Whole genome sequencing could have huge impact on medicine

The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion and analysis.

“It sounds kind of silly, right? Rare disease sounds rare,” Jacob said during his keynote speech at the Bio-IT World Conference & Expo in Boston. Yet 5-10% of the U.S. population lives with a rare, undiagnosed disease. Jacob believes that bringing whole genome sequencing to the clinic is the most accurate and economical option for these patients. And he has experience doing it.

Jacob is currently Executive Vice President for Genomic Medicine at the HudsonAlpha Institute for Biotechnology and CEO of Envision Genomics, both in Huntsville, Alabama. He previously pioneered patient-focused genome sequencing as the Director of the Human and Molecular Genetics Center at the Medical College of Wisconsin.

Whole exome sequencing covers approximately 1.5% of the genome, corresponding to gene-coding DNA. For many years, some researchers thought the rest of the genome was essentially “junk DNA” with no purpose. But the ENCODE project now shows that 80% of the genome is biochemically functional. This is why Jacob advocates for the use of whole genome sequencing, which he says results in a diagnosis 25% more often than whole exome sequencing. That percentage will rise as more genomic data is collected.

Jacob presented a study where 9 out of 22 instances of whole genome sequencing provided a diagnosis. Whole exome sequencing would have missed 3 out of those 9 diagnoses.

But there is a problem. “Insurance companies don’t pay for this, it is a major issue,” Jacob said.

Read full, original post: Howard Jacob on Why Whole Genome Sequencing is Best for Medicine