To: Dr Dusko Ilic
ACU
11th Floor Tower Wing
Guy's Hospital
Great Maze Pond
London
SE1 9RT

Genetics Centre
Guy's Hospital, 5th Floor Tower Wing
Great Maze Pond, London SE1 9RT
Tel: 020-7188-1709

ARRAY CGH REPORT

CELL LINE

Last Name : **IEBS**
First Names : **P10**

Specimen No: 18/04221
PRU No: 454870:01
Date Taken:
Date Rec'd: 05/02/2018

CHROMOSOME IMBALANCE DETECTED

arr[GRCh37] 1q31.3(196797104_196883398)x1,7p22.3(1087017_1129423)x1, Xp21.1(31735738_31972541)x1,Xp21.1(31972542_32011209)x0,Xp21.1(32011210_32102619)x1

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 four regions of imbalance:
i) deletion of approximately 86.3kb from band q31.3 in the long arm of chromosome 1, between base pair coordinates 196,797,104 and 196,883,398. This finding represents a known benign copy number variant.
ii) deletion of approximately 42.4kb from band p22.3 in the short arm of chromosome 7, between base pair coordinates 1,087,017 and 1,129,423. This finding is a rare variant, the functional significance of which is uncertain.
iii) deletion of approximately 0.367Mb from band p21.1 in the short arm of the X chromosome, between base pair coordinates 31,735,738 and 32,102,619, within which is
iv) an additional deletion of approximately 38.7kb between base pair coordinates 31,972,542 and 32,011,209. This is likely to represent a deletion on the second allele, which therefore results in nullisomy for this region. Both imbalance **iii)** and imbalance **iv)** are located within the *DMD* gene and encompass exonic regions.

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**: 28/02/2018**