General of the Genome – Francis Collins
(First in SERIES of 3)

VOICE: There's excitement whenever this man speaks. He describes his work as "an exhilarating experience, …like reading the world's most incredible literature classic that no one else had ever read before, and getting to write the first critical review."

PROF.: One article calls Dr. Francis Collins the "General of the Genome." He directed an international team of genetics researchers in a massive, three-billion-dollar project. They studied the complete genetic structure of the human body, researching the ways our genes are structured and how they affect our health.

VOICE: Now Collins and his colleagues are beginning to ***use*** their new knowledge to cure diseases that had previously been incurable. No wonder he's excited!

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PROF.: Dr. Francis Collins' boss said the Human Genome Project is probably the only achievement of its decade, that will be remembered a thousand years from now! The information that the project discovered is already being applied to maintain health and prevent disease.

VOICE: In July, 2009, President Obama appointed Dr. Collins to an even higher position. He is now director of America's "National Institutes of Health," composed of 23 different institutes and bureaus that deal with various aspects of human health.

PROF.: Dr. Collins asked us to clarify that the statements we're quoting today are not official positions of the United States government. They are his personal opinions and interpretations – based on his extensive research and analysis of the human genome.

VOICE: Isn't the genome the complete set of 23 chromosome pairs that we inherited from our parents?

PROF.: Yes. It contains all the information that an embryo needs, to develop into an adult human. And it continues to influence many things throughout our lifetimes.

VOICE: Isn't the genetic code similar to the zeroes and ones that make up computer code?

PROF.: Yes, but instead of zeroes and ones, the genome stores its information by a series of bases which are abbreviated A, C, G and T.

VOICE: How much genetic information is in the human body?

PROF.: An amazing amount! Dr. Collins illustrates, "You can think of the human genome as a book, a very large book which, if you printed it out and piled the pages on top of each other, would be as high as the Washington Monument."

VOICE: How tall is that monument?

PROF.: 169 meters tall! To figure how many pages that would be, I stacked one meter of paper and found that it contained 10,000 sheets. The 169-meter-high stack of printout would contain 1.7 million pages.

He illustrates the size another way. If we could read one letter every second, 24 hours a day without stopping, we would be reading for 31 years.

VOICE: Printouts full of A's, C's, G's and T's.

PROF.: Yes. And that information is in each cell of our bodies! Dr. Collins adds, "You find this incredibly elegant molecule as a hereditary material in all living things."

VOICE: Wasn't the ***first*** director of the Human Genome Project a Nobel Prize winner? If I remember correctly, Dr. James Watson shared a 1962 Nobel Prize for co-discovering the double-helix structure of DNA.

PROF.: That's right. Watson became the first director of the Human Genome Project, with the goal of learning everything about the genetic structure of the human body.

When Watson retired in 1993, Dr. Francis Collins became the project's director. He coordinated hundreds of researchers around the world, with a budget of three billion U.S. dollars.

And in July, 2009, President Obama appointed him as director of the National Institutes of Health.

VOICE: How many diseases are affected by our genes?

PROF.: Dr. Collins says our genes are involved in ***every*** disease, in one way or another. He explains, "Some of them are like cystic fibrosis that have a ***very strong*** genetic contribution. Others are like adult onset diabetes, where there is a ***mix*** of genes and environment that contributes to the risk of illness. Even infectious diseases like AIDS have host factors encoded by genes that determine the likelihood of becoming ill after an exposure to an infectious agent. So it is very much a mix of genes and environment, even in that circumstance."

VOICE: What have we learned from years of research and three billion dollars of investment?

PROF.: A lot. One thing we learned is how few genes the human body contains. Dr. Collins says, "We always thought we would have 100,000 or more, and instead it turns out we have only 22,000. This is a shockingly small number of instructions to be able to carry out all of the biological functions that human beings have to do, but it works. So our awe at the complexity of the system I think is increased by this, not decreased."

He adds, "We've also learned interesting things about what part of the genome is doing what. …There's an awful lot of stuff in there that is extremely important that we're just beginning to learn about."

VOICE: What will researchers do next?

PROF.: He answers, "Certainly for me as a physician, the idea of applying this to medicine is the strongest mandate, and we would like to get on with that.

"Having overseen the project for 12 years, I have to tell you this is the best part now. We have this foundation and we can start to build on it and apply that for medical benefit."

VOICE: How are these discoveries helping to cure patients?

PROF.: This type of research has already been utilized to identify a very powerful genetic contribution to a disease that most people didn't think had much heredity involved: age-related macular degeneration, the most common cause of blindness in the elderly.

Dr. Collins anticipates that researchers will identify the genes for diabetes, heart disease, cancer, hypertension, and other diseases in the next 2 or 3 years. As a result, if you want to know what your risk is for those common conditions, it will soon be possibleto offer you that opportunity.

VOICE: If we know our risk, we can plan our own program of prevention.

PROF.: And we'll be able to do that in a fashion that's individualized – that focuses on your specific risks. That will be a major improvement over what we currently do, which is to tell everybody to do the same thing.

VOICE: Why isn't it good to tell everyone to have the same tests?

PROF.: Dr. Collins answered with an example. In his words, "There's a family where several individuals have had colon cancer…at early ages, in their 40's and 50's. …This family came in for counseling to find out who else was at risk. A specific DNA test…was done, and doctors found…three people in this family who are currently healthy but who have about a 60% chance of developing colon cancer if nothing is done about it."

These high-risk people need to have a colonoscopy starting at age 35, instead of at age 50. And they need to have it every year, not every 5 years. By finding those small polyps while they're still easy to remove, they will never develop a fatal form of colon cancer.

VOICE: When two people have the same sickness and the doctor prescribes the same medicine for both, the medicine works well for one person but not for the other. Is genome research likely to solve that problem?

PROF.: Dr. Collins thinks so. He talks about developing an individualized drug therapy. He said that when a medical doctor prescribes a medicine, it doesn't always turn out well. The same medication that cures one patient, produces no response in another person, and some people get toxic side effects from it.

VOICE: Why is that?

PROF.: A lot of the reason is heredity – differences in DNA sequences that determine whether that drug is going to work at that dose or not. The United States Food and Drug Administration recently approved the first DNA chip that enables a physician to determine whether a particular patient is a rapid metaboliser, or an ultra-rapid or a poor metaboliser of a particular medication.

VOICE: A rapid metaboliser would probably need a smaller dose of medication than a slow metaboliser, wouldn't he?

PROF.: Yes. This is still in the exploratory stage, and even Dr. Collins isn't sure yet how it will work in practice. But it is starting us down a pathway. He says, "So don't be surprised if at some time in the not too distant future you need a drug for some purpose and the doc says before we prescribe this let's check your DNA and see if this is the right drug and the right dose. …There's a great deal of promise here that I believe will ultimately work out, but it's not right around the corner."

VOICE: It sounds as if Dr. Francis Collins is really a pioneer of new medical discoveries that can save many lives.

PROF.: Definitely! On our next program we'll talk about some of the precautions we need to take in order to make sure that we don't misuse these innovative new developments.

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