What is the generation study?

Genomics 101 explainer episode transcript

**Naimah:** What is the Generation Study? I'm Naimah Callachand, and today I'm joined by David Bick, the principal clinician for the Newborn Genomes Program at Genomics England, and he's going to explain more.

Okay, so first of all, David, please, can you tell me what is the generation study?

**David:** The Generation Study is a research study organised by Genomics England in partnership with the NHS. So what is the study exactly? We know that children are born every day with treatable genetic conditions. What we want to do in this study is we want to find those children and treat them before they become sick. We know that if we can find these children early in life, we can keep them healthy.

**Naimah:** Can you tell me a bit about how the study was designed?

**David:** Yes, this study was designed to look for genetic conditions that are treatable, and we went about looking for which conditions to include through an extensive evaluation that involves specialists, laboratory specialists, the NHS and patients from different support groups. And through this process, we identified more than 200 conditions that are treatable, and we are including those in the study.

**Naimah:** And David, you mentioned the conditions list there. You can also find an additional podcast on our website where you go into more detail as to why the conditions on that list were chosen initially for the study.

Can you tell me how this fits in with the current newborn screening program?

**David:** The current newborn screening program looks for a smaller number of conditions, nine conditions. It has been extremely successful and is an extremely important program, and so we're looking to see how our program could be an adjunct to the current screening process.

**Naimah:** How would people take part in this study?

**David:** What we're going to do is we're going to ask couples in mid-trimester, in the middle of pregnancy to join in the study, we're going give them a number of opportunities to learn about the study and those that sign up. We plan to obtain cord blood from the placenta after birth. And as you may recall, the placenta is discarded and so this cord blood would normally simply be discarded, but we can take this umbilical cord blood and test it for genes. We expect it to expand to perhaps 40 trusts across the country.

**Naimah:** And David, the NHS is already under quite a lot of pressure at the minute. What will this study mean for the NHS? What impact will it have?

**David:** That has been one of the main concerns as we went forward with thinking about the study was to make sure that we did not add additional burden to the NHS. But it's important to realise that the children that we're looking for already have the condition, they're going to become ill.

And our plan is, our hope is that if we can find them before they become ill, we will actually relieve stress on the NHS system. Let me give an example. There is a condition called biotinidase deficiency. Here is a condition where the child who has it, is unable to recycle a vitamin called biotin. Well, biotin is something you can pick up at the health food store, and so these children.

Need to be given extra biotin every day. Very, very inexpensive, very, very safe. But if you don't find these children before they become ill, they can become quite seriously ill. So if we can find these children before they get sick, get them started on this very simple, very inexpensive, very safe medication vitamin. In fact, this will actually save money for the NHS, but also help the NHS function more effectively. And most importantly, to allow parents to have the knowledge that they have done something for their child, which will prevent their child from becoming ill.

**Naimah:** Some parents might want to know if their child's data is being kept safe. What are you doing to ensure this?

**David:** Data safety is very important to us. We know that this information is extremely sensitive, and so our data protection is a very high priority. We're controlling very carefully who will have access to the data. There are groups that we do want to work with this data. We want researchers and the pharmaceutical industry to work with this data to improve treatments.

We know that there are many, many genetic conditions for which there is no treatment. Those conditions are not on our panel, and so we're hoping that the information that we learn from these genomes can be used to improve the care of conditions that exist, that are treatable, but most importantly, to find treatments for conditions that are currently not treatable.

And one way that we're doing this is we're going to ask families to stay with us for several years after the program starts. Once we have screened families, we're going to find some that are going to be screened positive, in which case we would look to see whether they have the condition or not, and then go ahead and start treatment.

We also, unfortunately, in a screening situation, we know that we're going to miss some cases, and so again, we want to stay in contact with families so we can know how to improve the testing, but most importantly, to understand how the test works, how screening works using genome and determine whether this makes sense to include all patients that are born in England going forward.

**Naimah:** And can you just finally tell me where you expect this study to go? In the coming months?

**David:** What we hope is that one day we'll reach a point where we can find and treat children who have treatable conditions before they get ill. this would really make our healthcare system a real healthcare system. We would find children before they get ill.

In this way we can keep children healthy and help them lead and live their best lives.

**Naimah:** So that was David Bick explaining ‘what is The Generation Study’. You can find out more about the study on our website and if you'd like to follow us on social media, you'll be able to keep up to date with any of the updates.

Thank you for listening.