Postnatal Chromosomal Microarray Report

Accreditation No.4033/50

Name: HN: - Gender: Female Tube ID:

DOB: Age: 35 Y Specimen: Other (Products of conception)

Collected Date: Received Date: Reported Date:

Requested by: Clinic/Ward/Hospital:

Clinical Information/Diagnosis: Pregnancy 10 wk with dead embryo in utero

Result: Female with trisomy 16

Interpretation	Change	Chromosome	Cytobands	Position (Start-Stop)	Size (Mbp)
Pathogenic	Gain	16	p13.3 (pter) - q24.3 (qter)	85,881 - 90,155,062	90.07

Sex chromosome complement: XX (Female)

Nomenclature (ISCN 2020): arr[GRCh37] 16p13.3q24.3(85881_90155062)x3 or arr(16)x3

Interpretation: Chromosomal microarray analysis detected a single copy gain of chromosome 16 in the specimen from this patient.

Recommendations: Genetic counseling

Method: Chromosomal microarray analysis was performed using CytoScan™ 750K microarray platform (Applied biosystem™, USA). This microarray consists of 750,436 oligonucleotide probes across the genome including 550,000 unique non polymorphic probes and 200,436 single-nucleotide polymorphism (SNP) probes with 4.0 kb overall median probe spacing were throughout the genome and with 1 kb in ISCA (International Standards for Cytogenomic Arrays) regions. DNAs were determined and results were analyzed using Chromosome Analysis Suite Software Version 4.4.0.63 (Applied Biosystems, USA). All data was analyzed and reported using the February 2009 NCBI Build 37.1 (hg19). Some copy number changes may not be reported if they are interpreted as clinically neutral. Duplications < 500 kb and deletions < 200 kb may not be reported if there is insufficient published information on gene content at the time of analysis. Regions with interstitial region of homozygosity (ROH) larger than 10.00 Mb and regions with terminal ROH larger than 5.00 Mb are

Reported by	Rechecked by	Approved by

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reported. The results were interpreted using the Database of Genomic Variants (DGV), The University of California Santa Cruz (UCSC) Genome Browser, Online Mendelian Inheritance in Man (OMIM) and additional available databases.

Limitations

Limitation of this assay includes the inability to detect balanced chromosome abnormalities (e. g. reciprocal translocations, Robertsonian translocations, inversions, balanced insertions), point mutations, copy number changes below the resolution of the array, low level mosaicism and complete uniparental heterodisomy. Normal microarray result do not rule out possibility of genetic disorder or syndrome or clinical implications due to an etiology not evaluated by this test.

References

- McGowan-Jordan J, Hastings RJ, Moore S,. ISCN 2020: An International System for Human Cytogenomic Nomenclature; Karger: Basel, Switzerland; Karger: Hartford, CT, USA, 2020.
- Riggs ER, Andersen EF, Cherry AM, Kantarci S, Kearney H, Patel A, Raca G, Ritter DI, South ST, Thorland EC, et al. Technical standards for the interpretation and reporting of constitutional copy-number variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics (ACMG) and the Clinical Genome Resource (ClinGen). Genet Med. 2020;22(2):245–57.

Laboratory note

This test was developed and its performance characteristics determined by Human Genetic Laboratory, Department of Pathology, Faculty of Medicine Ramathibodi Hospital.

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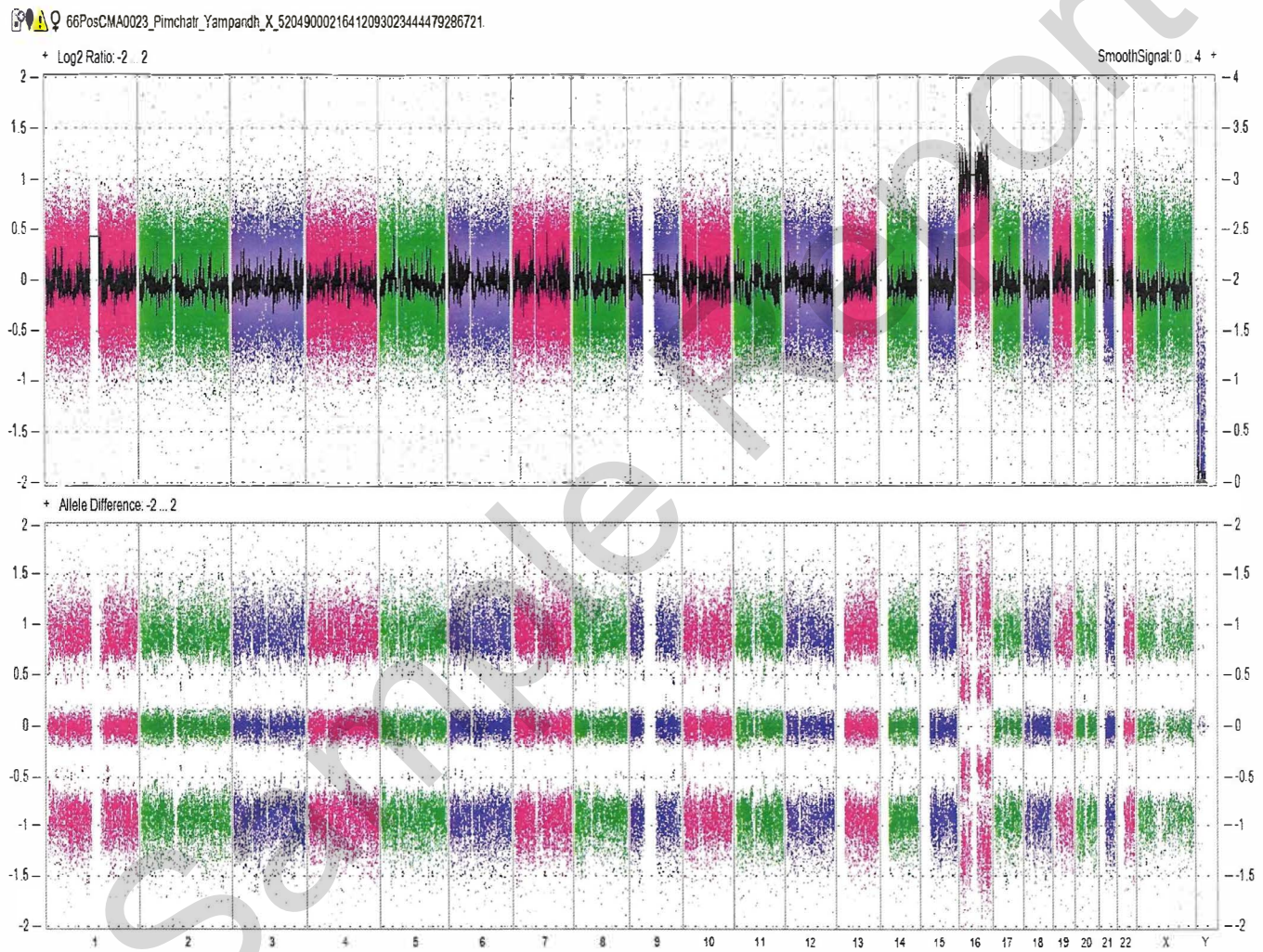
Whole genome view



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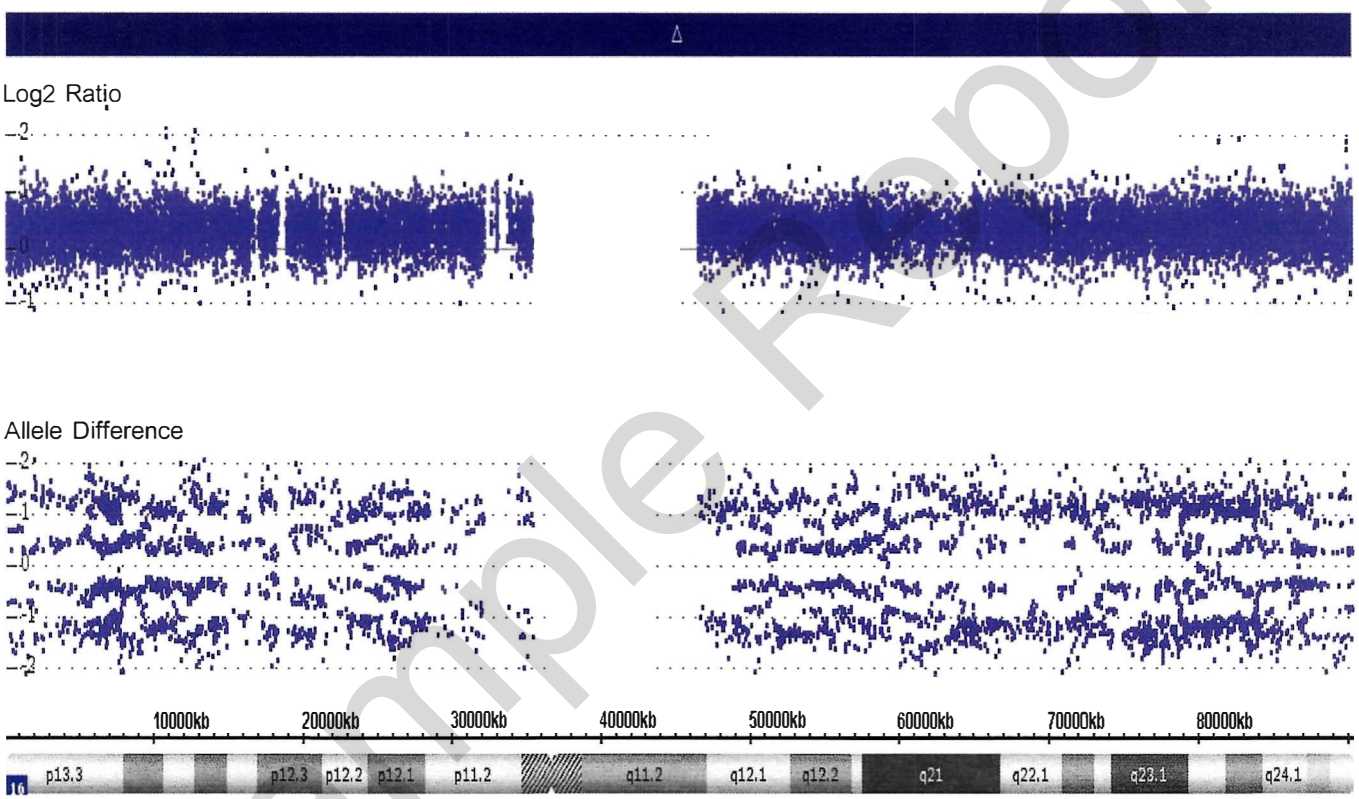
Result: Trisomy 16



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