viapath

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ARRAY CGH REPORT

CELL LINE

Last Name : **IOP101**
First Names : **P5**

Specimen No: 18/03471
PRU No: 454151:01
Date Taken: 30/01/2018
Date Rec'd: 30/01/2018
Hospital No:

CHROMOSOME IMBALANCE DETECTED

arr[GRCh37] 3q27.1(182749913_183145846)x1,17p13.1(8262727_8636715)x3

Array CGH analysis of DNA from this cell line has been carried out using oligonucleotide arrays with ~60,000 probes across the genome.

This test identified two regions of imbalance:
i) deletion of approximately 0.396Mb from band q27.1 in the long arm of chromosome 3, between base pair coordinates 182,749,913 and 183,145,846.
ii) duplication of approximately 0.374Mb of material originating from the short arm of chromosome 17; the duplicated region is from band p13.1, between base pair coordinates 8,262,727 and 8,636,715.

These findings are rare variants that are not commonly observed; however, the clinical significance of these imbalances is uncertain.

Other imbalance >3Mb has been excluded; however, due to the poor quality of DNA extracted from this sample, other small imbalances have not been excluded. Please send a repeat sample if further array CGH testing is required. Alternatively, we would accept a reference cell line for comparative hybridisation.

The results are consistent with a female chromosome complement.

Please see separate report for the results of the microsatellite analysis.

Array CGH is a technique for detecting abnormalities of genomic copy number. It has a higher resolution than karyotype analysis, and will therefore detect regions of imbalance too small to be detected by analysis of G-banded chromosomes. It will not detect balanced chromosome rearrangements or ploidy abnormalities such as triploidy, and low level mosaicism may not be detected. Interpretation of array CGH findings is based on current knowledge; future advances may provide further insight.

Array platform: Agilent design 085030. Median resolution: 120kb. Data analysis: Agilent GW. Positional information: GRCh37/hg19

Reported by**: AC** AC

Authorised by:

Date reported**: 21/02/2018**