Cerebrum DX *Mito*

Genekor Medical S.A. | 52, Spaton Ave., 15344, Gerakas, Athens, Greece ,G.E.MI. nr: 0007856001000
email: info@genekor.com, www.genekor.com | Tel. (+30) 210 6032138 Fax. (+30) 210 6032148
Scientific Director: George Nasioulas PhD

SAMPLE INFORMATION

Name :	-	**Date Received :**	-
Medical ID :	-	**Date of Report :**	-
Date of Birth :	-	**Req. Physician :**	-
Location :	-	**Barcode :**	-
Material :	WHOLE PERIPHERAL BLOOD	**Reason of referal:**	Mitochondrial disorders

CerebrumDX mito analysis by Next Generation Sequencing

Results associated with the reason of referral

NEGATIVE

No pathogenic/likely pathogenic variant related to the referral cause was identified.

Methodology

Genomic DNA was extracted from the sample under investigation and was analysed by a solution based capture approach using the target enrichment panel of mitochondrial DNA (KAPA HyperCap DS Human mtDNA Design ,Roche). These regions include exonic and flanking intronic regions of the analyzed genes. Sequencing was carried out using the DNBSEQ-G400 sequencing platform (MGI). The impact of missense changes on the structure and function of the protein is assessed using the prediction algorithm APOGEE (PMID: 28640805). The classification of mitochondrial variants was based on ACMG criteria (PMID: 32906214), and specific databases such as Mitomap and HmtVar were used for their interpretation. The mean coverage was …x with ..% of all targeted regions sequenced with >=100x depth.

The presence of LGRs is verified by use the MLPA method (Multiplex Ligation-dependent Probe Amplification, MRC Holland; PMID: 10978226) Probemix P125-C1.

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*Note:
Each molecular analysis has an internal error probability of 0.5-1%. This is due to rare molecular events and factors involved in the preparation and analysis of samples.
The method used achieves 99% sensitivity and specificity in detecting point nucleotide changes, duplications, and deletions <15bp.

Details about non-pathogenic variants

Non-pathogenic polymorphisms (findings without clinical significance) are not reported, as it is documented that they likely do not confer an increased risk of disease and therefore do not alter medical management beyond what is indicated based on family and personal history.

Genes analyzed (Table 1)

MT-ATP6	*MT-ATP8*	*MT-CO1*	*MT-CO2*	*MT-CO3*	*MT-CYB*	*MT-ND1*	*MT-ND2*	*MT-ND3*
MT-ND4	*MT-ND4L*	*MT-ND5*	*MT-ND6*	*MT-RNR1*	*MT-RNR2*	*MT-TA*	*MT-TC*	*MT-TD*
MT-TE	*MT-TF*	*MT-TG*	*MT-TH*	*MT-TI*	*MT-TK*	*MT-TL1*	*MT-TL2*	*MT-TM*
MT-TN	*MT-TP*	*MT-TQ*	*MT-TR*	*MT-TS1*	*MT-TS2*	*MT-TT*	*MT-TV*	*MT-TW*
MT-TY								

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