Prof. Francis S. Collins
Director, U.S. National Institutes of Health

Most important awards, prizes and academies

Dr. Collins is an elected member of the U.S. National Academy of Medicine and the National Academy of Sciences. In recognition of his revolutionary contributions to genetic research, he was awarded the Presidential Medal of Freedom in 2007 and the National Medal of Science in 2009. In 2020, he was elected a Foreign Member of the Royal Society (UK) and named the 50th winner of the Templeton Prize, which celebrates scientific and spiritual curiosity.

Summary of scientific research

Dr. Collins was appointed the 16th Director of the U.S. National Institutes of Health (NIH) by President Barack Obama and sworn in on August 17, 2009. In 2017, President Donald Trump asked Dr. Collins to continue to serve as the NIH Director, and President Joseph Biden did the same in 2021. In this role, he oversees the work of the largest supporter of biomedical research in the world, spanning the spectrum from basic to clinical research. Prior to that, he served as Director of NIH's National Human Genome Research Institute from 1993-2008. 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. In addition to his contributions to genetic research and scientific leadership, Collins is known for his close attention to ethical and legal

issues in biomedical research. He has been a strong advocate for protecting the privacy of genetic information and served as a national leader in efforts to guard against gene-based discrimination in employment and health insurance. Building on his experiences as a physician volunteer in a rural missionary hospital in Nigeria, Collins is also interested in opening avenues for biomedical research to benefit the health of people living in developing nations.

Main publications

Rommens J.M., Iannuzzi M.C., Kerem B., Drumm J.L., Melmer G., Dean M., Rozmahel R., Cole J.L., Kennedy D., Hidaka N., Zsiga M., Buchwald M., Riordan J.R., Tsui L.C., Collins F.S., Identification of the cystic fibrosis gene: chromosome walking and jumping, *Science*, 245:1059-1065, 1989; Wallace M.R., Marchuk D.A., Andersen L.B., Letcher R., Odeh H.M., Saulino A.M., Fountain J.W., Brereton A., Nicholson J., Mitchell A.L., Collins F.S., Type 1 neurofibromatosis gene: identification of a large transcript disrupted in three NF1 patients, *Science*, 249:181-186, 1990; Chandrasekharappa S.C., Guru S.C., Manickam P., Olufemi S.E., Collins F.S., Emmert-Buck M.R., Debelenko L.V., Zhuang Z., Lubensky I.A., Liotta L.A., Crabtree J.S., Wang Y., Roe B.A., Weismann J., Boguski M.S., Agarwal S.K., Kester M.B., Kim Y.S., Heppner C., Dong Z., Spiegel A.M., Burns A.L., Marx S.J., Positional cloning of the gene for multiple endocrine neoplasia-type 1, *Science*, 276:404-407, 1997; International Human Genome Sequencing Consortium, Initial sequencing and analysis of the human genome, *Nature*, 409:860-921, 2001; Eriksson M., Brown W.T., Gordon L.B., Glynn M.W., Singer J., Scott L., Erdos M.R., Robbins C.M., Moses T.Y., Berglund P., Dutra A., Pak E., Durkin S., Csoka A.B., Boehnke M., Glover T.W., Collins F.S., Recurrent de novo point mutations in lamin A cause Hutchinson-Gilford progeria syndrome, *Nature*, 423:293-298, 2003; The ENCODE Project Consortium, The ENCODE (ENCyclopedia of DNA Elements) Project, *Science,* 306:636-640, 2004; Crawford G.E., Holt I.E., Whittle J., Webb B.D., Tai D., Davis S., Margulies E.H., Chen Y., Bernat J.A., Ginsburg D., Zhou D., Luo S., Vasicek T.J., Daly M.J., Wolfsberg T.G., Collins F.S., Genome-wide mapping of DNase hypersensitive sites using massively parallel signature sequencing (MPSS), *Genome Res.*, 16:123-131, 2006; International HapMap Consortium, A second generation human haplotype map of over 3.1 million SNPs, *Nature*, 449:851-861, 2007; Scott L.J., Mohlke K.L., Bonnycastle L.L., Willer C.J., Li Y., Duren W.L., Erdos M.R., Stringham H.M., Chines P.S., Jackson A.U., Prokunina-Olsson L., Ding C.J., Swift A.J., Narisu N., Hu T., … Tuomilehto J., Collins F.S., Boehnke M., A genome-wide association study of type 2 diabetes in Finns detects multiple susceptibility variants, *Science*, 316:1341-1345, 2007; Cao K., Capell B.C., Erdos M.R., Djabali K., Collins F.S., 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; Capell B.C., Olive M., Erdos M.R., Cao K., Faddah D.A., Tavarez U.L., Conneely K.N., Qu X., San H., Ganesh S.K., 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; Stitzel M.L., Sethupathy P., Pearson D.S., Chines P.S., Song L., Erdos M.R., Welch R., Parker S.C.,

Boyle A.P., Scott L.J., NISC Comparative Sequencing Program, Margulies E.H., Boehnke M., Furey T.S., Crawford G.E., Collins F.S., Global epigenomics analysis of primary human pancreatic islets provides insights into type 2 diabetes susceptibility loci, *Cell Metabolism,* 12:443-455, 2010; Cao K., Blair C.D., Faddah D.A., Kieckhaefer J.E., Olive M., Erdos M.R., Nabel E.G., Collins F.S., Progerin and telomere dysfunction collaborate to trigger cellular senescence in normal human fibroblasts, *J. Clin. Invest.,* 121:2833-2844, 2011; Cao K., Graziotto J.J., Blair C.D., Mazzulli J.R., Erdos M.R., Krainc D., Collins F.S., Rapamycin reverses cellular phenotypes and enhances mutant protein clearance in Hutchinson-Gilford progeria syndrome cells, *Sci. Transl. Med.,* 3:89ra58, 2011; Parker S.C., Stitzel M.L., Taylor D.L., Orozco J.M., Erdos M.R., Akiyama J.A., van Bueren K.L., Chines P.S., Narisu N., NISC Comparative Sequencing Program, Black B.L., Visel A., Pennacchio L.A., Collins F.S., Chromatin stretch enhancer states drive cell-specific gene regulation and harbor human disease risk variants, *Proc. Natl. Acad. Sci. USA,* 110:17921-17926, 2013; Varshney A., Scott L.J., Welch R.P., Erdos M.R., Chines P.S., … Mohlke K.L., Boehnke M., Collins F.S., Parker S.C., Stitzel M.L., Genetic regulatory signatures underlying islet gene expression and type 2 diabetes, *Proc. Natl. Acad. Sci. USA,* 114:2301-2306, 2017; Di Gioia S.A., Connors S., Matsunami N., Cannavino J., Rose M.F., … Collins F.S., … Carey J.C., Robertson S.P., Manoli I., Engle E.C., A defect in myoblast fusion underlies Carey-Fineman-Ziter syndrome, *Nat Commun.,* 8:16077, 2017; Bar D.Z., Atkatsh K., Tavarez U., Erdos M.R., Gruenbaum Y., Collins F.S., Biotinylation by antibody recognition--a method for proximity labeling, *Nat. Methods,* 15:127-133, 2018; DuBose A.J., Lichtenstein S.T., Petrash N.M., Erdos M.R., Gordon L.B., Collins F.S., Everolimus rescues multiple cellular defects in laminopathy-patient fibroblasts, *Proc. Natl. Acad. Sci. USA,* 115:4206-4211, 2018; Taylor D.L., Jackson A.U., Narisu N., Hemani G., Erdos M.R., Chines P.S., Swift A., Idol J., Didion J.P., Welch R.P., Kinnunen L., Saramies J., Lakka T.A., Laakso M., Tuomilehto J., Parker S.C.J., Koistinen H.A., Davey Smith G., Boehnke M., Scott L.J., Birney E., Collins F.S., Integrative analysis of gene expression, DNA methylation, physiological traits, and genetic variation in human skeletal muscle, *Proc. Natl. Acad. Sci. USA,* 116:10883-10888, 2019; Koblan L.W., Erdos M.R., Wilson C., Cabral W.A., Levy J.M., Xiong Z.M., Tavarez U.L., Davison L.M., Gete Y.G., Mao X., Newby G.A., Doherty S.P., Narisu N., Sheng Q., Krilow C., Lin C.Y., Gordon L.B., Cao K., Collins F.S., Brown J.D., Liu D.R., *In vivo* base editing rescues Hutchinson-Gilford progeria syndrome in mice, *Nature,* 589:608-614, 2021.