnke M, Watanabe RM. Subsets of Finns with high HDL to total cholesterol ratio show evidence for linkage to type 2 diabetes on chromosome 6q. *Hum Hered.* **63**, 17-25. (2007).

371. Collins FS, Manolio TA. Merging and emerging cohorts: necessary but not sufficient. *Nature.* **445**, 259 (2007).

372. Willer CJ, Bonnycastle LL, Conneely KN, Duren WL, Jackson AU, Scott LJ, Narisu N, Chines PS, Skol A, Stringham HM, Petrie J, Erdos MR, Swift AJ, Enloe ST, Sprau AG,... Mohlke KL, Collins FS, Boehnke M. Screening of 134 single nucleotide polymorphisms (SNPs) previously associated with type 2 diabetes replicates association with twelve SNPs in nine genes. *Diabetes.* **56**, 256-264 (2007).

373. Baffoe-Bonnie AB, Kittles RA, Gillanders E, Ou L, George A, Robbins C, Ahaghotu C, Bennett J, Boykin W, Hoke G, Mason T, Pettaway C, Vijayakumar S, Weinrich S, Jones MP,... Collins FS,... Dunston GM, Powell I, Carpten JD. Genome-wide linkage of 77 families from the African American Hereditary Prostate Cancer Study (AAHPC). *Prostate.* **67**, 22-31 (2007).

374. The International Mouse Knockout Consortium. Collins FS, Rossant J, and Wurst W. A mouse for all reasons. *Cell.* **128**, 9-13 (2007).

375. Manolio TA, Collins FS. Genes, environment, health and disease: facing up to complexity. *Hum Hered.* **63**, 63-66 (2007).

376. Agarwal SK, Impey S, McWeeney S, Scacheri PC, Collins FS, Goodman RH, Spiegel AM, Marx SJ. Distribution of menin-occupied regions in chromatin specifies a broad role of menin in transcriptional regulation. *Neoplasia.* **9**, 101-107 (2007).

377. Sieving PA, Collins FS. Genetic ophthalmology and the era of clinical care. *JAMA.* **297**, 733-736 (2007).

378. Cao K, Capell BC, Erdos MR, Djabali K, and Collins FS. A lamin A protein isoform overexpressed in Hutchinson-Gilford progeria syndrome interferes with mitosis in progeria and normal cells. *Proc Natl Acad Sci USA.* **104**, 4949-4954 (2007).

379. Ozawa A, Agarwal SK, Mateo CM, Burns AL, Rice TS, Kennedy PA, Quigley CM, Simonds WF, Weinstein LS, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ. The parathyroid/pituitary variant of multiple endocrine neoplasia type 1 usually has causes other than p27Kip1 mutations. *J Clin Endocrinol Metab.* **92**, 1948-1951 (2007).

380. Scott LJ, Mohlke KL, Bonnycastle LL, Willer CJ, Li Y, Duren WL, Erdos MR, Stringham HM, Chines PS, Jackson AU, Prokunina-Olsson L, Ding CJ, Swift AJ, Narisu N, Hu T,... Tuomilehto J, Collins FS, Boehnke M. A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants. *Science.* **316**, 1341-1345 (2007).

381. Schwartz D, Collins FS. Environmental biology and human disease. *Science.* **316**, 695-696 (2007).

382. The ENCODE Project Consortium. The ENCODE pilot project: identification and analysis of functional elements in 1% of the human genome. *Nature.* **447**, 799-816 (2007).

383. NCI-NHGRI Working Group on Replication in Association Studies. Chanock SJ, Manolio T, Boehnke M, Boerwinkle E, Hunter DJ, Thomas G, Hirschhorn JN, Abecasis G, Altshuler D, Bailey-Wilson JE, Brooks LD, Cardon LR, Daly M, Donnelly P, Fraumeni JF Jr,... Wijsman EM, Winn DM, Collins FS. Replicating genotype–phenotype associations. *Nature.* **447**, 655-660 (2007).

384. Capell BC, Collins, FS, Nabel EG. Mechanisms of cardiovascular disease in accelerated aging syndromes. *Circ Res.* **101**, 13-26 (2007).

385. Lowrance WW, Collins FS. Identifiability in genomics research. *Science.* **317**, 600-602 (2007).

386. The GAIN Collaborative Research Group. New models of collaboration in genome-wide association studies: the Genetic Association Information Network. *Nat Genet.* **39**, 1045-1051 (2007).

387. International HapMap Consortium. A second generation human haplotype map of over 3.1 million SNPs. *Nature.* **449**, 851-861 (2007).

388. Ji Y, Prasad NB, Novotny EA, Kaur S, Elkahloun A, Chen Y, Zhang RZ, Chu ML, Agarwal SK, Marx SJ, Collins FS, Chandrasekharappa SC. Mouse embryo fibroblasts lacking the tumor suppressor menin show altered expression of extracellular matrix protein genes. *Mol Cancer Res.* **5**, 1041-1051 (2007).

389. McClintock D, Ratner D, Lokuge M, Owens DM, Gordon LB, Collins FS, Djabali K. The mutant form of lamin A that causes Hutchinson-Gilford Progeria is a biomarker of cellular aging in human skin. *PLoS One.* **2**, e1269 (2007).

390. Sabeti PC, Varilly P, Fry B, Lohmueller J, Hostetter E, Cotsapas C, Xie X, Byrne EH, McCarroll SA, Gaudet R, Schaffner SF, Lander ES, The International HapMap Consortium. Genome-wide detection and characterization of positive selection in human population. *Nature.* **449**, 913-918 (2007).

391. Merideth MA, Gordon LB, Clauss S, Sachdev V, Smith ACM, Perry MB, Brewer CC, Zalewski C, Kim HJ, Solomon B, Brooks BP, Gerber LH, Turner ML, Domingo DL, Hart TC,... Collins FS,... Cannon III RO, Gahl WA, Introne WJ. Phenotype and course of Hutchinson-Gilford progeria syndrome. *N Engl J Med.* **358**, 592-604 (2008).

392. Sanna S, Jackson AU, Nagaraia R, Willer CJ, Chen WM, Bonnycastle LL, Shen H, Timpson N, Lettre G, Usala G, Chines PS, Stringham HM, Scott L, Dei M, Lai S,... Collins FS,... Boehnke M, Abecasis GR, Mohlke KL. Common variants in the GDF5-UQCC region are associated with variation in human height. *Nat Genet.* **40**, 198-203 (2008).

393. Willer CJ, Sanna S, Jackson AU, Scuteri A, Bonnycastle LL, Clarke R, Heath SC, Timpson NJ, Najjar SS, Stringham HM, Strait J, Duren WL, Maschio A, Busonero F, Mulas A,... Collins FS,... Schlessinger D, Mohlke KL, Abecasis GR. Newly identified loci that influence lipid concentrations and risk of coronary artery disease. *Nat Genet.* **40**, 161-169 (2008).

394. Manolio TA, Brooks LD, Collins FS. A HapMap harvest of insights into the genetics of common disease. *J Clin Invest.* **118**, 1590-1605 (2008). [PMC2336881]

395. Zeggini E, Scott LJ, Saxena R, Voight BF, Marchini JL, Hu T, de Bakker PI, Abecasis GR, Almgren P, Andersen G, Ardlie K, Boström KB, Bergman RN, Bonnycastle LL, Borch-Johnsen K,... Collins FS,... McCarthy MI, Boehnke M, Altshuler D. Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. *Nat Genet.* **40**, 638-645 (2008). [PMC2672416]

396. Loos RJ, Lindgren CM, Li S, Wheeler E, Zhao JH, Prokopenko I, Inouye M, Freathy RM, Attwood AP, Beckmann JS, Berndt SI; The Prostate, Lung, Colorectal, and Ovarian (PLCO) Cancer Screening Trial, Jacobs KB, Chanock SJ, Hayes RB,... Collins FS, Boehnke M, Mohlke KL. Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nat Genet.* **40**, 768-775 (2008). [PMC2669167]

397. Chen WM, Erdos MR, Jackson AU, Saxena R, Sanna S, Silver KD, Timpson NJ, Hansen T, Orru M, Piras MG, Bonnycastle LL, Willer CG, Lyssenko V, Shen H, Kuusisto J,... Collins FS,... Boehnke M, Scuteri A, Watanabe RM. Variations in the G6PC2/ABCB11 genomic region are associated with fasting glucose levels. *J Clin Invest.* **118**, 2620-2628 (2008). [PMC2398737]

398. Capell BC, Olive M, Erdos MR, Cao K, Faddah DA, Tavarez UL, Conneely KN, Qu X, San H, Ganesh SK, Chen X, Avallone H, Kolodgie F, Virmani R, Nabel E, Collins F. A farnesyltransferase inhibitor prevents both the onset and late progression of cardiovascular disease in a progeria mouse model. *Proc Natl Acad Sci USA.* **105**, 15902-15907 (2008). [PMC2562418]

399. Sethupathy P, Collins FS. MicroRNA target site polymorphisms and human disease *Trends Genet.* **24**, 489-497 (2008).

400. Gaulton KJ, Willer CJ, Li Y, Scott LJ, Conneely KN, Jackson AU, Duren WL, Chines PS, Narisu N, Bonnycastle LL, Luo J, Tong M, Sprau AG, Pugh EW, Doheny KF, Valle TT,... Collins FS, Boehnke M, Mohlke KL. Comprehensive association study of type 2 diabetes and related quantitative traits with 222 candidate genes. *Diabetes.* **57**, 3136-3144 (2008). [PMC2570412]

401. Sagelius H, Rosengardten Y, Hanif M, Erdos MR, Rozell B, Collins FS, Eriksson M. Targeted transgenic expression of the mutation causing Hutchinson-Gilford progeria syndrome leads to proliferative and degenerative epidermal disease. *J Cell Sci.* **121**, 969-978 (2008).

402. Shen HC, Rosen JE, Yang LM, Savage SA, Burns AL, Mateo CM, Agarwal SK, Chandrasekharappa SC, Spiegel AM, Collins FS, Marx SJ, Libutti SK. Parathyroid tumor development involves deregulation of homeobox genes. *Endocr Relat Cancer.* **15**, 267-275 (2008). [PMC3133970]

403. Agalliu I, Suuriniemi M, Prokunina-Olsson L, Johanneson B, Collins FS, Stanford JL, Ostrander EA. Evaluation of a variant in the transcription factor 7-like 2 (TCF7L2) gene and prostate cancer risk in a population-based study. *Prostate.* **68**, 740-747 (2008). [PMC2765224]

404. Manolio T, Collins FS. The HapMap and genome-wide association studies in diagnosis and therapy. *Annu Rev Med.* **60**, 443-456 (2009). [PMC2336881]

405. Prokopenko I, Langenberg C, Florez JC, Saxena R, Soranzo N, Thorleifsson G, Loos RJ, Manning AK, Jackson AU, Aulchenko Y, Potter SC, Erdos MR, Sanna S, Hottenga JJ, Wheeler E,... Collins FS,... Wareham NJ, Meigs JB, Abecasis GR. Variants in MTNR1B influence fasting glucose levels. *Nat Genet.* **41**, 77-81 (2009). [PMC2682768]

406. Lindgren CM, Heid IM, Randall JC, Lamina C, Steinthorsdottir V, Qi L, Speliotes EK, Thorleifsson G, Willer CJ, Herrera BM, Jackson AU, Lim N, Scheet P, Soranzo N, Amin N, Aulchenko YS,... Collins FS,... Mohlke KL, Barroso I, McCarthy MI. Genome-wide association scan meta-analysis identifies three loci influencing adiposity and fat distribution. *PLoS Genet.* **5**, e1000508 (2009). [PMC2695778]

407. Novotny E, Compton S, Liu PP, Collins FS, Chandrasekharappa SC. In vitro hematopietic differentiation of mouse embryonic stem cells requires the tumor suppressor menin and is mediated by Hoxa9. *Med Dev.* **126**, 517-522 (2009). [PMC2717021]

408. Willer CJ, Speliotes EK, Loos RJ, Li S, Lindgren CM, Heid IM, Berndt SI, Elliott AL, Jackson AU, Lamina C, Lettre G, Lim N, Lyon HN, McCarroll SA, Papadakis K,... Collins FS,...Barroso I, Abecasis GR, Hirschhorn JN; Genetic Investigation on Anthropometric Traits Consortium. Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. *Nat Genet.* **41**, 25-34 (2009). [PMC2695662]

409. Kathiresan S, Willer CJ, Peloso GM, Demissie S, Musunuru K, Schadt EE, Kaplan L, Bennett D, Li Y, Tanaka T, Voight BF, Bonnycastle LL, Jackson AU, Crawford G, Surti A,... Collins FS,... Abecasis GR, Mohlke KL, Cupples LA. Common variants at 30 loci contribute to polygenic dyslipidemia. *Nat Genet.* **41**, 56-65 (2009). [PMC2881676]

410. Khoury MJ, McBride C, Schully SD, Ioannidis JPA, Feero WG, Janssens ACJW, Gwinn M, Simons-Morton DG, Bernhardt JM, Cargill M, Chanock SJ, Church GM, Coates RJ, Collins FS, Croyle RT,... Wanke KL, Witte JS, Xu J. The scientific foundation for personal genomics: recommendations from an NIH-CDC multidisciplinary workshop. *Genet Med.* **11**, 559-567 (2009). [PMC2936269]

411. Stancakova A, Kuulasmaa T, Paananen J, Jackson AU, Bonnycastle LL, Collins FS, Boehnke M, Kuusisto J, Laakso M. Association of 18 confirmed susceptibility loci for Type 2 diabetes with indices of insulin release, proinsulin conversion, and insulin sensitivity in 5,327 non-diabetic Finnish men. *Diabetes*. **58**, 2129-36 (2009). [PMC2731523]

412. Newton-Cheh C, Johnson T, Gateva V, Tobin MD, Bochud M, Coin L, Najjar SS, Zhao JH, Heath SC, Eyheramendy S, Papadakis K, Voight BF, Scott LJ, Zhang F, Farrall M,... Collins FS,... Abecasis GR, Caulfield M, Munroe PB. Genome-wide association study identifies eight loci associated with blood pressure. *Nat Genet*. **41(6)**, 666-76 (2009). [PMC2891673]

413. Prokunina-Olsson L, Welch C, Hansson O, Adhikari N, Scott L, Usher N, Tong M, Sprau A, Bonnycastle L, Erdos M, He Z, Saxena R, Harmon B, Kotova O, Hoffman E, Altshuler D, Groop L, Boehnke M, Collins F, Hall JL. Tissue-specific alternative splicing of TCF7L2. *Hum Mol Genet*. **18**, 3795-804 (2009). [PMC2748888]

414. Prokunina-Olsson L, Kaplan LM, Schadt EE, Collins FS. Alternative Splicing of *TCF7L2* Gene in omental and subcutaneous adipose tissue and risk of type 2 diabetes. *PLoS ONE*. **4(9)**, e7231 (2009). [PMC2747626]

415. Hindorff LA, Sethupathy P, Junkins HA, Ramos EM, Mehta JP, Collins FS, Manolio TA. Potential etiologic and functional implications for genome-wide association loci for human diseases and traits. *Proc Natl Acad Sci USA*. **106**, 9362-7 (2009). [PMC2687147]

416. Manolio TA, Collins FS, Cox NJ, Goldstein, DB, Hindorff LA, Hunter DJ, McCarthy MI, Ramos EM, Cardon LR, Chakravarti A, Cho JH, Guttmacher AE, Kong A, Kruglyak L, Mardis E, Rotimi CN, Slatkin M, Valle D, Whittemore AS, Boehnke M, Clark AG, Eichler EE, Gibson G, Haines JL, Mackay TFC, McCarroll, SA, Visscher PM. Finding the missing heritability of complex diseases. *Nature*. 461, 747-753 (2009). [PMC2831613]

417. Dupuis J, Langenberg C, Prokopenko I, Saxena R, Soranzo N, Jackson AU, Wheeler E, Glazer NL, Bouatia-Naji N, Lindgren CM, Magi R, Morris AP, Randall J, Rybin D, Johnson T, ...Collins FS,... McCarthy MI, Florez JC, Barroso I. New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. *Nat Genet*. **42**, 105-116 (2010). [PMC3018764]

418. Kong A, Steinthorsdottir V, Masson G, Thorleifsson G, Sulem P, Besenbacher S, Jonasdottir A, Sigurdsson A, Kristinsson KT, Jonasdottir A, Frigge ML, Gylfason A, Olason PI, Gudjonsson SA, Sverrisson S, Stacey SN, Sigurgeirsson B,...Collins FS,...Altschuler D, Boehnke M, McCarthy MI. Parental origin of sequence variants associated with complex diseases. *Nature*. **462**, 868-874 (2009). [PMC Journal – In Process]

419. Temple G, Gerhard DS, Rasooly R, Feingold EA, Good PJ, Robinson C, Mandich A, Derge JG, Lewis J, Shoaf D, Collins FS, ... Hopkins RF, Ohara O, Wiemann S. The completion of the Mammalian Gene Collection (MGC). *Genome Res*. **12**, 2324-2333 (2009). [PMC2792178]

420. Taimen P, Pfleghaar K, Shimi T, Möller D, Ben-Harush K, Erdos MR, Adam SA, Herrmann H, Medalia O, Collins FS, Goldman AE, Goldman RD. A progeria mutation reveals functions for lamin A in nuclear assembly, architecture, and chromosome organization. *Proc Natl Acad Sci USA*. **106**, 20788-93 (2009). [PMC2779830]

421. Saxena R, Hivert MF, Langenberg C, Tanaka T, Pankow JS, Vollenweider P, Lyssenko V, Bouatia-Naji N, Dupuis J, Jackson AU, Kao WHL, Li M, Glazer NL, Manning AK, Luan J, Stringham HM,... Collins FS,... Groop L, Wareham NJ, Watanabe RM. Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. *Nat Genet*. **42**, 142-148 (2010). [PMC2922003]

422. Collins FS. Opportunities for research and NIH. *Science*. **327**, 36-37 (2010).

423. Rockey SJ, Collins FS. Managing financial conflict of interest in biomedical research. *JAMA*. **303**, 2400-2 (2010).

424. Feero WG, Guttmacher AE, Collins FS. Genomic medicine—an updated primer. *N Engl J Med* **362**, 2001-11 (2010).

425. Lauer MS, Collins FS. Using science to improve the nation's health system: NIH's commitment to comparative effectiveness research. *JAMA.* **303**, 2182-3 (2010).

426. Hamburg MA, Collins FS. The path to personalized medicine. *N Engl J Med* **363,** 301-4 (2010).

427. Clancy C, Collins FS. Patient-Centered Outcomes Research Institute: the intersection of science and health care. *Sci Transl Med.* **2,** 37 (2010).

428. Hudson TJ, Anderson W, Aretz A, Barker AD, Bell C, Bernabe RR, Bhan MK, Calvo F, Eerola I, Gerhard DS, Guttmacher A, Guyer M, ... Collins FS, ...Wainwright BJ, Wilson RK, Yang H. International network of cancer genome projects. *Nature.* **464**, 993-998 (2010). [PMC2902243]

429. Ingelsson E, Langenberg C, Hivert MF, Prokopenko I, Lyssenko V, Dupuis J, Mägi R, Sharp S, Jackson AU, Assimes TL, Shrader P, Knowles JW, Zethelius B, Abbasi FA, Bergman RN, Bergmann A, ... Collins FS, ... Groop L, Watanabe RM, Florez JC. Detailed physiologic characterization reveals diverse mechanisms for novel genetic loci regulating glucose and insulin metabolism in humans. *Diabetes.* **59,** 1266-75 (2010). [PMC2857908]

430. McDaniell R, Bum-Kyu L, Song L, Liu Z, Boyle AP, Erdos MR, Scott LJ, Morken MA, Kucera KS, Batttenhouse A, Keefe D, Collins FS, Willard HF, Lieb JD, Furey TS, Crawford GE, Iyer VR, Birney E. Heritable individual-specific and allele-specific chromatin signatures in humans. *Science.* **328,** 235-9 (2010). [PMC2929018]

431. Olive M, Harten I, Mitchell R, Beers J, Djabali K, Cao K, Erdos MR, Blair C, Funke B, Smoot L, Gerhard-Herman M, Machan JT, Kutys R, Virmani R, Collins FS, Wight TN, Nabel EG, Gordon LB. Cardiovascular pathology in Hutchinson-Gilford Progeria: Correlation with the vascular pathology of aging. *Arterioscler Thromb Vasc Biol.* **30,** 2301-9 (2010). [PMC265471]

432. Lango AH, Estrada K, Lettre G, Berndt SI, Weedon MN, Rivadeneira F, Willder CJ, Jackson AU, Vedantam S, Raychaudhuri S, Ferreira T, Wood AR, Weyant RJ, Segre AV, Speliotes EK, Wheeler E, Soranzo N, Park JH, Yang J, Gudbjartsson D,...Collins FS,...Abecasis GR, Stefansson K, Frayling TM, Hirschhorn JN. Hundreds of variants clustered in genomic loci and biological pathways affect human heights. *Nature.* **467**, 832-8 (2010). [PMC2955183]

433. Teer JK, Bonnycastle LL, Chines PS, Hansen NF, Aoyama N, Swift AJ, Abaan HO, Albert TJ, NISC Comparative Sequencing Program, Margulies EH, Green ED, Collins FS, Mullikin JC, Biesecker LG. Systematic comparison of three genomic enrichment methods for massively parallel DNA sequencing. *Genome Res.* **20**, 1420-31 (2010). [PMC2945191]

434. 1000 Genomes Project Consortium, Durbin RM, Abecasis GR, Altshuler DL, Auton A, Brooks LD, Durbin RM, Gibbs RA, Hurles ME, McVean GA. A map of human genome variation from population-scale sequencing. *Nature.* **67,** 1061-73 (2010). [PMC3042601]

435. Speliotes EK, Willer CJ, Berndt SI, Monda KL, Thorleifsson G, Jackson AU, Allen HL, Lindgren CM, Luan J, Magi R, Randall JC, Vedantam S, Winkler TW, Qi L, Workalemahu T, Heid IM...Collins F,...McCarthy MI, Hirschhorn JN, Ingelsson E, Loos RJ. Association analyses of 249,796 individuals reveal of 18 new loci associated with body mass index. *Nat. Genet.* **42**, 937-8 (2010). [PMC3014648]

436. Heid IM, Jackson AU, Randall JC, Winkler TW, Qi L. Steinthorsdottire V, Thorleifsson G, Zillikens MC, Speliotes EK, Magi R, Workalemahu T. White CC, Bouatia-Naji N, Harris TB, Berndt SI, Ingelsson E, Willer CJ, ...Collins F, ...McCarthy MI, Fox CS, Mohlke KL, Lindgren CM. Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nat. Genet.* **42**, 949-60 (2010). [PMC3000924]

437. Tabara Y, Kohara K, Kita Y, Hirawa N, Katsuya T, Ohkubo T, Hiura Y, Tajima A, Morisaki T, Miyata T, Nakayama T, Takashima N, Nakura J, Kawamoto R, Takahashi N, Hata A, Soma M, Imai Y, Kokubo Y, Okamura T, Tomoike H, Iwai N, Ogihara T, Inoue, I, Tokunaga K, Johnson T, Caulfield M, Munroe P, Global Blood Pressure Genetics Consortium, Umemura S, Ueshima H, Miki T. Common variants in the ATP2B1 gene

are associated with susceptibility to hypertension: the Japanese Millennium Genome Project. *Hypertension.* **56,** 973-80 (2010). [PMC20921432]

438. Frieden TR, Collins FS. Intentional infection of vulnerable populations in 1946-1948: another tragic history lesson. *JAMA.* **304,** 2063-4 (2010). [PMC20937719]

439. Sudmant PH, Kitzman JO, Antonacci F, Aikan C, Malig M. Tsalenka A, Sampas N. Bruhn L, Shendure J, 1000 Genomes Project, Eichler EE. Diversity of human copy number variation and multicopy genes. *Science.* **330,** 641-6 (2010). [PMC3020103]

440. Stitzel ML, Sethupathy P, Pearson DS, Chines PS, Song L, Erdos MR, Welch R, Parker SC, Boyle AP, Scott LJ, NISC Comparative Sequencing Program, Margulies EH, Boehnke M, Furey TS, Crawford GE, Collins FS. Global epigenomics analysis of primary human pancreatic islets provides insights into type 2 diabetes susceptibility loci. *Cell Metabolism.* **12,** 443-55 (2010). [PMC3026436]

441. Ikram MK, Xueling S, Jensen RA, Cotch MF, Hewitt AW, Ikram MA, Wang JJ, Klein R, Klein BE, Breteler MM, Cheung N, Liew G, Mitchell P, UItterlinden AG, Rivadeneira F...Global BPgen Consortium...Siscovick DS, Vingerling JR, Wong TY. Four novel Loci (19q13, 6q24, 12q24, and 5q14) influence the microcirculation in vivo. *PLoS Genet.* **6,** (2010). [PMC2965750]

442. Collins FS, Glass RI, Whitescarver J, Wakefield M, Goosby EP. Public Health: Developing health workforce capacity in Africa. *Science.* **330,** 1324-5 (2010). [PMC21079014]

443. Voight BF, Scott LJ, Steinthorsdottir V, Morris AP,...Collins FS,...Altshuler D, Boehnke M, McCarthy MI. Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nat. Genet.* **42,** 579-89 (2010). [PMC3080658]

444. Mills RE, Walter K, Stewart C,...Korbel JO, 1000 Genomes Project. Mapping copy number variation by population-scale genome sequencing. *Nature.* **470,** 59-65 (2011). [PMC3077050]

445. Speliotes EK, Yerges-Armstrong LM, ...GOLD Consortium. Genome-wide association analysis identifies variants associate with nonalcoholic fatty liver disease that have distinct effect on metabolic traits. *PLos Genet.* **7,** e1001324 (2011). [PMC3053321]

446. Aylor DL, Valdar W,...Collins FS, Threadgill D, Pardo-Manuel de Villena F, Churchill GA. Genetic analysis of complex traits in the emerging collaborative cross. *Genome Res.* **21,** 1213-22 (2011). [PMC21406540]

447. Kelada SN, Wilson MS, Tavarez U, Kubalanza K, Borate B, Whitehead G, Maruoka S, Roy MG, Olive M, Carpenter DE, Brass DM, Wynn TA, Cook DA, Evans CM, Schwartz DA, Collins FS. Strain-dependent genomic factors affect allergen-induced airway hyper-responsiveness in mice. *Am J Respir Cell Mol Biol.* **45,** 817-24 (2011). [PMC21378263]

448. Verstraeten VL, Peckham LA, Olive M, Cappell BC, Collins FS, Nabel EG, Young SG, Fong LG, Lammerding J. Protein farnesylation inhibitors cause donut-shaped cell nuclei attributable to a centrosome separation defect. *Proc Natl Acad Sci USA.* **108,** 4997-5002 (2011). [PMC3064351]

449. Stancakova A, Paananen J, Soininen P, Kangas AJ, Bonnycastle LL, Morken MA, Collins FS, Jackson AU, Boehnke ML, Kuusisto J, Ala-Korpela M, Laakso M. Effects of 34 risk loci for type 2 diabetes or hyperglycemia on lipoprotein subclasses and their composition in 6,580 nondiabetic finnish men. *Diabetes.* **60,** 1608-1016 (2011). [PMC21421807]

450. Cao K, Blair CD, Faddah DA, Kieckhaefer JE, Olive M, Erdos MR, Nabel EG, Collins FS. Progerin and telomere dysfunction collaborate to trigger cellular senescence in normal human fibroblasts. *J Clin. Invest.* **121,** 2833-44 (2011). [PMC21670498]

451. Cao K, Graziotto JJ, Blair CD, Mazzulli JR, Erdos MR, Krainc D, Collins FS. Rapamycin reverses cellular phenotypes and enhances mutant protein clearance in Hutchinson-Gilford progeria syndrome cells. *Sci. Transl. Med.* **3**: 89ra58 (2011). [PMC21715679]

452. Collins FS. Reengineering translational science: the time is right. *Sci. Transl. Med.* **3**: 90cm17 (2011). [PMC21734173]

453. Jafar-Mohammadi B, Groves CJ, Gjesing AP, Herrera BM, Winckler W, Stringham HM, Morris AP, Lauritzen T, Doney ASF, Morris AD, Weedon MS, Swift AJ, Kuusisto J, Laakso M, Altshuler D, Hattersley AT, Collins FS, Boehnke M, Hansen T, Pedersen O, Palmer CNA, Frayling TM, DIAGRAM consortium, Gloyn AL, McCarthy MI. A role for coding functional variants in HNF4A in type 2 diabetes susceptibility. *Diabetologia.* **54,** 111-119 (2011). [PMC3119815]

454. Strawbridge RJ, Dupuis J, Prokopenko I, Barker A, Ahlqvist E, Rybin D, Petrie JR……Collins FS, Hopewell JC, Saleheen D, Chambers J, Parish S, Danesh J. Genome-Wide association identifies nine common variants associated with fasting proinsulin levels and provides new insights into the pathophysiology of the type 2 diabetes. *Diabetes.* **60**, 2624-34 (2011). [PMC21873549]

455. Wain LV, Verwoert GC, O'Reilly PF, Shi G, Johnson T, Johnson AD….. Tobin, MD, Elliott P, van Duijn CM, LifeLines Cohort, EchoGen consortium, AortaGen Consortium, CHARGE Consortium Heart Failure Working Group, KidneyGen consortium, CKDGen consortium, Cardiogenics consortium, CardioGram. Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nat. Genet.* **43**, 1005-11 (2011). [PMC21909110]

456. International Consortium for Blood Pressure Genome-Wide Association Studies, Ehret GB, Munroe PB, Rice KM, Bochud M…..Collins FS…..Abecasis GR, Lightstone L, Scott J. Navis G, Elliott P, Kooner JS. Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature.* **478**, 103-9 (2011). [PMC21909115]

457. Zhu H, Shyh-ChangN, Segre AV, Shinoda G, Shah SP, Einhorn WS, Takeuchi A, Engreitz JM, Hagan JP, Kharas MG, Urbach A, Thornton JE, Triboulet R, Gregory RI, DIAGRAM Consortium, MAGIC Investigators, Altshuler D, Dailey GQ. The Lin28/let-7 axis regulates glucose metabolism. *Cell.* **147**, 81-94 (2011). [PMC21962509]

458. Martin WJ 2nd, Glass RI, Balbus JM, Collins FS. Public health: A major environmental cause of death. *Science.* **334**, 180-1 (2011). [PMC21998373]

459. Chambers JC, Zhang W, Sehmi J, Li X, Wass MN, Van der Harst P, Holm H…..DIAGRAM consortium…..Scott J, Jarvelin MR, Elliott P, Kooner JS. Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. *Nat. Genet.* **43**, 1131-8 (2011). [PMC22001757]

460. Melgar MF, Collins FS, Sethupathy P. Discovery of active enhancers through bidirectional express of short transcripts. *Genome Biol.* **12**, R113 (2011). [PMC22082242]

461. Stancakova A, Civelek M, Saleem NK, Soininen P, Kangas AJ, Cederbert H, Paananen J, Pihiajamaki J, Bonnycastle LL, Morken MA, Boehnke M, Pajukanta P, Lusis AJ, Collins FS, Kuusisto J, Ala-Korpela M, Laakso M. Hyperglycemia and a Common Variant of GCKR are Associated with the Levels of Eight Amino Acides in 9,369 Finnish Men. *Diabetes.*

462. Johnson T, Gaunt TR, Newhouse SJ, Padmanabhan S, Tomaszewski M…..Cardiogenics Consortium…..Farrall M, Hingorani AD, Samani NJ, Caulfield MJ, Munroe PB. Blood pressure loci identified with gene-centric array. *Am. J. Hum. Genet.* **89**, 688-700 (2011). [PMC22100073]

463. Cho YS, Chen CH, Hu C, Long J, Ong RT...DIAGRAM Consortium.....Shu XO, Mohlke KL, Kato N, Han BG, Seielstad M. Meta-analysis of genome-wide association studies identifies eight new loci for type 2 diabetes in east Asians. *Nat. Genet.* **44**, 67-72 (2011). [PMC22158537]

464. Graziotto JJ, Cao K, Collins FS, Krainc D. Rapamycin activates authophagy in Hutchinson-Gilford progeria syndrome: implications for normal aging and age-dependent neurodegenerative disorders. *Autophagy.* **8**, 147-51 (2012). [PMC22170152]

465. Rees MG, Ng D, Ruppert S, Turner C, Beer NL.....Biesecker LG, Gloyn AL, Collins FS. Correlation of rare coding variants in the gene encoding human glucokinase regulatory protein with phenotypic, cellular, and kinetic outcomes. *J Clin Invest.* **122**, 205-217 (2012). [PMC 22182842]

466. Marquez M, Huyvaert M, Perry JR, Pearson RD, Falchi M.....DIAGRAM Consortium. Low-frequency variants in HMGA1 are not associated with type2 diabetes risk. *Diabetes.* **61**, 524-30 (2012). [PMC22210315]

467. Palmer ND, McDonough CW, Hicks PJ, Roh BH, Wing MR...DIAGRAM Consortium...Waterworth DM, Vollenweider P, Peltonen L, Mooser V, Sladek R. A genome-wide association search for type 2 diabetes genes in African Americans. *PLoS One.* **7**, e29202 (2012). [PMC22238593]

468. Conneely KN, Capell BC, Erdos MR, Sebastiani P, Solovieff N, Swift AJ, Baldwin CT, Budagov T, Barzilai N, Atzmon G, Puca AA, Perls TT, Geesaman BJ, Boehnke M, Collins FS. Human longevity and common variations in the LMNA gene: a meta-analysis. *Aging Cell.* Doi: 10.1111/j.1474-9726.2012.00808.x. (2012). [PMC22340368]

469. Demirkan A, van Duijn CM, Ugocsai P, Isaacs A, Pramstaller PP.....DIAGRAM Consortium.....Campbell H, Schmitz G; EUROSPAN consortium. Genome-wide association study identified novel loci associated with circulating phosphor-and sphingolipid concentrations. *PLoS Genet.* 8(2), e1002490 (2012). [PMC22359512]

470. Kelada SN, Aylor DL, Peck BC, Ryan JF, Tavarez U, Buus RJ, Miller CR, Chesler EJ, Threadgill DW, Churchill GA, Pardo-Manuel de Villena F, Collins FS. Genetic analysis of hematological paraments in incipient lines of the collaborative cross. *G3 (Bethesda).* **2**, 157-65 (2012). [PMC22384394]

471. Kristiansson K, Perola M, Tikkanen E, Kettunen J, Surakka I...Collins FS, Laakso M, Hurles ME, Palotie A, Peltonen L, Ripatti S, Salomaa V. Genome-wide screen for metabolic syndrome susceptibililty loci reveals strong lipid gene contribution but no evidence for common genetic basis for clustering of metabolic syndrome traits. *Cir. Cardiovasc. Genet.* **5,** 242-9 (2012). [PMC22399527]

472. Manolio TA, Weis BK, Cowie CC, Hoover RN, Hudson K...Sprosen T, Walport M, Collins FS. New models for large prospective studies: Is there a better way? *Am. J. Epidemiol.* 2012 Mar 12 [Epub ahead of print] [PMC22411865]

473. Scott RA, Chu AY, Grarup N, Manning AK, Hivert MF...Chines PS, Collins FS.....Florez JC, Franks PW, Dupuis J, Wareham NJ, Langenberg C. No interactions between previously associated 2-h glucose gene variants and physical activity or BMI on 2-h glucose levels. *Diabetes.* 2012 Mar 13 [Epub ahead of print] [PMC22415877]

474. Stefanovski D, Youn JH, Rees M, Watanabe RM, Ader M, Ionut V, Jackson AU, Boehnke M, Collins FS, Bergman RN. Estimating hepatic glucokinase activity using a simple model of lactate kinetics. *Diabetes Care.* 2012 Mar 28 [Epub ahead of print] [PMC 22456868]

475. Dastani Z, Hivert MF, Timpson N, Perry JR...DIAGRAM Consortium...Cupples LA, Sandhu MS, Ridker PM, Rader DJ, Kathiresan S. Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45, 891 individuals. *PLoS Genet.* 8(3), e1002607 (2012). [PMC22479202]

476. Kelly MA, Rees SD, Hydrie MZ, Shera AS, Bellary S. O'Hare JP, Kumar S, Taheri S, Basit A, Barnett AH; DIAGRAM Consortium; SAT2D Consortium. Circadian gene variants and susceptibility to type 2 diabetes: a pilot study. *PLoS One.* 7(4), e32670 (2012). [PMD 22485135]

Non Peer Reviewed

1. Forget BG, Tuan D, Newman MV, Feingold EA, Collins FS, Fukumaki Y, Jagadeeswaran P, Weissman SM. Molecular studies of mutations that increase Hb F production in man in *Globin Gene Expression and Hematopoietic Differentiation: Proceedings of the Third Conference on Hemoglobin Switching, September 12-15, 1982* (eds Nienhuis AW, Stamatoyannopoulos G) 65-76 (Alan R. Liss, 1983).

2. Fukumaki Y, Collins FS, Kole R, Stoeckert CJ, Jagadeeswaran P, Duncan CH, Weissman SM. Sequences of human repetitive DNA, non-alpha globin genes and major histocompatibility locus genes. *Cold Spring Harbor Symp Quant Biol.* **47**, 1079-1086 (1983).

3. Reddy ESP, Stoeckert CJ, Collins FS, Weissman SM, Yoshie O, Schmidt H, Lengyel P. New aspects of gene conversion and expression in the globin and MHC systems in *Gene Expression in Normal and Abnormal Hematopoeisis: ICN-UCLA Symposia on Molecular and Cellular Biology.* 101-112 (Alan R. Liss, 1983).

4. Feingold EA, Collins FS, Metherall JE, Stoeckert CJ, Weissman SM, Forget BG. Analysis of mutations associated with deletion and non-deletion hereditary persistence of fetal hemoglobin in *Experimental Approaches for the Study of Hemoglobin Switching: Proceedings of the Fourth Conference on Hemoglobin Switching, October 1-3, 1984* (eds Stamatoyannopoulos G, Ingénues AW) (Alan R. Liss, 1985).

5. Collins FS, Weissman SM. Cloning at a distance with a circulization technique in *Cell Biology of the Major Histocompatibility Complex* (eds Pernis B, Vogel HJ) 37-40 (P & S Biomedical Sciences Symposia Series, Academic Press, 1985).

6. Feingold EA, Collins FS, Metherall JE, Stoeckert CJ Jr, Weissman SM, Forget BG. Molecular analysis of deletion and nondeletion hereditary persistence of fetal hemoglobin and identification of a new mutation causing beta-thalassemia. *Ann NY Acad Sci.* **445**, 159-169 (1985).

7. Duceman BW, Srivastava R, Collins FS, Chorney M, Greenspan D, Biro PA, Sood A, Weissman SM. Complexity of class I genes of the human major histocompatibility complex. *Advances Inflamm Res.* **9**, 25-31 (1985).

8. Collins FS. Chromosome hopping and human genetic disease in *DNA Probes: Applications in Genetic and Infectious Disease and Cancer* (ed Lerman LS) 67-72 (Cold Spring Harbor Laboratory, 1986).

9. Lawrance SK, Srivastava R, Chorney MJ, Rigas B, Vasavada H, Gillespie GA, Smith C, Cantor C, Collins FS, Weissman SM. The human MHC: Approaches to characterization of megabase regions of DNA. *Cold Spring Harbor Symp Quant Biol.* **51**, 123-130 (1986).

10. Shows TB, et al, McAlpine PJ, Boucheix C, Collins FS, Conneally PM, Frezal J, Gershowitz H, Goodfellow PN, Hall JG, Issitt P, Jones CA, Knowles BB, Lewis M, McKusick VA, Meisler M, Morton NE, Rubinstein P, Schanfield MS, Schmickel RD, Skolnick MH, Spence AM, Sutherland GR, Traver M, Van Cong N, Willard HF. Guidelines for human gene nomenclature: An international system for human gene nomenclature (ISGN, 1987). *Cytogenet Cell Genet.* **46**, 11-28 (1987).

11. Collins FS, Martin JB. Huntington's chorea in *McGraw-Hill Yearbook of Science and Technology,* 164-167 (McGraw-Hill, 1989).

12. Prochownik EV, Collins FS. cDNA and genomic cloning in *Molecular Genetics: Methods in Hematology, v. 20* (ed Benz EJ) 72-87 (Churchill Livingstone, 1989).

13. Dean M, Stewart C, Perry A, Gerrard B, Beck T, Rapp U, Drumm ML, Iannuzzi MC, Collins FS, O'Brien S. Genetic markers for oncogenes, growth factors, and cystic fibrosis in *Modern Trends in Human Leukemia VIII: New Results in Clinical and Biological Research Including Pediatric Oncology, Wilsede Joint Meeting on Pediatric Oncology V, Hamburg, June 17, 1988.* (ed Neth R) 360-5 (Springer-Verlag; 1989).

14. Iannuzzi MC, Collins FS. Reverse genetics and cystic fibrosis. *Am J Respir Cell Mol Biol.* **2**, 309-316 (1990).

15. Collins FS, Riordan JR, Tsui LC. The cystic fibrosis gene. isolation and significance. *Hosp Pract.* **25**, 47-57 (1990).

16. Collins FS. Identification of the type I neurofibromatosis gene. *Neuroscience Forum.* **1**, 5 (1991).

17. Tanaka M, Nolan JA, Bhargava AK, Rood K, Collins FS, Weissman SM, Forget BG, Chamberlain JW. Expression of human globin genes in transgenic mice carrying the beta-globin gene cluster with a mutation causing G gamma beta + hereditary persistence of fetal hemoglobin. *Ann NY Acad Sci.* **612**, 167-178 (1990).

18. Collins FS. The Genome Project and human health. *FASEB J.* **5**, 77 (1991).

19. Collins FS. Identification of disease genes: recent successes. *Hosp Pract.* **26**, 93-98 (1991).

20. Collins FS. Of needles and haystacks: finding human disease by positional cloning. *Clin Res.* **39**, 615-623 (1991).

21. Collins FS. Physician-scientists: a vanishing breed. *Yale Medicine.* Fall/Winter, 5-8 (1991-92).

22. Collins FS, Wilson JM. Cystic fibrosis: a welcome animal model. *Nature.* **358**, 708-709 (1992).

23. Wilson JM, Collins FS. Cystic fibrosis: More from the modellers. *Nature.* **359**, 195-196 (1992).

24. Collins FS. Identifying human disease genes by positional cloning in *The Harvey Lectures,* **86**, 149-164 (Wiley-Liss, 1992).

25. Nakamura T, Ishikawa T, Collins FS. NF1 gene and its alteration. *CRC.* 96-102. (1992) (Written in Japanese)

26. Tagle DA, Swaroop M, Lovett M, Collins FS. Magnetic bead capture of expressed sequences encoded within large genomic segments. *Nature.* **361**, 751-753 (1993).

27. Collins FS, Galas D. A new five-year plan for the U.S. Human Genome Project. *Science.* **262**, 434-436 (1993).

28. Brown MS, Collins FS, Goldstein JL, Watson JD, Wexler NS. Roundtable: The Human Genome Project. *Issues in Science & Technology.* **10.1**, 43-50 (1993).

29. Gottesman MM, Collins FS. The role of the human genome project in disease prevention. *Prev Med.* **23**, 591-594 (1994).

30. Caskey CT, Collins FS, Juengst ET, McKusick VA. Human genes: the map takes shape. *Patient Care.* **28**, 28-34 (1993).

31. Collins FS. Genetic protocols in human genetics in *Current Protocols in Human Genetics.* (J. Wiley and Sons, 1994).

32. Collins FS. Colon cancer screening. response (letter). *Science.* **264**, 13-14 (1994).

33. Hudson K, Collins FS (letter to editor). *Nature.* **270**, 1423 (1995).

34. Collins FS, Fink L. The Human Genome Project. *Alcohol Health & Research World.* **19**, 190-195 (1995).

35. Jordan E, Collins FS. A march of genetic maps. *Nature.* **380**, 111-112 (1996).

36. Collins FS (letter to editor). *New Engl J Med.* **334**, 1205 (1996).

37. NAPBC Hereditary Susceptibility Working Group. Genetic screening for breast cancer. *J Clin Oncology.* **14**, 1738-1740 (1996).

38. Collins FS. Sequencing the human genome. *Hosp Pract.* **32**, 35-43, 46-49, 53-54 (1997).

39. Fink L, Collins FS. The Human Genome Project: view from the National Institutes of Health. *J Am Med Women's Assoc.* **52**, 4-7, 15 (1997).

40. Kahn MJ, Jamison KR, Collins FS. Protecting our family secrets. *The Washington Post,* A15, July 31, 1997.

41. Collins FS. Preparing health professionals for the genetic revolution. *JAMA.* **278**, 1285-1286 (1997).

42. Francomano CA, Collins FS. The Human Genome Project: implications for medical practice. *Today's Internist.* **38**, 11-15 (1997).

43. Collins FS, Jenkins JF. Implications of the Human Genome Project for the nursing profession in *The Genetics Revolution: Implications for Nursing.* (ed Lashley FR) (American Academy of Nursing; 1997).

44. Collins FS. Foreword in *Inherited Susceptibility to Cancer: Clinical, Predictive, and Ethical Perspectives* (eds Foulkes WD, Hodgson SV) xii-iv (Cambridge University Press, 1998).

45. Marx SJ, Agarwal SK, Kester MB, Heppner C, Kim YS, Skarulis MC, James LA, Goldsmith PK, Saggar SK, Park SY, Spiegel AM, Burns AL, Debelenko LV, Zhuang Z, Lubensky IA, Liotta LA, Emmert-Buck MR, Guru SC, Manickam P, Crabtree J, Erdos MR, Collins FS, Chandrasekharappa SC. Multiple endocrine neoplasia type 1. clinical and genetic features of the hereditary endocrine neoplasias. *Recent Prog Horm Res.* **54**, 397-439 (1999). Presented at the 54th Recent Progress in Hormone Research conference, Stevenson, WA, August 1-5, 1998.

46. Collins FS. Genetics: not just in there somewhere, but at the very center of medicine. *Genetics in Medicine*, **1**, 3 (1998).

47. Collins FS. In *Proceedings: The Genetics of Disease Symposium and the RCMI Program Director's Meeting.* (Howard University Press, 1998).

48. Thomson ET, Collins FS. Clinical genetics in nursing practice. (ed Lashley FR) ix-x (Springer, 1998).

49. Marx SJ, Agarwal SK, Heppner C, Kim YS, Kester MB, Goldsmith PK, Skarulis MC, Spiegel AM, Burns AL, Debelenko LV, Zhuang Z, Lubensky IA, Liotta LA, Emmert-Buck MR, Guru SC, Manickam P, Crabtree JS, Collins FS, Chandrasekharappa SC. The gene for multiple endocrine neoplasia type 1: recent findings. *Bone.* **25**, 119-133 (1999). Presented at the combined meeting of the American Society for Bone and Mineral Research and of the International Bone and Mineral Society.

50. Collins FS. Microarrays and macroconsequences. *Nat Genet,* **21 S2** (1999).

51. Marx SJ, Agarwal SK, Heppner C, Kim YS, Skarulis MC, Doppman JL, James L, Park S, Bilimoria KY, Knapp JL, Spiegel AM, Burns AL, Debelenko LV, Lubensky IA, Zhuang Z, Liotta LA, Emmert-Buck MR, Guru SC, Manickam P, Crabtree J, Erdos M, Collins FS, Chandrasekharappa SC. Clinical features and pathophysiology of multiple endocrine neoplasia type 1. Presented at the Seventh International Workshop on Multiple Endocrine Neoplasia (1999).

52. Collins FS, Bochm K. Avoiding casualties in the genetic revolution: the urgent need to educate physicians about genetics. *Acad Med.* **74**, 48-49 (1999).

53. Collins FS. The human genome project and the future of medicine. *Ann N Y Acad Sci.* **882**, 42-55 (1999). Talk given at "Great Issues for Medicine in the 21st Century: A Consideration of the Ethical and Social Issues Arising Out of Advances in the Biomedical Sciences" Symposium, Dartmouth Medical School, September 1997.

54. Collins FS. Genetics: an explosion of knowledge is transforming clinical practice. *Geriatrics.* **54**, 41-47 (1999).

55. Collins FS. Shattuck lecture—medical and societal consequences of the Human Genome Project. *N Engl J Med.* **341**, 28-37 (1999).

56. Collins FS. The Human Genome Project: Tool of atheistic reductionism or embodiment of the Christian mandate to heal? *Science & Christian Belief.* **11**, 99-111 (1999).

57. Collins FS, Jegalian KG. Deciphering the code of life. *Sci Am.* **281**, 86-91 (1999).

58. Collins FS, Morgan M, Patrinos A. International cooperation and the Human Genome Project. *Harvard International Review.* Fall (1999).

59. Collins FS, Jegalian KG. Soon the Human Genome Project will be completed: What then? *The Chronicle of Higher Education.* **XLVI**, B12 (2000).

60. Collins FS. Proceedings. GAAC 4th Public Symposium, *The Changing Character, Use and Protection of Intellectual Property.* 99-107 (German-American Academic Council Foundation Press, 2000).

61. Collins FS. Realizing the Human Genome Project's potential. *Genetics Engineering News* (2000).

62. Collins FS. Future trends in research in human genetics and genomics. *Discover* (2000).

63. Guttmacher AG, Collins FS. Genetic resources on the web (GROW). *Genetics in Medicine.* **2**, 296-299 (2000).

64. Collins FS, Mansoura M. The promise of the Human Genome Project for the Huntington's disease community. Huntington's Disease Society of America's *Toward a Cure* Newsletter. (2000).

65. Collins FS, Mansoura M. The Human Genome Project: revealing the shared inheritance of all humankind. *American Cancer Society's Intercultural Cancer Council Supplement.* **91**, 221-225 (2001).

66. Collins FS, McKusick VA. Implications of the Human Genome Project for medical science. *JAMA.* **285**, 540-544 (2001).

67. Collins FS. Contemplating the end of the beginning. *Genome Res.* **11**, 641-643 (2001).

68. Collins FS, Weiss L, Hudson K. Heredity and humanity. Have no fear. Genes aren't everything. *New Republic.* 27-29 (June 2001).

69. Collins FS. Reflections from the Director of the National Human Genome Research Institute. *Dignity the Newsletter of the Center for Bioethics and Human Dignity* (2001).

70. Collins FS, Guttmacher AE. Genetics moves into the medical mainstream. *JAMA.* **286**, 2322-2324 (2001).

71. Collins FS. Translation of Genomic Research Into Health Care. *JAMA.* **285**, 2447-2448 (2001).

72. Collins, FS. Foreword in *Pharmacogenomics: Social, Ethical, and Clinical Dimensions.* (ed Rothstein MA) ix-x. (Wiley-Liss, 2003).

73. Collins FS. Genomics: the coming revolution in medicine. *Global Agenda, Magazine of the World Economics Forum Annual Meeting.* 152-154 (2003).

74. Grady PA, Collins FS. Genetics and nursing science: realizing the potential. *Nurs Res.* **52**, 69 (2003).

75. Insel TR, Collins FS. Psychiatry in the genomics era. *Am J Psychiatry.* **160**, 616-620 (2003).

76. Collins FS, Morgan M, Patrinos A. The Human Genome Project: lessons from large-scale biology. *Science.* **300**, 286-290 (2003).

77. Collins FS, Green ED, Guttmacher AE, Guyer MS. A vision for the future of genomics research. *Nature.* **422**, 835-847 (2003).

78. Jenkins J, Collins FS. Are you genetically literate? *Am J Nurs.* **103**, 13 (2003).

79. Collins FS. Faith and the human genome: *Perspectives on Science and Christian Faith.* September, 142-153 (2003). Plenary address, American Scientific Affiliation annual meeting (2002).

80. Collins FS. Genome research: the next generation in *LXVIII Cold Spring Harbor Symposium on Quantitative Biology: the genome of Homo sapiens.* (ed. Rogers J) (Cold Spring Harbor Laboratory, 2003).

81. Guttmacher AE, Collins FS. Welcome to the genomic era. *N Engl J Med.* **349**, 996-998 (2003).

82. Collins FS. The Genome Era and Mental Illness. *Nami Advocate,* 29-30 (2003).

83. Collins FS, Watson JD. Genetic Discrimination: Time to Act. *Science.* **302**, 745 (2003).

84. Collins FS, Jordan E: Human genome project in *2004 Yearbook of Science and Technology.* (eds.) 147-150 (McGraw-Hill, New York, 2004).

85. Collins FS. The case for a US prospective cohort study of genes and environment. *Nature.* **429**, 475-477 (2004).

86. Collins FS, Guttmacher AG. Foreword In: Nursing care in the genomic era: A case based approach. By Jean Jenkins and Dale Lea. Jones & Bartlett Publishers, pp. xiii-xiv (2004).

87. Collins FS. Foreword In: Genetic Testing for Cancer. By Andreas Farkas Patenaude. American Psychological Association, Washington, D.C. pp. vii-viii (2004).

88. Agarwal SK, Burns LA, Sukhodolets KE, Kennedy PA, Obungu VH, Hickman AB, Mullendore ME, Whitten I, Skarulis M, Simons WF, Mateo C, Crabtree J, Scacheri PC, Ji Y, Novotny EA, Garrett-Beal L, Ward JM, Libutti SK, Alexander RH, Cerrato A, Parisi MH, Samta SAA, Oliver B, Chandrasekharappa SC, Collins FS, Spiegel AM, Marx SJ. Molecular pathology of the MEN1 gene. *Ann NY Acad Sci.* 1014, 189-198 (2004).

89. Collins FS. News & Analysis: An audience with Francis S. Collins. *Nat Rev Drug Discov.* **3**, 640 (2004).

90. Collins FS, Tabak L. A call for increased education in genetics for dental health professionals. *J Dent Educ.* **68**, 807-808 (2004).

91. Collins FS. Genomics and the Family Physician: Realizing the potential. *Am Fam Physician.* **70**, 1637-1642 (2004).

92. Guttmacher AE, Collins FS, Carmona RH. Sounding board: the family history: more important than ever. *N Engl J Med.* **351**, 2333-2336 (2004).

93. Collins FS, et al. Advanced Placement Program Professional Development for Biology Special Topic: The Importance of Laboratory Work. *College Board,* 5-8 (2004-2005)

94. Guttmacher AE, Collins FS, Carmona RH. Family history (letter). *N Engl J Med.* **352**, 732-733 (2005).

95. Collins FS. Preface (letter). *J Sci, Math, Tech Teknos.* **14**, 3 (2005).

96. Jenkins J, Grady PA, Collins FS. Nurses and the genomic revolution. *J Nurs Scholarsh.* **37**, 98-101 (2005).

97. Collins, FS. Personalized medicine: a new approach to staying well. *The Boston Globe,* July 17, 2005.

98. Collins, FS. Exploring the frontiers of life. *Seattle Times,* August 7, 2005.

99. Collins FS, Kington R, Bonham V, Berg K, Boyer J, Brody L, Brooks L, Guttmacher A, McEwen J, Muenke M, Olson S, Rodriguez LL, Vydelingum N, Ota Wang V, Warshauer-Baker E for the Race and Genetics Working Group of the National Human Genome Research Institute. The use of racial, ethnic, and ancestral categories in human genetics research. *Am J Hum Genet.* **77**, 519-532 (2005).

100. Agarwal SK, Kennedy PA, Scacheri PC, Novotny EA, Hickman AB, Cerrato A, Rice T, Moore JB, Rao S, Ji Y, Mateo C, Libutti SK, Chandrasekharappa SC, Burns AL, Collins FS, Spiegel AM, Marx SJ. Menin molecular interactions: Insights into normal functions and tumorigenesis. *Horm Metab Res.* **37**, 369-374 (2005).

101. Collins FS, von Eschenbach A. Targeting the genetic basis of cancer. *Am Urol Assoc* (2005).

102. Collins FS. Heritage of humanity. *Nature Human Genome Collection.* June 1 (2006).

103. Collins FS. 2005 William Allan Award Address: no longer just looking under the lamppost. *Am J Hum Genet.* **79**, 421-426 (2006).

104. Collins FS, Barker AD. Mapping the cancer genome. *Scientific American.* **296**, 50-7 (2007).

105. Collins FS, Finnell RH, Rossant, J, Wurst W. A new partner for the international knockout mouse consortium. *Cell.* **129**, 235-235 (2007).

106. Feero WG, Manolio TA, Guttmacher AE, Collins FS. Validity of reported genetic risk factors for acute coronary syndrome. *JAMA.* **298**, 1757-1759 (2007).

107. Hudson KL, Holohan MK, Collins, FS, Keeping pace with the times --- The Genetic Information Nondiscrimination Act of 2008. *N Engl J Med.* **358**, 2661-2663 (2008).

108. Collins FS. Retrospective: Victor A. McKusick (1921-2008). *Science.* **321**, 925 (2008).

109. Collins F. Francis Collins interview. Departing U.S. Genome Institute Director takes stock of personalized medicine. *Science.* **320**, 1272 (2008).

110. Manolio TA, Brooks LD, Collins FS. A HapMap harvest of insights into the genetics of common disease. *J Clin Invest.* **118**, 1590-1605 (2008). [PMC2336881]

111. Feero WG, Gttmacher AE, Collins FS. The genome gets personal—almost. *JAMA.* **299**, 1351-1352 (2008).

112. Collins FS, Gray GM, Bucher JR. Toxicology. Transforming environmental health protection. *Science.* **319**, 906-907 (2008). [PMC2679521]

113. Collins FS. Francis Collins: Director of the US National Institutes of Health. Interview by Tony Kirby. *Lancet,* **374**, 969 (2009)

114. Collins FS. Opportunities and challenges for the NIH—an interview with Francis Collins. Interview by Robert Steinbrook. *N Engl J Med.* **361**, 1321-1323 (2009).

115. Collins FS. Genetic pioneer eager for NIH challenge. Interview by Deborah Levenson. *Am J Med Genet A.* **149A**, vii-ix (2009).

116. Goode EL, Szabo C, Prokunina-Olsson L, Vierkant RA, Fredreicksen ZS, Collins FS, White KL, Schmidt M, Fridley BL, Couch FJ. No association between a candidate *TCF7L2* variant and risk of breast or ovarian cancer. *BMC Cancer.* **9,** 312 (2009). [PMC2749057]

117. Guttmacher AE, Nabel EG, Collins FS. Why data-sharing policies matter. *Proc Natl Acad Sci USA.* **106**, 16894 (2009). [PMC2761326]

118. Collins FS. Revealing the body's deepest secrets. *Parade Magazine*, pp 6-7 (Feb 21, 2010).

119. Collins FS. Has the revolution arrived? *Nature.* **464**, 674-675 (2010).

120. Collins FS. NIH Chief Francis Collins: Medical Research 'Ought to tell us what works.' Interview by Jennifer Evans. *Kaiser Health News* (April 2010).

121. Collins FS. Francis S. Collins: NIH director discusses challenges, opportunities driven by health care reform, stimulus spending, and fast pace of scientific discovery. Interview by Britt E. Erickson. *Chemical & Engineering News.* **88**, 12-18 (2010).

122. Collins FS. Change, change, change: heeding the call. *Mol Biol Cell.* **21**, 3793-4 (2010). [PMC2982095]

123. Collins FS, Fauci AS. How we're living with HIV. *Parade Magazine*, pp 10-12 (May 23, 2010).

124. Collins FS. Genome-sequencing anniversary: Faces of the Genome. *Science.* **331**, 566 (2011).

ONLINE PUBLICATIONS

1. Brown WT, Gordon LB, Collins FS. Hutchinson-Gilford Progeria Syndrome, *GeneReviews*, GeneTests Medical Genetics Information Resource, www.genetests.org (University of Washington, 1997-2010).

2. Chen G, Adeyemo AA, Johnson T, Zhou J, Amoah A, Owusu S, Acheampong J, Agyenim-Boateng K, Eghan Jr BA, Oli J, Okafor G, Abbiyesuku F, Dunston GM, Chen Y, Collins FS, Rotimi C. A genome-wide scan for quantitative trait loci linked to obesity phenotypes among West Africans. *Int J Obes.* (2004). online publication paper.

3. Bonilla C, Panguluri RK, Taliaferro-Smith L, Argyropoulos G, Chen G, Adeyemo AA, Amoah A, Owusu S, Acheampong J, Agyenim-Boateng K, Eghan BA, Oli J, Okafor G, Abbiyesuku F, Johnson T, Rufus T, Fasanmade O, Chen Y, Collins FS, Dunston GM, Rotimi C, Kittles RA. Agouti-related protein promoter variant associated with leanness and decreased risk for diabetes in West Africans. *Int J Obes.* (2005). online publication paper.

4. Cerato A, Parisi M, Anna SS, Missirlis F, Guru S, Agarwal S, Sturgill D, Talbot T, Spiegel A, Collins F, Chandrasekharappa S, Marx S, and Oliver B. Genetic interactions between Drosphila melanogaster menin and Jun/Fos. www.sciencedirect.com (Developmental Biology, 2006).

CHAPTERS IN BOOKS

1. Collins FS. Chromosome jumping in *Genome Analysis: A Practical Approach.* (ed Davies KE) 73-94 (IRL Press, 1988).

2. Gumucio DL, Gray TA, Rood KL, Blanchard KL, Collins FS. Nuclear proteins which bind the human gamma-globin gene in *Hemoglobin Switching, Part A: Transcriptional Regulation* (eds Stamatoyannopoulos G, Nienhuis AW) 129-38 (Alan R. Liss, 1989).

3. Wallace MR, Collins FS. Molecular genetics of von Recklinghausen neurofibromatosis. *Adv Hum Genet,* **20**, 267-307, (1991).

4. Gumucio DL, Blanchard-McQuate KL, Heilstedt-Williamson H, Tagle DA, Gray TA, Tarle SA, Gragowski L, Goodman M, Slightom J, Collins FS. Gamma-globin gene regulation: evolutionary approaches in *The Regulation of Hemoglobin Switching* (eds Stamatoyannopoulos G, Nienhuis AW) 277-89 (The Johns Hopkins University Press, 1991).

5. Collins FS, Iannuzzi MC. Genetic defect in cystic fibrosis in *Update, Pulmonary Diseases and Disorder.* (ed Fishman AP) 83-92 (McGraw-Hill, 1992).

6. Chandrasekharappa SC, Marchuk DA, Collins FS. Analysis of yeast artificial chromosome clones in *Pulsed-field Gel Electrophoresis: Protocols, Methods, and Theories* (eds Burmeister M, Ulanovsky L) 235-57, (Humana Press, 1992).

7. Marchuk DA, Collins FS. The use of YACs to identify expressed sequences: cDNA screening using total YAC insert in *YAC Libraries: A User's Guide* (eds Nelson DL, Brownstein BH) 113-26 (W.H. Freeman and Co., 1994).

8. Collins FS, Gelehrter TD. Molecular genetics in *The Genetic Basis of Common Diseases* (eds King RA, Rotter JI, Motulsky AG) 19-33 (Oxford University Press, 1992).

9. Wallace MR, Collins FS. Neurofibromatosis in *Molecular Basis of Neurology* (ed Conneally PM) 160-80 (Blackwell Scientific Publications, 1993).

10. Strong TV, Collins FS. The structure of the cystic fibrosis gene in *Cystic Fibrosis—Current Topics* (eds Dodge JD, Brock DJH, Widdicombe JH) 3-28 (J. Wiley, 1993).

11. Sferra TJ, Collins FS. The molecular biology of cystic fibrosis in *Annual Review of Medicine: Selected Topics in the Clinical Sciences* (eds Creger WP, Coggins CH, Hancock EW) 133-44 (Annual Reviews, 1993).

12. Marchuk DA, Collins FS. Molecular genetics of NF1 in *The Neurofibromatoses: A Pathogenetic and Clinical Overview* (eds Huson SM, Hughes RAC) 23-49 (Chapman & Hall Medical, 1994).

13. Drumm ML, Collins FS. Molecular biology of cystic fibrosis in *Molecular Genetic Medicine* (ed Friedmann T) 33-64 (Academic Press, 1993).

14. Orenstein DM, Collins FS. *Cystic Fibrosis: A Guide for Patient and Family,* update (1992).

15. Gutmann DH, Cole JL, Collins FS. Expression of the neurofibromatosis type 1 (NF1) gene during mouse embryonic development. *Prog Brain Res* **105**, 327-35 (1995).

16. Tagle DA, Swaroop M, Elmer L, Valdes J, Blanchard-McQuate K, Bates G, Baxendale S, Snell R, MacDonald M, Gusells J, Lehrach H, Collins FS. Magnetic bead capture of cDNAs. a strategy for isolating expressed sequences encoded within large genomic segments in *Advances in Biomagnetic Separation* (eds Uhlen M, Hornes E, Olsvik O) 91-106 (Eaton Publishing, 1993).

17. Gutmann DH, Collins FS. Von Recklinghausen neurofibromatosis in *The Metabolic and Molecular Basis of Inherited Disease* (ed Scriver CR) 677-96 (McGraw-Hill, Health Profession Div., 1995).

18. Couch FJ, Weber BJ, Collins FS, Tagle DA. Isolation of expressed sequences from the chromosome 17q21 BRCA1 region by magnetic bead capture in *Identification of Transcribed Sequences* (eds Hochgeschwender U, Gardiner KJ) 51-63 (Plenum Press; 1994).

19. Abel KJ, Castilla L, Buckler AJ, Ho P, Couch FJ, Collins FS, Weber BL. Isolation of gene sequences from the BRCA1 region on chromosome 17q21 by exon amplification in *Identification of Transcribed Sequences* (eds Hochgeschwender U, Gardiner KJ) 183-98 (Plenum Press, 1994).

20. Collins FS. The Human Genome Project in *Life at Risk: The Crises in Medical Ethics* from the 1993 Christian Life Commission's Annual Seminar on Medical Ethics. (eds Moore LA, Land RD) 100-175 (Broadman Holman Publishers, 1995).

21. Francomano CA, Collins FS. The Human Genome Project, update 1995 in *Molecular and Functional Aspects of Blood Group Antigens* (ed Silberstein LE) 229-37 (American Association of Blood Banks, 1995).

22. Orenstein DM, Collins FS. Genetics in *Cystic Fibrosis: A Guide for Patient and Family* (ed Orenstein DM) 203-16 (Lippincott Williams & Wilkins, 1997).

23. Hajra A, Liu PP, Collins FS. Transforming properties of the leukemic Inv(16) fusion gene CBFB-MYHII in *Molecular Aspects of Myeloid Stem Cell Development* (eds Wolff L, Perkins AS) 289-98 (Springer, 1996) This chapter has been retracted.

24. Jenkins J, Collins FS. Human genome research: essentials for nurses in *Maternity & Women's Health Care* (eds Lowdermilk DL, Perry SE, Bobak IM) (Mosby, 1997).

25. Gutmann DH, Collins FS. Von Recklinghausen neurofibromatosis in *The Metabolic and Molecular Basis of Inherited Disease,* CD-ROM (eds Scriver CR, Beaudet AL, Valle DV) (McGraw-Hill; 1997).

26. Biesecker LG, Biesecker BB, Collins FS. The genome project and molecular diagnosis in *Textbook of Internal Medicine* (ed Kelley WN) 13-15 (Lippincott-Raven, 1997).

27. Collins FS. Discovering genes that cause disease in *Gene Therapy for Diseases of the Lung* (ed Brigham KL) 17-26 (M. Dekker; 1997).

28. Collins FS, Trent JM. Cancer genetics *in Harrison's Principles of Internal Medicine, 15th edition* (eds Braunwald MD, Fauci AS, Kasper DL, Hauser SL, Longo DL, Jameson JL) 503-9 (McGraw-Hill Professional; 2001).

29. Collins FS. Essay in *Biology: Science and Life* (ed Cummings M) 166-68 (West Educational Publishing, 1996).

30. Collins FS. Essay in *Human Heredity: Principles and Issues* (ed Cummings M) 304 (West Educational Publishing, 1997).

31. Jenkins J, Collins FS. Genes and chromosomes: the Human Genome Project in *Maternal Child Nursing Care* (eds Wong DL, Perry SE, Hess CS) 52-53 (Mosby, 1998).

32. Marx SJ, Agarwal SK, Kester MB, Kim YS, Heppner C, Spiegel AM, Burns AL, Emmert-Buck MR, Debelenko LV, Zhuang Z, Lubensky I, Liotta LA, Crabtree JS, Wang Y, Roe BA, Weisemann J, Boguski MS, Doppman JL, Skarulis MC, Alexander RH, Guru SC, Manickam P, Olufemi SE, Collins FS, Chandrasekharappa SC. Multiple endocrine neoplasia type 1: from clinical physiology to the gene in *Parathyroid Diseases from the Gene to the Cure* (ed Brandi ML) 43-52 (See-Firenze, 1997).

33. Fink L, Collins FS. The Human Genome Project: evolving status and emerging opportunities for disease prevention in *Genetics and Public Health in the 21st Century: Using Genetic Information to Improve Health and Prevent Disease* (eds Khoury MJ, Burke W, Thomson EJ) 45-49 (Oxford University Press, 2000).

34. Smith JR, Gelehrter TD, Collins FS. Molecular genetics of common disease in *The Genetic Basis of Common Diseases, 2nd edition* (eds King RA, Rotter JI, Motulsky AG) (Oxford University Press, 2002).

35. Woodage T, Collins FS. The genomic basis of medicine in *The Oxford Textbook of Medicine, 4th edition* (eds Warrell DA, Cox TM, Firth JD, Benz EJ) 88-87 (Oxford University Press, 2003).

36. Collins FS, Jegalian K. Human Genome Project in *Encyclopedia of Public Health.* (ed Breslow L) (Macmillan Reference, 2002).

37. Collins FS, Gutmann DH. Neurofibromatosis 1. Peripheral neurofibromatosis in the genetic basis of human cancer in *The Metabolic and Molecular Basis of Inherited Disease, 8th Edition* (eds Scriver CR, Sly WS) 877-96 (McGraw-Hill Professional, 2000).

38. Biesecker LG, Biesecker BB, Collins FS. The Genome Project and molecular diagnosis in *Kelley's Textbook of Internal Medicine, 4th Edition* (ed Humes HD) (Lippincott Williams & Wilkins, 2000).

39. Collins FS. Essay in *Human Heredity: Principles and Issues* (ed Cummings M) (West Educational Publishing, 2001).

40. Collins FS. Human genetics in *Cutting-edge Bioethics: A Christian Exploration of Technologies and Trends* (eds Kilner JF, Hook CC, Uustal DB) 3-17 (W.B. Eerdmans, 2002).

41. Collins FS. Genome Research: The Next Generation in *Cold Spring Harbor Symposia on Quantitative Biology, Volume LXVIII.* 49-54 (Cold Spring Harbor Laboratory Press, 2003).

42. Collins FS. A word from Francis S. Collins, M.D., Ph.D. in *2004-2005 Workshop Materials for AP Biology,* Chptr. II, 5-8 (College Entrance Examination Board, 2004).

43. Guttmacher, AE, Collins FS. Foreword in *The Renaissance of Sickle Cell Disease in the Human Genome Era* (eds) . XXIX-XXX (Imperial College Press, 2007).

44. Collins FS, Medical and Societal Implications of the Human Genome Project in *PRIM&R Through the Years,* pages (Public Responsibility in Medicine & Research, 2006).

45. Collins FS. Foreword in *Discovering Genomics, Proteomics, & Bioinformatics, 2nd Edition* (Campbell AM, Heyer LJ) ix (Benjamin Cummings).

46. Collins FS. Foreword in *Medicine After the Holocaust: From the Master Race to the Human Genome and Beyond.* Sheldon Rubenfeld., Ed. Hampshire: Palgrave Macmillan Ltd. (in press).

BOOKS

1. Gelehrter TD, Collins FS, Ginsburg D. *Principles of Medical Genetics, 2nd Edition* (Williams & Wilkins, 1998).
2. Collins FS. *The Language of God: A Scientist Presents Evidence For Belief* (Free Press, 2006).
3. Collins FS. *The Language of Life: DNA and the Revolution in Personalized Medicine* (HarperCollins, 2010).
4. Collins FS. *Belief: Readings on the Reason for Faith* (HarperCollins, 2010).
5. Giberson KW, Collins FS. *The Language of Science and Faith* (IVP Books, 2011).