Genetic Science Spotlight

The European Society of Human Genetics Recommends WES as Fist-line Diagnostic Tool in Cases with No Clear Differential Diagnosis

Provided with the advances in sequencing techniques, Whole Exome Sequencing (WES) has been employed in clinical diagnostic use but mostly limited to recessive disease screening in consanguineous families and highly selected groups of patients with homogenous disease presentations. Only until recently has a study been published in the European Journal of Human Genetics, the official journal of The European Society of Human Genetics, validating WES as an effective first-line diagnostic tool, particularly in cases with nonspecific and heterogeneous phenotypes. The research project was led by Centogene AG in Germany and included 1,000 unrelated cases from 54 counties, presented with 19 major types of symptoms as categorized per the Human Phenotype Ontology (HPO) system. 307 of the 1,000 (30.7%) cases had a positive genetic finding, among which 165 patients received a definitive molecular diagnosis and 142 had likely pathogenic variants detected in them. A positive correlation was also observed between greater phenotypic complexity and higher diagnostic yield. Notably, 3 patients received a molecular diagnosis of 2 pathogenic or likely pathogenic variants associated with either non-overlapping clinical presentations or contributing to one major phenotype, namely non-incidental dual findings. In short, when there is no clear differential diagnosis for a case, his physicians, medical geneticists and the diagnostic laboratory should consider diagnostic WES as a first-line option and work closely together, provided that the patient has been appropriately counseled in the implications and limitations of the test, as well as the potential disclosure of medically actionable incidental findings.

http://www.nature.com/ejhg/journal/vaop/ncurrent/full/ejhg2016146a.html

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