



## Francis S. Collins



**Date of Birth** 14 April 1950

**Place** Staunton, VA (USA)

**Nomination** 10 February 2009

**Field** Genetics

**Title** Professor

### Most important awards, prizes and academies

Professor Collins' accomplishments have been recognized by numerous awards and honors, including election to the Institute of Medicine and the National Academy of Sciences. On November 5, 2007, he received the Presidential Medal of Freedom from U.S. President George W. Bush, the nation's highest civil award, for his revolutionary contributions to genetic research. He was also present at the bill signing for the Genetic Information Nondiscrimination Act in the Oval Office, in recognition of his work in genetics, and his early papers and commentary on the need for such protections.

### Summary of scientific research

Professor Collins' research has led to the identification of genetic variants associated with type 2 diabetes and the genes responsible for cystic fibrosis, neurofibromatosis, Huntington's disease and Hutchinson-Gilford progeria syndrome. As director of the National Center for Human Genome Research, which became NHGRI in 1997, he oversaw the International Human Genome Sequencing Consortium and many other aspects of what he has called "an adventure that beats going to the moon or splitting the atom". In 1994, Collins founded NHGRI's Division of Intramural Research (DIR), a collection of investigator-directed laboratories that conduct genome research on the NIH campus and that has developed into one of the nation's premier research centers in human genetics. With new tools arising from the human genome project and technology development studies supported by the genome institute, Collins is optimistic about the chances of uncovering hereditary contributors to common diseases, such as heart disease, cancer and mental illness. In the overall research agenda of NHGRI, this interest is reflected in the highly ambitious effort to construct a haplotype map of the human genome. The now-completed "hap map" project produced a catalog of genetic variations – called single nucleotide polymorphisms (SNPs) – which is now being widely used to discover genetic variations correlate with disease risk. There was a dramatic increase of published scientific papers linking genetic variations to common illnesses in 2007. Collins's work in his highly active lab demonstrates that research emphasis, which is devoted to finding the genes that contribute to adult-onset, Type 2 diabetes. In addition to his long list of contributions to basic genetic research and scientific leadership, Collins is known for his close attention to ethical and legal issues in genetics. He has been a strong advocate for protecting the privacy of genetic information and has served as a national leader in efforts to prohibit gene-based insurance discrimination. Building on his own experiences as a physician volunteer in a rural missionary hospital in Nigeria, Collins is also very interested in opening avenues for genome research to benefit the health of people living in developing nations.

### Main publications

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Green E.D., Wolfsberg T.G., Collins F.S., Identifying gene regulatory elements by genome-wide recovery of DNase hypersensitive sites, *Proc Natl Acad Sci USA* 101:992-997, 2004; Goldman R.D., Shumaker D.K., Erdos M.R., Eriksson M., Goldman A.E., Gordon L.B., Gruenbaum Y., Khuon S., Mendez M., Varga R., Collins F.S., 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; Silander, K., Mohlke K.L., Scott L.J., Peck E.C., Hollstein P., Skol A.D., Jackson A.U., Deloukas P., Hunt S., Stavrides G., Chines P.S., Erdos, M.R., Narisu N., Conneely K.N., Li C., Fingerlin T.E., Dhanjal S.K., Valle T.T., Bergman R.N., Tuomilehto, J., Watanabe R.M., Boehnke M., Collins F.S. Genetic variation near the hepatocyte nuclear factor-4a gene predicts susceptibility to type 2 diabetes, *Diabetes*, 53:1141-1149, 2004; Capell BC, Erdos MR, Madigan JP, Fiordalisi JJ, Varga R, Conneely KN, Gordon LB, Der CJ, Cox AD, Collins FS, Inhibiting farnesylation of progerin prevents the characteristic nuclear blebbing of Hutchinson-Gilford progeria syndrome, *PNAS*, 102: 12879-12884, 2005; Guttmacher A.E., Collins, F.S., Realizing the promise of genomics for biomedical research, *JAMA*, 294(11):1399-402, 2005; 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 Research*, 16(1) 123-131, 2006; 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(9) 3250-3255, 2006; Scacheri PC, Davis S, Odom DT, Crawford GE, Perkins S, *et al.*, Genome-Wide Analysis of Menin Binding Provides Insights to MEN1 Tumorigenesis, *PLoS Genet*, 2(4)e51, 2006; 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(7) 503-509, 2006; 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, Reibow NL, Buchanan TA, Valle TT, Tuomilehto J, Bergman RN, Mohlke KL, Boehnke M, and Collins FS, Common Variants in MODY Contribute to Risk of Type 2 Diabetes in Finns, *Diabetes*, (9):2534-40, 2006; Feero, WG, Guttmacher, AE, Collins, The Genome Gets Personal - Almost, *JAMA*, 299(11):1351-1352, 2008. Books Guttmacher, Alan E., Collins, Francis S., Drazen, Jeffrey M. eds., Foreword by Elias Zerhouni, M.D., *Genomic Medicine: Articles from the New England Journal of Medicine*, Baltimore: Johns Hopkins University Press, 2004; Gelehrter, Thomas D., Collins, Francis S., Ginsberg, David, *Principles of Medical Genetics*, 2nd ed. Baltimore: Lippincott, Williams & Wilkins, 1998.