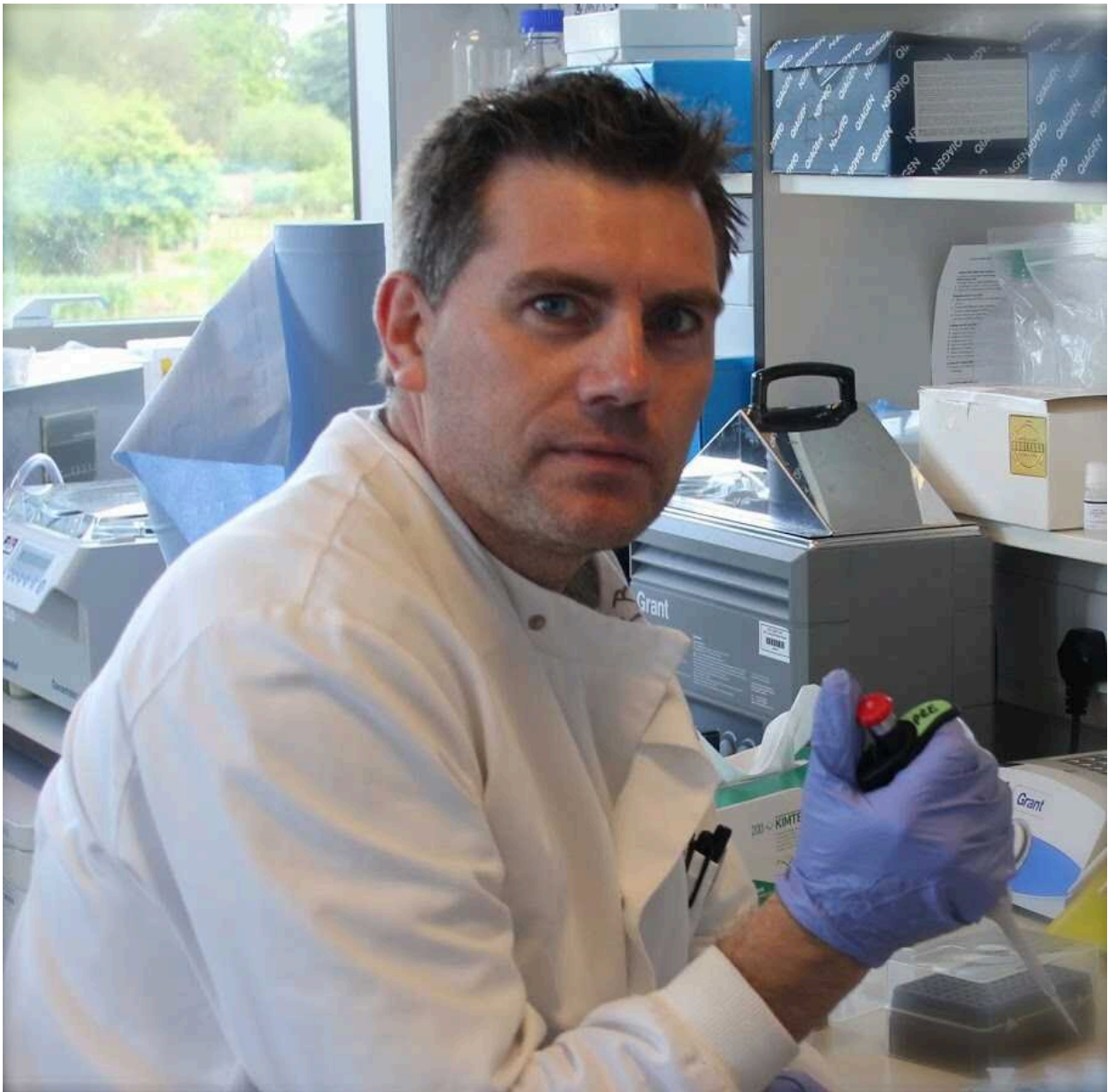


Pilot study reveals the benefits of 'whole genome sequencing' for children with cancer

November 8 2021



Dr Patrick Tarpey. Credit: NCRI/Patrick Tarpey

Unraveling the entire genetic code for each child who has been diagnosed with cancer can give a more accurate diagnosis or reveal new treatment options, according to research presented at the NCRI Festival. These are the results of a pilot study of the process known as whole genome sequencing.

The study also shows how [whole genome](#) sequencing, which is currently being rolled out for all childhood [cancer](#) patients in England, could be refined to bring maximum benefits to [children](#) with cancer.

The study was presented by Dr. Patrick Tarpey, lead scientist for solid cancer in the East Genomic Laboratory Hub, based at Cambridge University Hospitals NHS Foundation Trust, UK.

He said: "Whole genome sequencing can sometimes unravel unexpected results that may not have been considered via routine investigations. We've already learned a lot about using this type of test in our pilot study, both in terms of its benefits for children and in terms of how to optimize the results."

The study, part of the 100,000 Genomes Project, involved 36 children treated at Addenbrooke's hospital in Cambridge and included 23 different types of solid tumor. For each [child](#), a sample of their tumor and a sample of their blood were sent to a lab where the full DNA sequence could be read. Scientists then looked for genetic differences, called variants, that might offer clues about how the cancer developed, the specific cancer type and which treatments might be most effective.

This process revealed several potentially important variants. In two

cases, the information refined children's diagnosis, and in four cases, it changed their diagnosis. In eight cases, it revealed new information about children's prognoses (the likely course of their disease). In two cases, it showed possible hereditary causes of the cancers. In seven cases, it revealed treatments that might not have been considered but were likely to be effective for treating the children.

For example, in one child, scientists found that two genes had become stuck together (known as gene fusion), and this was probably encouraging the tumor to grow. Knowing this meant that doctors could offer a different [treatment](#), called a MEK inhibitor, that would not normally be used to treat this type of cancer.

Dr. Tarpey said: "Our results from this relatively small pilot group of children with cancer, show how diagnosis and treatment can be improved. It suggests that offering [whole genome sequencing](#) to all children with cancer will provide more accurate information on diagnosis and prognosis, show whether there could be any hereditary cancer risk and help inform treatment options."

The [pilot study](#) also showed that some of the important variants were difficult to spot among all the data produced by whole genome sequencing. Their significance might have been missed without the careful analysis and interpretation provided by an expert panel called a Genomic Tumor Advisory Board.

Dr. Tarpey added: "As we scale up whole genome sequencing to complement current standard-of-care testing for children with cancer, we need to ensure we optimize all the steps in this process to reduce turnaround times and keep financial costs down."

Dr. Julia Chisholm is Chair of the NCRI's Children's Group, based at The Royal Marsden NHS Foundation Trust and The Institute of Cancer

Research, London, UK, and was not involved in this research. She said: "Cancer is rare in children and thanks to research and better treatments, survival rates in the UK have doubled since the 1970s.

"This research shows that it's feasible and beneficial to analyze the whole genetic code in children diagnosed with cancer. Whole [genome](#) sequencing helps us to be more precise in tumor diagnosis and to tailor treatments to suit individual patients as accurately as possible. As this innovation is introduced more widely, we hope that even more children will survive cancer."

Provided by National Cancer Research Institute

Citation: Pilot study reveals the benefits of 'whole genome sequencing' for children with cancer (2021, November 8) retrieved 26 April 2024 from <https://medicalxpress.com/news/2021-11-reveals-benefits-genome-sequencing-children.html>

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