potentially related to the proband's medical condition are reported. Rare polymorphisms may lead to false negative or positive results. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. If results obtained do not match the clinical findings, additional testing should be considered. Specific genetic events like copy number variants, translocations and repeat expansions may not be reliably detected with Exome Sequencing. In addition, due to limitations in technology, certain regions may either not be covered or may be poorly covered, where variants cannot be confidently detected.

ADDITIONAL INFORMATION

This test was developed and its performance validated by Breakthrough Genomics. The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts. In line with ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing (Genetics in Medicine, 2016), we report incidental findings, i.e. pathogenic variants (class 1) and likely pathogenic variants (class 2) only in the recommended genes for the recommended phenotypes. To also exclude mistaken identity in your clinic, several guidelines recommend testing a second sample that is independently obtained from the proband. Please note that any further analysis will result in additional costs. The classification of variants can change over the time. Please feel free to contact Breakthrough Genomics (info@btgenomics.com) in the future to determine if there have been any changes in classification of any reported variants.

ENLITER v3.0.1

Assembly (GRCh37.p13)	gnomAD (r2.1)	ExAct (r1)	samtools (1.6)
ESP (20141103)	ClinVar (2019-08)	RefSeq (2015-01)	bedtools (2.17.0)
SIFT (5.2.2)	PolyPhen (2.2.2)	GENCODE (19)	bcftools (1.6)
1000 Genomes (Phase 3)	HGMD (2017.4)	BWA (0.7.17-r1188)	Picard (2.8.2-4-g2105a1e)
dbSNP (151)	genebuild (2011-04)	GATK (4.0.1)	

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04/05/2020

Date