New gene sequencers slash disorder detection time

Cutting-edge gene sequencers installed at a children's hospital will cut detection times for rare genetic disorders from a year to weeks.

About 15 of the £500,000 Next Generation Sequencers at The Children's Hospital, Sheffield, are in operation at NHS trusts around the country but the two installed at Sheffield are even more efficient because they have been combined with the latest robotic technology to help spot faulty genes as early as possible.

The Children's Hospital said blood samples were usually processed one gene at a time and, therefore, an accurate diagnosis for the cause of illness could take up to a year.

But the new machines allow up to 100 genes to be tested at once and the results will be back to the patient within a maximum of eight weeks and as little as a fortnight.

"This is going to revolutionise genetic science immediately for our patients," said Dr Ann Dalton, consultant clinical director of genetics at Sheffield Children's NHS Foundation Trust.

"What makes us unique from the majority of the other facilities in the country is that we have an excellent system built around the sequencer, including robotic machines.

"This allows us to fully process a sample and produce everything the scientist needs thanks to our in-house experts who interpret the data and generate clinically relevant reports."

Inherited genetic disorders were highlighted earlier this year when Hollywood superstar Angelina Jolie announced she had opted to have a double mastectomy after finding out she carried a faulty gene, called BRCA1.

This made her likelihood of developing breast cancer 87 per cent, which the operation cut to 5 per cent, but specialists at Sheffield said Jolie could have had her DNA tested on their "gene machine" and it would have shown which gene was responsible.

The new technology, which is funded by The Children's Hospital Charity, can test DNA from any living organism and the trust said it could help researchers understand more about many areas of biology, including diseases in animals and plants.

Darren Grafham, head of laboratory services for genetics at Sheffield Children's NHS Foundation Trust, has been working on the first phase of testing for patients suspected of having Glycogen Storage Diseases (GSDs), a set of serious but treatable metabolic disorders.

"The GSDs can be caused by gene mutations that affect different tissues: the liver, muscles, or the heart," he said. "This new technology allows us to test the many genes that may cause the disease all at once instead of one at a time, speeding up the process."

The machines will eventually be used to test for a wide spectrum of diseases including connective tissue disorders, inherited diseases such as some cancers and for newborn screening.