

LIMS Report #: 981309

Patient: 862033213 CLN 1 862033213 Cln 1

Ms. Annabelle Suter  
MCRI  
Flemington Rd  
PARKVILLE VIC 3052

DOB: Unknown  
Sex:  
VCGS sample ID: 23C101701  
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