Francis Sellers Collins (1950-) [1]

By: Carvalho, Tito Keywords: genetic sequencing [2]

Francis Sellers Collins helped lead the International Human Genome Sequencing Consortium, which helped describe the DNA sequence of the human genome [3] by 2001, and he helped develop technologies used in molecular genetics while working in the US in the twentieth and twenty-first centuries. He directed the US National Center for Human Genome Research (NCHGR), which became the National Human Genome Research Institute (NHGRI), of the US National Institutes of Health [4] (NIH), located in Bethesda, Maryland, from 1993 to 2008. Collins led teams of researchers to use data on human genomes to investigate the genetic aspects of diseases and treatments, the variations among people in terms of their DNA sequences, and the evolution [5] of humans [6]. Collins became director of the NIH in 2009. Some criticized him for his Christian faith and its possible impacts on science funding through the NIH, such as for stem cell research, cloning [7], and embryonic genetic testing. As a director of the NHGRI and the NIH, Collins helped shape the structures and aims of projects in biology that pursue what he called big science, and he helped relate those projects to federal governments and to private companies.

Collins was born in Staunton, Virginia, on 14 April 1950. He grew up on a farm and was home schooled until the sixth grade by his mother, Margaret Collins, a playwright, and by his father, Fletcher Collins, a drama professor. He attended the Robert E. Lee High School and planned a career in physical chemistry, in which he majored as an undergraduate at the University of Virginia in Charlottesville, Virginia. During college, Collins identified himself as an agnostic, later writing that his family life never involved the practice of faith. Collins married Mary Lynn Harman in his junior year at the Virginia. Collins later divorced Harman and married Diane Baker, a genetic counselor.

After he graduated from Virginia in 1970, Collins entered a PhD program in physical chemistry at Yale University [8] in New Haven, Connecticut. At Yale, Collins studied quantum mechanics and mathematical descriptions of collisions of atoms and molecules. He later reported that a graduate seminar in biochemistry, taught by Peter Lengyel and Bill Summers, sparked Collins's interest in molecular biology and genetics in 1972. Collins began to study biology and medicine, but he finished his PhD in physical chemistry in 1974 with R. James Cross, Jr. as his advisor.

Collins then went to medical school at the University of North Carolina [9] in Chapel Hill, North Carolina. Collins later said that as a medical student working at the bedside with patients, he started to contemplate questions about the existence of God, and he became a Christian. After he graduated with his MD in 1977, Collins completed a residency in internal medicine at Chapel Hill, and then he returned to New Haven for a postdoctoral fellowship in human genetics at Yale Medical School [10]. During his fellowship he worked with Sherman Morton Weissman, with whom he developed the technique of chromosome jumping in 1984. Chromosome jumping is a molecular biology technique where scientists cut and rearrange sections of large strands of DNA to identify and map genes [11].

After his fellowship at Yale, Collins joined the University of Michigan [12] in Ann Arbor, Michigan, in 1984, later securing a position as professor of internal medicine and human genetics. While at Michigan, Collins developed another technique in genome [3] mapping, called positional cloning [7], in which researchers identify a gene for a specific phenotype by its approximate location on the chromosome. Drawing on the techniques of chromosome jumping and positional cloning [7], Collins worked with several other scientists to identify the genes [11] relevant to cystic fibrosis [13], Huntington's disease, neurofibromatosis, multiple endocrine neoplasia type 1, and the M4 type of adult acute leukemia.

Some observers noted that Collins's reputation as a geneticist made him a good fit for the position of director of the NCHGR, a job he began in 1993, but his nuanced view on gene patents and intellectual property also influenced his nomination. He replaced <u>James Watson</u> [14] as director of the NCHGR, who left over disagreements about gene patenting. Collins said that researchers should patent <u>genes</u> [11] that they've sequenced in instances where an intellectual property claim on a gene would provide incentives to develop a product that the public needs, and without which that product would likely not happen. On the other hand, he said that patents should not be allowed on <u>genes</u> [11] whose functions are unknown, or in situations where there is no clear method for how the patent would benefit the public. Collins continued to direct the NCHGR as it evolved into the the National Human Genome Research Institute (NHGRI) in 1997.

Collins testified before the United States Congress in 1998 on the ethical and social policy of mapping the human genome [3], and he argued that the US federal government should continue to fund the NHGRI. In 1998 a private company, Celera Genomics, headquartered in California and led by Craig Venter, began to map the human genome [3] with techniques that differed from those

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used by the NHGRI, funded by the NIH. As a result, the US Congress considered defunding the public effort of the Human Genome Project. Testifying before a congressional subcommittee in June 1998, Collins explained to Congress members that Venter's approach to sequencing the human genome ^[3] differed from that of the NHGRI. On one hand, the techniques developed by Venter, such as shotgun sequencing, were faster and focused only on genes ^[11], which are the parts of the genome ^[3] that produce proteins, then estimated as roughly five percent of the human genome ^[3]. On the other hand, the NHGRI's approach attempted to map the whole genome ^[3]. During the testimonials to Congress, the Director of the NIH, Harold Varmus, alongside Collins, made the case that the human genome ^[3] effort had room for both the NHGRI's and Celera's approaches. The NIH administrators privately expressed worries that Venter's approach was sloppy, not having the quality standards that the NIH's project demanded. Furthermore, the NIH administrators were concerned about the public availability of the human genome ^[3] data, keeping in mind the for-profit orientation of Celera.

With Congress convinced in 1998 that the NIH should continue its efforts to map the human <code>genome</code> ^[3], the NHGRI had funding for the next years of the project. However, Venter's approach pressured the NHGRI, accelerating the human <code>genome</code> ^[3] effort. Venter and Collins appeared alongside US President Bill Clinton and UK Prime Minister Tony Blair to announce the completion of the preliminary draft of the human <code>genome</code> ^[3] on 26 June 2000. Clinton called the human <code>genome</code> ^[3] map "the language of God." On 16 February 2001, ahead of schedule and under budget, both NHGRI and Celera published working drafts of the human <code>genome</code> ^[3] in the journals *Nature* and *Science*, respectively. Collins and Venter discussed their findings in the <u>American</u> Association for the Advancement of Science ^[15]'s annual meeting in April 2001 as keynote speakers in San Francisco, California.

Using the phrase used by Clinton to describe the human genome [3], Collins published the book, *The Language of God: A Scientist Presents Evidence for Belief*, in 2006. In his book, Collins argues that a scientific worldview can co-exist with religious faith, and that scientific discoveries give humans [6] knowledge of God's work, and reasons for worship. For Collins, science ought to explain as much as it can about the natural world, and it provides the only means to understand the creation of God.

While at the NCHGR, Collins studied a number of ethical, legal and social issues around the Human Genome Project. Collins, a proponent of legislation on genetic privacy, suggested as early as 1995 that the Ethical Legal and Social Implications (ELSI) research program of the NCHGR, which later became the NHGRI, should devote attention to policy. Noting that some in 1995 had considered policy on genetic privacy to be a premature endeavor, Collins later described the 13-year period to pass the Genetic Information Non-discrimination Act of 2008 (GINA) as a necessary task before the end of his tenure at the NHGRI. Collins left the NHGRI on 1 August 2008, after GINA passed on 21 May 2008.

Collins's outspoken religious faith caused controversy when US President Barack Obama [16] appointed him director of the NIH on 8 July 2009. A number of scientists criticized Collins's appointment because of his religious beliefs. Collins said that he supports stem cell research, though with the caveat that it should happen via somatic cell nuclear transfer [17]. Collins argues that there is an important moral distinction between this technique, which he has calls a purely manmade event, and the development of an embryo via the union of sperm [18] and egg [19]. In October 2009, Pope Benedict XVI appointed Collins to The Pontifical Academy of Sciences [20] of the Catholic Church in the Vatican City.

While a number of scientist criticized Collins's appointment to the head of the NIH, others, including Harold Varmus, praised Collins, citing his record as a geneticist, his management skills during his administration of the NHGRI, and his preoccupation with the ethically and socially responsible conduct of science. In *The Language of God*, Collins writes that it would be a mistake to leave questions of ethics and science policy solely to scientists.

As the director of the NIH, Collins advocated for a new institute to address translational medicine, including efforts to translate findings of the human genome [3] project into clinical practices. He also advised Obama administration in 2009 as it lifted former President George W. Bush [21] is 2001 ban on federal funds allocated to human embryonic stem cell research [22].

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Arizona State University. School of Life Sciences. Center for Biology and Society. Embryo Project Encyclopedia.

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Last Modified

Wednesday, July 4, 2018 - 04:40

DC Date Accessioned

Tuesday, April 15, 2014 - 22:28

DC Date Available

Tuesday, April 15, 2014 - 22:28

DC Date Created

2014-04-10

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