# New blood test accelerates diagnosis of rare genetic disorders in children



The development of a groundbreaking blood-based test offers new hope for the swift diagnosis of children suffering from rare genetic disorders. Researchers at the University of Melbourne have designed a method that leverages the analysis of proteins in specific blood cells, which could not only hasten the identification of such disorders but also provide crucial treatment options sooner than current methods allow.

Rare genetic disorders encompass a wide range of conditions, including cystic fibrosis and diseases linked to mitochondrial dysfunction. Diagnosing these disorders has often been a painstaking process. Dr David Stroud, a co-author of the study, explained that while genomic testing has significantly improved diagnostic rates, it remains effective in only 30-50% of suspected cases. This can leave families in a state of uncertainty as they embark on a "diagnostic odyssey" — a protracted journey through various testing protocols over months or even years. The strain of this lengthy process is further compounded when invasive procedures, such as muscle biopsies, become necessary, particularly affecting young patients who must endure general anaesthesia.

In their research, published in the journal Genome Medicine, Stroud and his colleagues propose a complementary approach to genomic testing. By examining the proteins produced in response to specific gene alterations, this method promises a more comprehensive view of which genetic mutations are pathogenic, thereby expediting the diagnostic process. “Since genes are the instructions to make proteins, we use this information to ascertain which genetic changes lead to damages and which are benign,” Stroud commented.

The new proteomic test could potentially boost diagnostic accuracy to between 50-70%, addressing many cases where genomic testing alone falls short. Notably, this innovative method requires merely 1ml of blood from a newborn, greatly reducing the need for more invasive techniques. While primarily designed for mitochondrial diseases, Stroud mentioned its applicability extends to approximately half of the 7,000 known rare diseases, underscoring its utility across a broader clinical landscape.

The financial implications of this test are also significant. Since it is not restricted to specific disorders, it is expected to be more cost-effective, ultimately benefitting both patients and the healthcare system by minimising unnecessary tests. A definitive diagnosis can illuminate care pathways, paving the way for possible treatments and aiding parents in making informed decisions about future pregnancies.

The research has garnered praise from experts in the field. Michal Minczuk, a professor of mitochondrial genetics at the University of Cambridge, articulated the importance of the study, asserting that it represents a "very significant step forward in diagnostic practices." By introducing a rapid and minimally invasive method, this advancement can transform patient care and empower healthcare providers with better tools for genomic medicine.

In parallel to these advancements, organisations like the Centers for Disease Control and Prevention (CDC) continue to innovate in the realm of newborn screening. Recent developments by the CDC involve tests that screen for conditions such as homocystinuria (HCU), where elevated homocysteine levels in blood samples enable quicker diagnoses and interventions. Such proactive measures also emphasise the critical role early testing plays in improving infant health outcomes across the board.

Emerging technologies in this field, including machine learning combined with sequencing techniques, are further enhancing the accuracy of newborn screenings, diminishing the likelihood of false positives, and alleviating the stress associated with unnecessary further testing. Major breakthroughs in rapid genome sequencing, exemplified by records in speedy diagnoses of rare genetic diseases, further highlight the evolving landscape of neonatal diagnostics.

The ongoing commitment to refining diagnostic methodologies signals a promising future for patients and families affected by rare genetic disorders, ensuring they receive timely care and empowering them with knowledge that can significantly impact their lives. As researchers continue to push the boundaries of genomic and proteomic understandings, the hope is that these innovations translate into better outcomes for vulnerable populations.

## Reference Map:

* Paragraph 1 – [[1]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies), [[2]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies)
* Paragraph 2 – [[1]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies), [[6]](https://primaryimmune.org/understanding-primary-immunodeficiency/diagnosis/newborn-screening), [[5]](https://medicine.yale.edu/news-article/new-approach-improves-detection-of-diseases-at-birth/)
* Paragraph 3 – [[1]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies), [[2]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies), [[4]](https://www.nationwidechildrens.org/family-resources-education/health-wellness-and-safety-resources/helping-hands/newborn-screen-blood-test)
* Paragraph 4 – [[1]](https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies), [[5]](https://medicine.yale.edu/news-article/new-approach-improves-detection-of-diseases-at-birth/)
* Paragraph 5 – [[3]](https://www.cdc.gov/newborn-screening/featurestory/newborn-screening-is-a-lifesaver.html), [[7]](https://www.statnews.com/2022/01/12/researchers-shatter-speed-record-for-diagnosing-rare-genetic-diseases-with-dna-sequencing/)

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## Bibliography

1. <https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies> - Please view link - unable to able to access data
2. <https://www.theguardian.com/science/2025/may/23/blood-test-could-speed-diagnosis-rare-diseases-babies> - A new blood-based test developed by researchers aims to expedite diagnoses for children born with rare genetic disorders. This test analyzes proteins in blood cells to identify disease-causing mutations, potentially increasing diagnostic yield from 50% to 70%. Unlike traditional methods, it requires only 1ml of blood, reducing the need for invasive procedures like muscle biopsies. The approach is applicable to about half of the 7,000 known rare diseases, offering a cost-effective and less invasive diagnostic tool.
3. <https://www.cdc.gov/newborn-screening/featurestory/newborn-screening-is-a-lifesaver.html> - The Centers for Disease Control and Prevention (CDC) has developed a new test to screen newborns for homocystinuria (HCU), a rare genetic disorder. This test measures homocysteine levels directly from a blood sample, enabling faster diagnosis and treatment initiation, potentially preventing serious health issues or death. The CDC's study, published in the Clinical Chemistry journal, represents a significant advancement in newborn screening, improving the quality of life for infants with HCU.
4. <https://www.nationwidechildrens.org/family-resources-education/health-wellness-and-safety-resources/helping-hands/newborn-screen-blood-test> - Ohio law mandates that all newborns undergo a blood test to screen for rare genetic, hormone-related, or metabolic disorders. This test, performed within 24 to 48 hours after birth, can identify conditions that, if left undiagnosed, may lead to serious health problems or death. Early diagnosis and treatment are crucial for the child's healthy development. The blood sample is typically collected before the baby leaves the hospital, ensuring timely intervention.
5. <https://medicine.yale.edu/news-article/new-approach-improves-detection-of-diseases-at-birth/> - Yale researchers have developed a new sequencing technique combined with machine learning to enhance the detection of diseases in newborns. This approach aims to reduce false-positive results in newborn blood tests, which can cause unnecessary anxiety and additional testing. By improving the accuracy of these screenings, the method ensures that infants receive appropriate care without undue delays, marking a significant advancement in neonatal diagnostics.
6. <https://primaryimmune.org/understanding-primary-immunodeficiency/diagnosis/newborn-screening> - Newborn screening involves three tests: a blood test (heel stick), hearing screen, and pulse oximetry test, all conducted within 24-48 hours after birth. The blood test identifies potential serious health conditions, including severe combined immunodeficiency (SCID), by analyzing a small blood sample. Early detection through these screenings is vital for initiating timely treatments, ensuring better health outcomes for newborns.
7. <https://www.statnews.com/2022/01/12/researchers-shatter-speed-record-for-diagnosing-rare-genetic-diseases-with-dna-sequencing/> - Researchers have achieved a new speed record in diagnosing rare genetic diseases using DNA sequencing. In a case involving a child with heart issues, rapid genome sequencing identified a genetic cause within hours, leading to timely intervention. This breakthrough demonstrates the potential of ultra-rapid sequencing to expedite diagnoses, offering hope for patients with rare conditions who previously faced prolonged diagnostic odysseys.