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Scientists develop genome-wide mutation hunting computational software for genomic medicine

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Phen-Gen is the first computer analysis software that cross-references a patient's symptoms and a person's genome sequence, to better aid doctors in diagnosing diseases.

This software was created by a team of scientists at A*STAR's Genome Institute of Singapore (GIS), led by Dr. Pauline Ng. Results from the research were published in the prestigious journal *Nature Methods* on 4th August 2014.

Phen-Gen can detect faulty genes responsible for diseases by up to 88 per cent, yielding results in 15 to 30 minutes. It has been proven to be faster and more efficient compared to current methods analysing genomes for this purpose.

One area that Dr Ng is currently working on is incorporating the Phen-Gen technique in the diagnosis of rare diseases. Rare diseases are often hard to diagnose based on symptoms alone. By using Phen-Gen, doctors are able to make a more accurate diagnosis based on a patient's unique genetic code.

Dr Ng's team is working with doctors in local and international hospitals to incorporate Phen-Gen to diagnose patients with rare disorders. "We aim to translate scientific research to help people directly," said senior author Pauline Ng. "To this end, GIS has created a programme to help

diagnose patients with rare disorders. Phen-Gen works with both exome and whole genome sequencing data. It is the first algorithm to leverage disease symptoms and give genome-wide predictions."

Most rare diseases, such as those that affect neurological, brain or cardiac development, manifest early in life. "There is little else more satisfying than the opportunity to help a sick patient, and through our research at GIS, we want others in the world to benefit as well," said first author Dr. Asif Javed. "The program is also downloadable online for those who prefer to keep their DNA information private."

The Executive Director of the GIS, Prof. Ng Huck Hui, commented, "As we enter the genomics era with more powerful Next-Generation Sequencing technologies that can analyse the human genomes at a reduced cost, data analytics becomes a bottleneck. Dr. Pauline Ng's group has taken on this exciting challenge to develop analytics capabilities. In partnership with the Singapore hospitals, the GIS has initiated a research project on sequencing patients with undiagnosed conditions or congenital disorders. The Phen-Gen method is timely as it fills an urgent gap in hospitals for accurate diagnosis of rare diseases."

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