VIO TCU - Saumya.

Reg No 198904227G

KK Women's and Children's Hospital
SingHealth

DNA Diagnostic and Research Laboratory
Genetics Service
Department of Paediatrics

Chromosome Microarray Analysis Report

Patient and Sample particulars:

Name:	ROBERT MIOTKE	Sample ID:	#141191
IC No.:	Y0756094645C	Lab ID:	CM14050
Sex:	Male	Sample Type:	Peripheral Blood
DOB:	05/04/2013	Date collected:	09/05/2014
		Date Received:	10/05/2014

Requesting Doctor:

Name:	Dr Saumya Jamuar	Location:	Clinic K
Institution:	KKH		

Indication: Neuroregression, short nose, hypotonia, seizures

Date of Report: 11 June 2014

RESULT: NORMAL - ISCN arr(1-22)x2,(XY)x1

Interpretation:

Chromosome Microarray Analysis (CMA) showed a male profile with no copy number changes that are associated with known microdeletion or microduplication syndromes.

AOH (Absence of heterozygosity), suggestive of uniparental disomy, was NOT detected.

The clinical phenotype of the patient cannot be explained by this result.

Further clinical evaluation is recommended.

METHODOLOGY:

CMA was carried out using the Agilent 4x180K CGH+SNP array (G4890A, slide no. 252983020864_1_2). The referred sample was hybridized against the Agilent Euro Male reference. Data were scanned at a resolution of 5 μ and analysed with the Agilent Cytogenomics software version 2.5 using the ADM-2 setting which provides a practical average resolution of 50 kb. Benign CNVs are not listed and can be provided upon request. CNVs less than 300 kb in size will not be reported unless they contain genes known to be associated with disease. Only CNVs relevant to the phenotype will be reported but a complete list of CNVs is available to the referring clinician upon request. Interpretation of the clinical significance of detected CNVs is based on currently available medical literature. CMA will not detect balanced translocations, inversions, point mutations and low-level mosaicism.

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