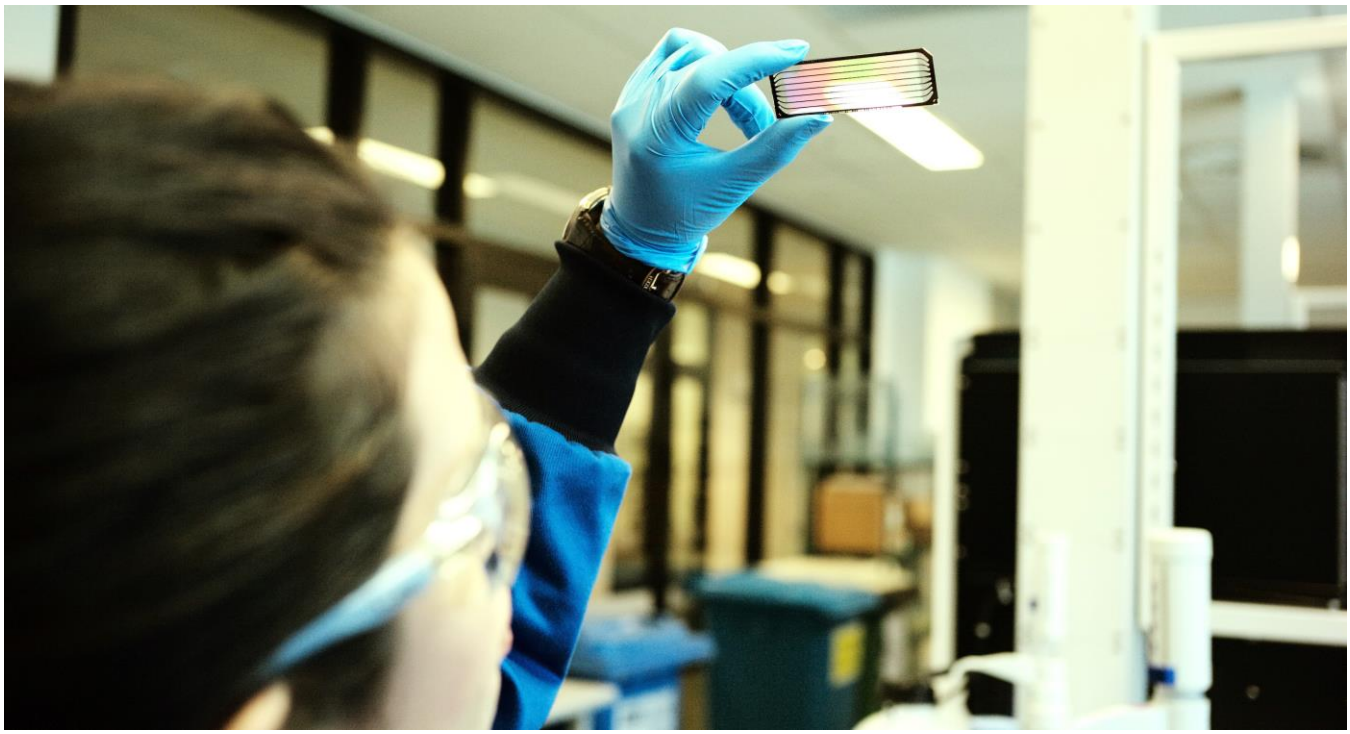
Advancing Health and Accelerating Discovery with Genome Sequencing



Genome sequencing has led to dramatic improvements in the diagnostic rate of genetic disease and is helping to advance medical research
Picture: Genome.One

Genetic disorders can go undiagnosed in patients leading to people enduring lengthy and invasive tests to try and find answers. Genome.One has developed a genomic test for rare and genetic conditions to assist Australians who are searching for a diagnosis.

The NSW-based health information company aims to advance the field of genomics by sequencing entire genomes for clinicians and researchers. The company captures and analyses information about the patient and reports it in a way that is accessible and meaningful for clinicians. By conducting whole genome sequencing, Genome.One offers people with genetic disorders a clearer, more reliable and less drawn out path to diagnosis. As a subsidiary of the Garvan Institute of Medical Research, Genome.One brings together clinical and research excellence, allowing individuals who remain undiagnosed to transition to research projects and clinical trials for further investigation.

Once a diagnosis is formed from the testing, Genome.One hopes that it can give families the answers that they need to work with their doctors to make informed choices about their loved ones' care and treatment. Many people with rare genetic disorders endure months or years of testing before receiving a diagnosis, with up to 40% of people waiting between 5-30 years for answers.

Genomic testing has led to dramatic improvements in the diagnostic rate of genetic disease, enabling access to new therapies, treatment, support opportunities and leading to reduction in unnecessary consultations, tests and treatments. Genome.One's high reporting rates have also been demonstrated in validation studies for specific disorders, with rates as high as 86% in patients with autosomal dominant polycystic kidney disease.

CEO of Genome.One Marcel Dinger explains that genome sequencing is essential both for streamlining current genetic diagnosis and to facilitate discoveries that will accelerate diagnoses in the future.

"For families, receiving an accurate and timely diagnosis could result in access to new treatments and therapies as well as a clearer understanding of the journey ahead," Dr Dinger said. "With the existence of thousands of Mendelian disorders known to affect human health, each traditionally requiring an independent test to diagnose, genome sequencing provides a rapid and efficient means to conclusively and rapidly provide a precise molecular diagnosis of such disorders."

Genome.One's whole genome sequencing test has reported variants in 54% of cases, with a testing turnaround time of 8-12 weeks, often avoiding the lengthy diagnostic "odysseys" that may occur with single gene testing.

One family who had two girls with severe epilepsy endured years of different medical specialists and therapists, undergoing highly invasive testing and trialling different drugs, diets and supplements. Whole genome sequencing gave the family "some real hope" with a diagnosis. From this success, the family has connected with more than 100 families around the world who have children with the same condition.

Genome.One's ongoing discovery and genomics collaborations with research and biotech organisations are helping to advance knowledge in the field, which will lead to more robust testing services. Additionally, the development of applications to integrate genomic sequences with rich phenotypic information will help to realise the benefits of genomics and improve testing and treatment options

"Through our Discovery program, we are actively partnering with local and international clinical and research communities – including industry – to grow our knowledge of the human genome. This will ultimately lead to further breakthroughs in diagnostics, drug development and targeted treatments and a greater understanding of how each person's unique genome affects their individual health," Dr Dinger said.

For more information visit www.genome.one or contact Genome.One's head of Discovery Andrew Stone via andrew.stone@genome.one.