ALTUM CHROMOSOMAL MICROARRAY REPORT

Name:	Age/Gender: /
Patient ID:	Sample ID:
Specimen type:	Sample Quality:
Referring Doctor:	Referring Center:
Test:	Collection Date:
Recieving Date:	Reporting Date:
Indication:	

SUMMARY

No clinically relevant copy number gains or losses were found in the targeted regions of the given specimen.

RESULT

Clinically relevant Copy Number Variations: **Not Detected**

INTERPRETATION

Chromosomal microarray analysis did not reveal any copy number variations of clinical significance.

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TEST METHODOLOGY

This microarray consists of 750K oligonucleotide probes across the genome, including 550K unique non-polymorphic probes, and 200K bi-allelic SNP (single nucleotide polymorphism) probes. The minimum resolution for detection is ~200 kb for losses, ~400 kb for gains and >5 Mb for LOH. However, LOH will be reported depending upon chromosomal location, significance and likelihood of imprinting disorder.

Genomic DNA was digested with Nsp1 and then ligated by Nsp1 adaptor followed by PCR amplification. Amplified PCR products were then purified and fragmented. The fragmented products were labelled w ith biotin and hybridised overnight onto the array. The array was washed using a fluidics station and then scanned on an Affymetrix GeneChip scanner. The data file generated was analysed using Chromosome Analysis Suite (ChAS). The analysis is based on the Human reference genome (GRCh38/hg38). All findings are correlated with clinical history before reporting. All VOUS (variants of unknown significance) are reported if they are found relevant to clinical history. An unrelated pathogenic or likely pathogenic finding is reported if there is sufficient empirical evidence for its involvement in a disorder.

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REFERENCES

1. South *et. al.* , Constitutional Microarray Guidelines, Genetics in medicine, Volume 15, Number 11, November 2013.
2. CytoScan Suite- Data sheet.

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- End of Report -

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