Dr Katerina Vlahos
Murdoch Childrens Research Institute
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PARKVILLE VIC 3052

Report for Patient Cim001.4 CIM001.4 is attached.
Sample: 17C108285 (422060)
Report #: 394006

Date Printed: 29/11/2017 11:53:30 AM

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LIMS Report #: 394006

Patient: CIM001.4 Cim001.4

DOB: Unknown
Sex: Unknown
VCGS sample ID: 17C108285
Date collected: 02-Nov-2017
Date received: 02-Nov-2017
Date reported: 29-Nov-2017
Source: Cell Pellet
Ext. sample ID:
Ext. patient ID:

Dr. Andrew Elefanty
Institute Of Medical Research
Royal Parade
PARKVILLE VIC 3052

CC: Dr. Katerina Vlahos

Cytogenetics Laboratory

Specimen source Cell Pellet

Molecular karyotype

Array type	Illumina Infinium GSA-24 v1.0
Resolution	0.50Mb
Assembly	hg19 / GRCh37 (Feb 2009)
Molecular karyotype	arr(1-22,X)x2
Result	NO ANEUPLOIDIES DETECTED

Interpretation
Female 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 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. Copy number changes that do not contain genes, are well established polymorphisms, or are assessed as being of unlikely clinical significance (based on available evidence), will not be reported. Reporting of regions of homozygosity (>5Mb) is dependent on referral setting and clinical indication. Please contact the laboratory if a recessive disorder is suspected. Interpretation is based on the UCSC GRCh37/hg19 human reference sequence.

Validated: 29-Nov-2017 by Kathy Butler

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PATHOLOGY REPORT

END OF TEST REPORT

FINAL REPORT