Whole-exome sequencing IDs disorders when other tests fail

When all else fails, whole-exome sequencing (WES) may succeed at nailing down a tough diagnosis, say investigators.

A new review of 1200 clinical cases in which patients presented with nonspecific or unusual symptoms suggestive of a genetic disorder showed that WES was positive for a specific condition in 26% of unselected clinical samples.

WES is an efficient and comparatively inexpensive alternative to whole genome sequencing that looks at exons, the protein-coding regions of genes, and at adjoining sequences to aid in the diagnosis of Mendelian disorders, pharmacogenetic traits, and potentially complex traits. WES currently costs around $1000 vs about $5000 for whole genome sequencing, although the price of the latter continues to fall.

Read the full, original story here: Whole-Exome Sequencing Unmasks Rare Disorders