Cerebrum DX Arrays

Confidential
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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:**	e.g Autism

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Results

Genetic analysis using GenetiSure Cyto revealed a male genetic profile, with no indications of significant copy number variations (CNVs) in the chromosomal regions studied that are directly correlated with the clinical phenotype.

Karyotype: arr(X,Y)x1,(1-22)x2

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Methodology

Genomic DNA was extracted from the sample under investigation. The comparative genomic hybridization method using the GenetiSure Cyto 8 x 60K CGH Microarrays from Agilent Technologies was applied. This method allows for the detection of copy number variations (CNVs) and/or large deletions/duplications. The results were analyzed using Agilent CytoGenomics 5.2.0.20 software and according to the specifications of the Genome assembly NCBI Build 38. This methodology covers 3644 clinically significant genes, as documented in ClinGen, ClinVar, OMIM, and Development Disorder Genotype-Phenotype Database (DDG2P) databases (https://www.agilent.com/cs/library/brochures/GenetiSure%20Cyto%20Brochure%205994-1727EN.pdf). Additionally, this analysis provides greater coverage of telomeric and pseudo-autosomal chromosomal regions. The average resolution of the microarrays used is at least 200 Kb across the entire genome, with higher resolution in critical chromosomal regions and telomeric regions. Moreover, the high-quality probes used enable the detection of mosaicism with greater specificity and sensitivity while limiting the detection of non-clinically significant/random findings.

Notes:

1) Results were evaluated according to the guidelines for the use of microarrays for postnatal constitutional cytogenomic analysis (European guidelines for constitutional cytogenomic analysis, 2017, ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013). Databases Decipher, DGV, ClinVar, Ecaruca, OMIM were utilized for analysis.
2) The average resolution of the microarrays used is at least 200 Kb across the entire genome, potentially higher in critical chromosomal regions and telomeric regions. A negative result does not exclude the presence of deletions/duplications beyond the sensitivity limits of this analysis, which could be detected by other techniques with higher sensitivity.
3) Molecular karyotype is suitable only for detecting imbalances (duplications or deletions - CNVs) and not for detecting balanced rearrangements, low-level mosaicism, and point mutations, which could be detected by other techniques such as conventional karyotyping, Clinical Exome Sequencing (CES), Whole Exome Sequencing (WES), or Whole Genome Sequencing (WGS).
4) False-negative or false-positive results may arise due to biological or technical limitations (e.g., maternal contamination).
5) 5) The presence of mutations or polymorphisms in the tested chromosomal regions may affect the final result.