Chromosomal Microarray (CMA) Report

Personal Information	Specimen Information	Test Information
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Relation: -	**Medical record No:**	**Ordering physician:** Dr.Smith
Sex/Birth: M / 2014-01-03	**Date received:** 2023-03-09	**Institution:** Hospital A

TEST PERFORMED

Chromosomal Microarray (CMA)

REASON FOR REFERRAL

Autism

RESULT

Inconclusive

ISCN	Type	Size	Classification
arr 6q16.3(103,520,024-104,231,738) X 3	Gain	711 kb	B
arr 7q21.3q31.31(97,284,382-121,033,191) X2 hmz	LOH	23.7 Mb	VOUS
arr 7q34q36.2(138,571,709-152,648,941)X2 hmz	LOH	14.1 Mb	VOUS
arr 8q24.13q24.22(124,129,205-136,000,923)X2 hmz	LOH	11.9 Mb	VOUS
arr 11p12q14.3(36,857,170-92,667,730)x2 hmz	LOH	55.8 Mb	VOUS
arr 20p13p12.3(61,794-7,742,326)x2 hmz	LOH	7.7 Mb	VOUS

Abbreviations: P (pathogenic), LP (likely pathogenic), VOUS (variant of uncertain significance), LB (likely benign), B (benign)
ISCN: International System for Human Cytogenomic Nomenclature

INTERPRETATION

In this chromosomal microarray test, a heterozygous duplication in 6q16.3 and several large regions of homozygousity were detected in 7q21.3q31.31, 7q34q36.2, 8q24.13q24.22, 11p12q14.3 and 20p13p12.3.

A heterozygous duplication in 6q16.3 is considered as a benign CNV due to absence of protein coding genes and lack of pathogenic cases in ClinGen and Decipher.
Several large regions of loss of heterozygosity (LOH) encompassing about 7.4% of the autosome are not diagnostic of a specific condition. However it raises the possibility of uniparental disomy (UPD) disorder such as UPD7, UPD11, and UPD20 and a recessive disorder with a causative gene located within one of these LOH regions. Additionally, these results could indicate a familial relationship between this individual's parents. A genetics consultation is recommended.

Clinical correlation is recommended and methylation study such as MS-MLPA is recommended to confirm or rule out the conditions such as UPD disorders and sequencing analysis such as DES or WES is recommended to diagnose mendelian genetic disorders.

*The quality control metrics of DNA and data were all passed compared with the thresholds of internal quality control.

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METHODS	
Test Chip	CytoScan DX Assays
Reference genome	GRCh37/hg19
ISCN	ISCN2020

TEST DESCRIPTIONS

Chromosomal microarray test (CytoScan Dx) can detect copy number variation (CNV) (deletion >25kb, duplication >50kb) and loss of heterozygosity (LOH) (>3Mb) across the entire genome. We report all CNVs of >400kb and clinical significant CNVs with any sizes. And we also consider to report LOH associated with clinical significant situations such as uniparental disomy (UPD) or consanguinity.

REFERENCES

1. DGV (Database of Genomic Variants)
2. DECIPHER (DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources)
3. ClinGen (Clinical Genome Resource)
4. G-CMA Database (GC Genome In-house CMA Database with ~5,000 Cases)

LIMITATIONS

1. Low levels of mosaicism, balanced translocation, inversion, and point mutations cannot be detected.
2. If the LOH area is small or is a heterodisomic UPD, UPD may not be detected on the test principle.
3. Undetectable CNV area: 13p, 14p, 15p, 21p, 22p, Yq11.23, Yq12 and pericentric heterochromatin regions of all chromosomes

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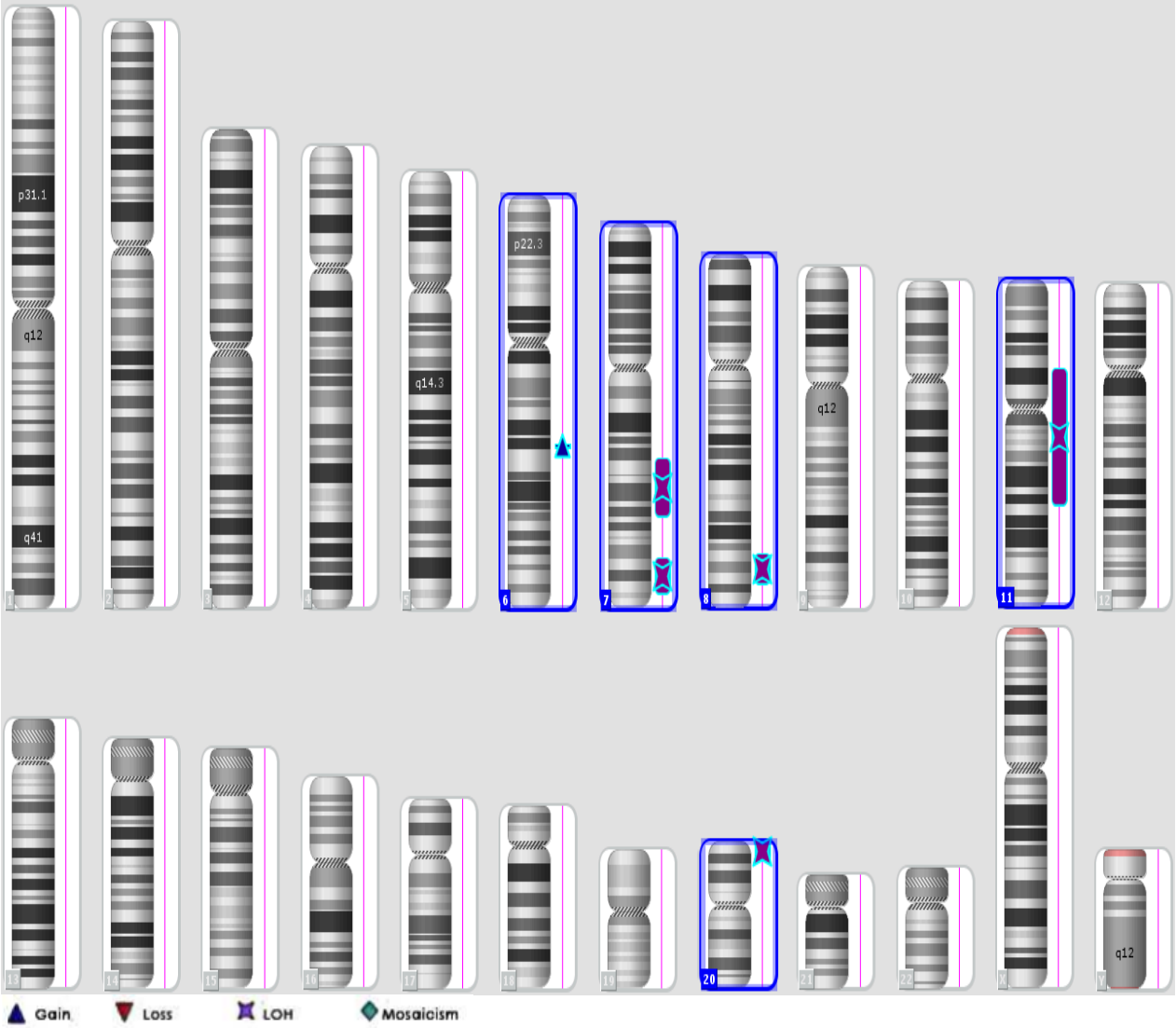

The Centromic region of each chromosome is polymorphic, so it can be marked gain or loss.

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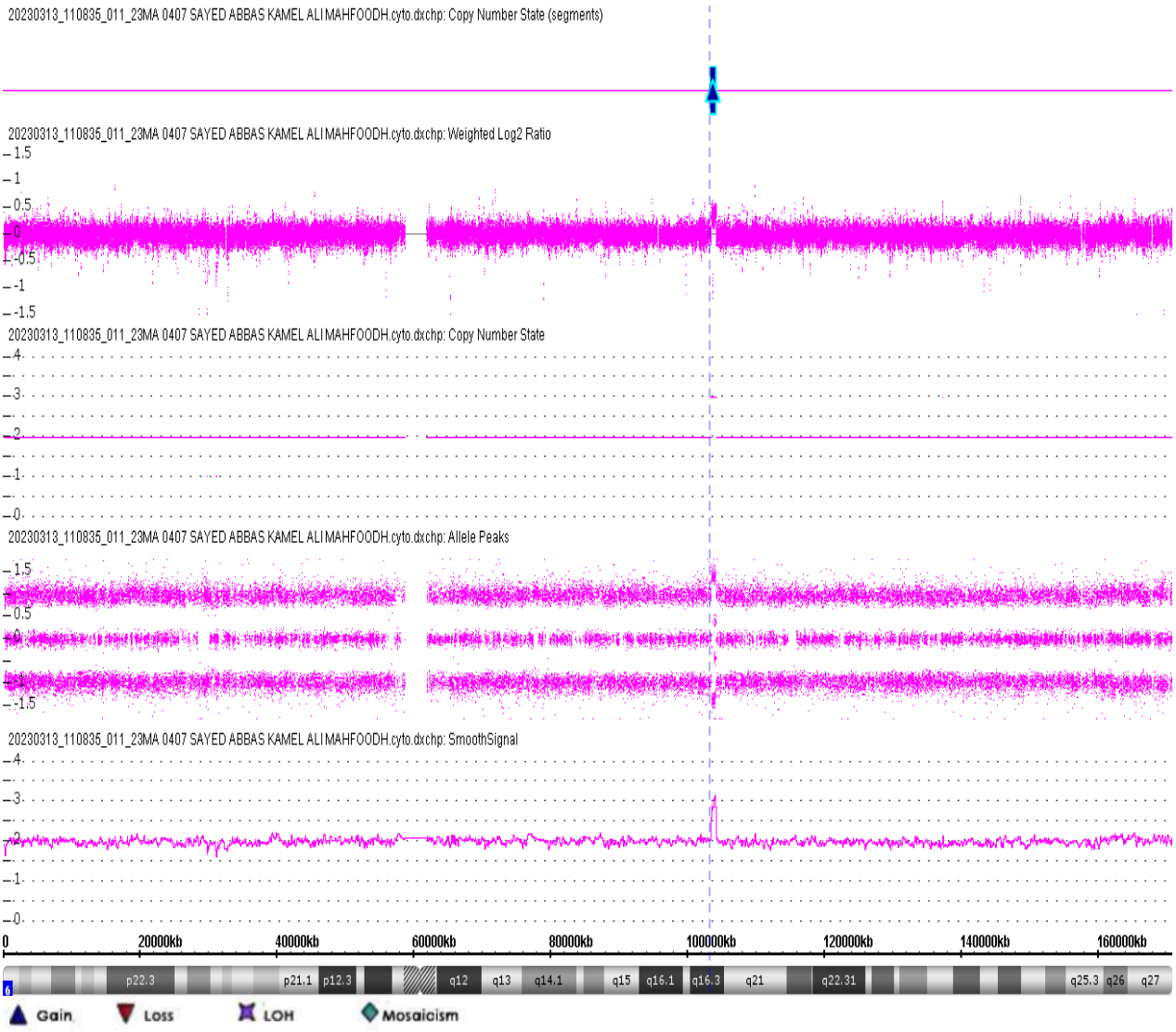

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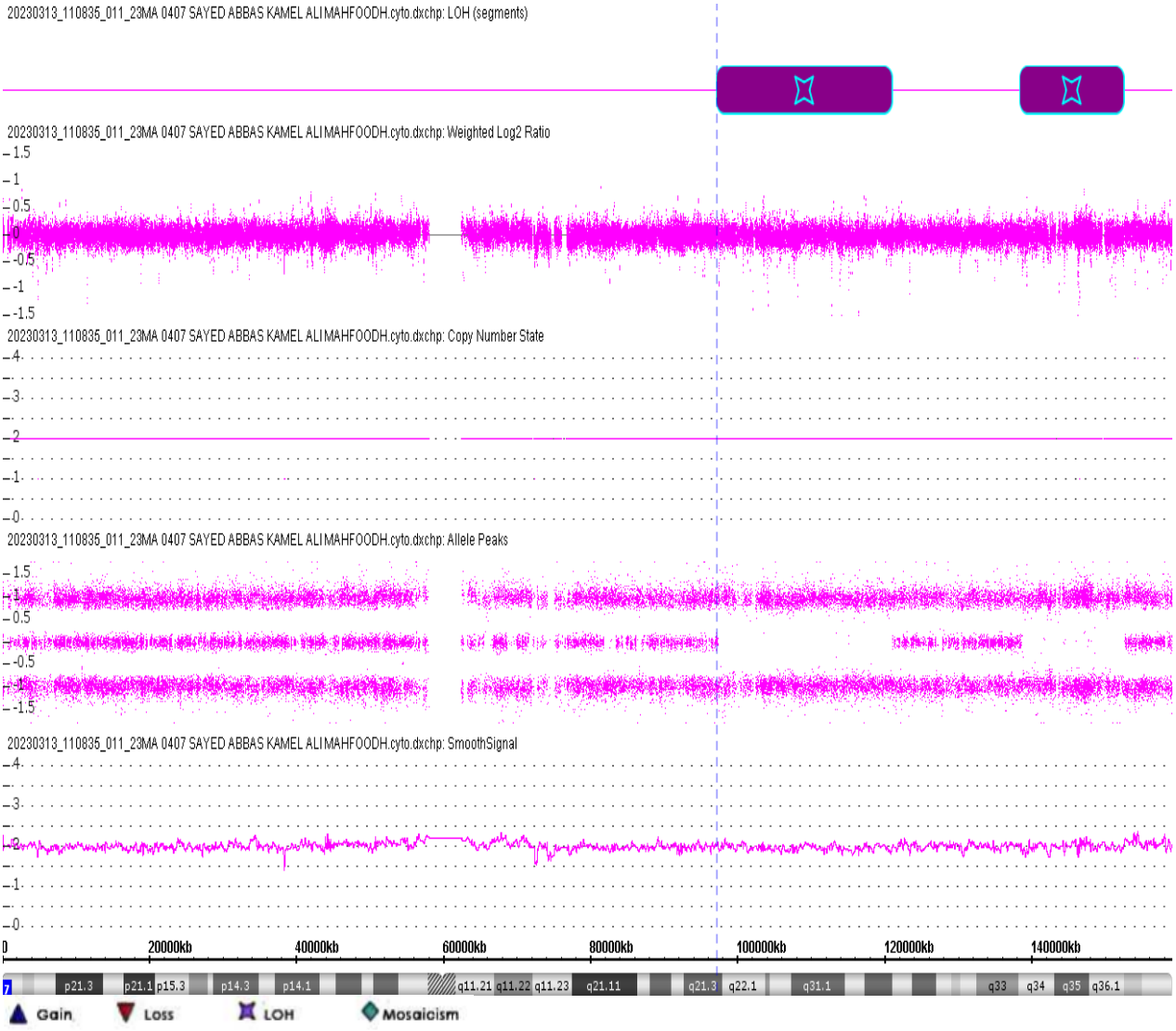

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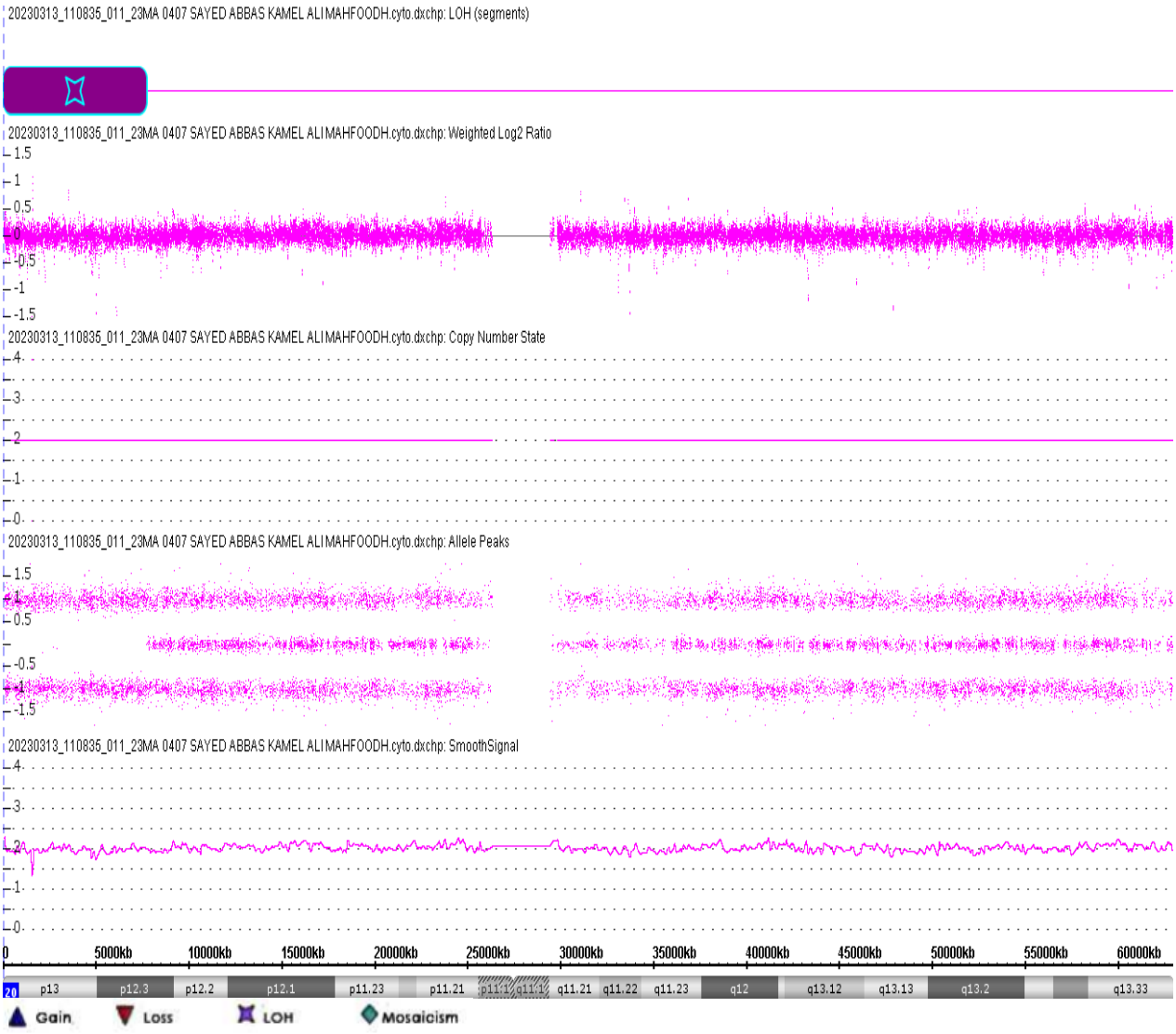

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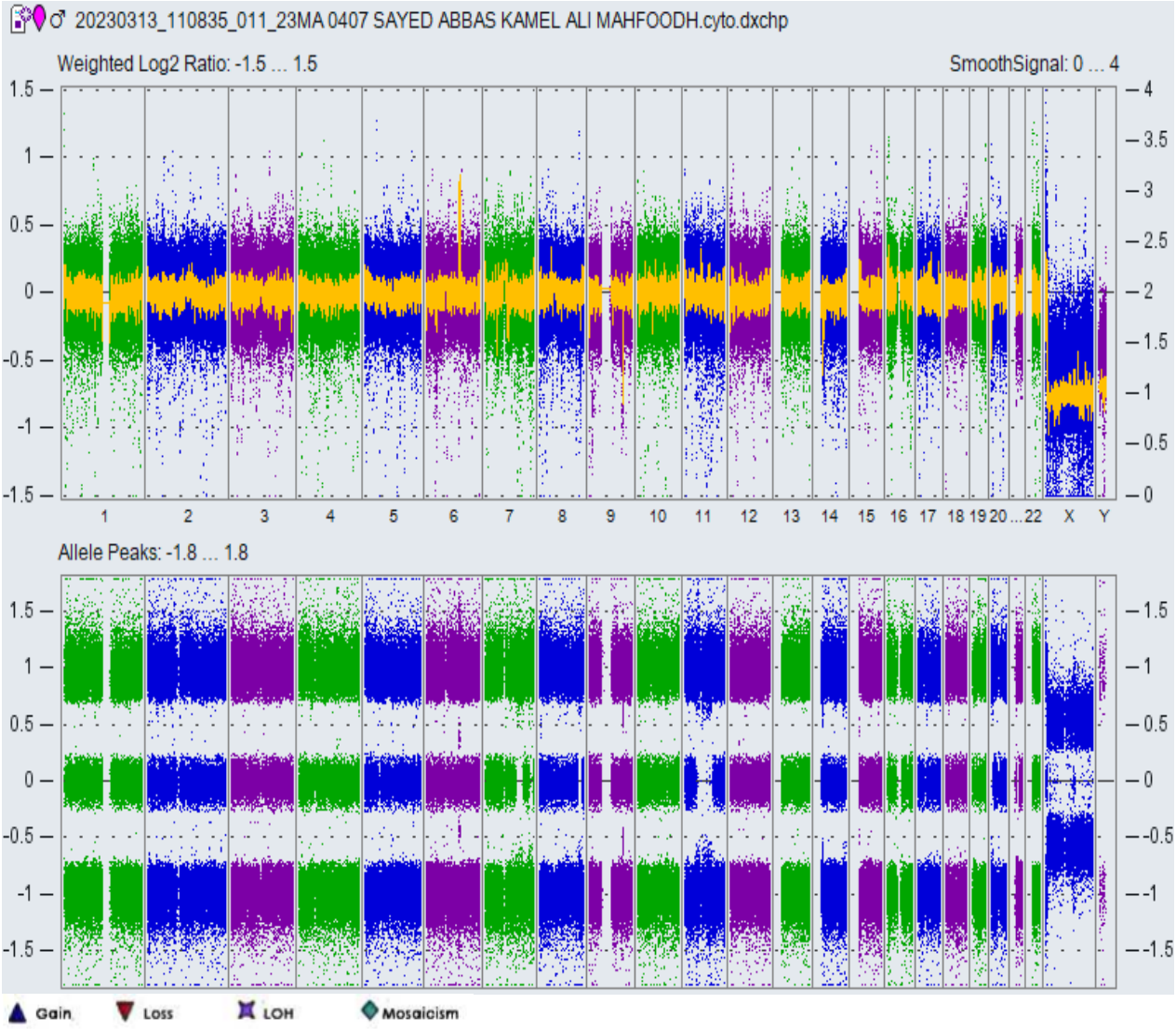

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