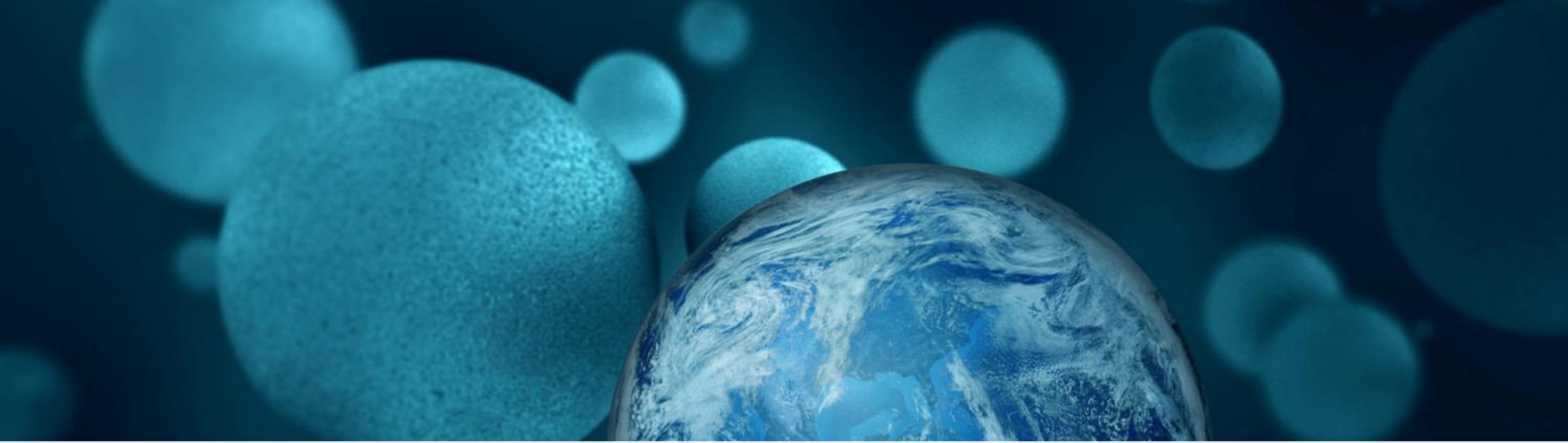


CytoScan HD Sample Data

2019-08-22 Release

The world leader in serving science

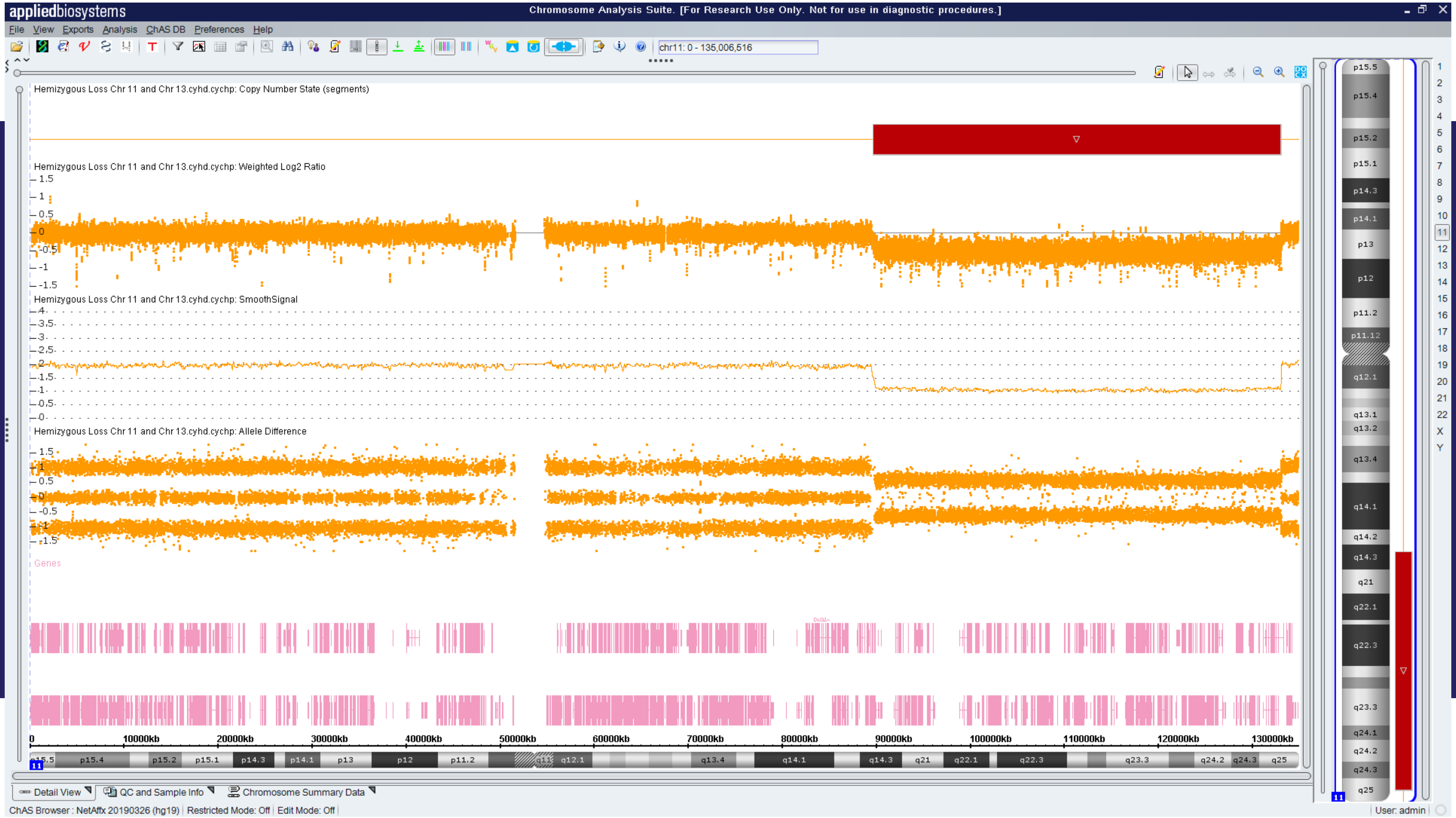
Summary

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.

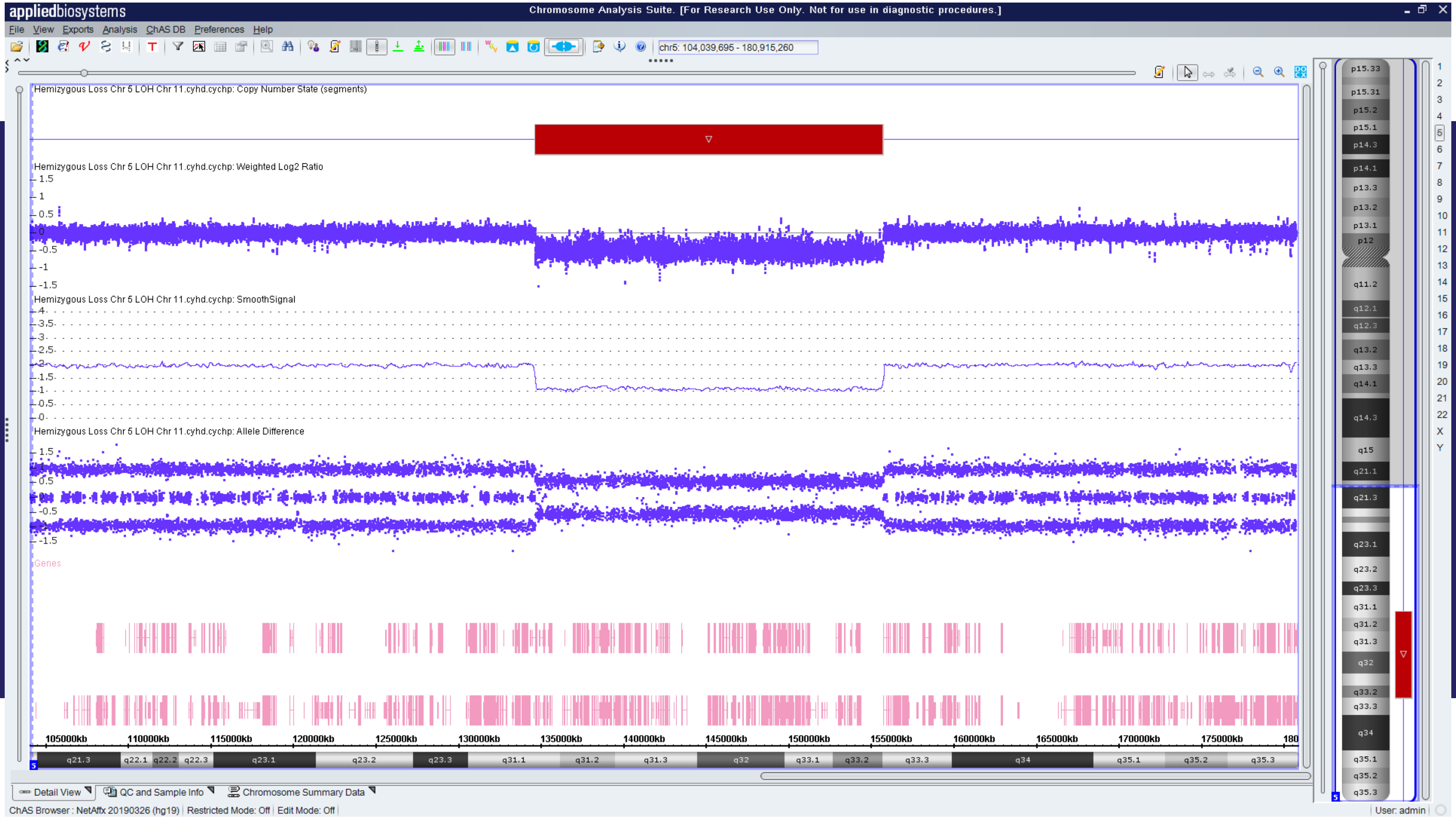
Gains and Losses

Hemizygous Loss on Chromosome 11



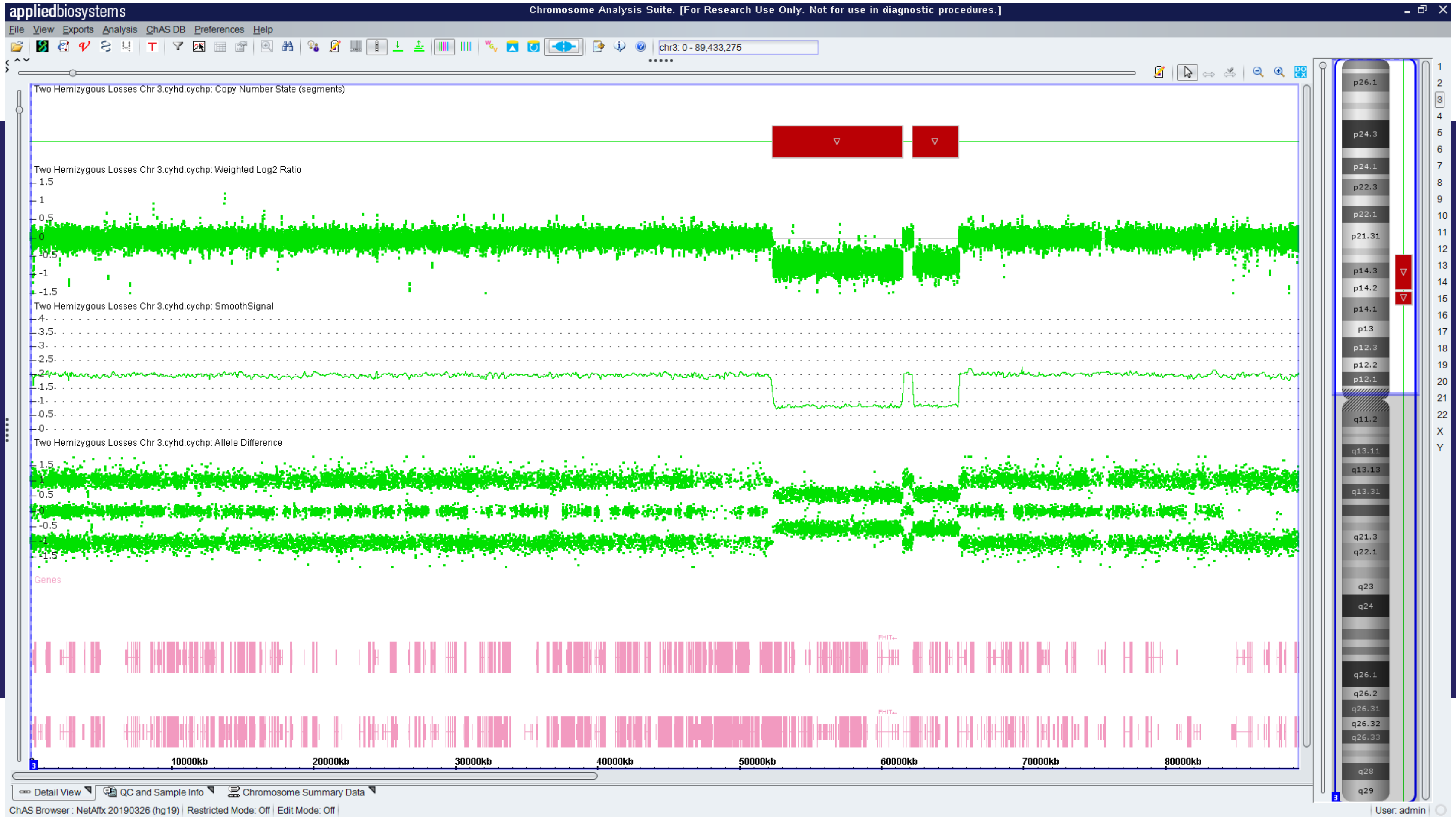
- This example illustrates of a hemizygous loss on chromosome 11.
- The allelic peaks track shows a pattern change from 3 to 2 bands, confirming the hemizygous loss.

Hemizygous Loss on Chromosome 5



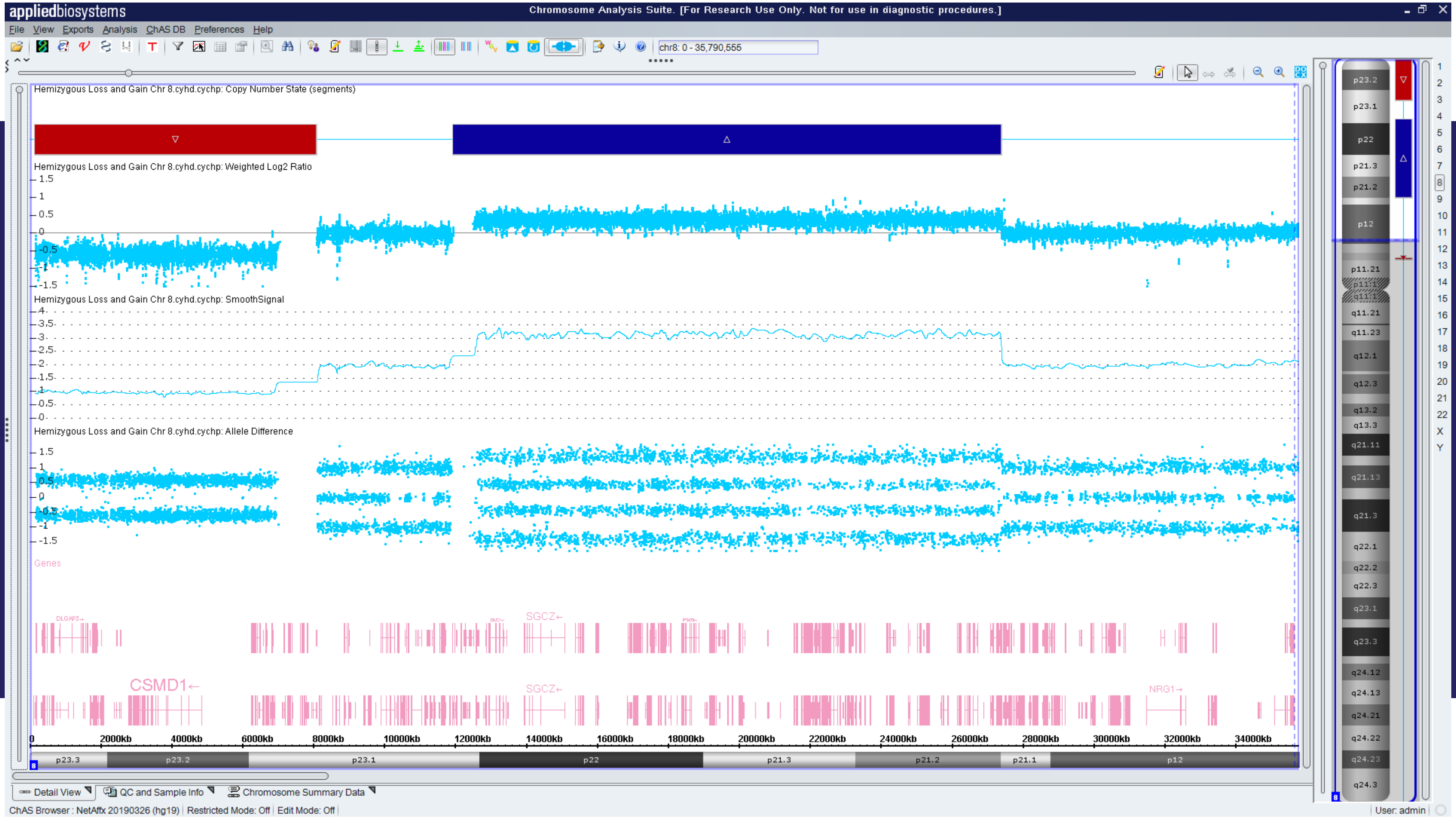
- Another example, this time an interstitial hemizygous loss on chromosome 5.

Two Hemizygous Losses



- This example illustrates of two hemizygous losses on chromosome 3.
- These losses and the neutral structural region in the middle were all confirmed by FISH.

Hemizygous Loss and Gain



Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

chr8: 0 - 35,790,555

Hemizygous Loss and Gain Chr 8.cyhd.cychp: Copy Number State (segments)

Hemizygous Loss and Gain Chr 8.cyhd.cychp: Weighted Log2 Ratio

Hemizygous Loss and Gain Chr 8.cyhd.cychp: SmoothSignal

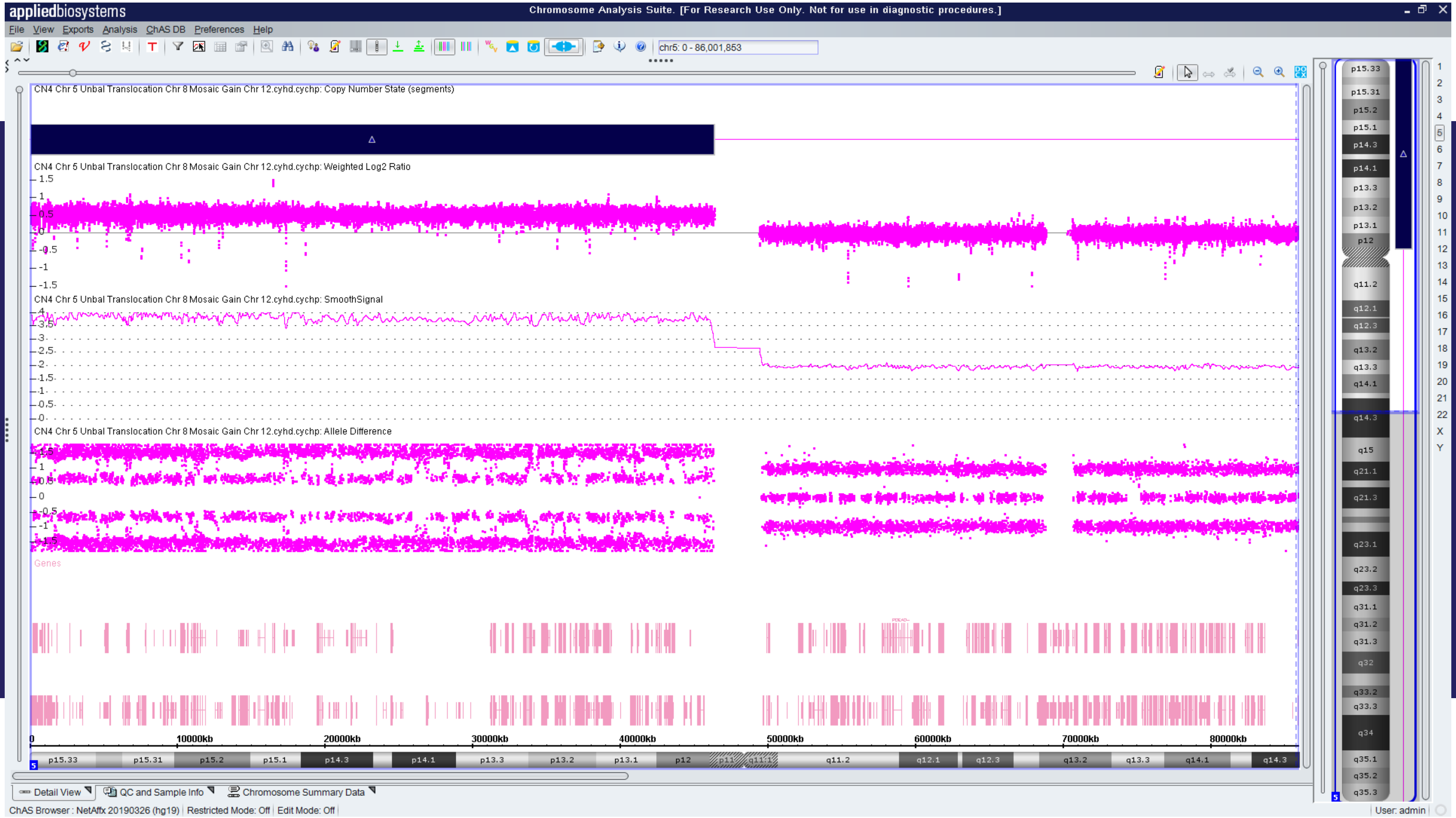
Hemizygous Loss and Gain Chr 8.cyhd.cychp: Allele Difference

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off

User: admin

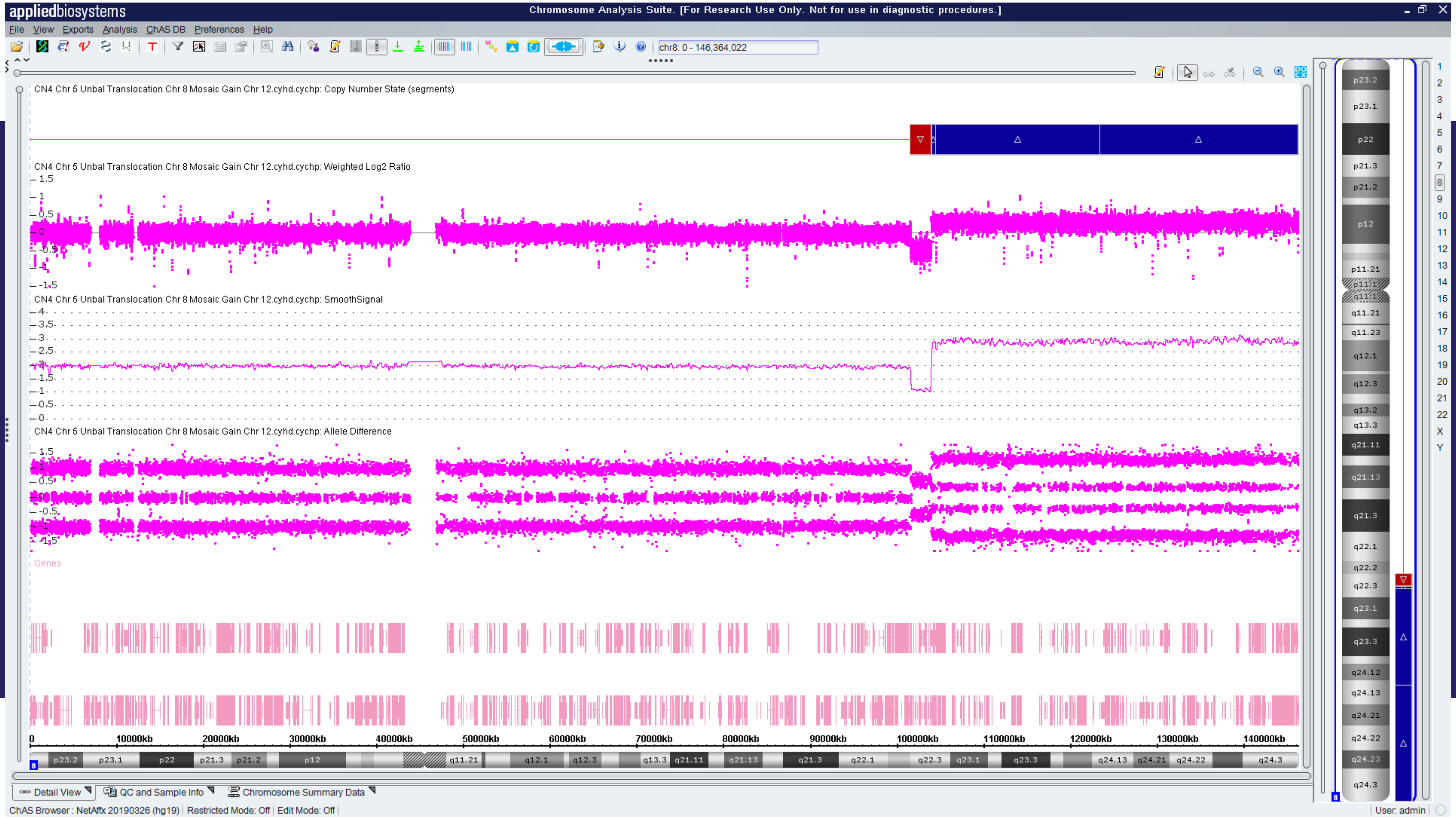
- This example illustrates copy numbers of 1, 2, and 3 on chromosome 8.
- These copy number changes were all confirmed by FISH.

Copy Number 4 Gain



- This example illustrates a segment with a copy number of 4.
- The allelic peaks track shows that one allele has been triplicated and the other is a single copy.

Unbalanced Translocation



- This example shows a hemizygous loss followed by a big gain segment. This pattern is usually shown in unbalanced translocations.

Fresh-Frozen Solid Tumor with Complex Chromosomal Aberrations

Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

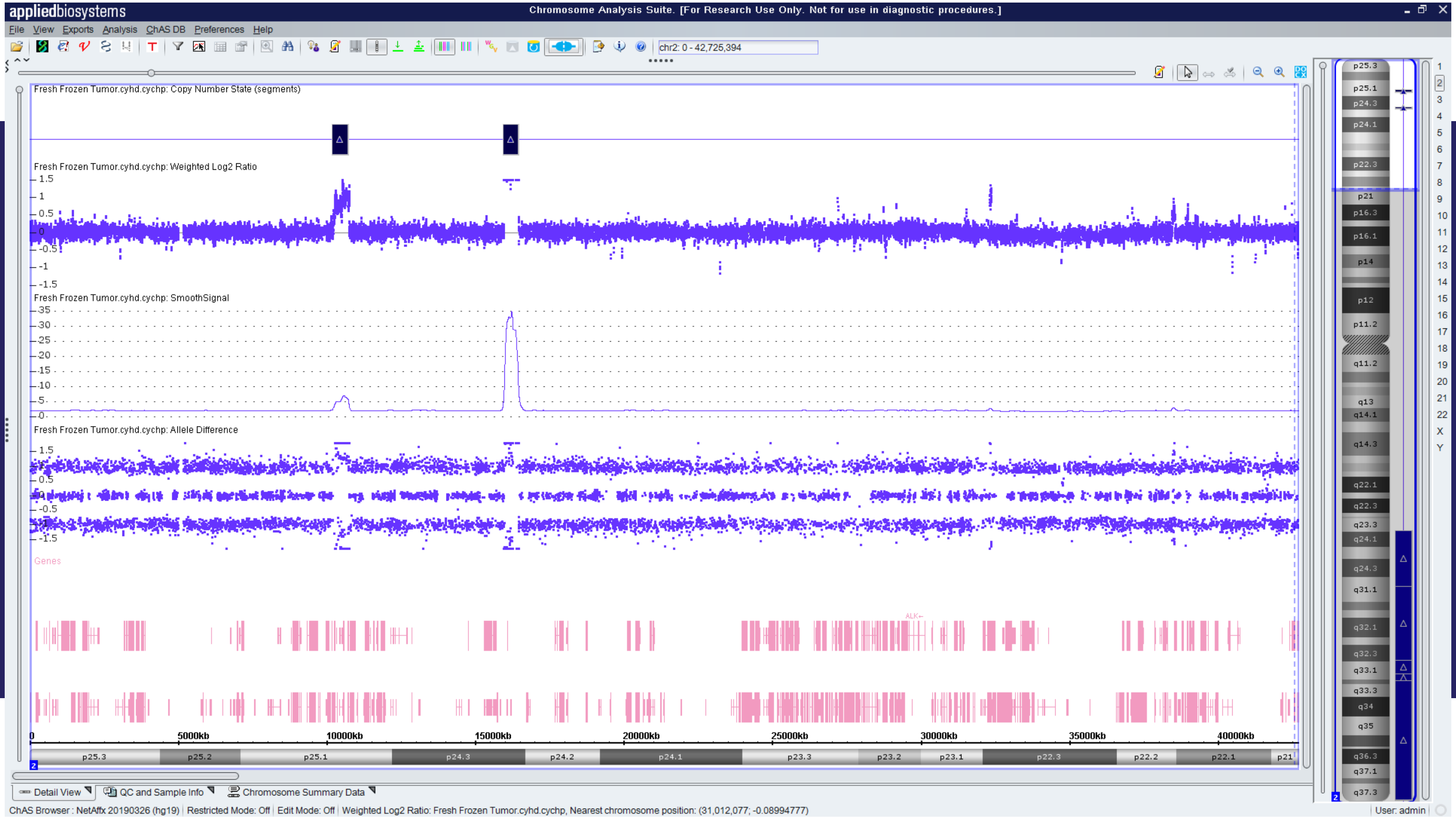
chr17: 0 - 81,195,210

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

User: admin |

- This sample shows high complexity with several types and sizes of chromosomal aberrations.

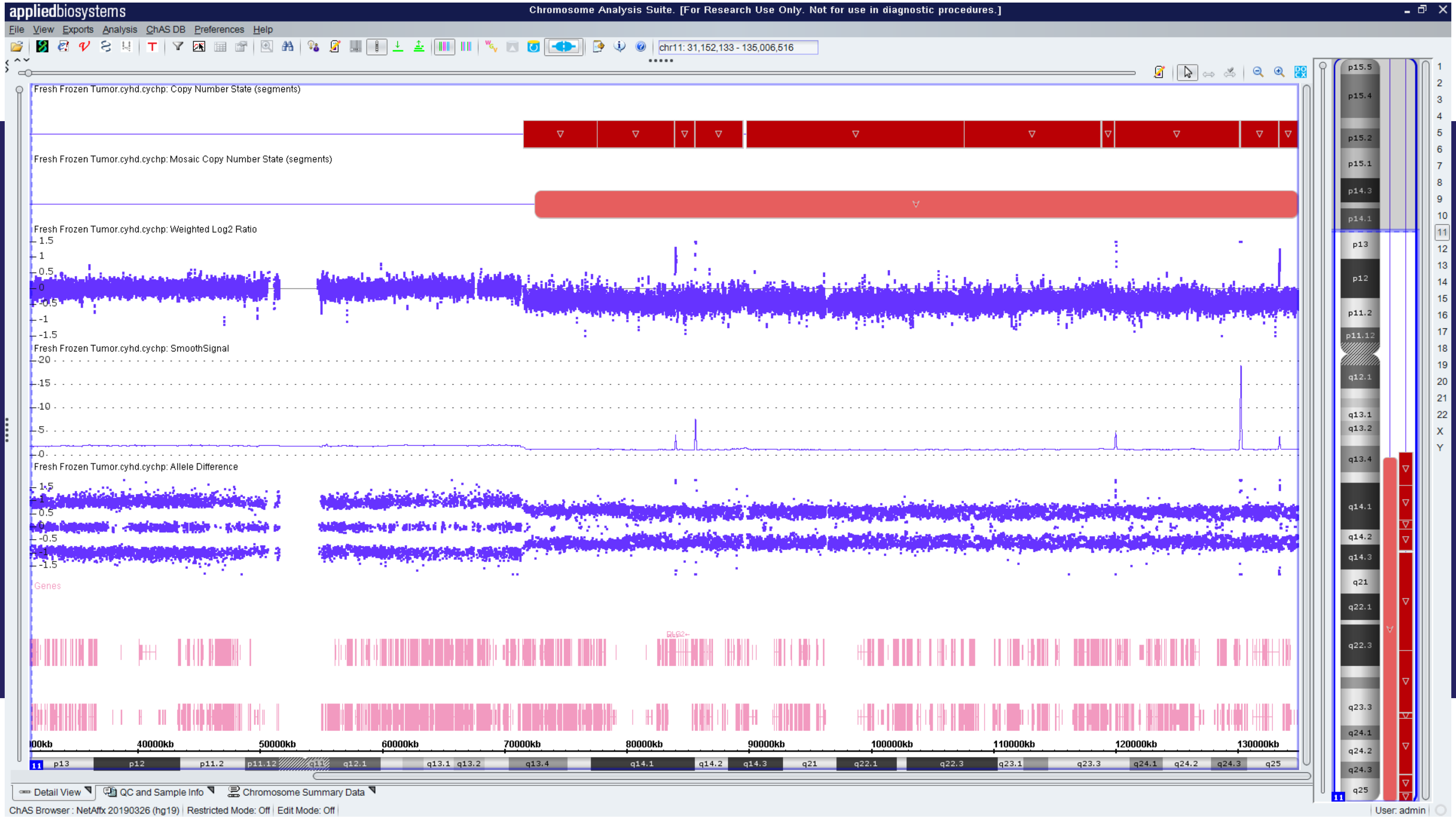
High-Level Copy Number Gains on Chromosome 2



ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off | Weighted Log2 Ratio: Fresh Frozen Tumor.cyhd.cychp, Nearest chromosome position: (31,012,077; -0.08994777)

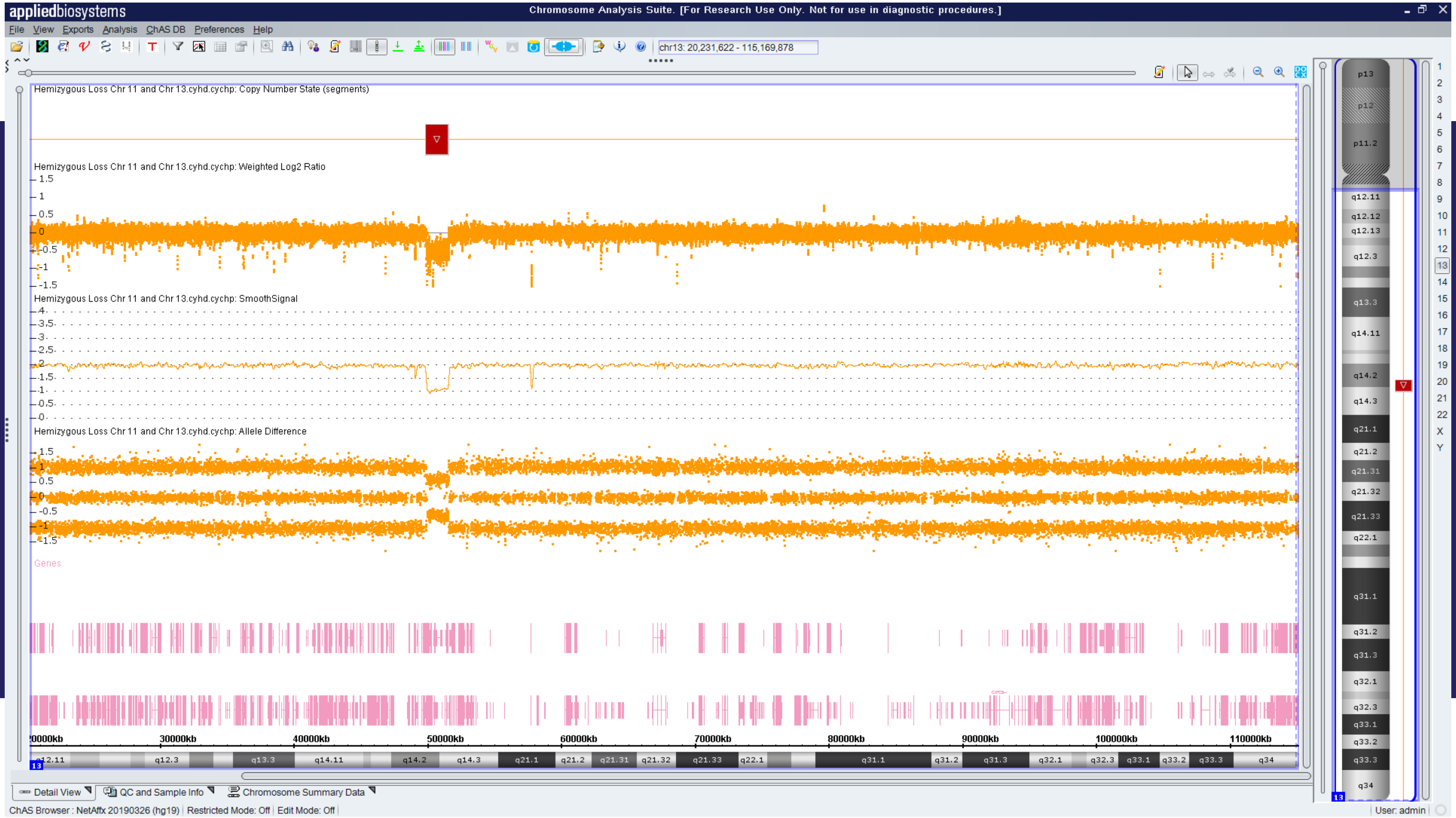
- Two amplifications (8 and >30 copies).
- These are visualized by increasing the scale of the smooth signal track.

Complex Rearrangement on Chromosome 11



- A mosaic loss (estimated at 85–90%) interrupted by copy-number gain segments and high-level amplifications.
- The smooth signal track scale has been increased for better visualization.

Hemizygous Loss on Chromosome 13



Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

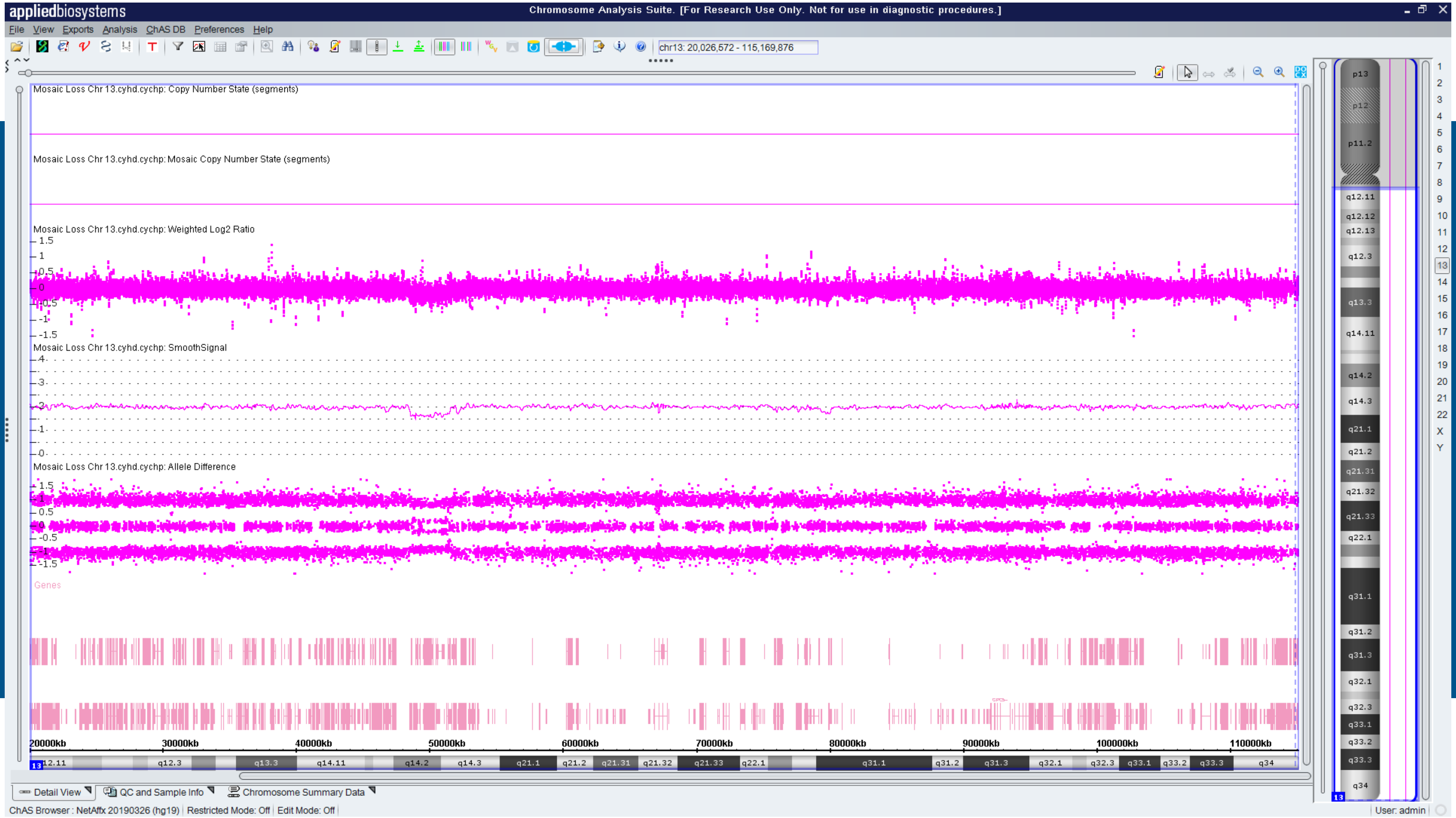
chr13: 20,231,622 - 115,169,878

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

- A full hemizygous loss on chromosome 13.
- This aberration was confirmed with interphase FISH.

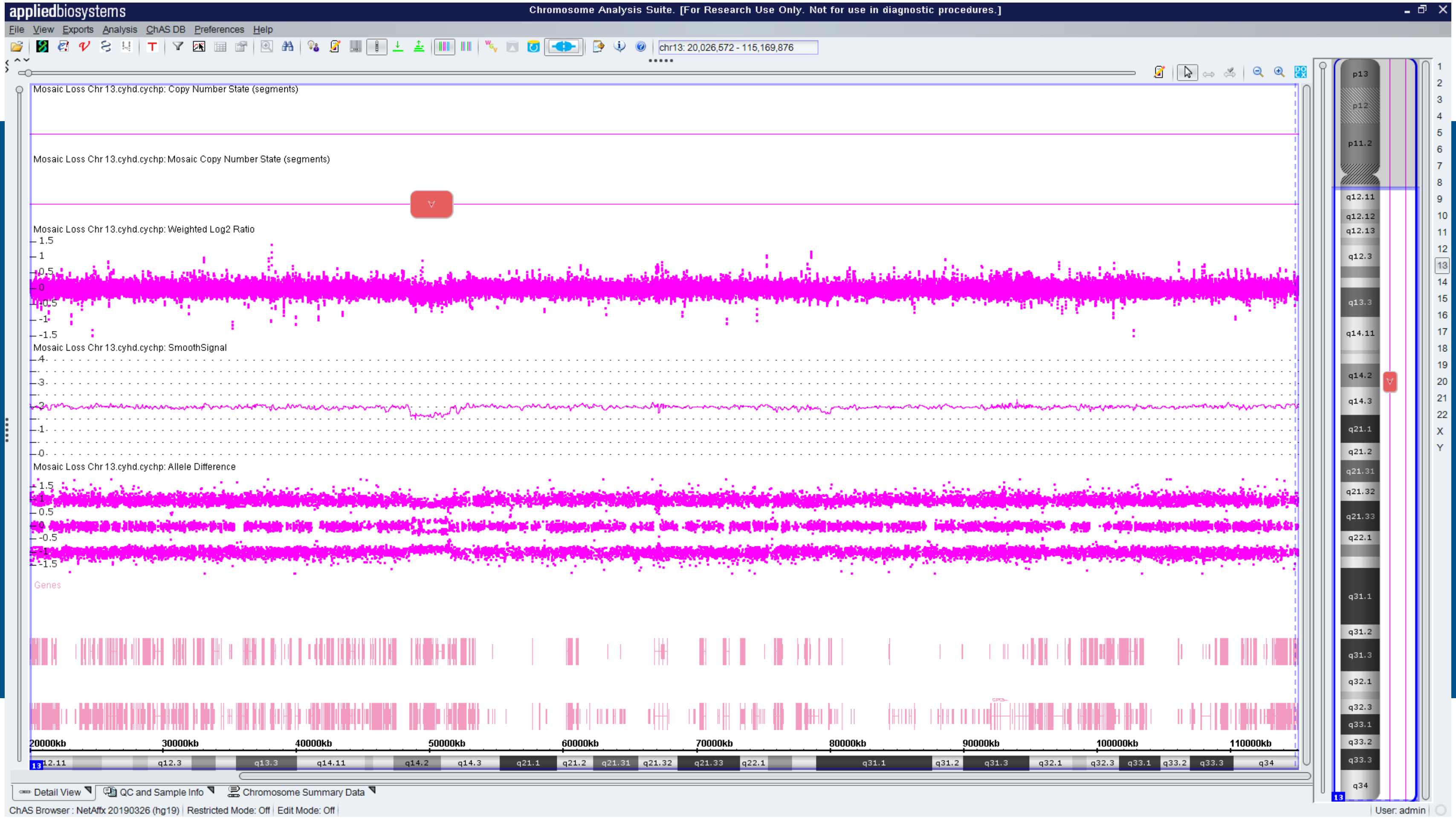
Mosaic Segments

Mosaic Loss on Chromosome 13 (1/2)



- This sample represents a mosaic loss in the same region as in the previous sample.
- 20% mosaicism is visible on the smooth signal and allele peaks tracks.
- Confirmed by interphase FISH.

Mosaic Loss on Chromosome 13 (2/2)



Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

chr13: 20,026,572 - 115,169,876

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off | User: admin

- A mosaic segment/flag was drawn with the “Edit” mode for simplified identification.

CLL Sample with Many Mosaic Segments

Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

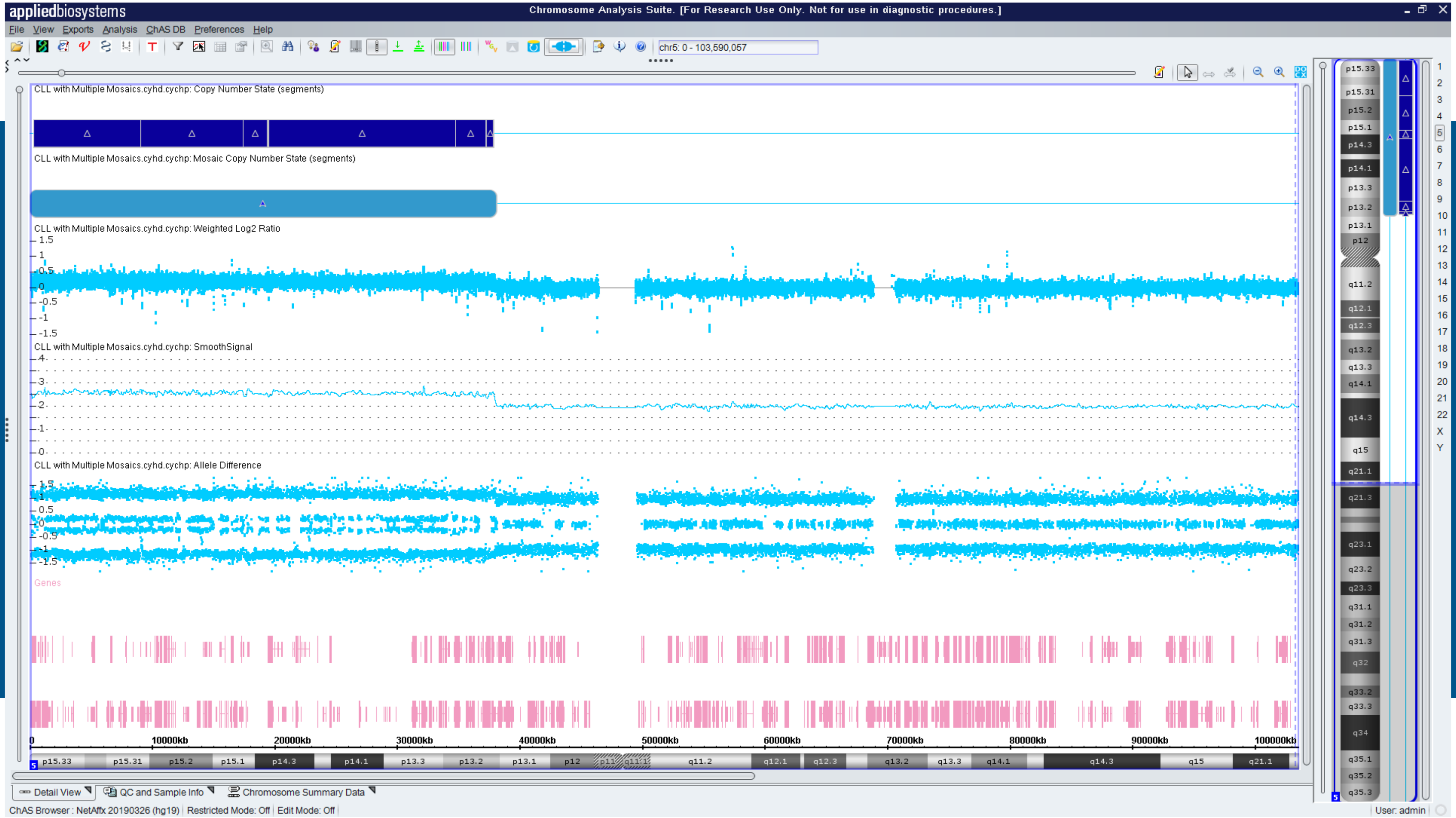
chr5: 0 - 103,590,057

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

User: admin |

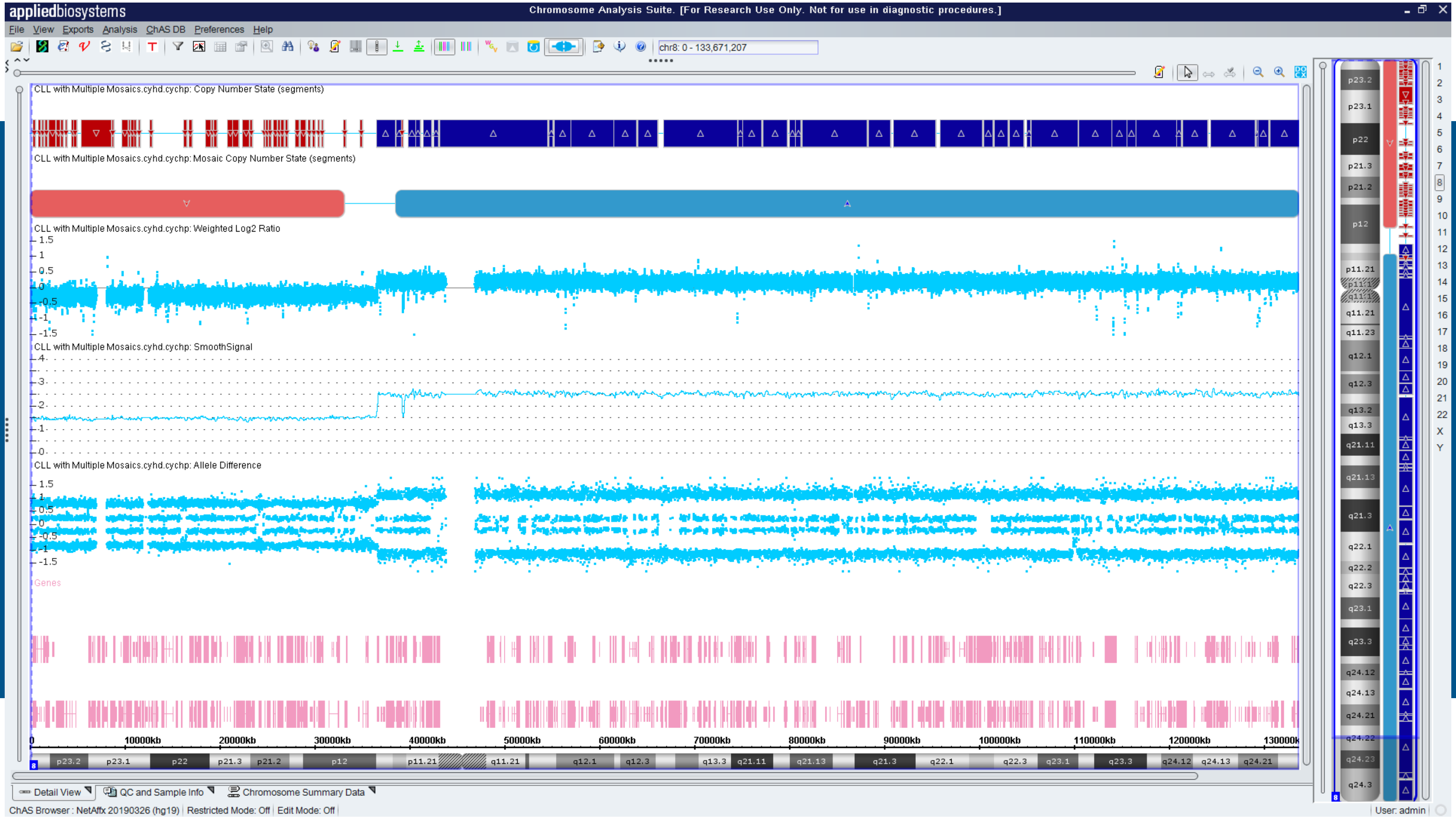
- This CLL sample has many informative aberration types.

Mosaic Gain



- Chromosome 5 illustrates a mosaic gain. The mosaic can be identified with smooth signal and/or the mosaic segment/flag. The mosaic level is estimated at 60%.
- The split in the allelic peaks track can confirm the finding.

Mosaic Loss and Mosaic Gain



Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

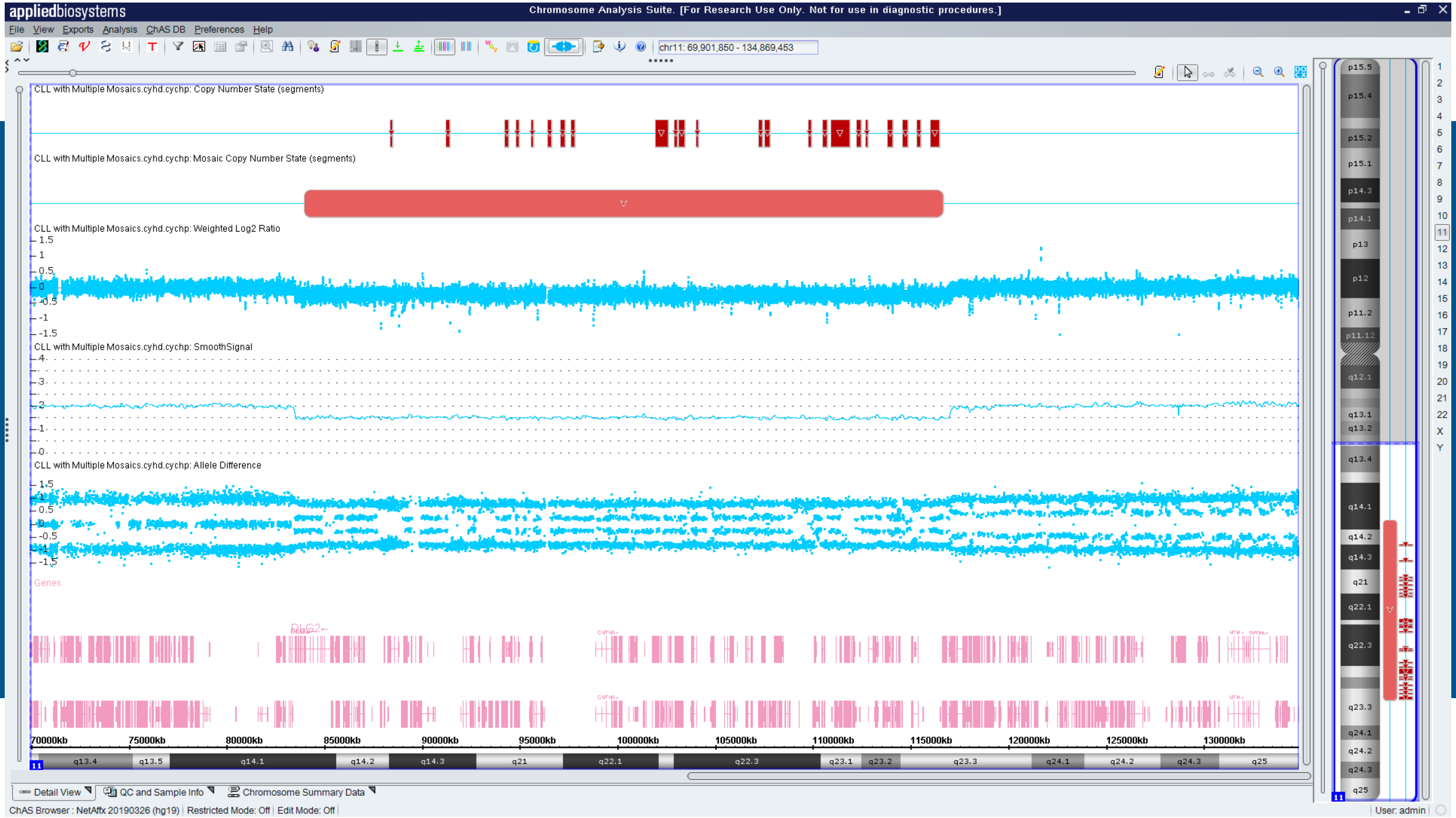
chr8: 0 - 133,671,207

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

User: admin

- Chromosome 8 illustrates a mosaic loss and a mosaic gain.

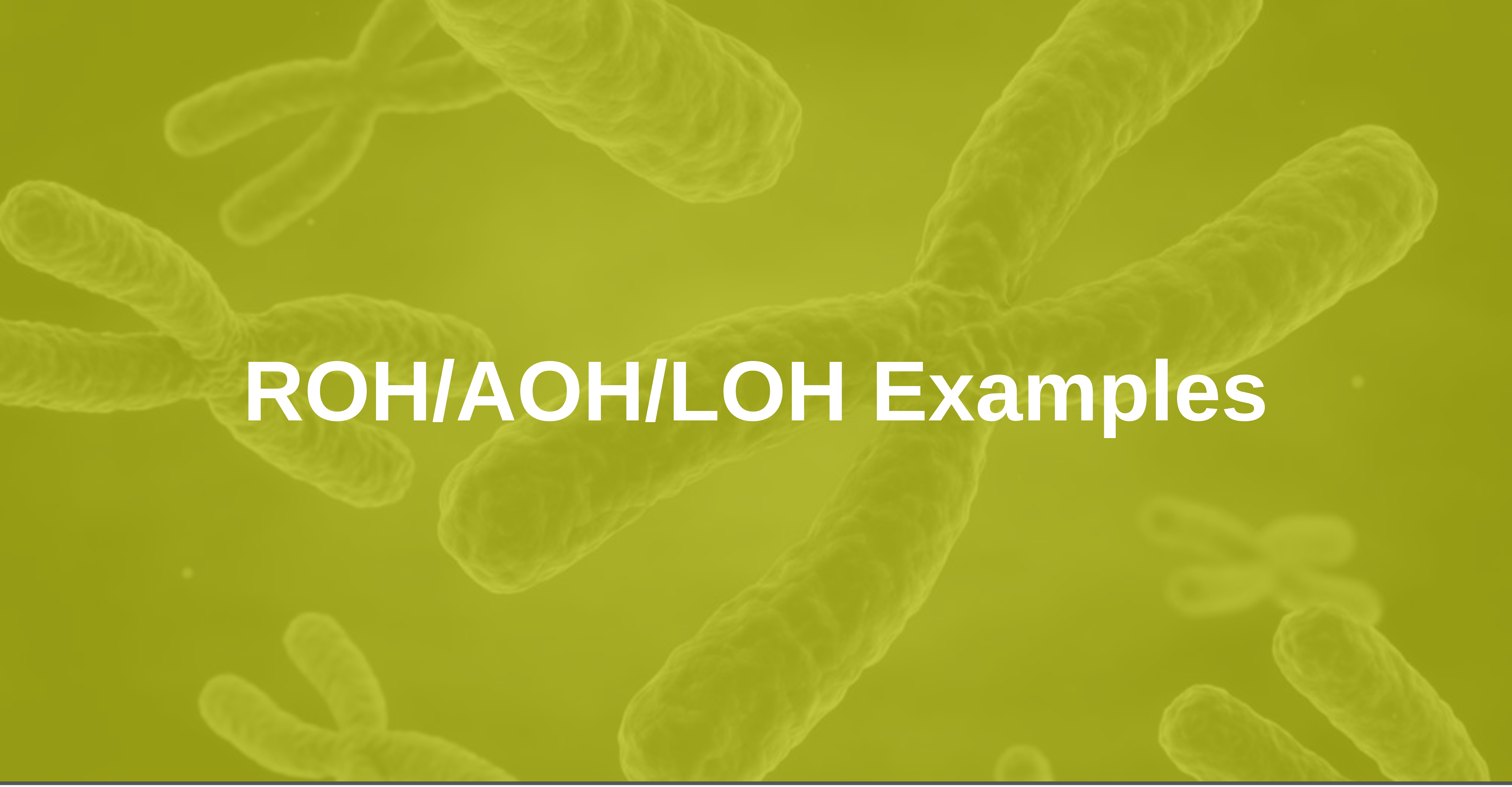
Mosaic Loss and Mosaic CN LOH



- Mosaic loss is highlighted by the light red segment/flag.
- Mosaic LOH/AOH can be identified by the absence of change in the smooth signal and the change in the inner bands of the allelic peaks track.

Mosaic Gain

- This sample illustrates a mosaic gain on chromosome 12.
- The percent mosaicism has been estimated at 85–90% using the smooth signal and mosaic segment/flag.



ROH/AOH/LOH Examples

Regions Identical by Descent: Genomic Profile of Consanguinity

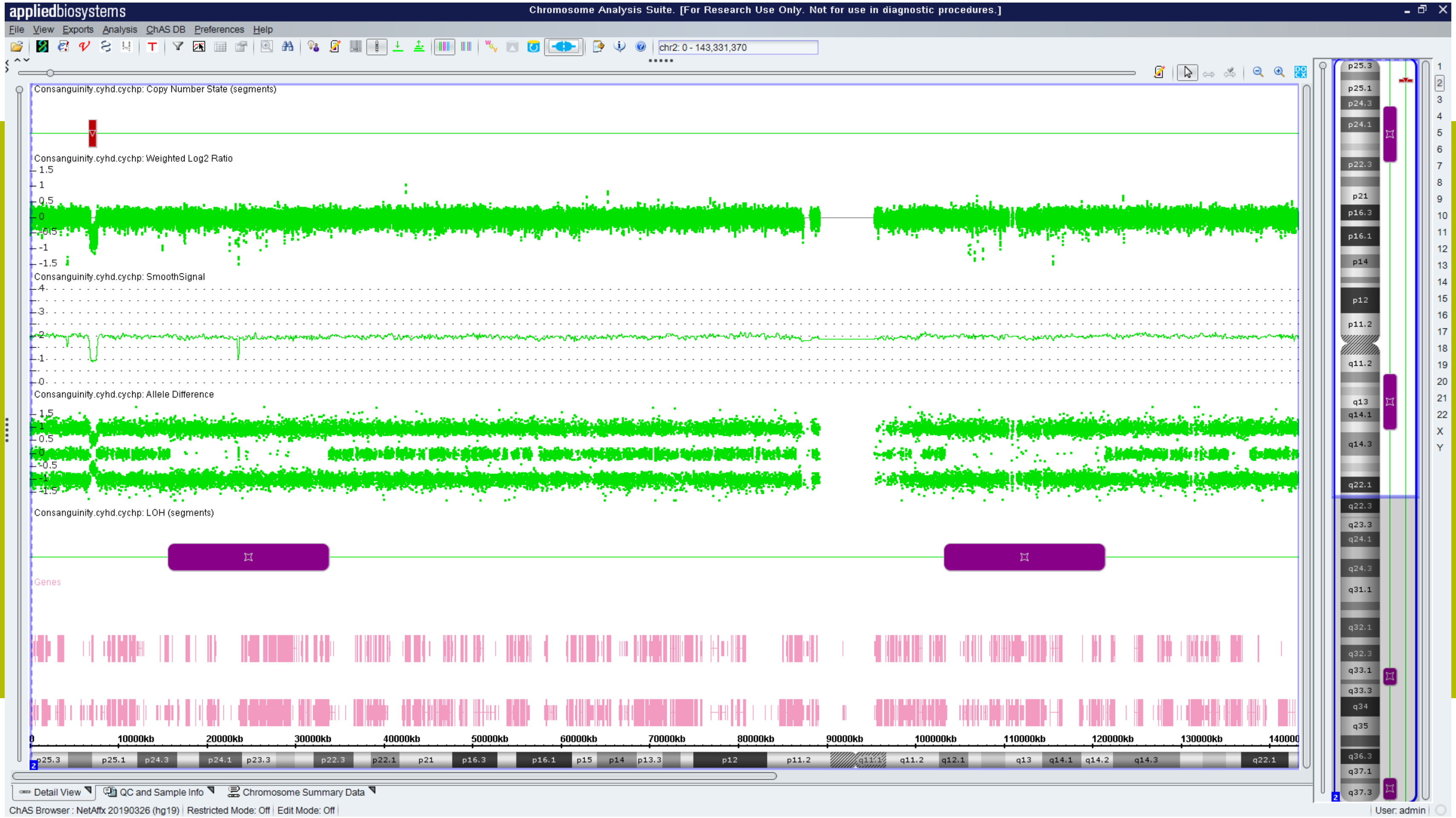
Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

User: admin |

- This example illustrates blocks of LOH >10 Mb across a majority of the chromosomes.

Regions Identical by Descent: Detailed View of Chromosome 2



- This is an example of two blocks of LOH >10 Mb on chromosome 2.
- There is also a hemizygous loss on this chromosome, illustrated by the red segment.

Copy-Neutral LOH on Chromosome 11: Karyoview

Chromosome Analysis Suite. [For Research Use Only. Not for use in diagnostic procedures.]

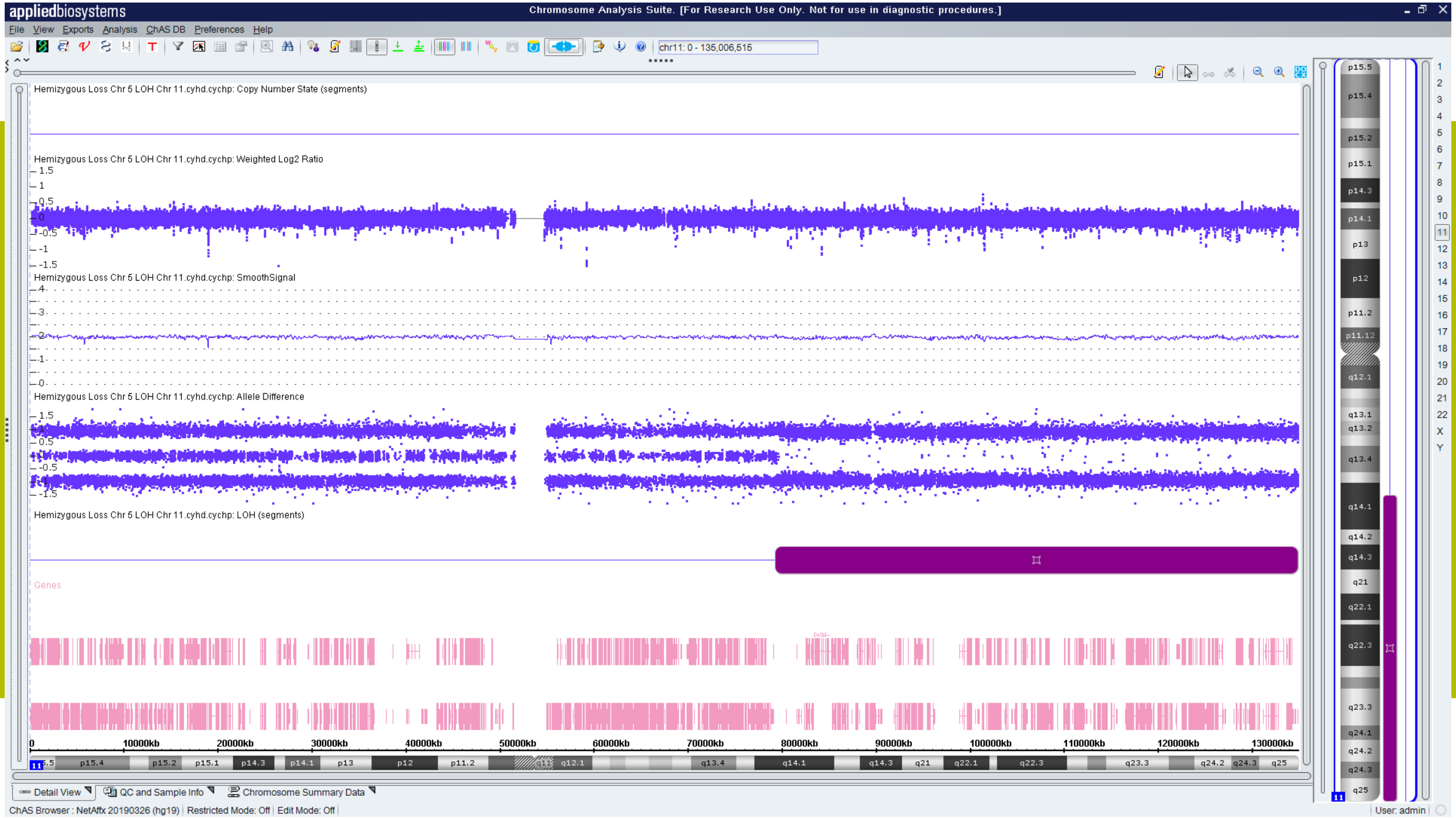
chr1: 0 - 249,250,621

ChAS Browser : NetAffx 20190326 (hg19) | Restricted Mode: Off | Edit Mode: Off |

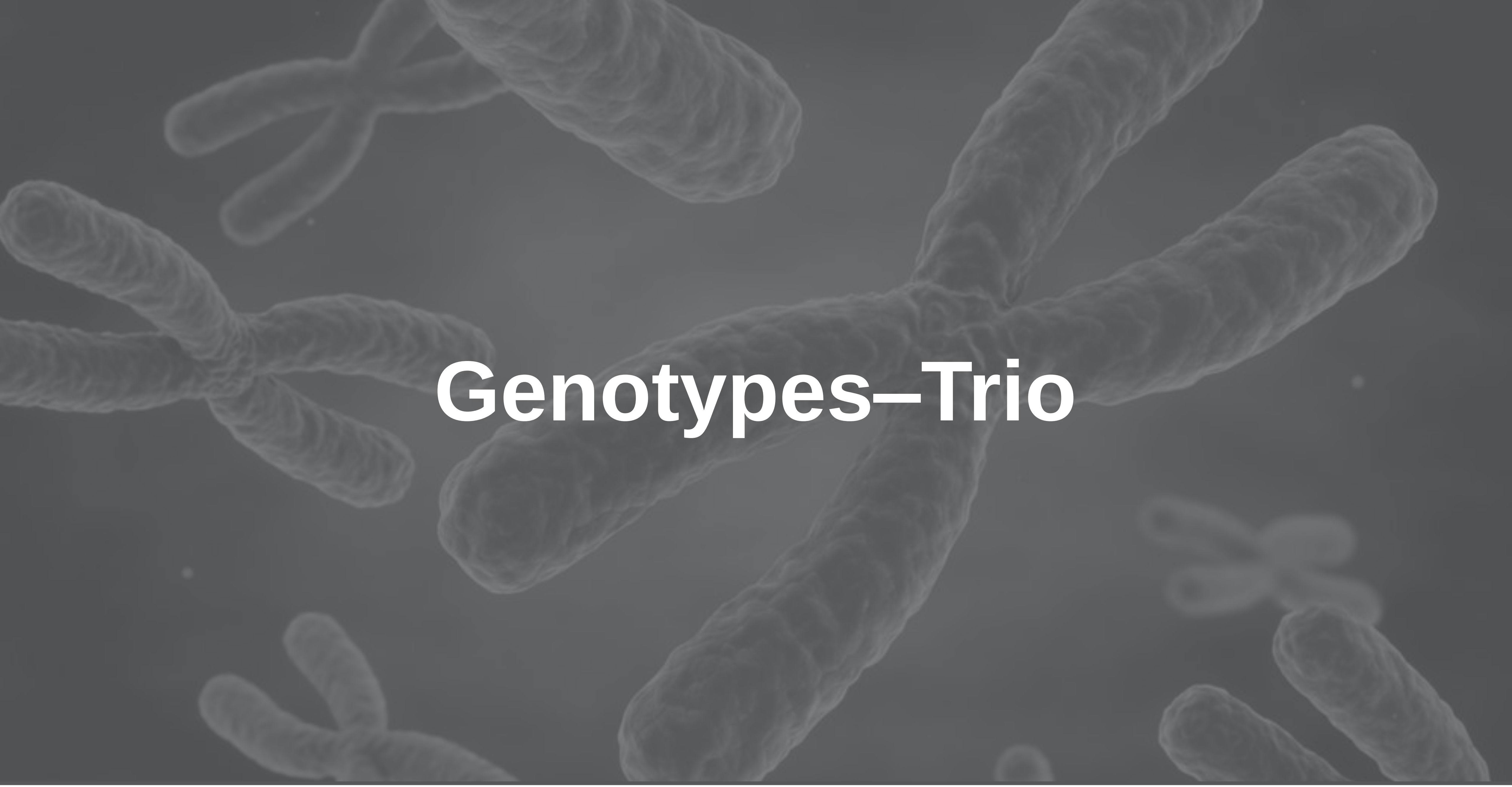
User: admin

- Only one autosome (Chr 11) has a block of LOH greater than 10 Mb.
- The X chromosome shows LOH because this is a male sample.

Copy-Neutral LOH on Chromosome 11: Detailed View

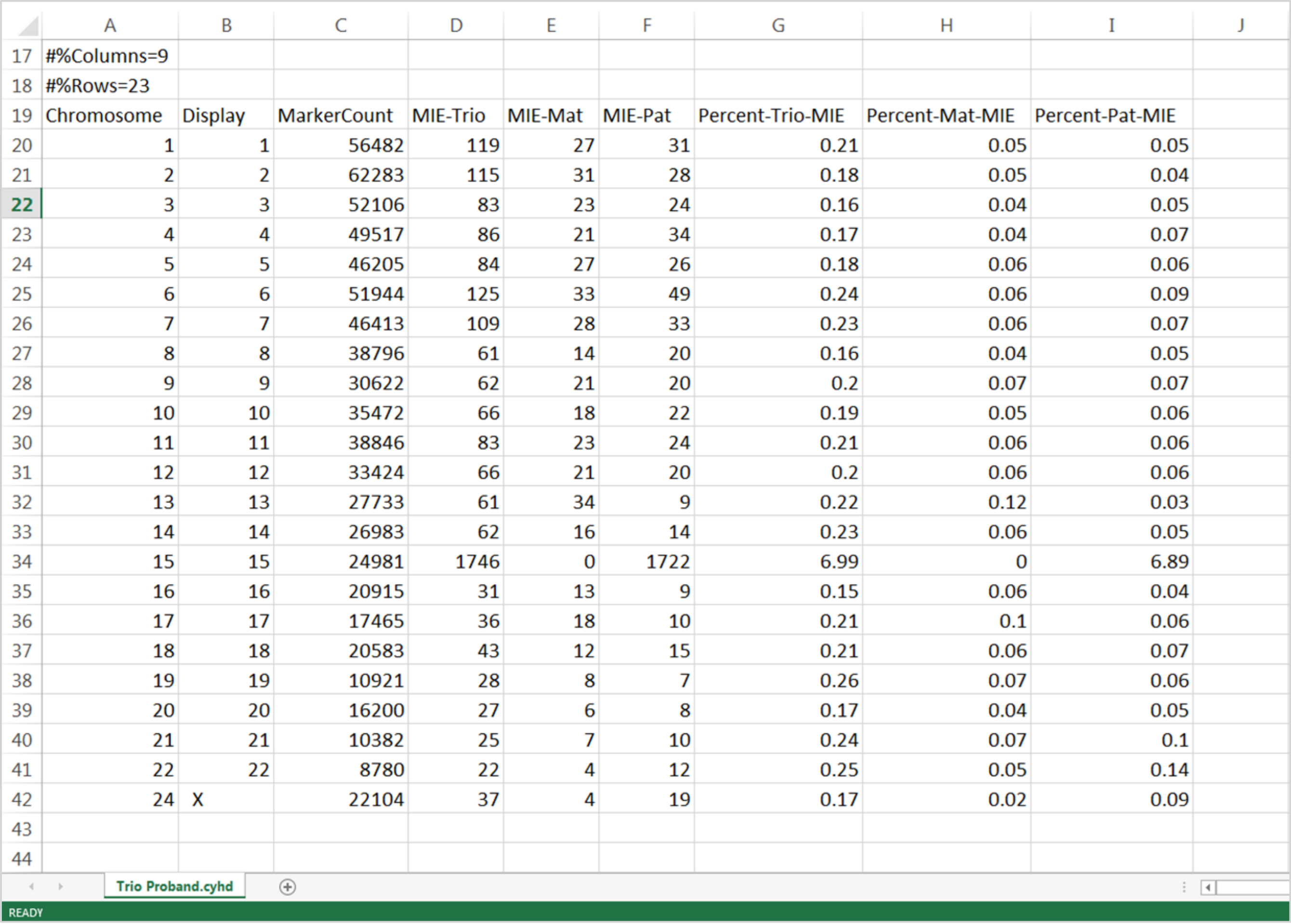


- This bone marrow sample has 55 Mb of copy-neutral LOH on chromosome 11q.



Genotypes–Trio

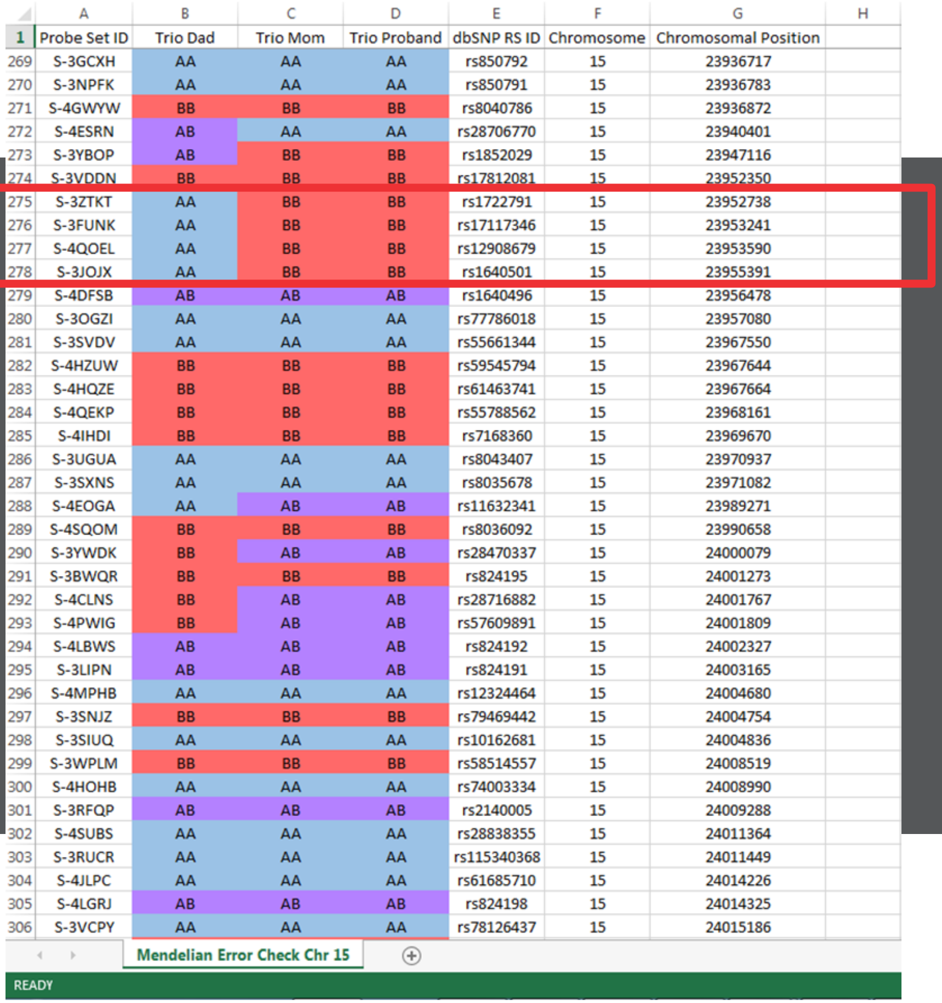
Mendelian Error Checking



	A	B	C	D	E	F	G	H	I	J
17	#%Columns=9									
18	#%Rows=23									
19	Chromosome	Display	MarkerCount	MIE-Trio	MIE-Mat	MIE-Pat	Percent-Trio-MIE	Percent-Mat-MIE	Percent-Pat-MIE	
20	1	1	56482	119	27	31	0.21	0.05	0.05	
21	2	2	62283	115	31	28	0.18	0.05	0.04	
22	3	3	52106	83	23	24	0.16	0.04	0.05	
23	4	4	49517	86	21	34	0.17	0.04	0.07	
24	5	5	46205	84	27	26	0.18	0.06	0.06	
25	6	6	51944	125	33	49	0.24	0.06	0.09	
26	7	7	46413	109	28	33	0.23	0.06	0.07	
27	8	8	38796	61	14	20	0.16	0.04	0.05	
28	9	9	30622	62	21	20	0.2	0.07	0.07	
29	10	10	35472	66	18	22	0.19	0.05	0.06	
30	11	11	38846	83	23	24	0.21	0.06	0.06	
31	12	12	33424	66	21	20	0.2	0.06	0.06	
32	13	13	27733	61	34	9	0.22	0.12	0.03	
33	14	14	26983	62	16	14	0.23	0.06	0.05	
34	15	15	24981	1746	0	1722	6.99	0	6.89	
35	16	16	20915	31	13	9	0.15	0.06	0.04	
36	17	17	17465	36	18	10	0.21	0.1	0.06	
37	18	18	20583	43	12	15	0.21	0.06	0.07	
38	19	19	10921	28	8	7	0.26	0.07	0.06	
39	20	20	16200	27	6	8	0.17	0.04	0.05	
40	21	21	10382	25	7	10	0.24	0.07	0.1	
41	22	22	8780	22	4	12	0.25	0.05	0.14	
42	24	X	22104	37	4	19	0.17	0.02	0.09	
43										
44										

- This set of samples represents a trio. The Mendelian Error Check function was used to check for relatedness.
- The trio is consistent, but there is an increased rate of errors on Chr15, compatible with a maternal hetero UPD.

Genotype Calls on Chromosome 15



1	Probe Set ID	Trio Dad	Trio Mom	Trio Proband	dbSNP RS ID	Chromosome	Chromosomal Position
269	S-3GCXH	AA	AA	AA	rs850792	15	23936717
270	S-3NPFK	AA	AA	AA	rs850791	15	23936783
271	S-4GWYW	BB	BB	BB	rs8040786	15	23936872
272	S-4ESRN	AB	AA	AA	rs28706770	15	23940401
273	S-3YBOP	AB	BB	BB	rs1852029	15	23947116
274	S-3VDDN	BB	BB	BB	rs17812081	15	23952350
275	S-3ZTKT	AA	BB	BB	rs1722791	15	23952738
276	S-3FUNK	AA	BB	BB	rs17117346	15	23953241
277	S-4QOEL	AA	BB	BB	rs12908679	15	23953590
278	S-3JOJX	AA	BB	BB	rs1640501	15	23955391
279	S-4DFSB	AB	AB	AB	rs1640496	15	23956478
280	S-3OGZI	AA	AA	AA	rs77786018	15	23957080
281	S-3SVDV	AA	AA	AA	rs55661344	15	23967550
282	S-4HZUW	BB	BB	BB	rs59545794	15	23967644
283	S-4HQZE	BB	BB	BB	rs61463741	15	23967664
284	S-4QEKP	BB	BB	BB	rs55788562	15	23968161
285	S-4IHDI	BB	BB	BB	rs7168360	15	23969670
286	S-3UGUA	AA	AA	AA	rs8043407	15	23970937
287	S-3SXNS	AA	AA	AA	rs8035678	15	23971082
288	S-4EOGA	AA	AB	AB	rs11632341	15	23989271
289	S-4SQOM	BB	BB	BB	rs8036092	15	23990658
290	S-3YWDK	BB	AB	AB	rs28470337	15	24000079
291	S-3BWQR	BB	BB	BB	rs824195	15	24001273
292	S-4CLNS	BB	AB	AB	rs28716882	15	24001767
293	S-4PWIG	BB	AB	AB	rs57609891	15	24001809
294	S-4LBWS	AB	AB	AB	rs824192	15	24002327
295	S-3LIPN	AB	AB	AB	rs824191	15	24003165
296	S-4MPHB	AA	AA	AA	rs12324464	15	24004680
297	S-3SNJZ	BB	BB	BB	rs79469442	15	24004754
298	S-3SIUQ	AA	AA	AA	rs10162681	15	24004836
299	S-3WPLM	BB	BB	BB	rs58514557	15	24008519
300	S-4HOHB	AA	AA	AA	rs74003334	15	24008990
301	S-3RFQP	AB	AB	AB	rs2140005	15	24009288
302	S-4SUBS	AA	AA	AA	rs28838355	15	24011364
303	S-3RUCR	AA	AA	AA	rs115340368	15	24011449
304	S-4JLPC	AA	AA	AA	rs61685710	15	24014226
305	S-4LGRJ	AB	AB	AB	rs824198	15	24014325
306	S-3VCPY	AA	AA	AA	rs78126437	15	24015186

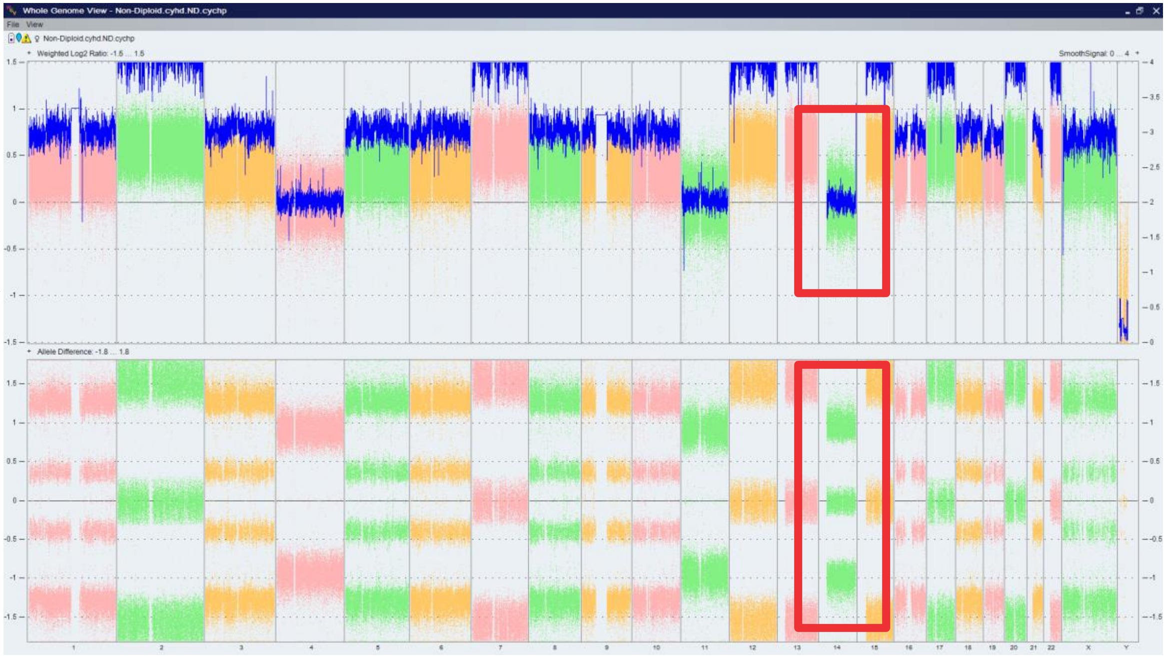
- Chromosome 15 genotypes show how the UPD is of maternal origin and is hetero UPD (most genotypes are the same in mother and proband, and different in father).

Non-diploid Normalization

A Non-diploid Sample Processed with the ND Algorithm—WGV

.cyhd

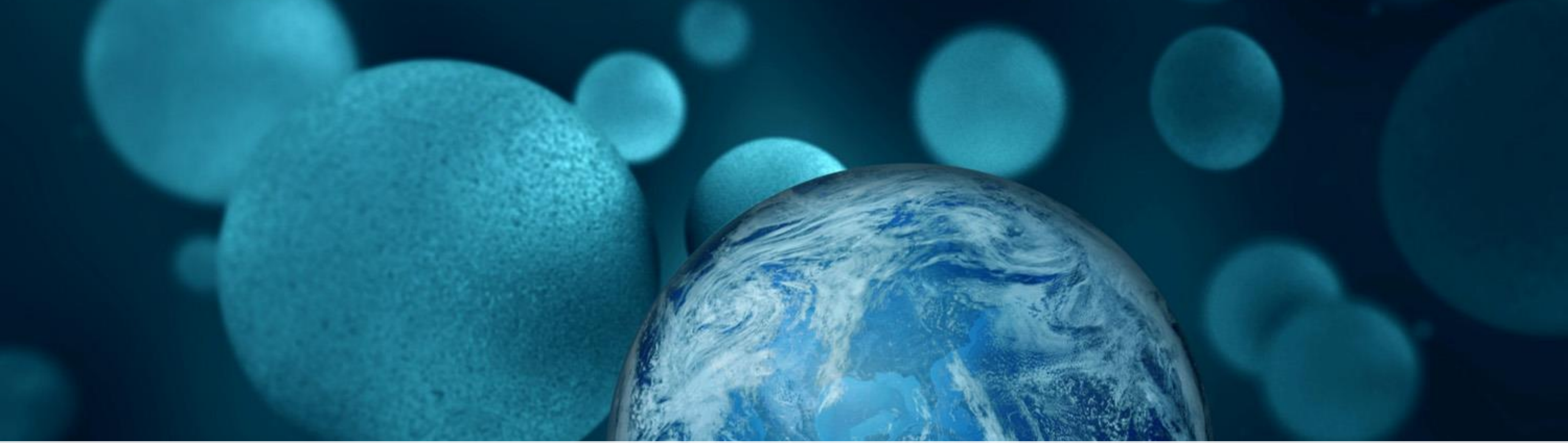
.cyhd.ND



This sample (T12-24) had several aberrations and hyperdiploid status. The baseline was not set correctly. The ND algorithm successfully corrects the baseline and calls.

A Non-diploid Sample Processed with the ND Algorithm—Karyoview

This sample (T12-24) had several aberrations and hyperdiploid status. The baseline was not set correctly. The ND algorithm successfully corrects the baseline and calls.



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