VCGS
leaders in genetic health

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LIMS Report #: 981295

Patient: 86200001 CLN 1 86200001 Cln 1

Ms. Annabelle Suter
MCRI
Flemington Rd
PARKVILLE VIC 3052

DOB: Unknown
Sex:
VCGS sample ID: 23C101705
Date collected: 20-Feb-2023
Date received: 20-Feb-2023
Date reported: 03-Mar-2023
Source: Cell Pellet
Ext. sample ID:
Ext. patient ID:

Cytogenetics Laboratory

Clinical details	Cell line
Specimen source	Cell Pellet

Molecular karyotype

Array type	Illumina Infinium GSA-24 v3.0
Resolution	0.50Mb
Reference genome	GRCh38 / hg38 (Dec 2013)
Molecular karyotype	arr(X,Y)x1,(1-22)x2
Result	NO ANEUPLOIDIES DETECTED

Interpretation

Male molecular karyotype. No aneuploidies were detected in this sample.

Molecular karyotyping is limited in its ability to detect low grade mosaicism and genomic copy number changes below the resolution stated. Balanced rearrangements and Robertsonian translocations will not be detected. This test does not exclude single gene disorders caused by sequence mutations or trinucleotide repeat expansions (such as fragile X syndrome, Huntington disease, some spinocerebellar ataxias, Friedreich ataxia and myotonic dystrophy). Testing for fragile X syndrome should be considered in individuals with developmental delay/ intellectual disability. Further genomic based testing may be considered if there remains a high suspicion of a monogenic disorder. Copy number variants that do not contain genes, are well established polymorphisms, or are assessed as being of unlikely clinical significance (based on "ACMG Technical Standards for the Interpretation and Reporting of Constitutional Copy Number Variants"), will not be reported. The classification is based on the current scientific evidence available at the time of reporting. Reporting of regions of homozygosity (>5Mb) is dependent on referral setting and clinical indication. CNVs that contain autosomal recessive genes will not be reported unless there is specific clinical relevance, high carrier population frequency or a history of consanguinity. Please contact the laboratory if there is a family history of a known recessive disorder or a clinically suspected recessive condition. CNVs involving moderate to high risk cancer susceptibility genes may be reported. However, incidentally ascertained CNVs involving genes assessed as having low to moderate cancer risk will not generally be reported unless they are part of a larger CNV. This testing was performed on a standard SNP microarray platform which may not have sufficient probe coverage to detect clinically relevant CNVs related to this patient's specific clinical features. Please contact the laboratory if a higher resolution microarray may be required for a specific gene or genetic condition. This test does not exclude the possibility of tissue limited mosaicism and further testing of an alternative tissue may be considered if clinically indicated. Interpretation is based on the UCSC GRCh38/hg38 human reference sequence.

Validated: 03-Mar-2023 by David Francis

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END OF TEST REPORT

FINAL REPORT

PATHOLOGY REPORT

RCPA
NATA
Accredited for compliance with NPAAC Standards and ISO 15189
Accreditation number: 3171