New genome analysis method detects genetic diseases more efficiently, scientists claim

A team of scientists has developed *Phen-Gen*, the first computer analysis software that cross-references a patient's symptoms and a person's genome sequence. The results from the research have been published in the journal *Nature Methods*.

Phen-Gen can detect faulty genes responsible for diseases by up to 88 percent, 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.

Led by Dr. Pauline Ng from the Genome Institute of Singapore (GIS) under the Agency for Science, Technology and Research (A*STAR), the team 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.

"We aim to translate scientific research to help people directly," said Ng, who was the senior author of the study. "To this end, GIS has created a program 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."

Read the full, original story: Detecting rare genetic diseases in under 30 minutes