Book of Life: the true wonder emerges

Wellcome Trust, 2001

A-Roll interviewees:
Isobel and Mark Emms
Professor Kay Davies, Head of Human Anatomy and Genetics, University of Oxford
Dr Michael Dexter, Director Wellcome Trust, London
Sir John Sulston, Director, Sanger Centre
Dr Mike Stratton, Head, Human Cancer Genome Project, Sanger Centre

Produced by APTN Productions.

Colour
Duration: 00:12:49:18

00:00:00:00

<Opening titles>

<Intertitles>

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> **The Book of Life: The true wonder emerges**
> **Following identification by the Human Genome Project of mankind's genetic code, research papers now reveal the first astonishing overview of how human beings are made. New medical breakthroughs could follow.**

Wellcome Film Project

A-ROLL: 5 mins 19 secs

B-ROLL: 6 mins 29 secs>

<Opening scene of a physiotherapist lifting a boy, who is wearing leg braces, up from the floor. Physiotherapist speaks>

One, two, three, up you go.

<Narration over film, unspecified narrator>

A physiotherapy session for Theo. He's a happy six year old but his muscles are weak and wasting away. He has a hereditary disease called spinal muscular atrophy known as SMA. Unless there's a breakthrough in treatment, he'll always be in a wheelchair. For his parents there seems little chance of reversing the disease and now that's changing.

<Isobel Emms to camera>

I feel very, very hopeful. You know, I can see it in the distance as something that could well happen in five, ten years time and I think I'm realistic in that, you know, maybe Theo could have some sort of treatment that could enable him to weight-bear so he could stand and that would make a tremendous difference to his life and to our life.

<Narrator over film>

This new optimism stems directly from the groundbreaking human genetic code – the human genome or so-called 'Book of Life' that's now being published for the first time.

Professor Kay Davies, a leading researcher into muscular and nervous diseases, is using the genetic code to study particular genes connected with SMA. This could bring a cure much closer.

<Davies to camera>

If we can actually explore five or six different therapies in the next ten years, there's a possibility that might affect those children born today, whereas without the human genome sequence that would never be possible.

<Narrator over film>

Inherited muscular and nervous diseases are among the first to benefit directly from mapping the human genetic code. They'll soon be many others. Now researchers from several countries in the human genome project, who produced the draft blueprint, are already working on the next steps to refine the draft and analyse how it could lead to breakthroughs in everyday medicine with treatment tailored to a patient's personal genetic make-up.

The world's largest medical research charity, the Wellcome Trust, spend 300 million dollars to fund the Sanger Centre near Cambridge in England. Here researchers contributed a third of the entire genome data. Dr Michael Dexter heads the Wellcome Trust.

<Dexter to camera>

What this is going to lead to is a whole new era of medicine – it's truly individualised medicine, where we will be treating you as an individual for the right disease with the right medicine at the right dose at the right time.

<Narrator over film>

To reach that stage, the process of refining the human DNA sequence continues as vital as ever. Every one of the body's hundred trillion cells is tightly packed with DNA. If unravelled each DNA molecule would be six feet long – enough DNA in the whole body to stretch from the earth to the sun and back, hundreds of times. Within the

DNA, combinations of four letters represent the base chemicals that determine our genes.

Special machines read and analyse colour-coded sequences produced from DNA fragments. Afterwards, giant computers reassemble all the fragments into the entire sequence of three billion letters in the DNA code. The publication now of research outlining the whole DNA sequence is the very first overview of how human beings are made. It brings together sequence information that's been made public every day as soon as it becomes available.

For Sir John Sulston, Director of the Sanger Centre, this free release of the data is vital.

<Sulston narrates over film>

All of this should be in the public domain; it should not be something which is essentially based in profit making. Profit making may come into it for particular applications but overall I think we need a public social welfare attitude to the use of this information and I believe that we have to drive medicine forward in this way.

<Narrator over film>

With this research given freely to the world, scientists can now begin to identify what genes go wrong when we develop a given disease. Among the target diseases – Alzheimer's, diabetes, heart disease and cancer. Dr Mike Stratton is a leading cancer geneticist.

<Stratton to camera>

I would be extremely surprised if children being born today did not have a much better chance of surviving their cancer than people of our generation. There will be a significant, a major transformation of cancer treatment over the next twenty years.

<Narrator over film>

But for some, the revolution in medicine can't come soon enough. Theo's physiotherapy helps him still to use his muscles, just in case a cure for SMA is found. Behind the dry science of the now published research papers lie these most enduring of images: hope for one life transformed, perhaps millions more to follow, a result of the astonishing breakthrough in identifying the human genetic code.

<00:05:57:12>

<Intertitles>

B-ROLL

Sir John Sulston
Director, Sanger Centre

<Sulston to camera>

I'm concerned that we pay attention to the possibility of creating genetic underclasses, of excluding some people from proper medical care on the grounds – oh, they have bad genes, you know, they're not going to live very long. We must not do that; we must keep this thing open and we must not allow it to become subject to short-term market forces in a way that will fundamentally diminish human rights for any individuals.

<Intertitle>

Dr Jane Rogers
Head of Sequencing, Sanger Centre

<Rogers flicking through journal and then to camera>

What we're able to tell people in these papers is how the genome looks for the very first time. We have sequence covering 90% of the whole of the human genome. We can now start to look into how it's organised

<Intertitle>

Professor Kay Davies
Head of Human Anatomy and Genetics
University of Oxford

<Davies to camera>

We will be able to develop treatments more quickly and so those taking twenty years are now going to take five years to disprove or prove that they will work, and that now brings that into the realms of the lifetime of the child, particularly in SMA for example.

<Intertitle>

Dr Mike Stratton
Head, Human Cancer Genome Project,
Sanger Centre

<Stratton to camera>

So there are a considerable number of genes which are involved in driving human cancers which are yet to be discovered and the aim of the Cancer Genome Project is to implement systematic searches, gene by gene, through the human genome sequence to identify the genes that are abnormal.

<Intertitle>

Dr David Bentley,
Head of Human Genetics,
Sanger Centre

<Bentley working at his desk and then to camera>

Areas such as diabetes, heart disease, many forms of cancer, all undoubtedly do have a very important genetic component. There are genes which contribute either to susceptibility or to the actual development and progress of such diseases. And having knowledge of the genes involved will enable us to be much more precise about the implications for every individual, even in a first or early visit to the clinic.

<Intertitle>

Preparation of DNA:

Isolating pure DNA from the bacteria in which it is cloned

<Film of scientists in laboratory dispensing samples into 96-well cell plates>

<Intertitle>

Adding DNA to chemical solution, in preparation for sequencing

<Film of scientist operating machine in which DNA is added to chemical solution>

<Intertitle>

Thermocycler – a special incubator producing the DNA sequence from DNA copies, tagged and coloured with ultra-violet dye

<Film of scientist operating thermocycler>

<Intertitle>

Quality control of DNA sequence

<Film of scientist viewing DNA sequence on VDU monitor>

<Intertitle>

Loading DNA plates into sequence analyser

DNA being extracted from plates, and inserted into analyser

<Film of scientist operating sequence analyser>

<Intertitle>

DNA sequence analysis

<Film of two scientists viewing DNA sequence analysis on VDU monitor>

<Intertitle>

Previously published Genome research

<Film of pages being turned in booklet showing DNA sequence information and then pages of journal, Nature>

<Intertitle>

Isobel and Mark Emms

<Isobel Emms to camera>

I would say, over the last twelve months, I've come to believe that there is a lot of hope. Things are moving quickly.

<Intertitle>

Theo Emms

In physiotherapy class.

In School class.

<Voice of physiotherapist as she raises Theo's leg>

Bug's life, did you see the film? There you go.

<Theo>

Well, guess what? I've got a friend called Fraser and he's got a *<unclear word>*.

<Physiotherapist>

Has he?

<Film of Theo playing with construction toys at a table with other children, general chatter of children>

<End credits>