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Australia develops new blood test to rapidly diagnose rare genetic diseases

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The blood test will eventually be offered as a diagnostic service at the Victorian Clinical Genetic Services

Researchers from the University of Melbourne and Murdoch Children's Research Institute (MCRI) in Australia have developed a blood test capable of rapidly diagnosing rare genetic diseases in babies and children, eliminating the need for costly and invasive procedures and giving families earlier access to treatment.

Genome sequencing has advanced the diagnosis of rare diseases, however this test is only successful for half of all cases. The remaining half of patients must undergo additional functional tests to confirm if a gene mutation is causing the disease. This diagnostic process can take months or years with no guarantee of a result, as most functional tests are only applicable to a single or handful of rare diseases.

Now new research reveals a new blood test can rapidly detect abnormalities in up to 50 percent of all known rare genetic diseases in a matter of days by analysing the pathogenicity of thousands of gene mutations at once, potentially replacing thousands of other functional tests.

The research team benchmarked their test against an existing clinically accredited enzyme test offered by the Victorian Clinical Genetics Services at MCRI, focussing on mitochondrial diseases. These are a group of severe rare disorders that rob the body’s cells of energy, causing single or multiple organ dysfunction or failure, and potentially death.

The team found, comparatively, their new test is more effective in confirming a mitochondrial disease diagnosis as it’s much more sensitive and accurate and can produce faster results.

Dr Daniella Hock at University of Melbourne said, “A recent health economics analysis in collaboration with the Melbourne School of Population and Global Health showed that our test could be offered at a similar cost to the enzyme test that is currently offered clinically for mitochondrial diseases, but our test is much more cost-effective as it can test for thousands of different genetic diseases, whereas other functional tests are mostly targeted to a small number of genetic disorders."