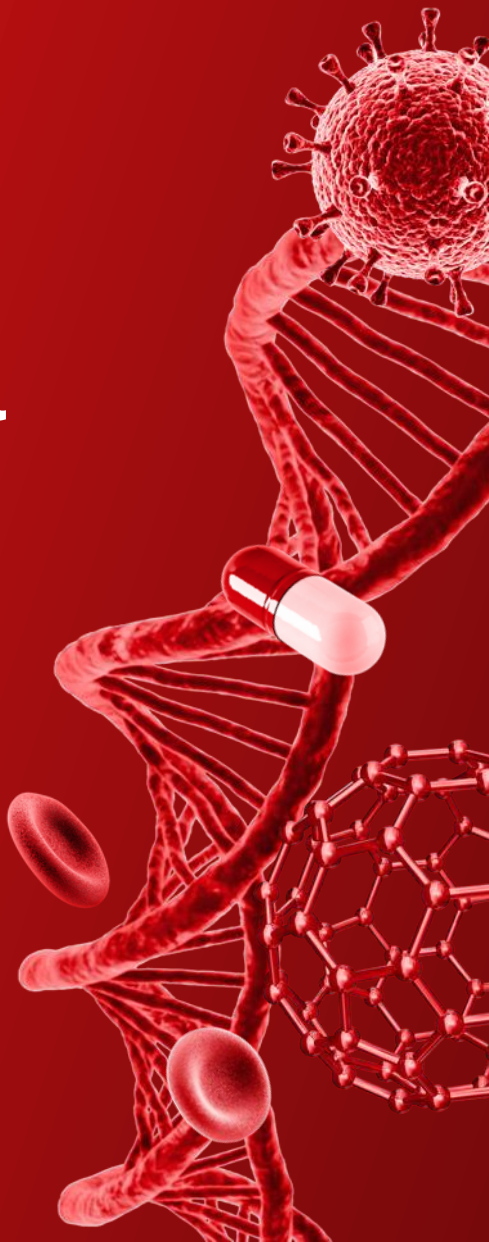
CytoScan™ HD Accel Sample Data

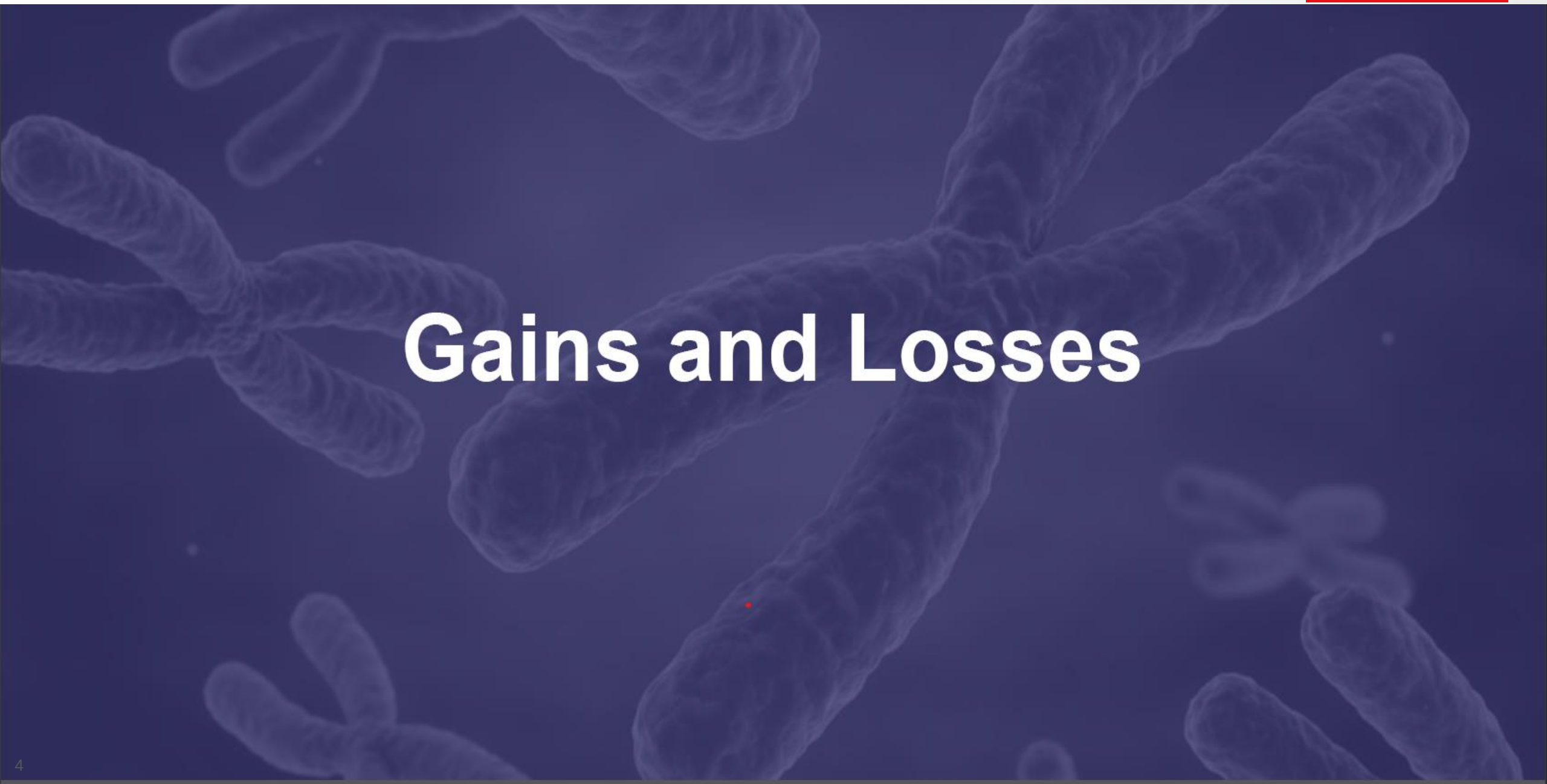


Aberrations found in Sample Data Set

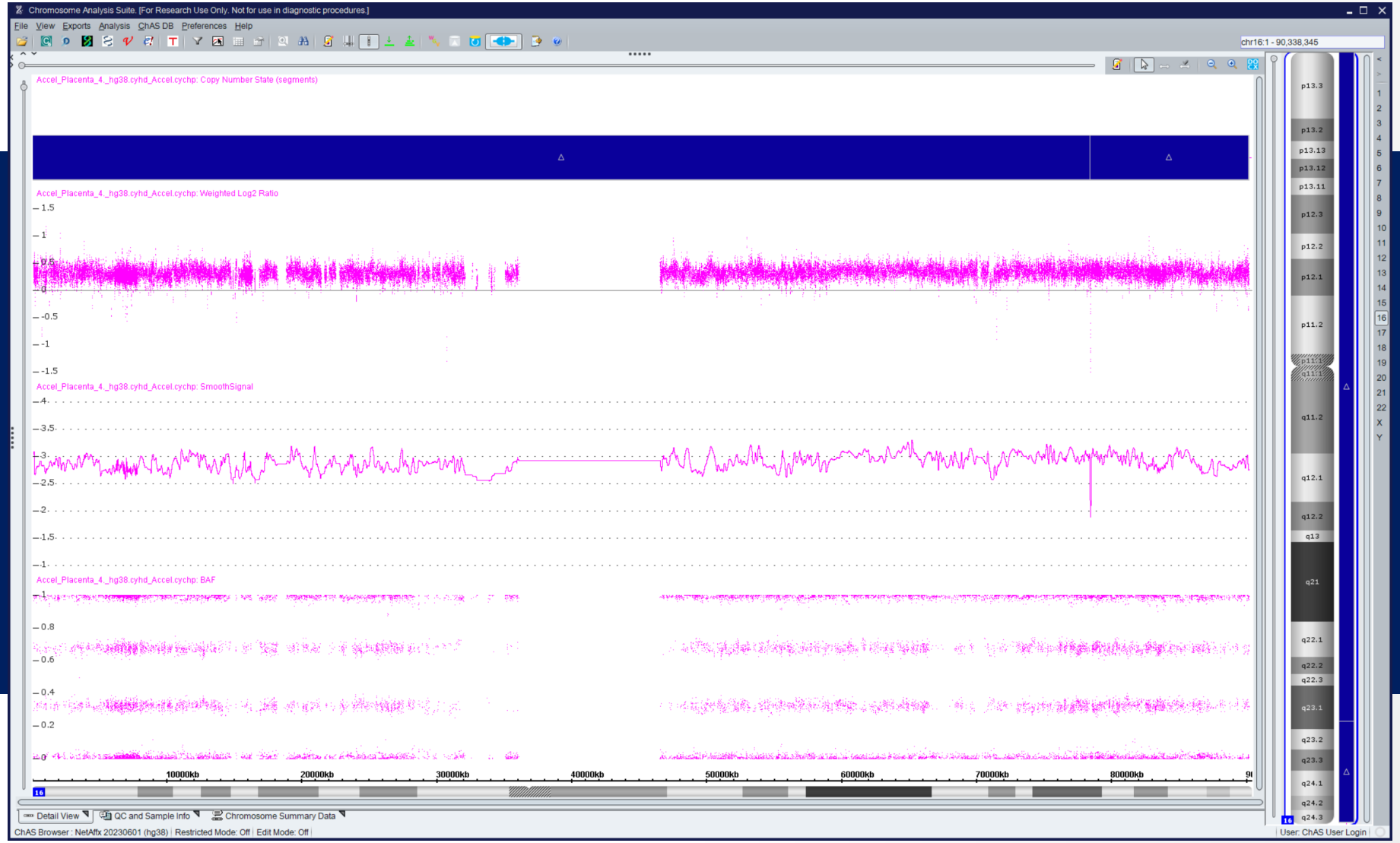
Sample Name	Sample Source	Detailed Aberrations	Additional Aberrations	Notes
Accel_Whole Blood_1	Whole Blood - EDTA	Copy Neutral Mosaic AOH of chr 14		
Accel_Whole Blood_3	Whole Blood - EDTA	22q12.2 x4	16p13.2 x3 21q22.3 x4	
Accel_Whole Blood_4	Whole Blood - Heparin	15q13.2 x1 (CHRNA7 del)		
Accel_Pacenta_3	Placenta	Mosaic loss of chr Y	X x1 Y x0-1	Sex of sample is unknown. ChAS assigns sex as female which generates mosaic gain segments on chr Y. The segments in the image are changed to mosaic losses to reflect the data track. The sample can be reanalyzed as a male.
Accel_Placenta_4	Placenta	Trisomy of chr 16	17q11.2 x3	
Accel_Cultured Amnio_3	Cultured Amnio	Numerous CN AOH	5q14.6 x1	
Accel_Bone Marrow_3	Bone Marrow - Heparin	Mosaic gain of chr 10 15q26.2 x1 and 15q26.2 x3 22q11.22 complex loss Xq25-q28 x2	5q21.1 x3 9p21.3 complex loss 12p13.2 x1 13q41.2 x1 chr 21 x2-3	Complex loss is observed by turning off marker count and size, keeping default settings for Smoothing and Joining, and turning on “Skip smoothing/joining for copy number less than 1”.
Accel_Genomic DNA Control_3	Cell Line	N/A		Example of Genomic Control DNA

A set of different samples analyzed with the Applied Biosystems™ CytoScan™ Cytogenetics Suite is provided for educational proposes.

Sample display color might be different in this presentation when compared to your software.


Gains and Losses

Trisomy chr 16



The Placenta_4 sample illustrates a segment with a chr 16 trisomy.

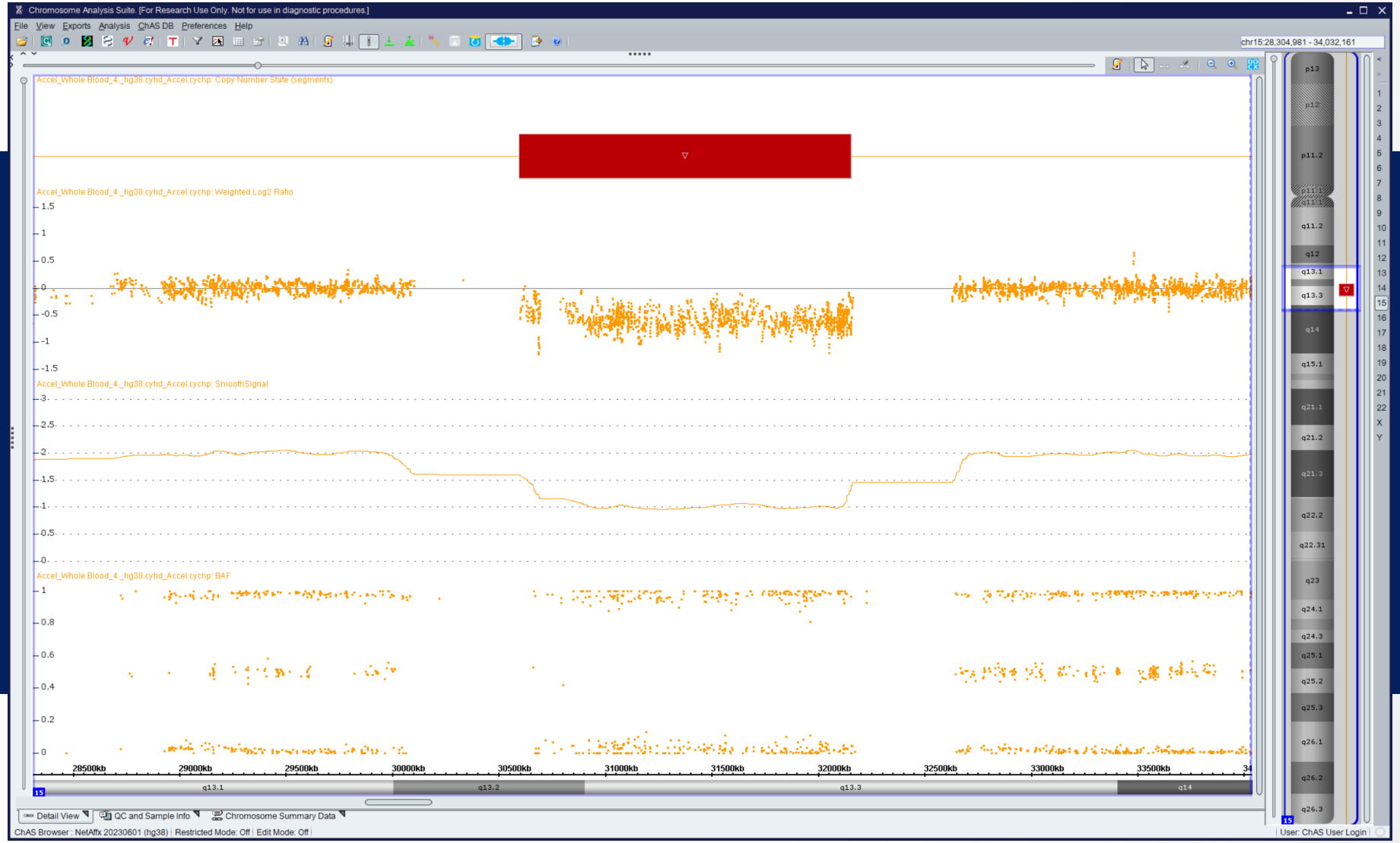
The data tracks support CN=3.

Copy number gain of 4

The Whole Blood_3 sample illustrates a small segment with CN = 4.

The segment is 56 kb.

Hemizygous loss



The Whole Blood_4 sample illustrates a segment with CN = 1.

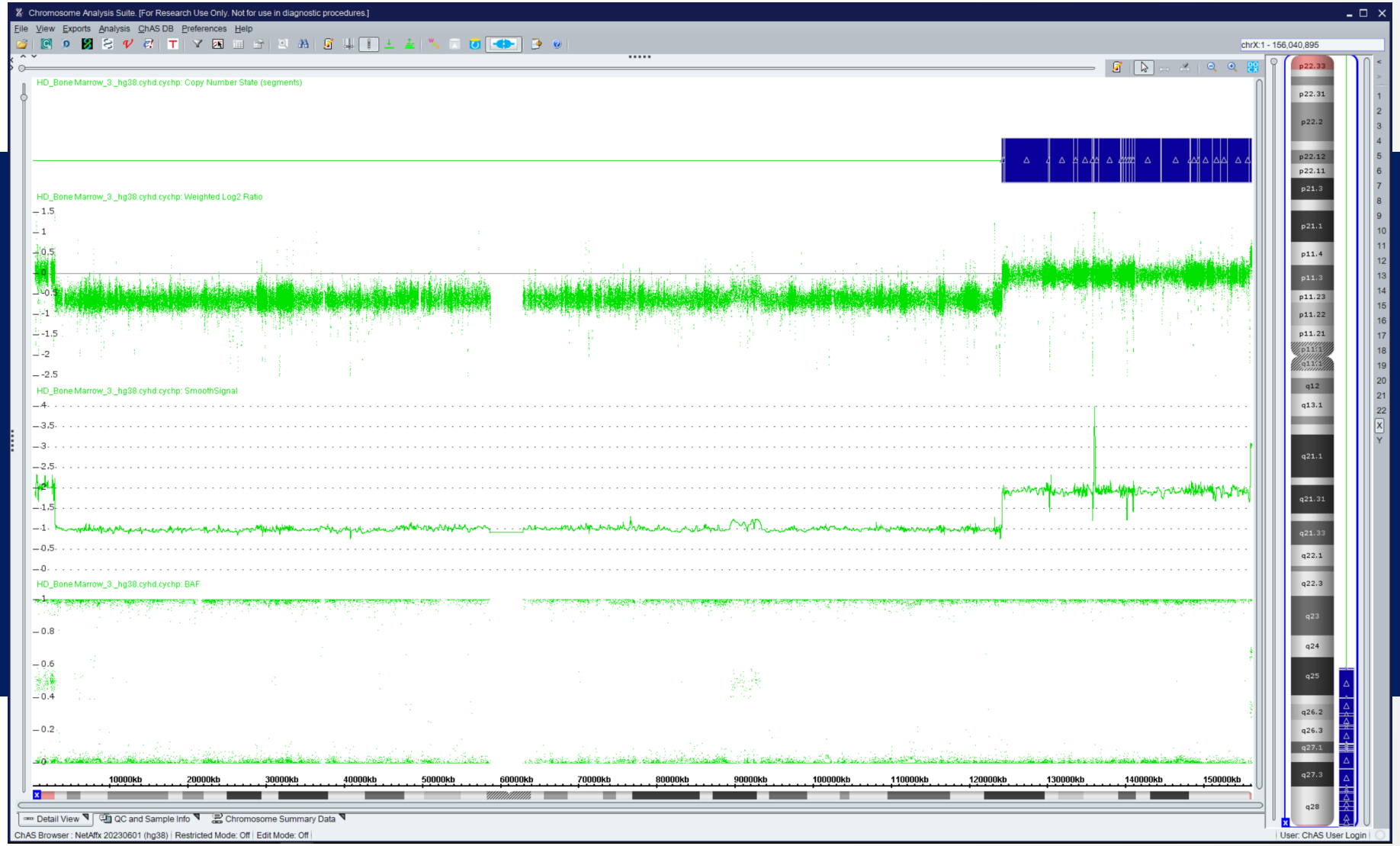
The segment is 1,559 kb.

Complex homologous loss

The Bone Marrow_3 sample illustrates a segment with a complex homologous loss.

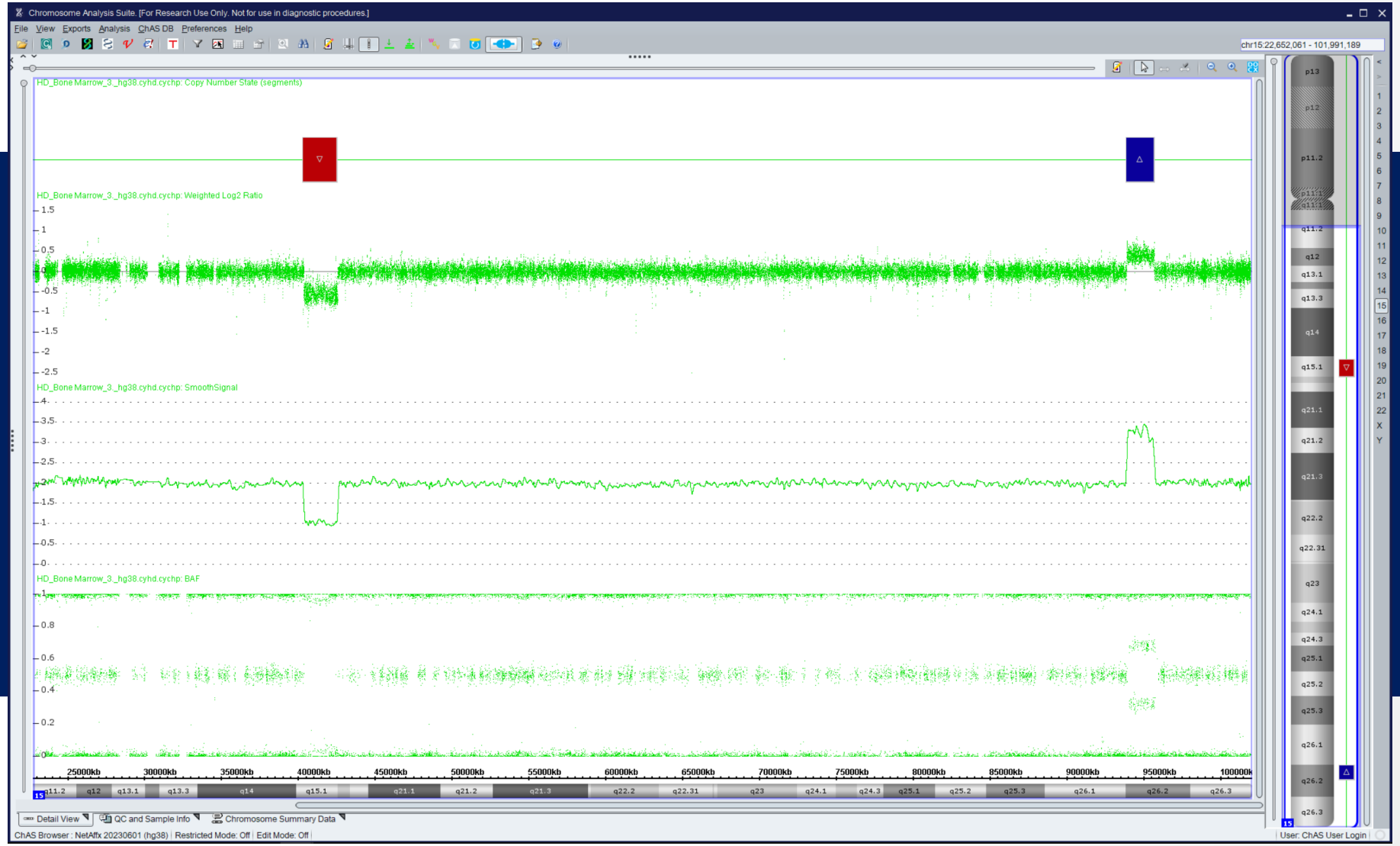
A CN=0 segment is nested between two CN=1 segments.

Gain on chr X



The Bone Marrow_3 sample shows gain on chr X.

Loss and gain on chr 15

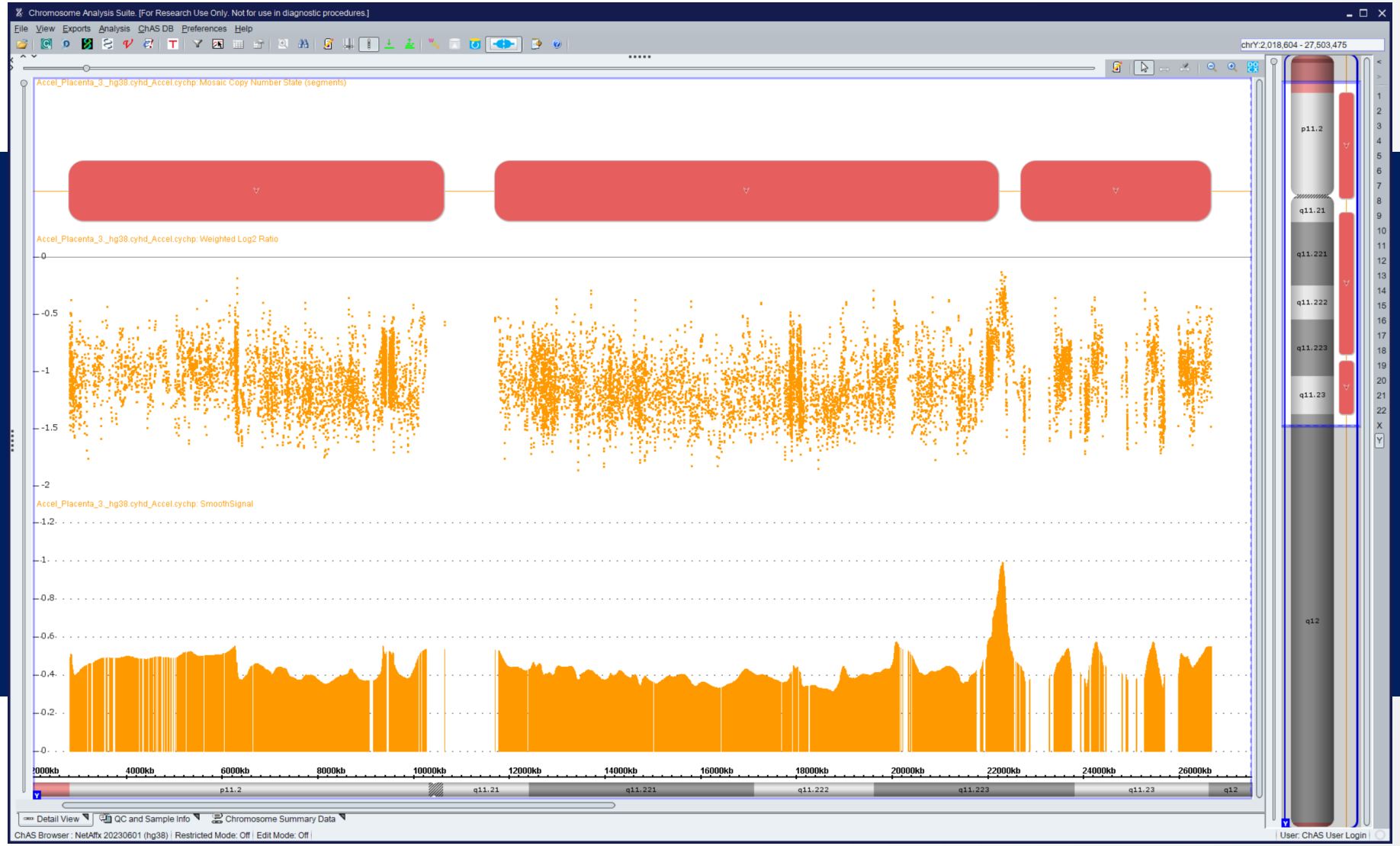


The Bone Marrow_3 sample shows a 2,201 kb loss and a 1,798 kb gain on chr 15.

The data track changes support the different aberration types.

Mosaic Segments

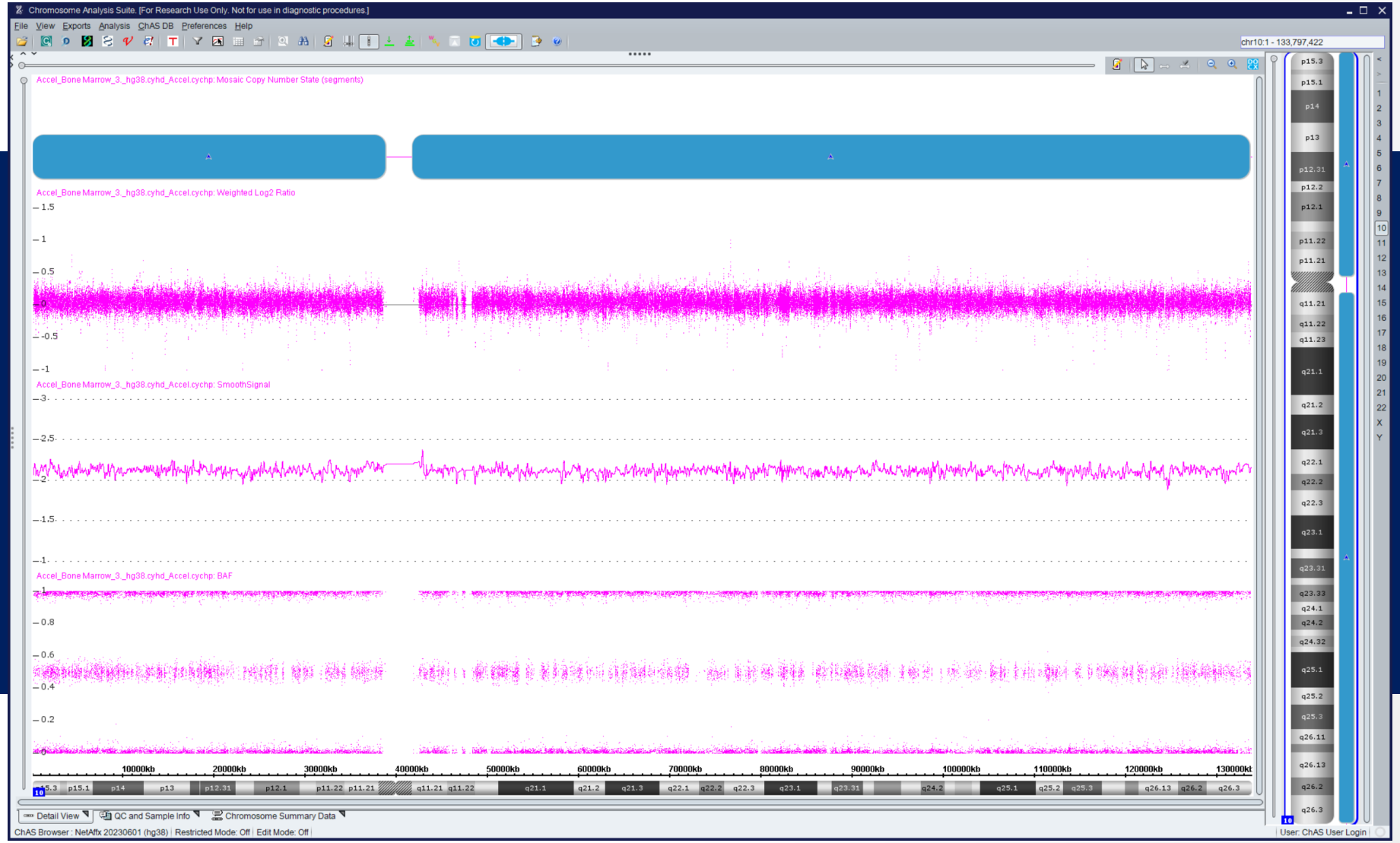
Mosaic loss of chr Y



The Placenta_3 sample shows a mosaic loss on chr Y.

The Smooth Signal track indicates CN = 0.42.

Mosaic gain of chr 10



The Bone Marrow_3 sample shows a low-level mosaic gain on chr 10.

The Smooth Signal track indicates CN = 2.11.

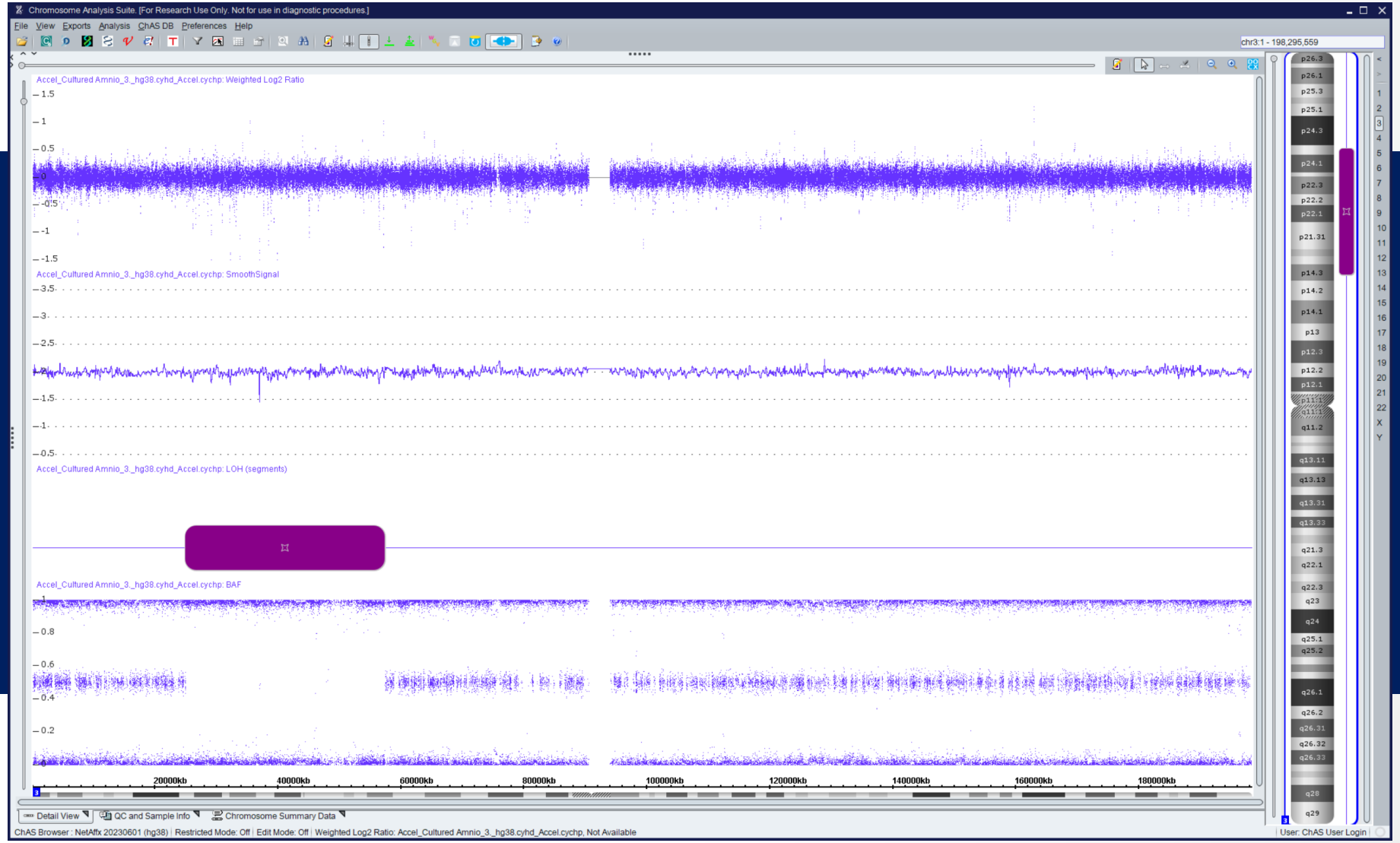
ROH/AOH/LOH Examples

Copy Neutral Absence of Heterozygosity

The Cultured Amnio_3 sample shows numerous 3 Mb AOH segments.

The Autosomal LOH is 6.28%.

Copy Neutral AOH on chr 3



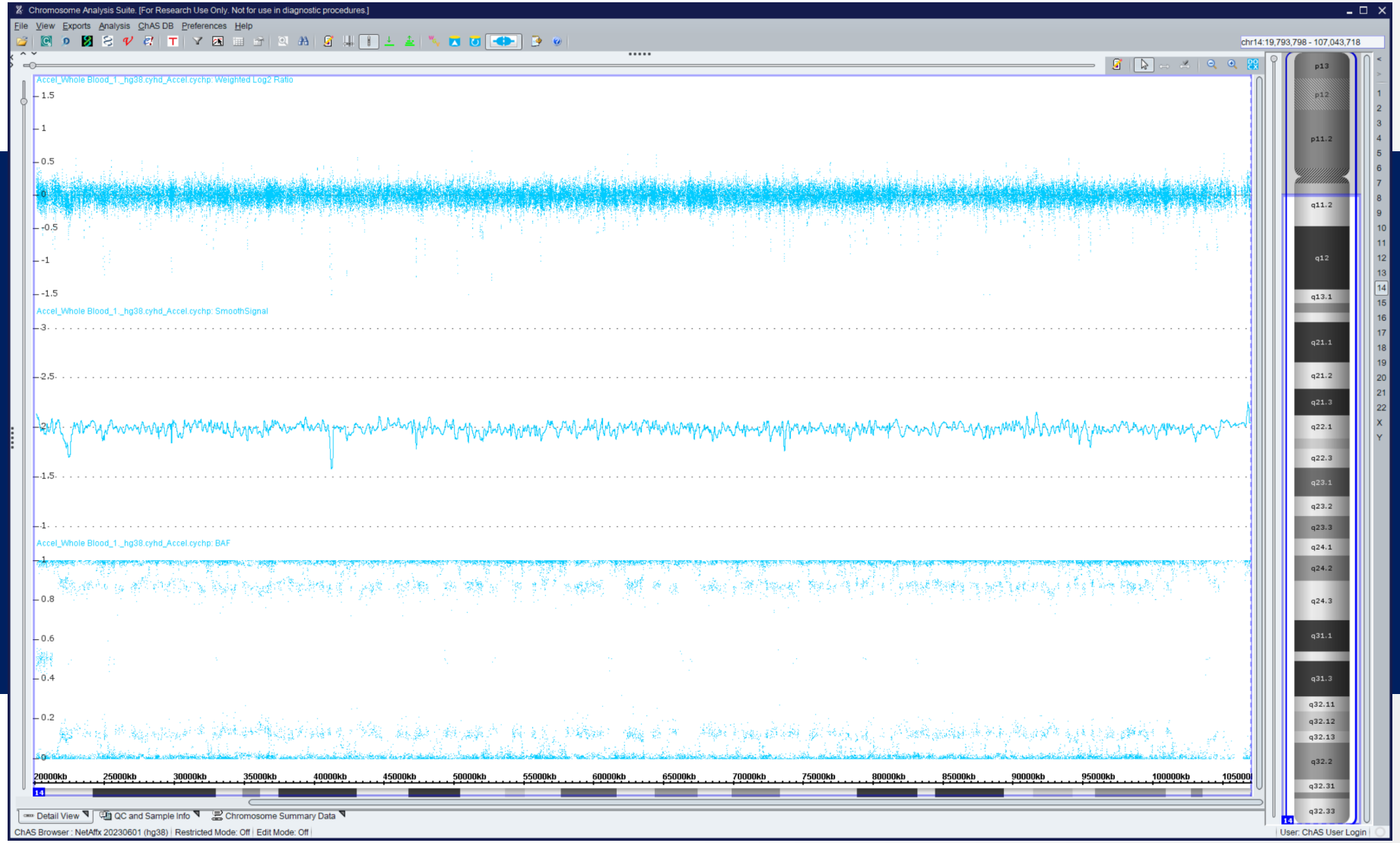
A focused view of chr 2 shows the Weighted Log2 Ratio = 0 and Smooth Signal =2. This indicates CN=2.

The BAF shows AOH.

Copy Neutral Mosaic AOH of chr 14

The Whole Blood_1 sample indicates a Copy Neutral Mosaic AOH on chr 14.

Copy Neutral Mosaic AOH of chr 14



A focused view of chr 14 shows the Weighted Log2 Ratio = 0 and Smooth Signal =2. This indicates CN=2.

The BAF shows mosaic AOH.

Thank you