RESULTS AND DISCUSSION

Among the 118 cases in which a clinically-relevant diagnostic finding was detected, 3% of cases were identified as having multiple diagnoses. Inheritance patterns ranging from autosomal dominant, X-linked dominant, X-linked recessive, mitochondrial DNA inheritance, and autosomal recessive with uniparental isodisomy were represented.

Disorders resulted from 2 SNVs, a SNV plus aneuploidy, a nuclear DNA SNV plus a mitochondrial DNA SNV, and a case of multiple diagnoses resulting from an SNV, CNV, aneuploidy, and uniparental isodisomy.

50% of patients with multiple diagnoses had a previous analysis that identified one of their diagnoses suggesting that the diagnostic evaluation should not always end with the identification of an initial molecular diagnosis and that genome-wide analysis can still be beneficial in some cases.