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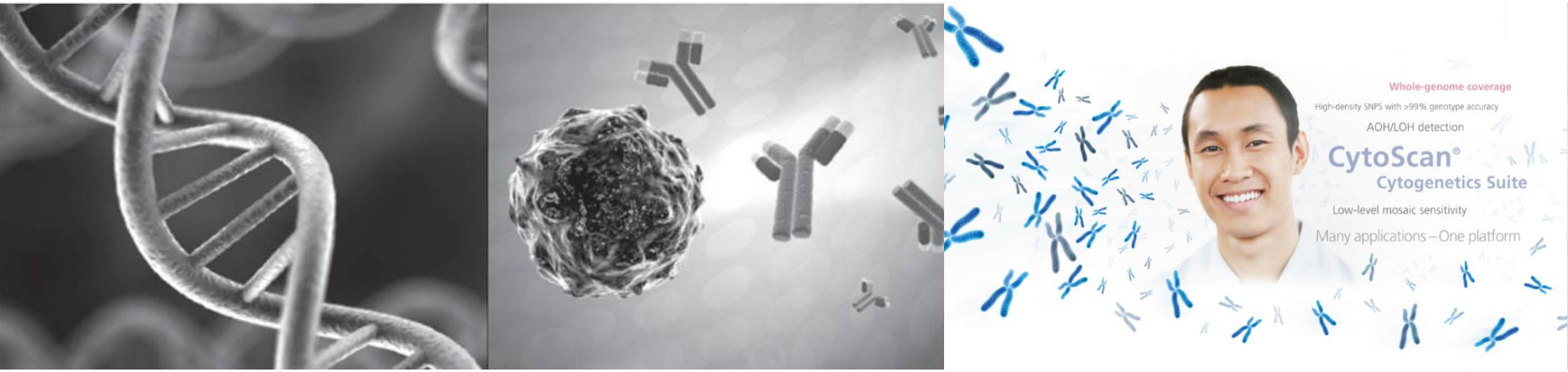

CytoScan® Cytogenetics Suite

CytoScan 750K

Educational Examples of Different Aberration Types

eBioscience GeneChip Panomics USB

Summary

- A set of different samples analyzed with CytoScan 750K is provided for educational propose
- A set of samples analyzed with CytoScan HD is provided for additional information.
- *Sample display color might be different in this presentation when compared to your software.*

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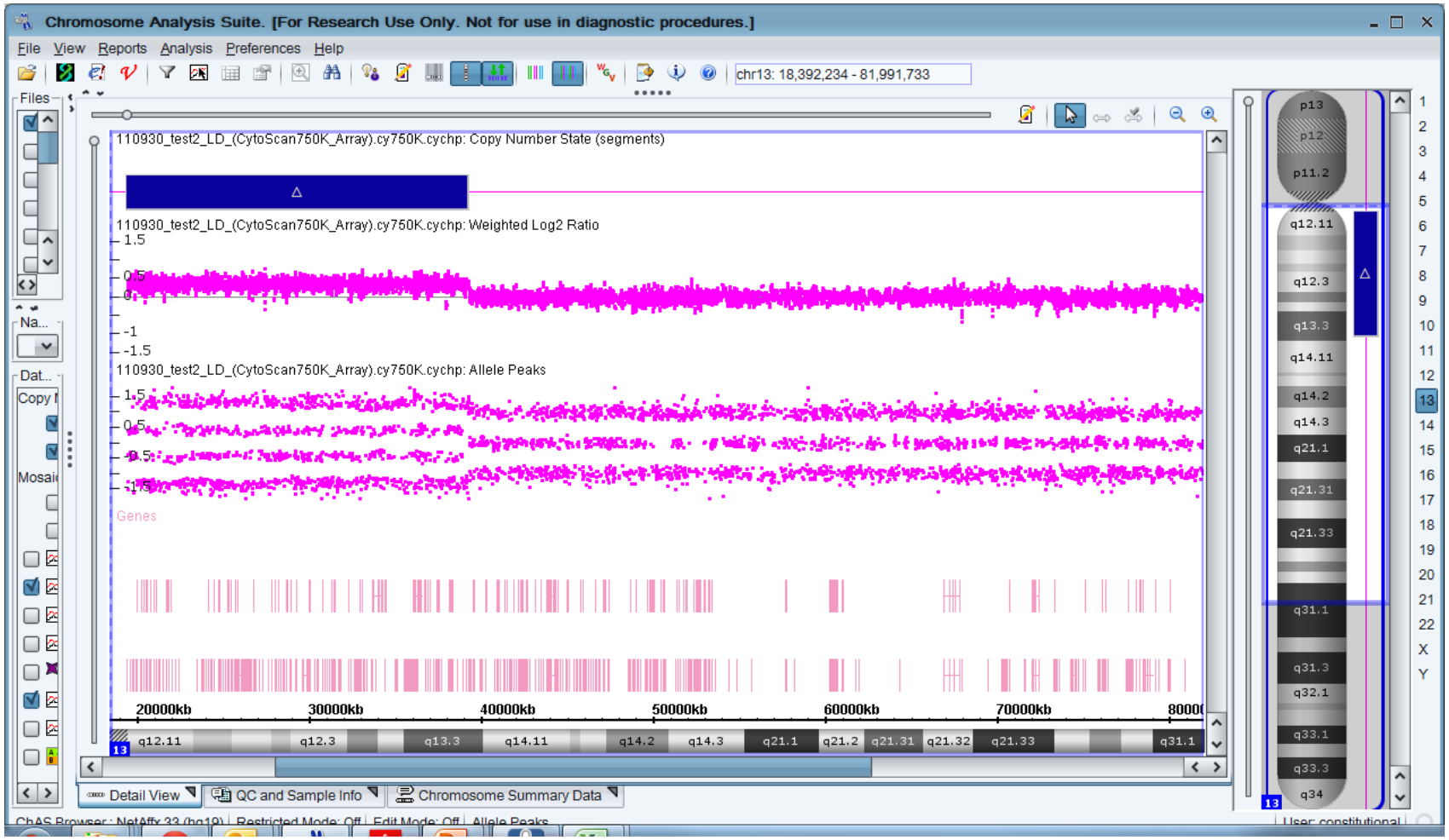
Hemizygous Gain on Chr13



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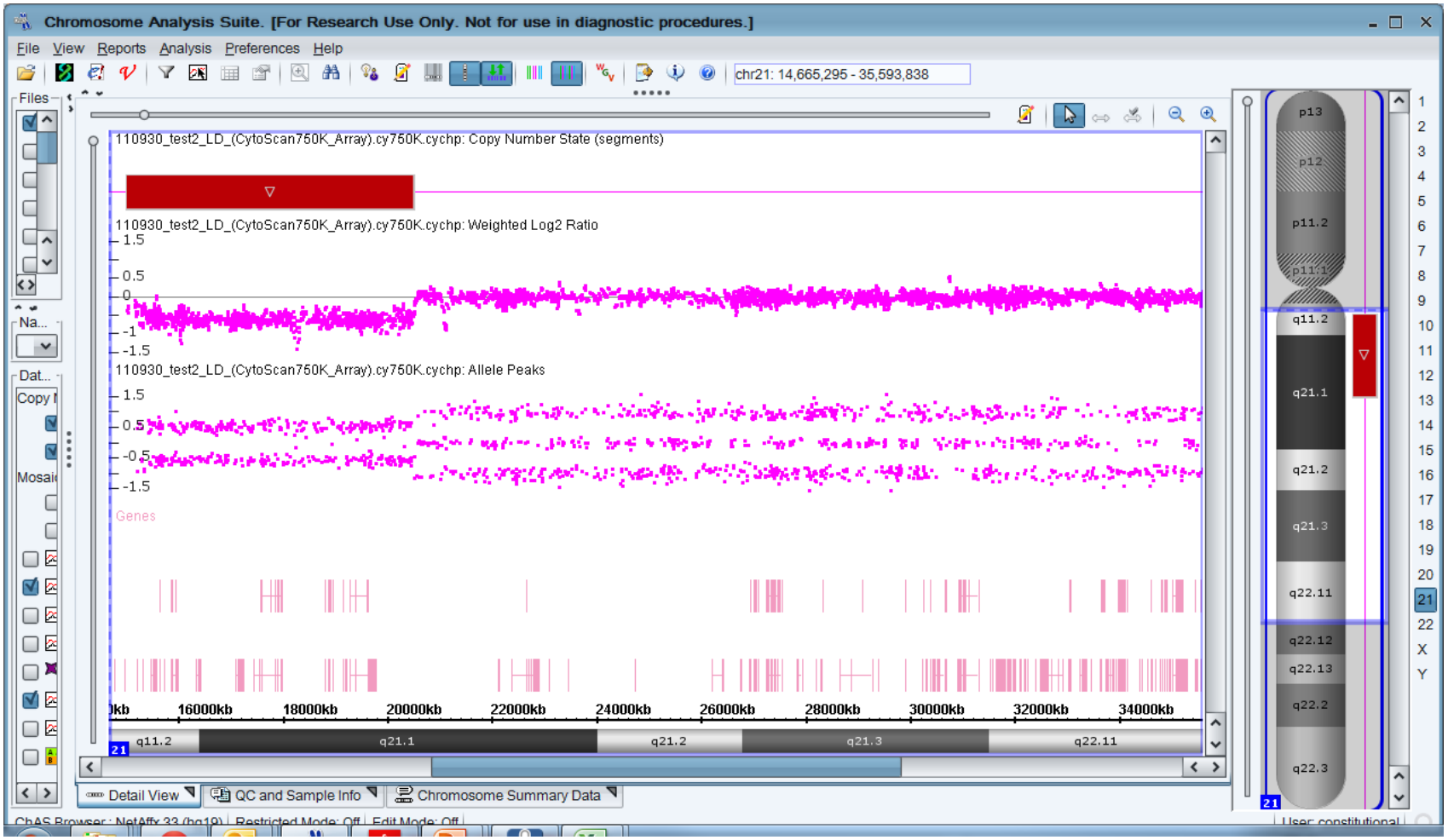
Hemizygous loss on Chr21



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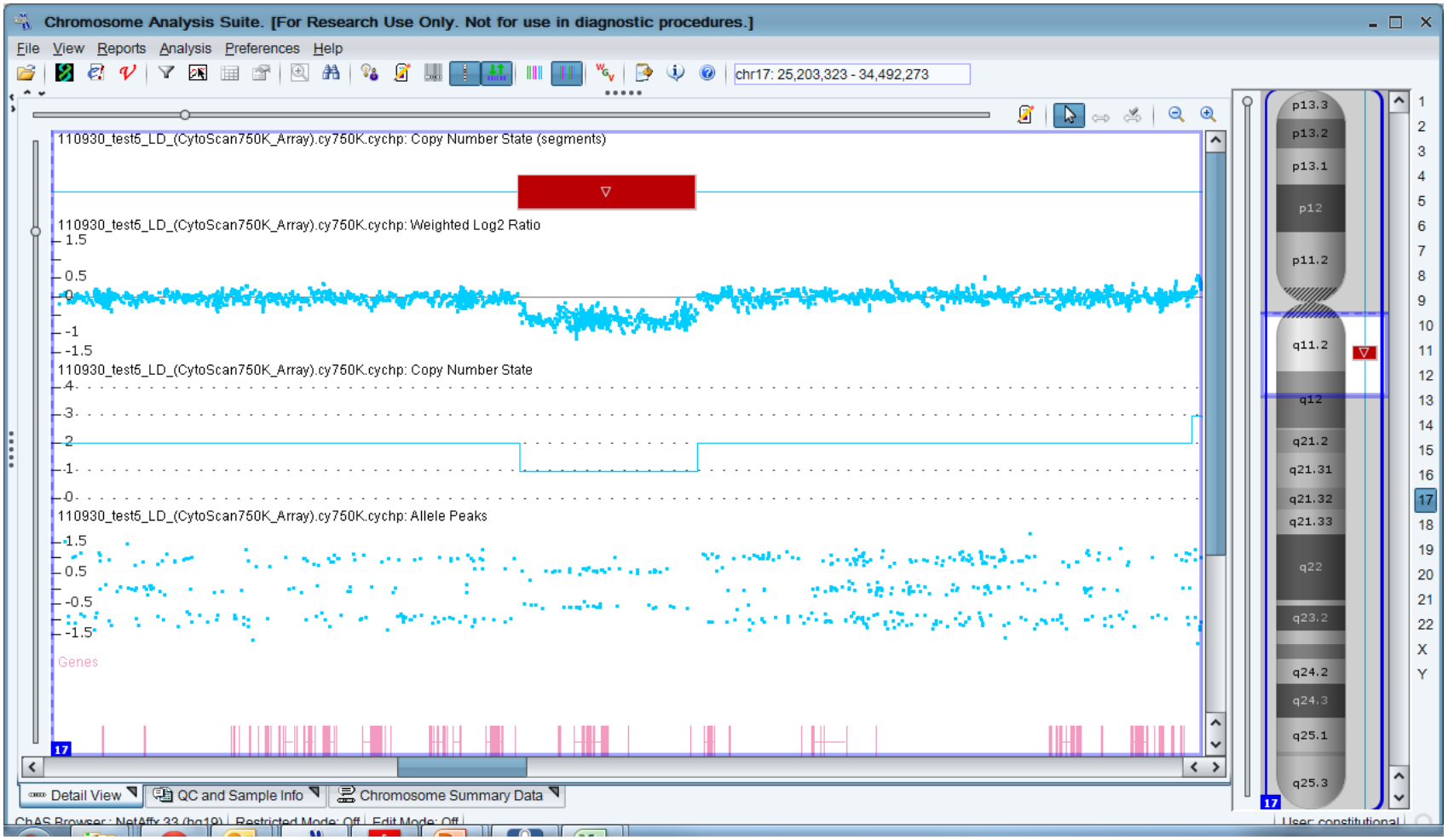
Hemizygous loss on Chr17



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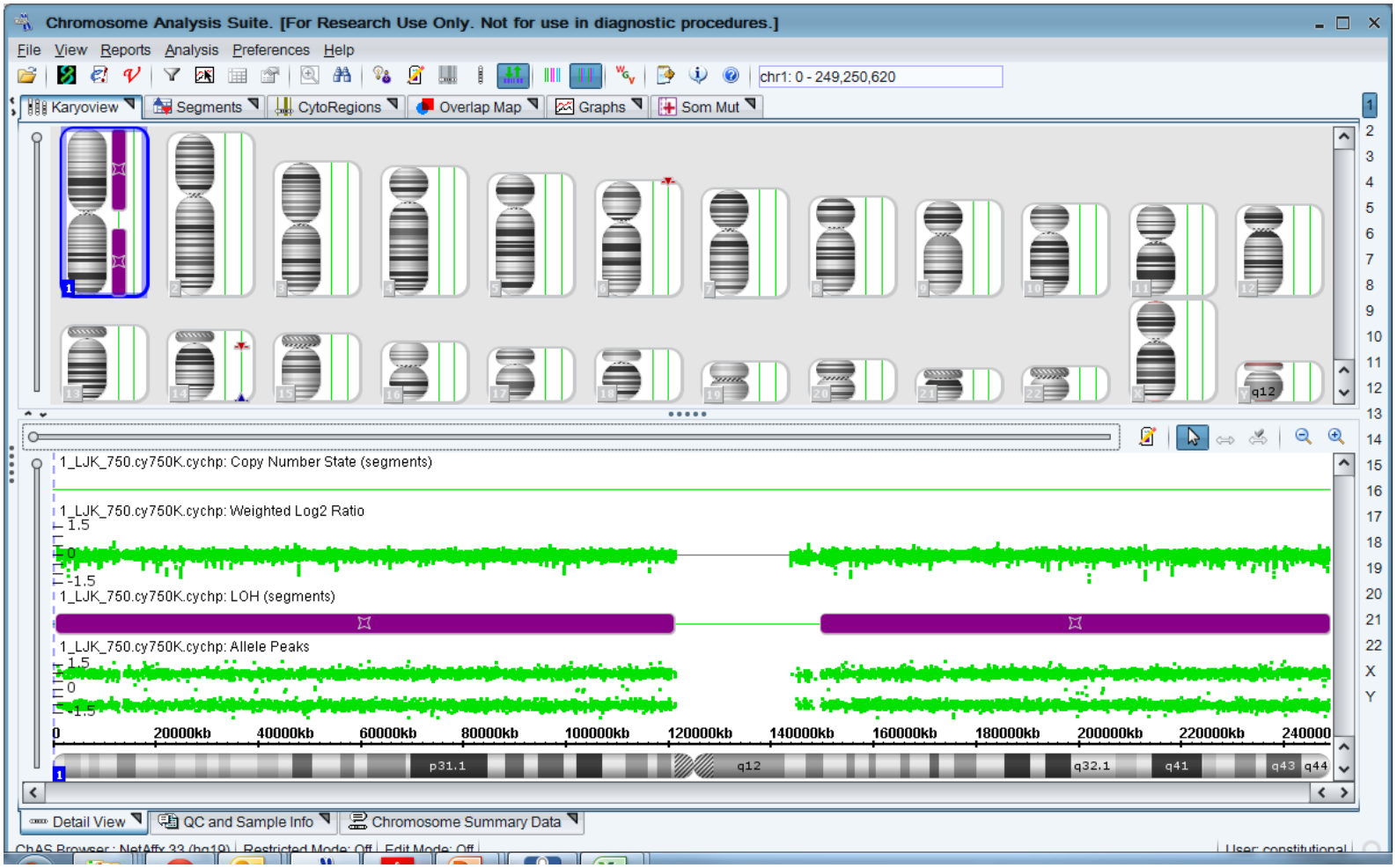
Copy neutral LOH on chromosome 1 illustrating AOH/LOH >10 Mb



- **This LOH/AOH was due to uniparental Isodisomy**

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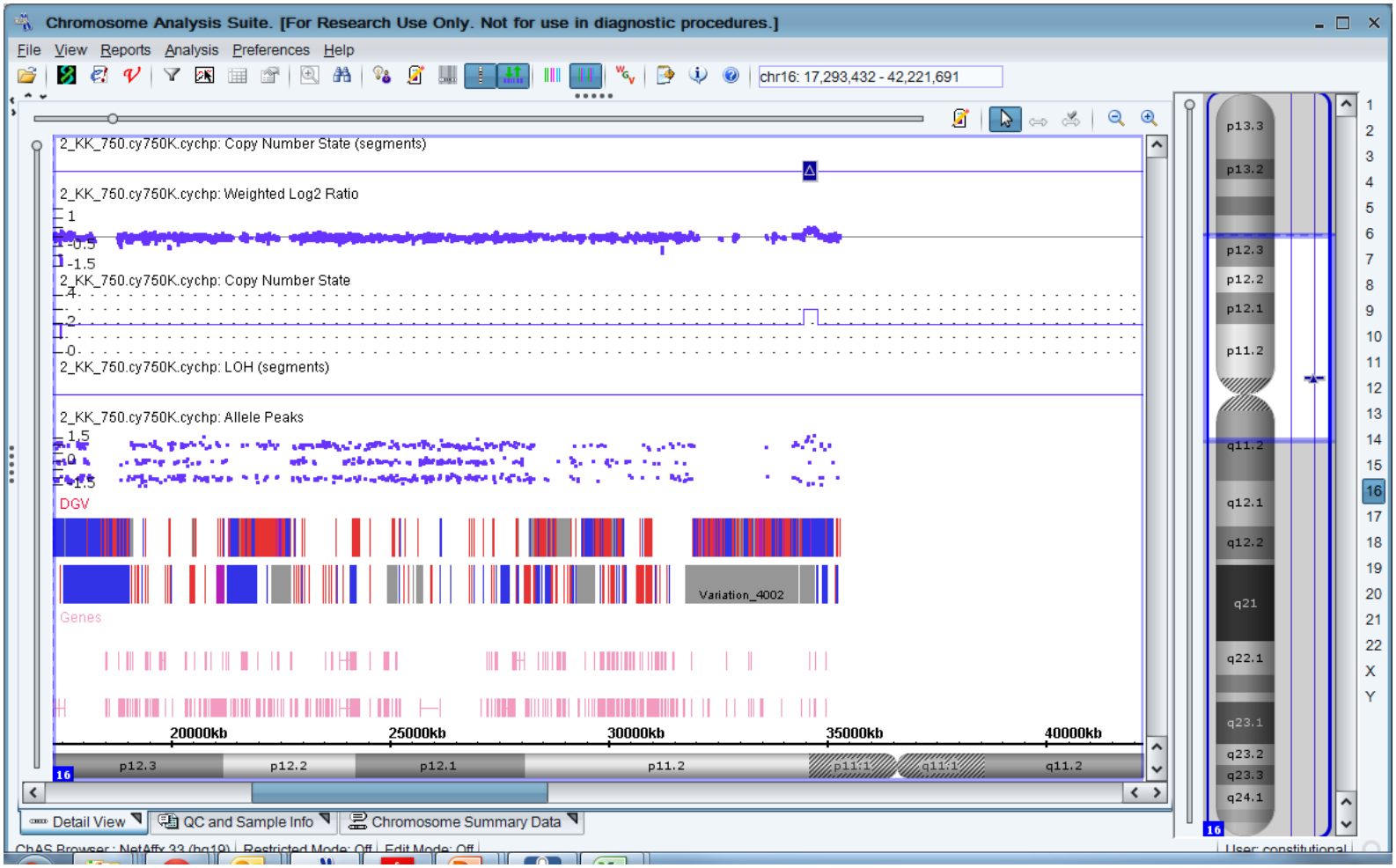
16p11.2 hemizygous gain of 317kb



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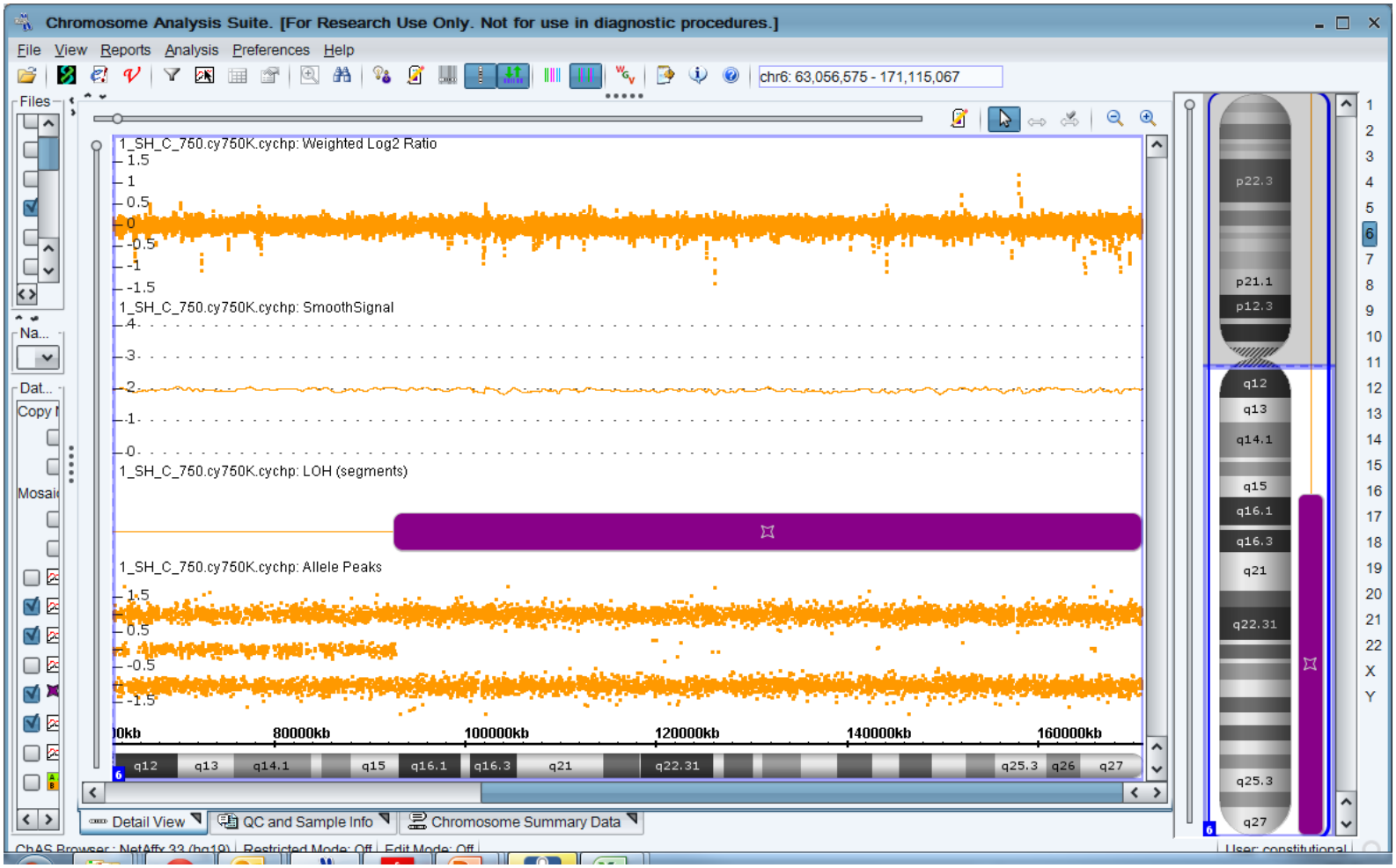
Copy neutral AOH/LOH on chromosome 6
Closer detail illustrating LOH>10 Mb



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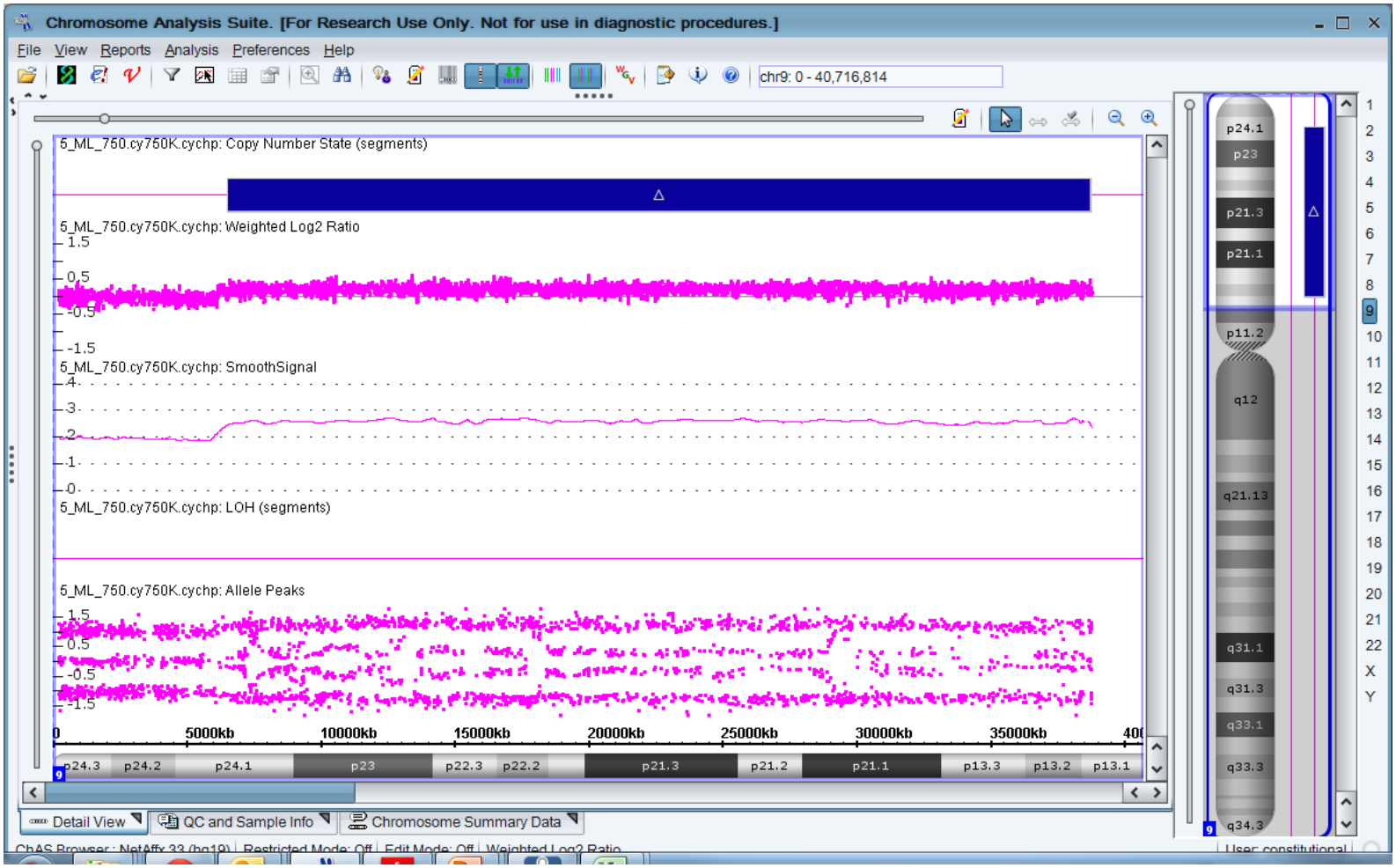
Mosaic gain on chromosome 9



- **Mosaic was estimated at ~60% using the smooth signal values.**

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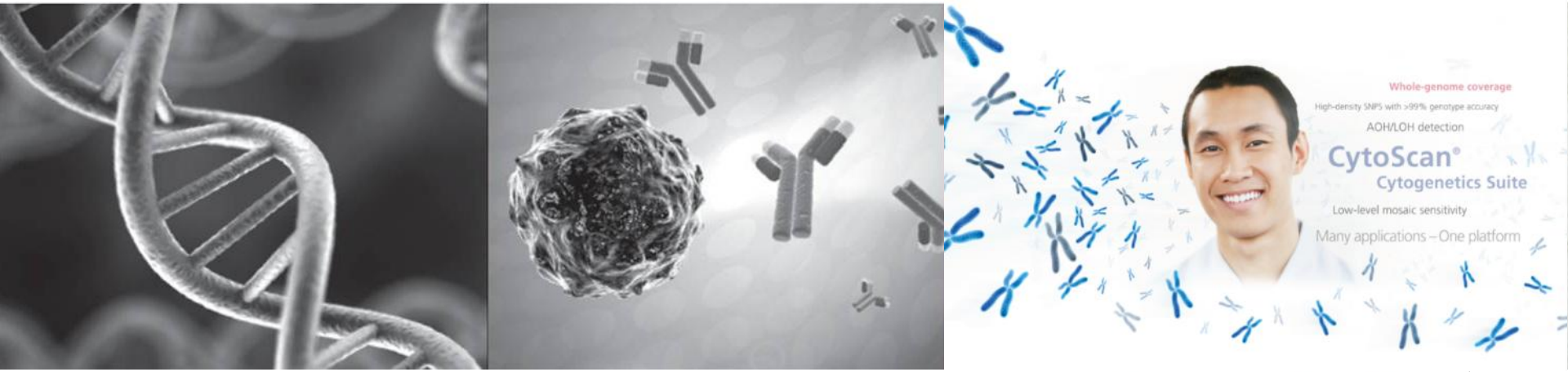

CytoScan® Cytogenetics Suite

CytoScan HD

Educational Examples of Different Aberration types

eBioscience GeneChip Panomics USB

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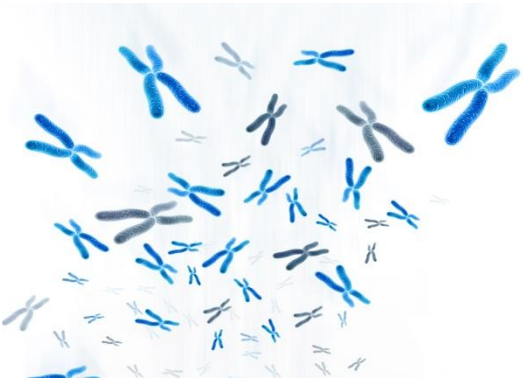
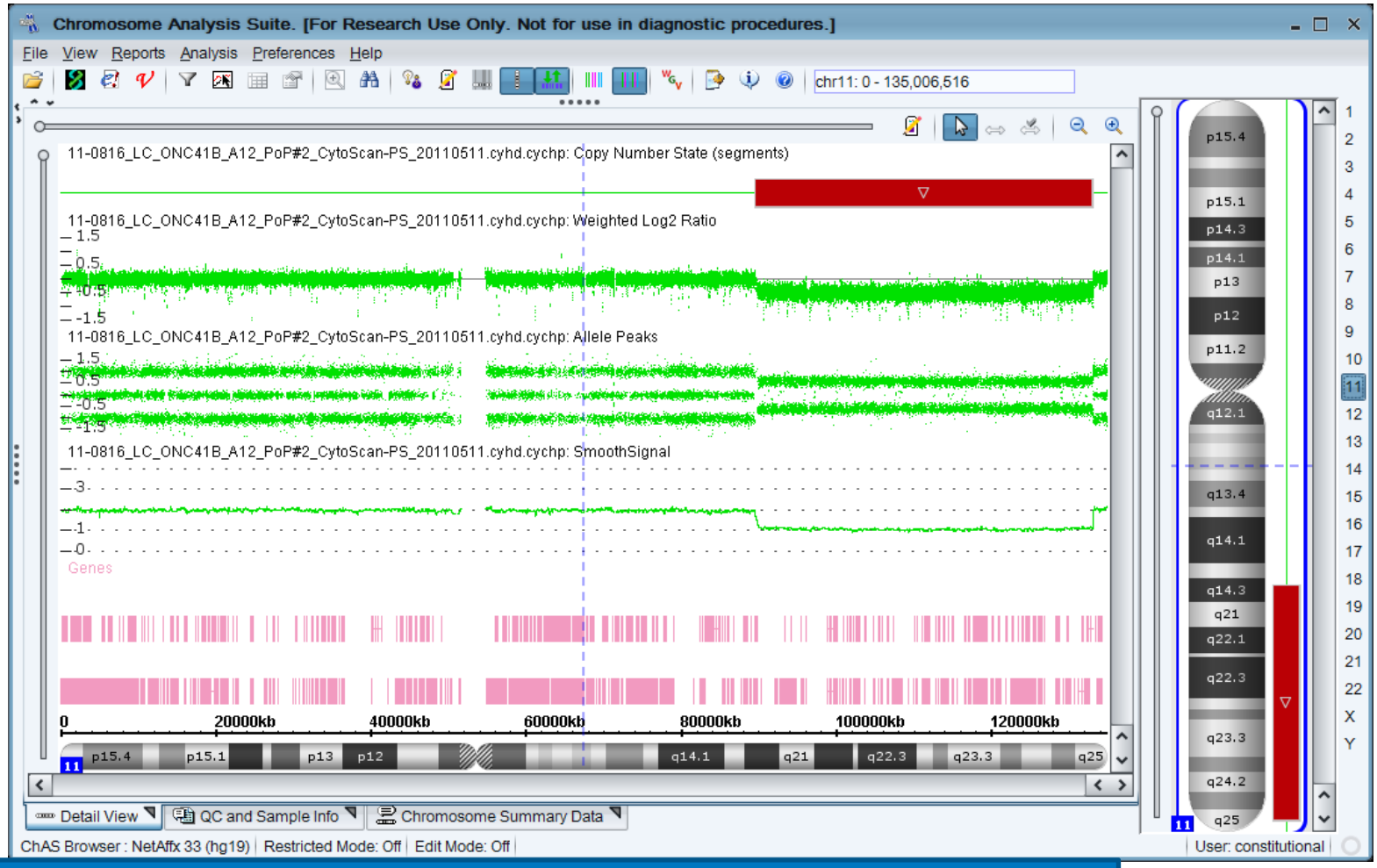


Gains and losses

Hemizygous loss on chromosome 11



- **This example provides an illustration of an hemizygous loss on chromosome 11.**
- **The allelic peaks track shows a pattern change from 3 to 2 bands confirming the hemizigous loss.**

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Hemizygous loss on chromosome 5

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

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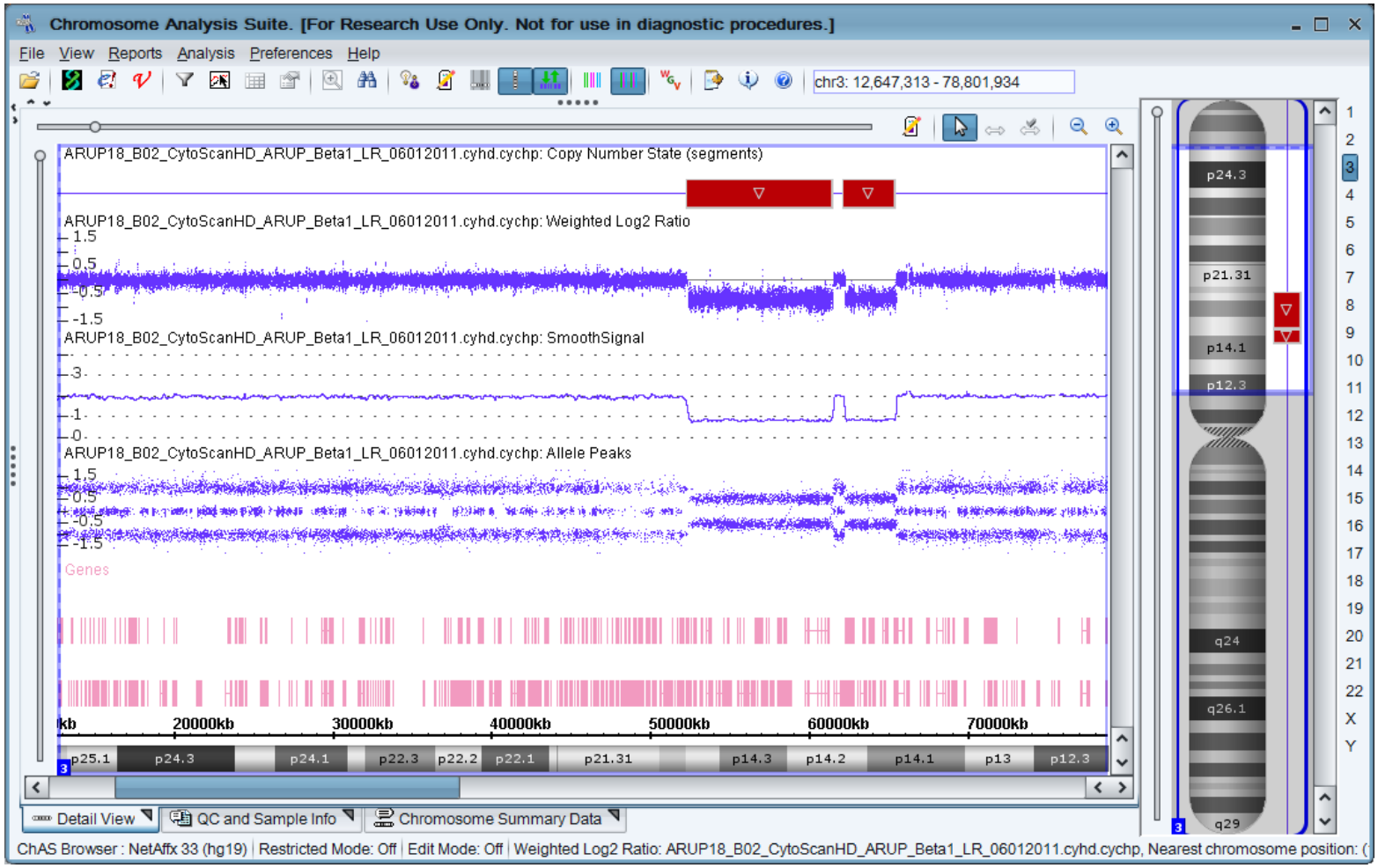
Example of two hemizygous losses



- **This example provides an illustration of two hemizygous losses on chromosome 3.**
- **These losses and the neutral structural region in the middle were all FISH confirmed**

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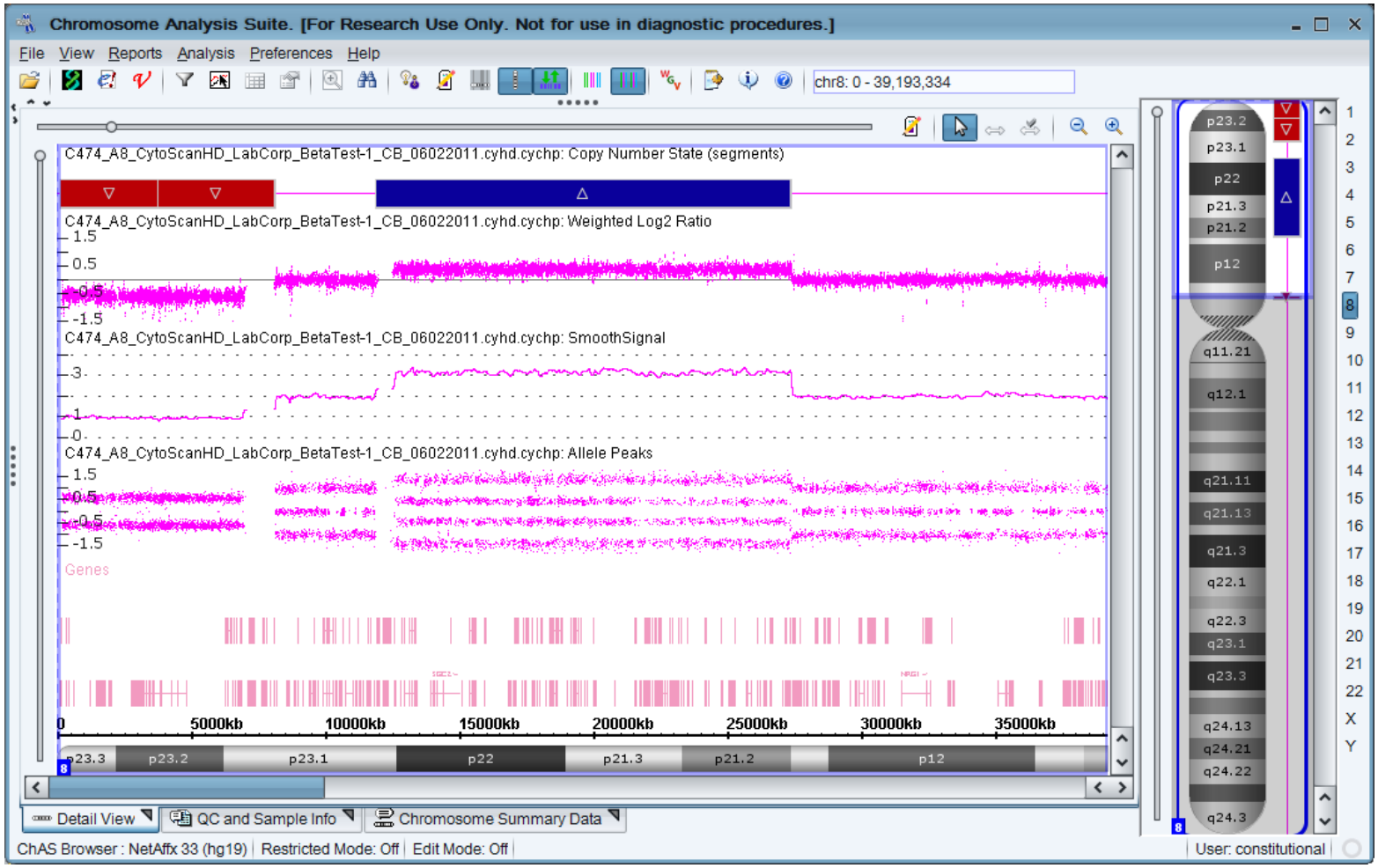
Example of hemizygous loss and gain



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

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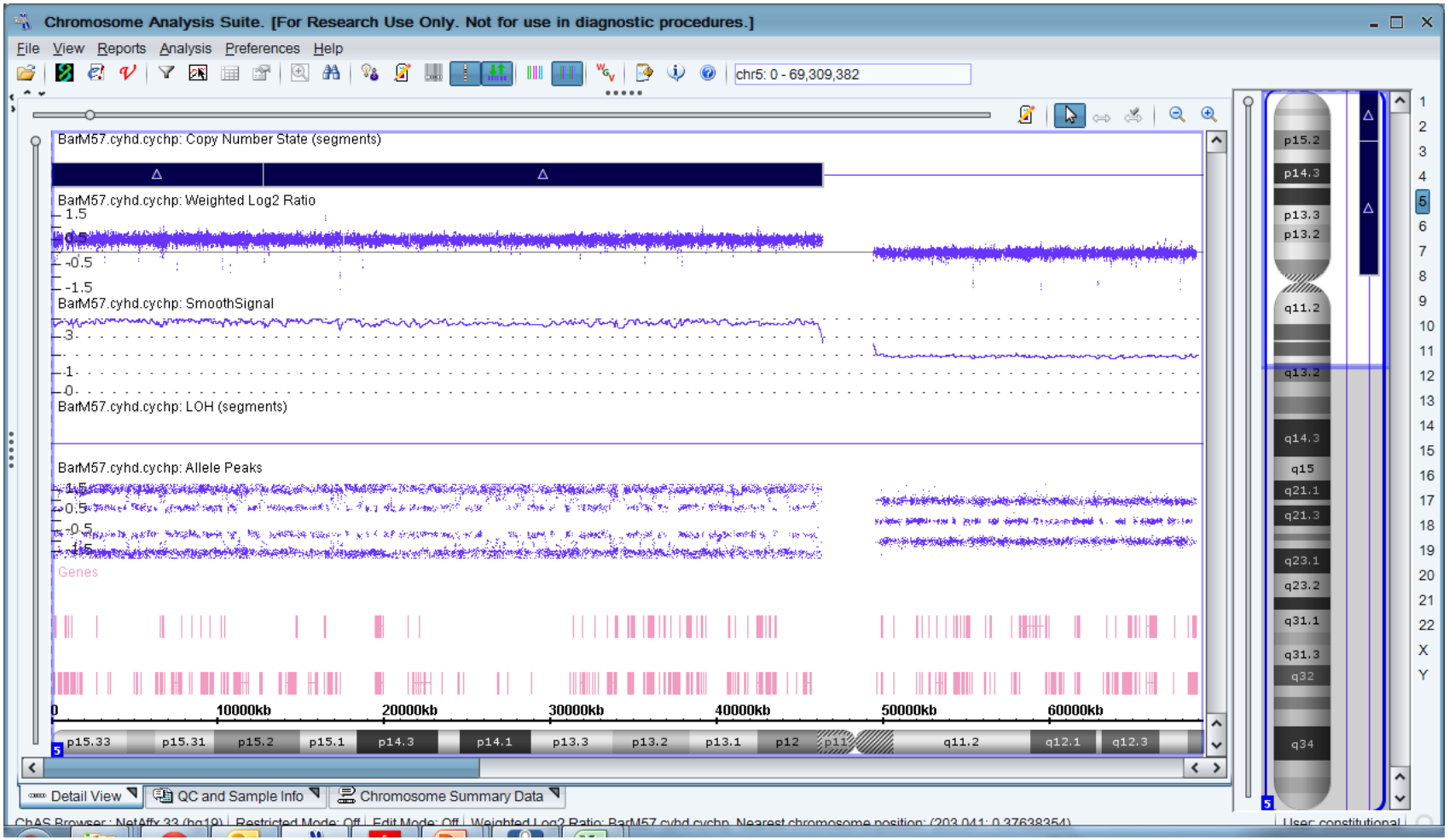
Copy number 4 Gain



- **This example provides an illustration of Copy number 4 Segment.**
- **The allelic peaks track shows that 1 allele has been triplicated and the other is in one copy**

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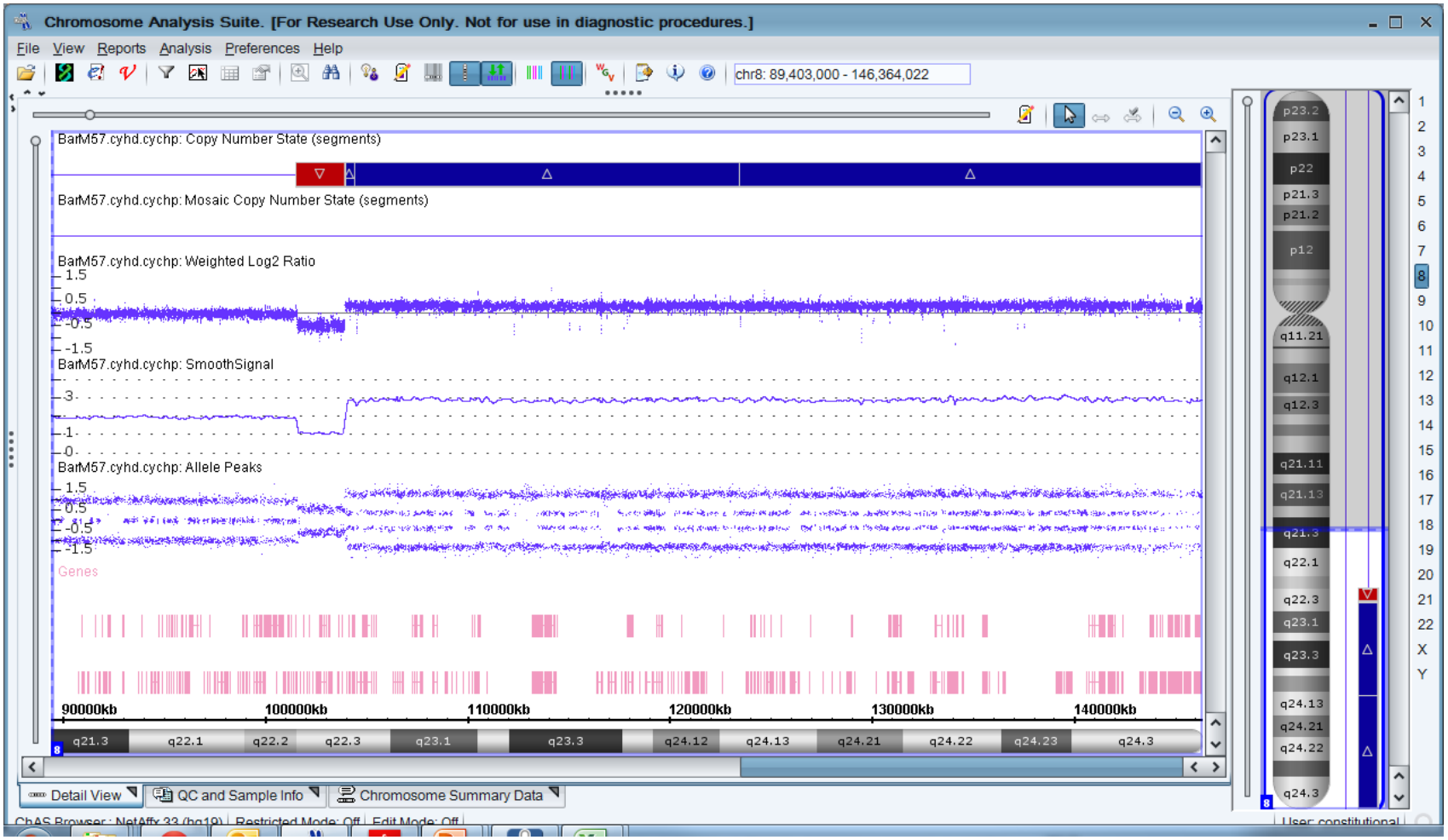
An unbalanced translocation



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

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A fresh-frozen solid tumor sample with complex chromosomal aberrations

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

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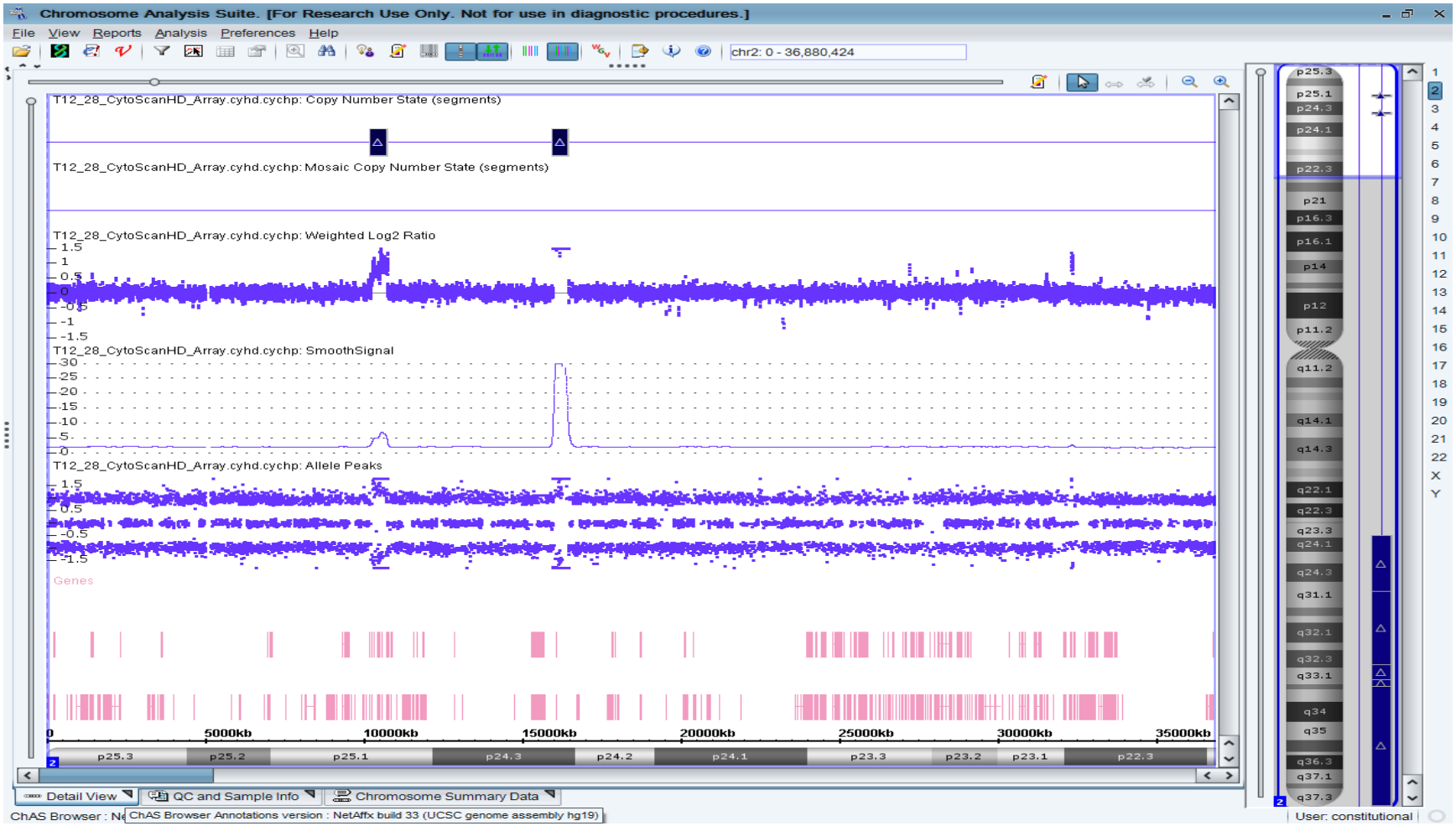
high level copy number gains/ amplifications on chromosome 2



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

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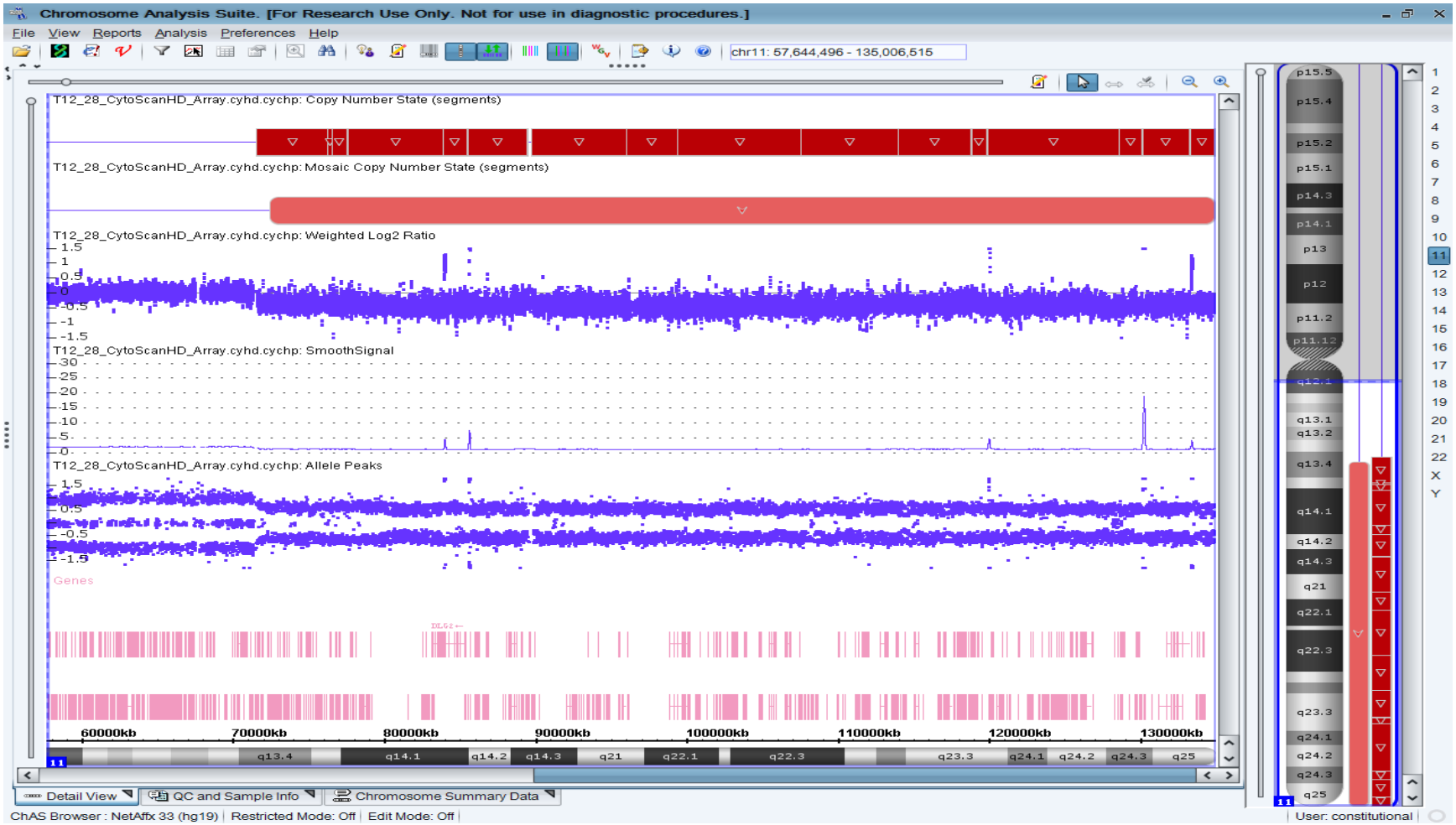
Complex rearrangement on chromosome 11



- 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

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Hemizygous loss on chromosome 13

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

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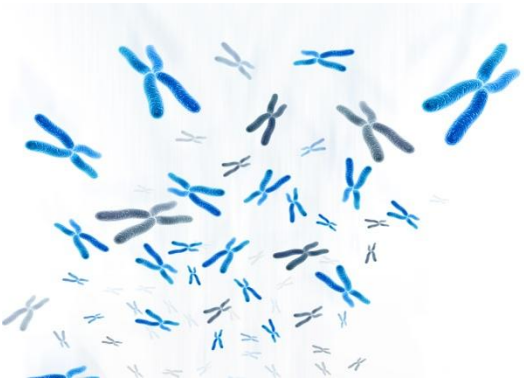
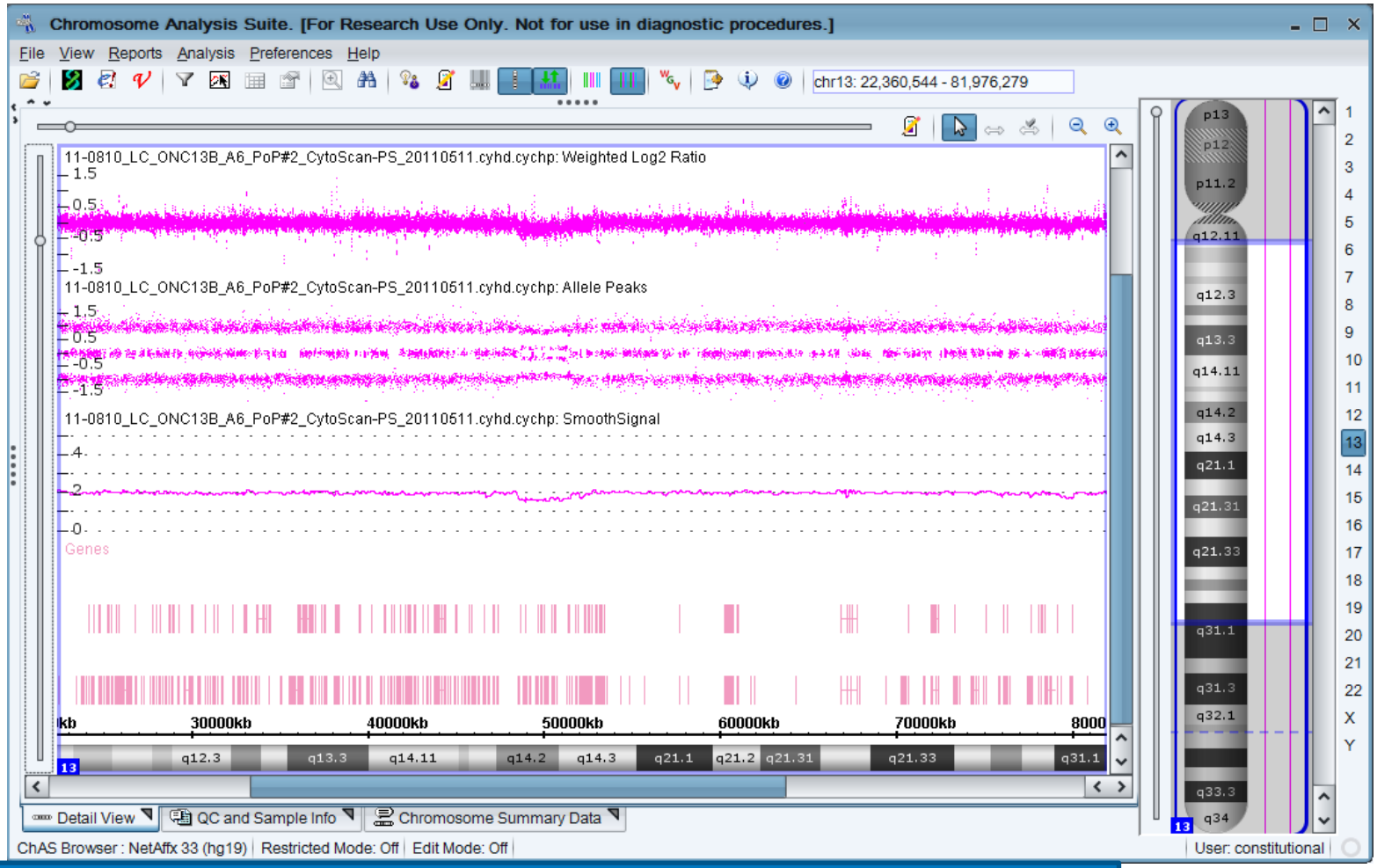


Mosaic Segments

Mosaic Loss on chromosome 13 (1/2)



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

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Mosaic Loss on chromosome 13 (2/2)

- **A mosaic segment/flag was drawn with the "Edit" mode for simplified identification**

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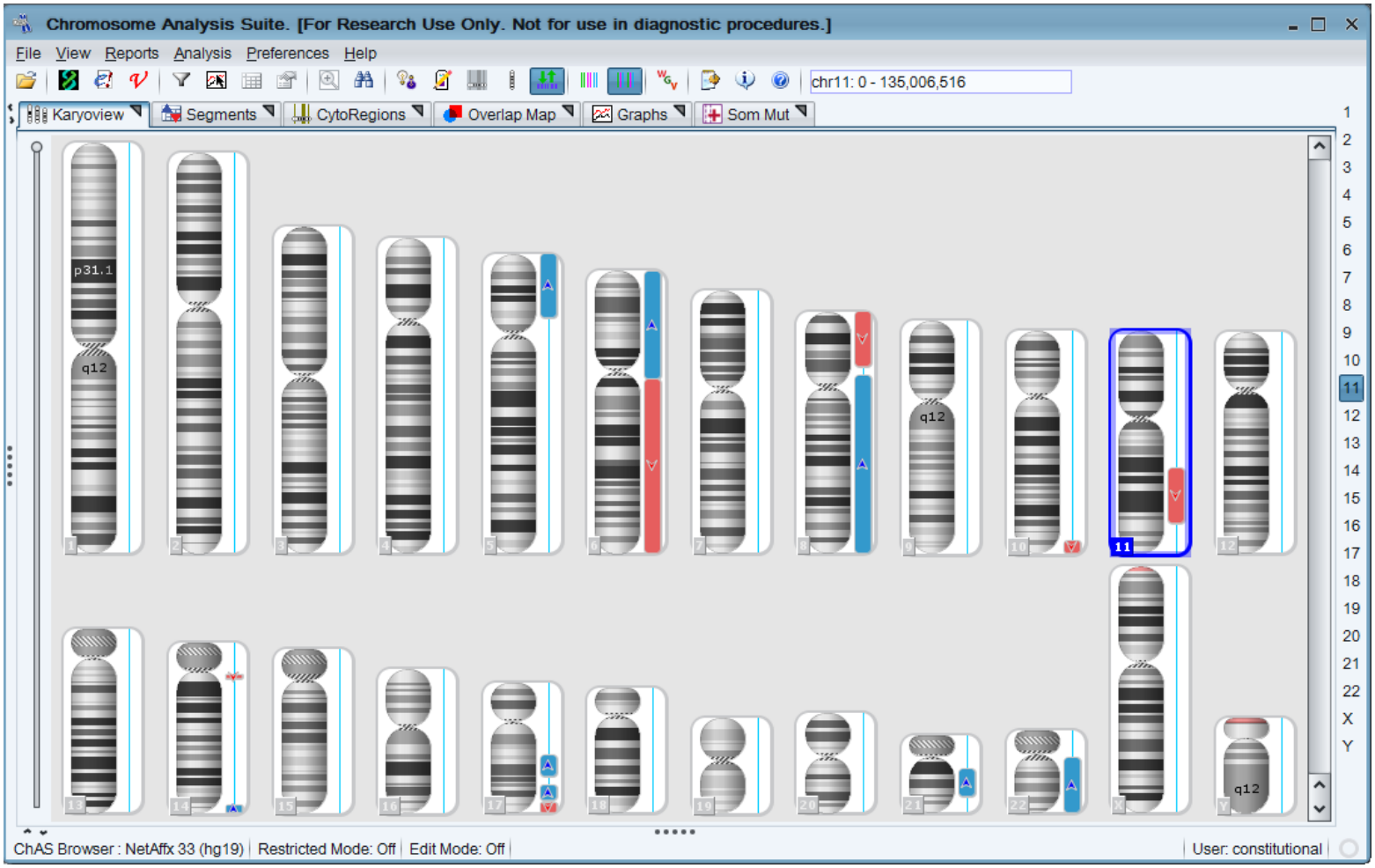
A CLL sample with many mosaic aberrations



- **This CLL sample has many educational aberration types.**

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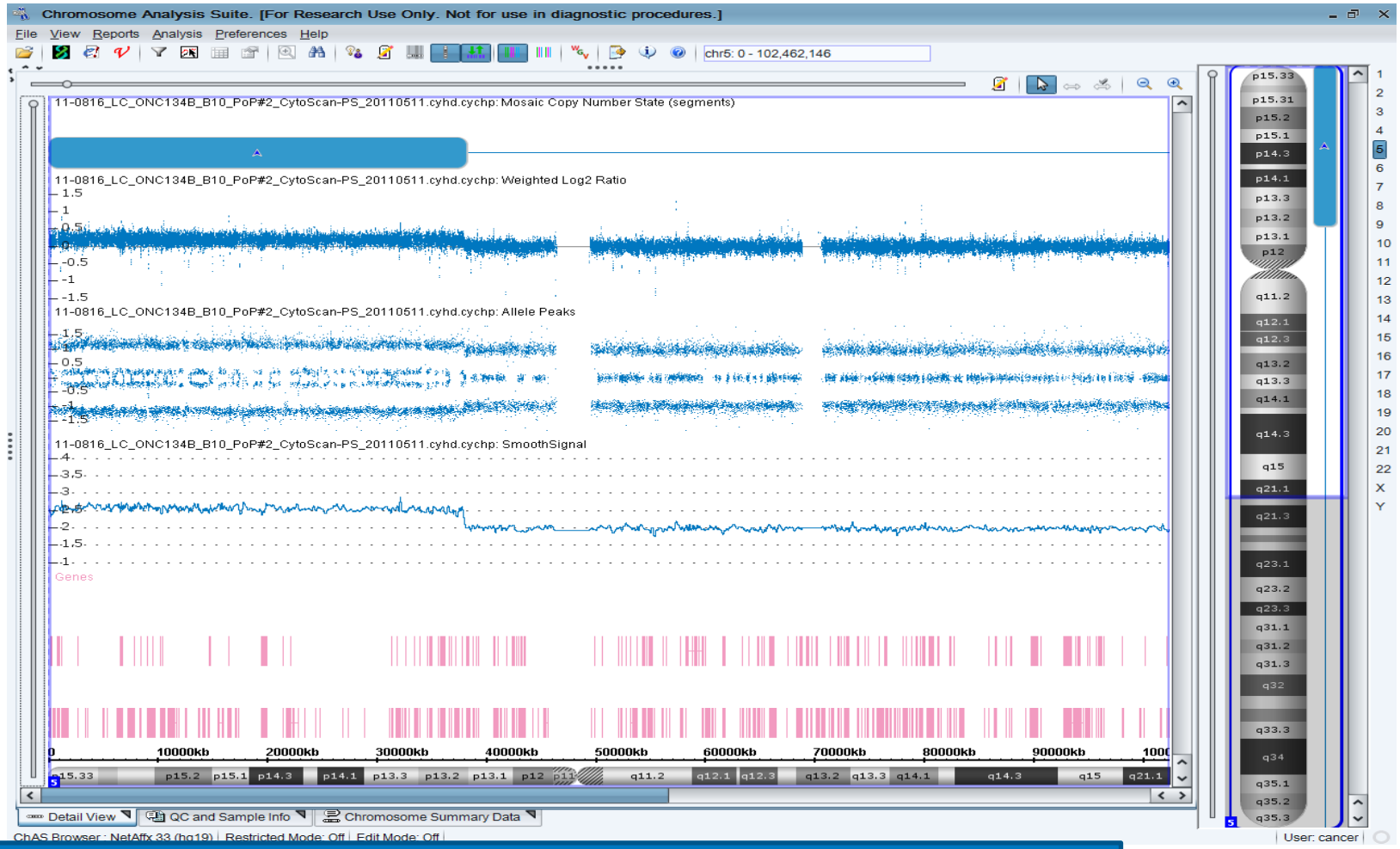
Mosaic Gain



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

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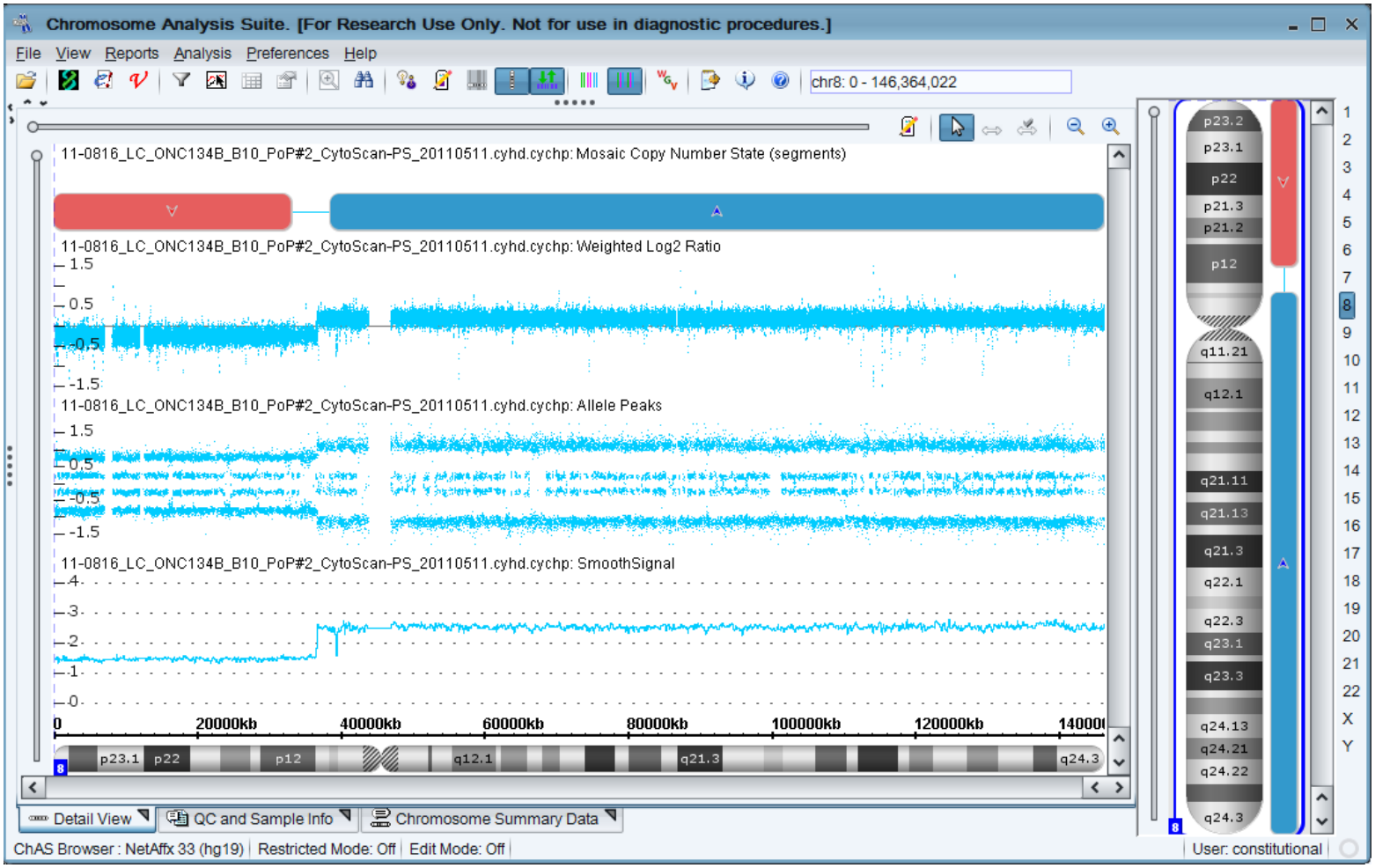
Mosaic loss and mosaic gain



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

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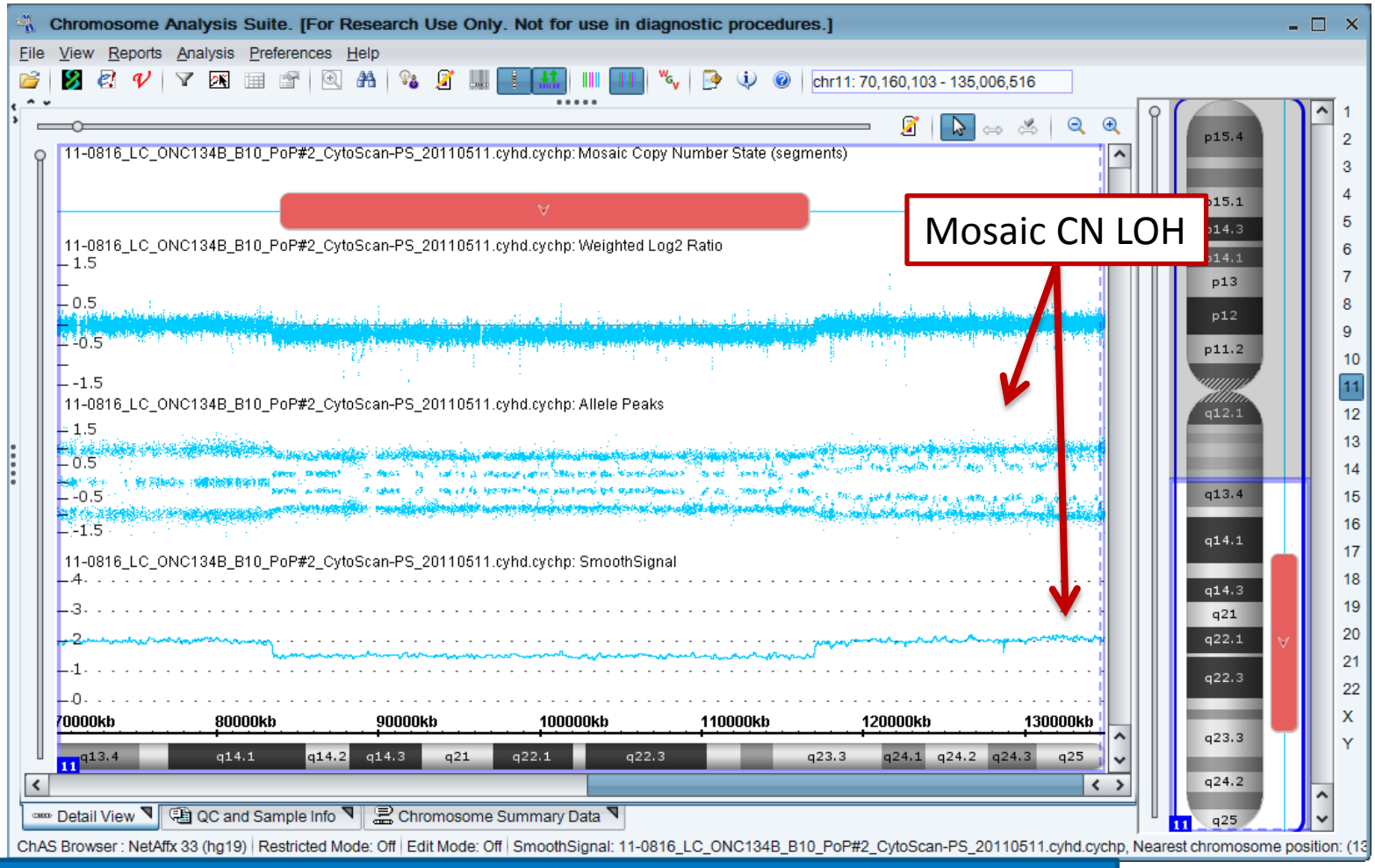
Mosaic loss and mosaic copy neutral LOH



- **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**

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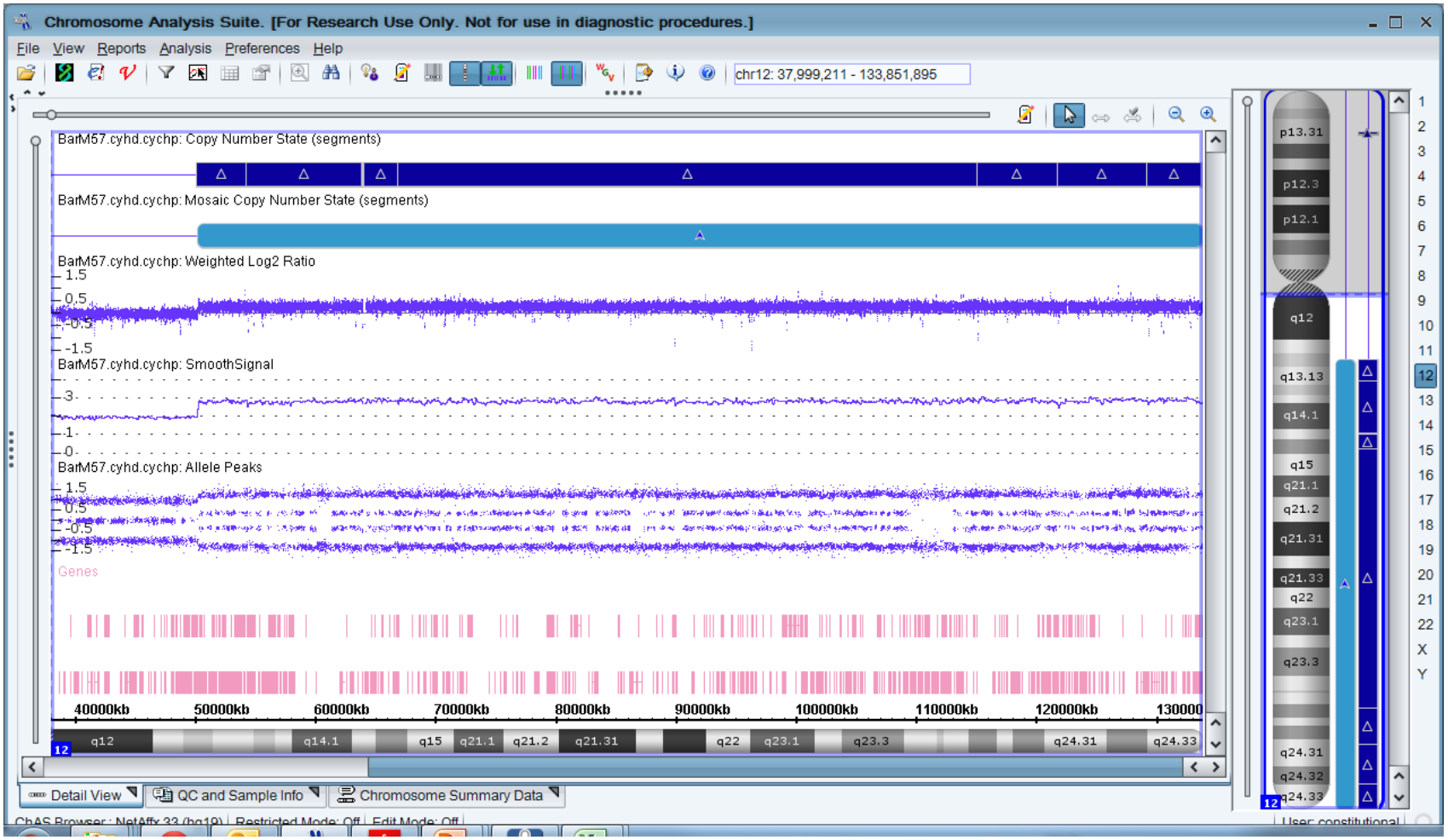
Mosaic Gain



- **This sample illustrates a mosaic gain on chromosome 12.**
- **Mosaic has been estimated at 85-90% using smooth signal and mosaic segment/flag.**

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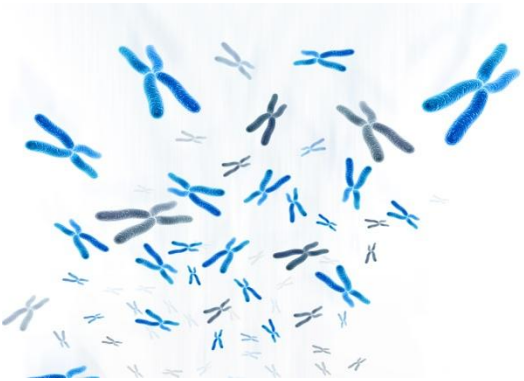

ROH/AOH/LOH Examples

Regions identical by descent
Genomic profile of consanguinity

09-420_B2_Phase4CustomerPanel_CytoScan_PS_20110228

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

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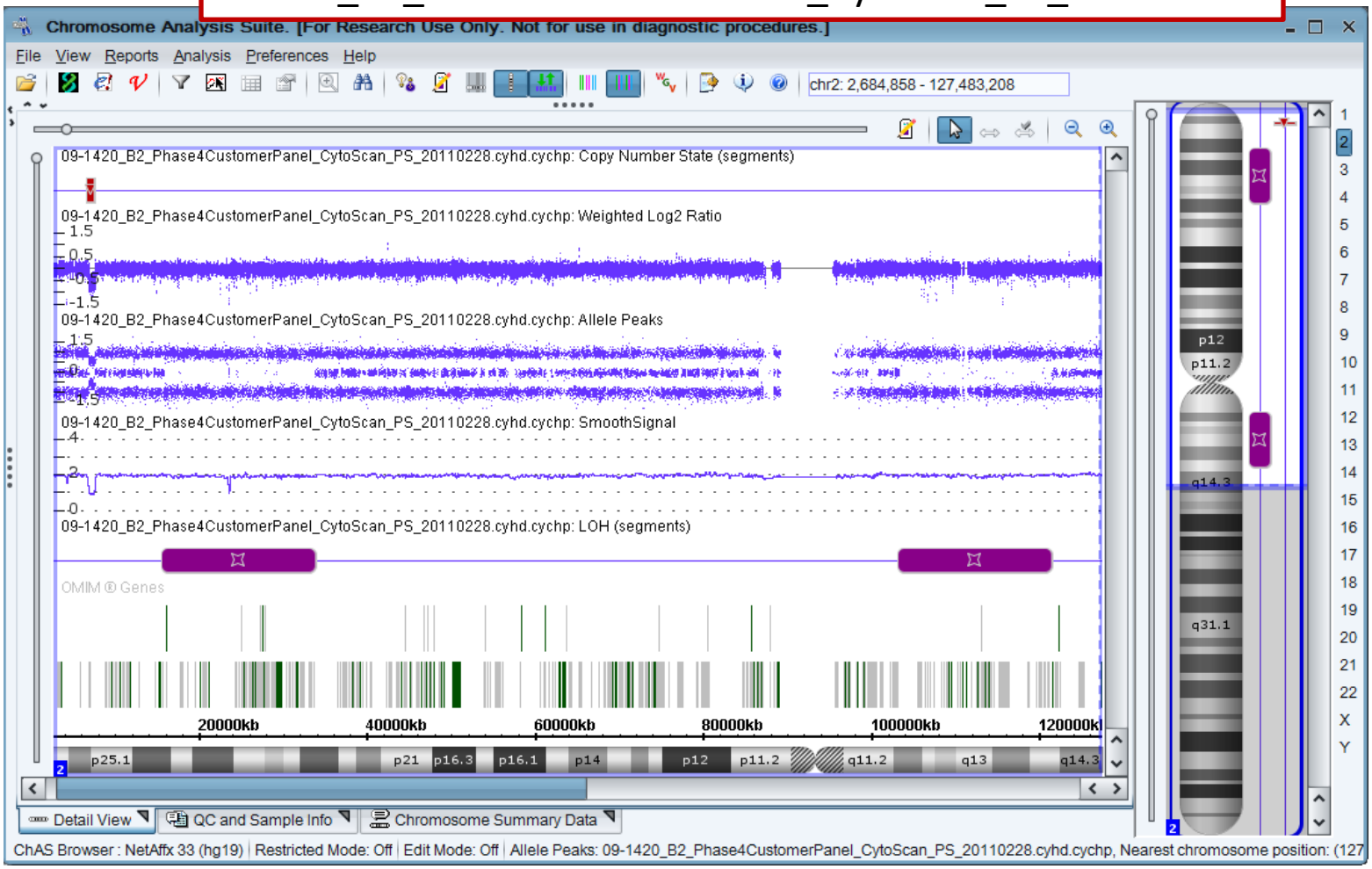
Regions identical by descent
Detailed view of chromosome 2

09-420_B2_Phase4CustomerPanel_CytoScan_PS_20110228



- **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**

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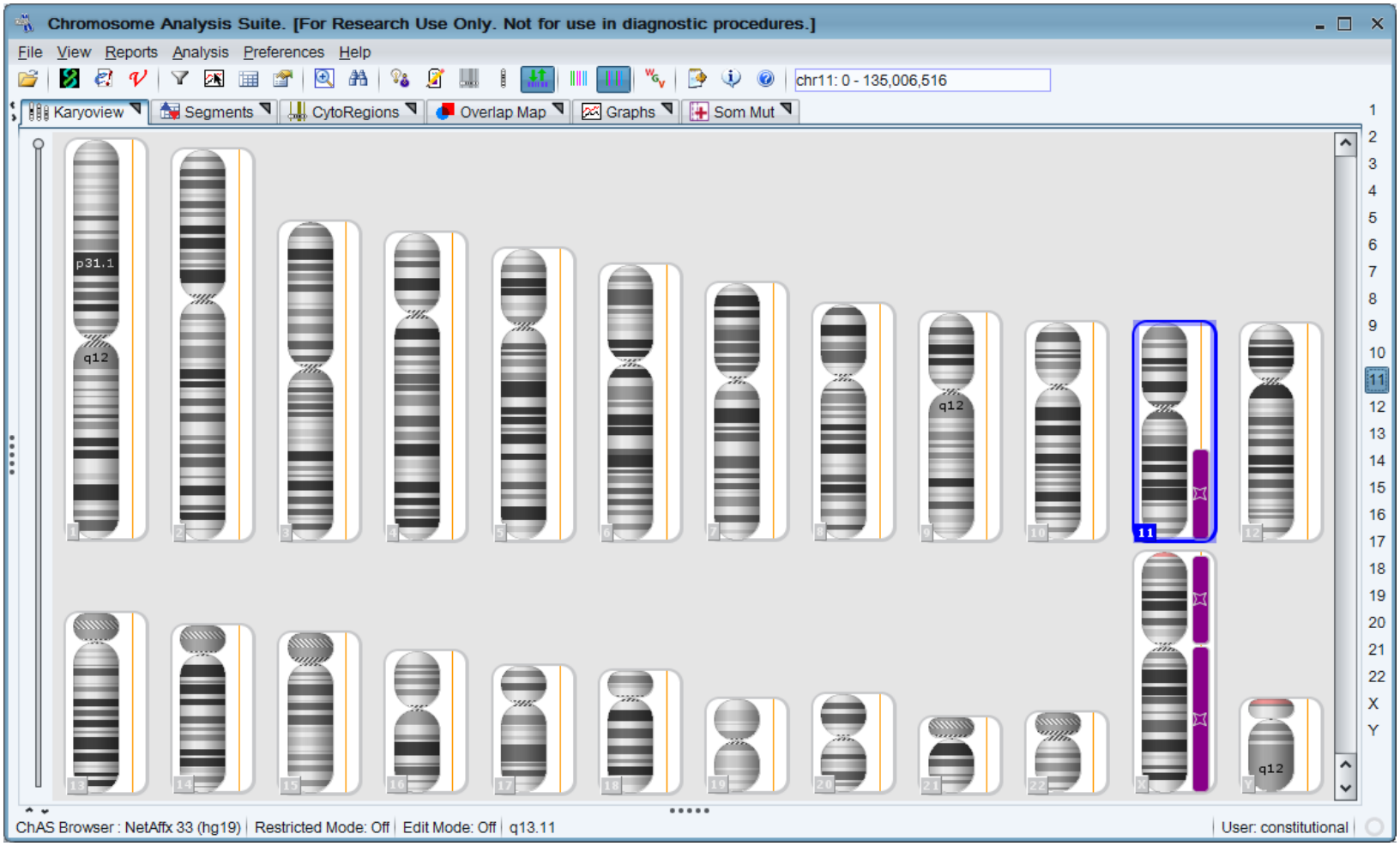
Copy neutral LOH on chromosome 11 Karyoview illustrating LOH >10 Mb



- **Only one autosomal chromosome (Chr 11) has a block of LOH greater than 10 Mb.**
- **The chromosome has blocks because is a male sample.**

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Copy neutral AOH/LOH on chromosome 11. Closer detail illustrating LOH >10 Mb

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

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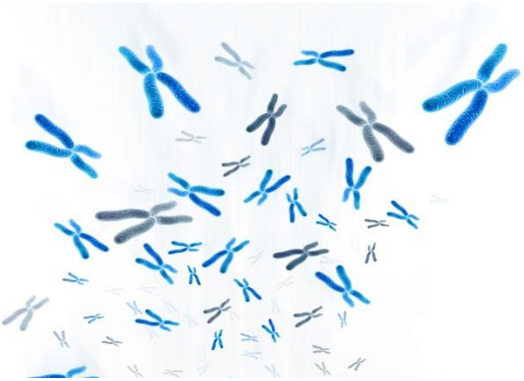

Genotypes - Trio

Mendelian Consistency Checking

Dad_Trio1.cyhd

Mom_Trio1.cyhd

Proband_Trio1.cyhd

AnalysisTy	Reference	FamilialSa	RoleValid	RoleIndexScore				
0	0	2	1	97601.66				
1	0	1	1	89006.27				
2	0	2	1	53284.64				
#%SetName=MIE								
#%Columns=9								
#%Rows=23								
Chromosc	Display	MarkerCo	MIE-Trio	MIE-Mat	MIE-Pat	Percent-T	Percent-M	Percent-Pat-MIE
1	1	56482	119	27	31	0.210687	0.047803	0.054885
2	2	62283	115	31	28	0.184641	0.049773	0.044956
3	3	52106	83	23	24	0.159291	0.044141	0.04606
4	4	49517	86	21	34	0.173678	0.04241	0.068663
5	5	46205	84	27	26	0.181799	0.058435	0.056271
6	6	51944	125	33	49	0.240644	0.06353	0.094332
7	7	46413	109	28	33	0.234848	0.060328	0.071101
8	8	38796	61	14	20	0.157233	0.036086	0.051552
9	9	30622	62	21	20	0.202469	0.068578	0.065313
10	10	35472	66	18	22	0.186062	0.050744	0.062021
11	11	38846	83	23	24	0.213664	0.059208	0.061782
12	12	33424	66	21	20	0.197463	0.062829	0.059837
13	13	27733	61	34	9	0.219955	0.122598	0.032452
14	14	26983	62	16	14	0.229774	0.059297	0.051885
15	15	24981	1746	0	1722	6.989312	0	6.893239
16	16	20915	31	13	9	0.148219	0.062156	0.043031
17	17	17465	36	18	10	0.206127	0.103063	0.057257
18	18	20583	43	12	15	0.20891	0.058301	0.072876
19	19	10921	28	8	7	0.256387	0.073253	0.064097
20	20	16200	27	6	8	0.166667	0.037037	0.049383
21	21	10382	25	7	10	0.240801	0.067424	0.096321
22	22	8780	22	4	12	0.250569	0.045558	0.136674
X	X	22104	37	4	19	0.167391	0.018096	0.085957

Errors trio | **Errors Mat** | **Errors Pat**

- **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 het-UPD**

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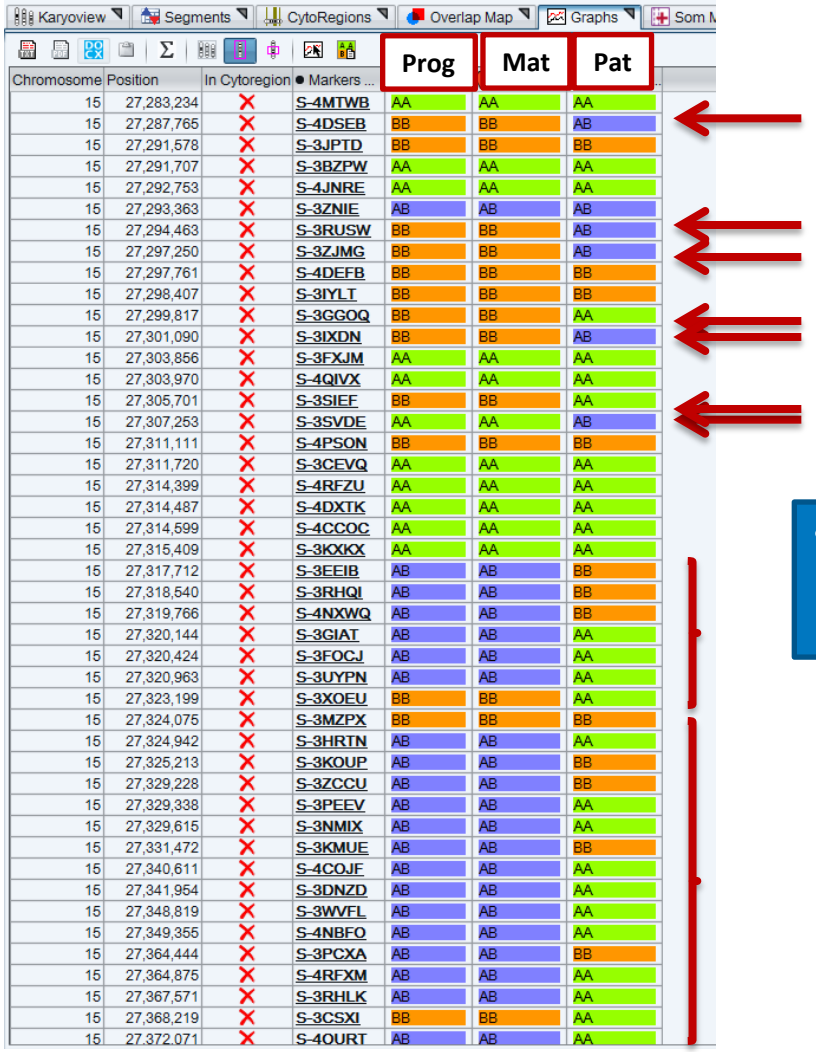
Genotype calls on chromosome 15



Chromosome	Position	In Cytoregion	Markers ...	Prog	Mat	Pat
15	27,283,234	X	S-4MTWB	AA	AA	AA
15	27,287,765	X	S-4DSEB	BB	BB	AB
15	27,291,578	X	S-3JPTD	BB	BB	BB
15	27,291,707	X	S-3BZPW	AA	AA	AA
15	27,292,753	X	S-4JNRE	AA	AA	AA
15	27,293,363	X	S-3ZNIE	AB	AB	AB
15	27,294,463	X	S-3RUSW	BB	BB	AB
15	27,297,250	X	S-3ZJMG	BB	BB	AB
15	27,297,761	X	S-4DEFB	BB	BB	BB
15	27,298,407	X	S-3IYLT	BB	BB	BB
15	27,299,817	X	S-3GGOQ	BB	BB	AA
15	27,301,090	X	S-3IXDN	BB	BB	AB
15	27,303,856	X	S-3FXJM	AA	AA	AA
15	27,303,970	X	S-4QIVX	AA	AA	AA
15	27,305,701	X	S-3SIEF	BB	BB	AA
15	27,307,253	X	S-3SVDE	AA	AA	AB
15	27,311,111	X	S-4PSON	BB	BB	BB
15	27,311,720	X	S-3CEVQ	AA	AA	AA
15	27,314,399	X	S-4RFZU	AA	AA	AA
15	27,314,487	X	S-4DXTK	AA	AA	AA
15	27,314,599	X	S-4CCOC	AA	AA	AA
15	27,315,409	X	S-3KXKX	AA	AA	AA
15	27,317,712	X	S-3EEIB	AB	AB	BB
15	27,318,540	X	S-3RHQI	AB	AB	BB
15	27,319,766	X	S-4NXWQ	AB	AB	BB
15	27,320,144	X	S-3GIAT	AB	AB	AA
15	27,320,424	X	S-3FOCJ	AB	AB	AA
15	27,320,963	X	S-3UYPN	AB	AB	AA
15	27,323,199	X	S-3XOEU	BB	BB	AA
15	27,324,075	X	S-3MZPX	BB	BB	BB
15	27,324,942	X	S-3HRTN	AB	AB	AA
15	27,325,213	X	S-3KOUP	AB	AB	BB
15	27,329,228	X	S-3ZCCU	AB	AB	BB
15	27,329,338	X	S-3PEEV	AB	AB	AA
15	27,329,615	X	S-3NMIX	AB	AB	AA
15	27,331,472	X	S-3KMUE	AB	AB	BB
15	27,340,611	X	S-4COJF	AB	AB	AA
15	27,341,954	X	S-3DNZD	AB	AB	AA
15	27,348,819	X	S-3WVFL	AB	AB	AA
15	27,349,355	X	S-4NBFO	AB	AB	AA
15	27,364,444	X	S-3PCXA	AB	AB	BB
15	27,364,875	X	S-4RFXM	AB	AB	AA
15	27,367,571	X	S-3RHLK	AB	AB	AA
15	27,368,219	X	S-3CSXI	BB	BB	AA
15	27,372,071	X	S-4OURT	AB	AB	AA

Dad_Trio1.cyhd

Mom_Trio1.cyhd

Proband_Trio1.cyhd

- **Chromosome 15 genotypes show how the UPD is from Mat origin and is hetero UPD (most genotypes are the same in Mom and proband and different in Dad)**

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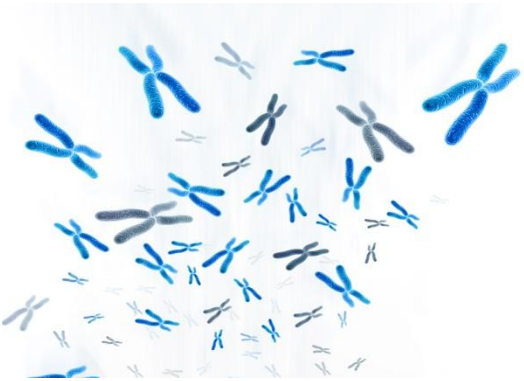
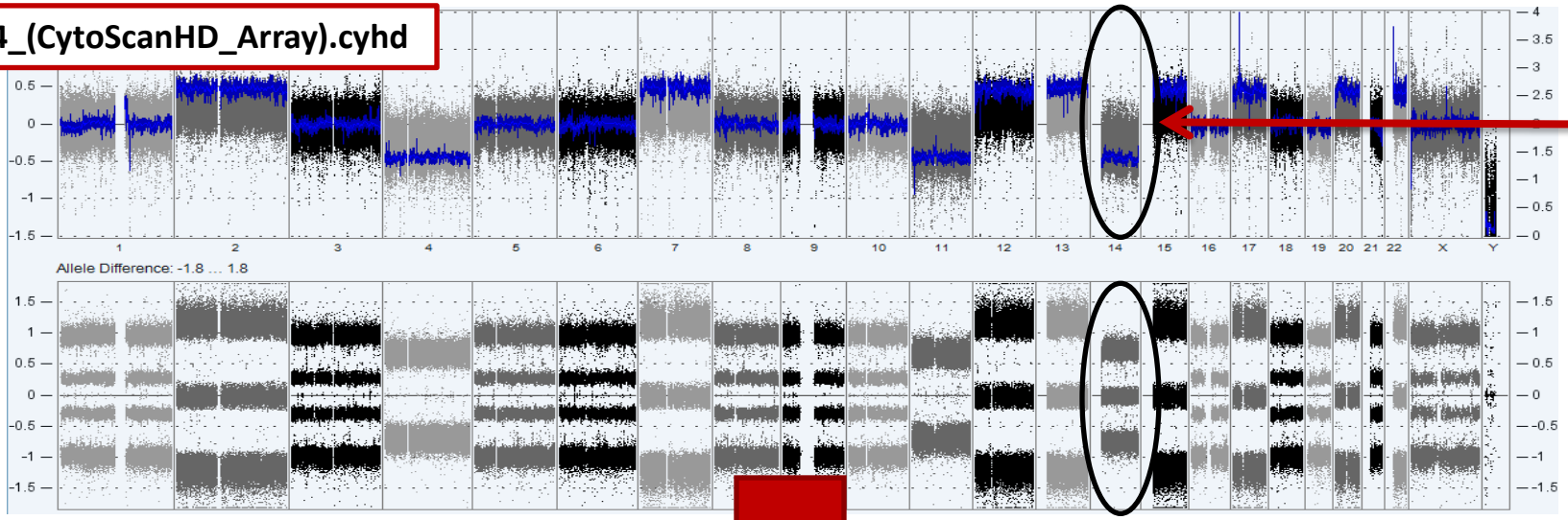


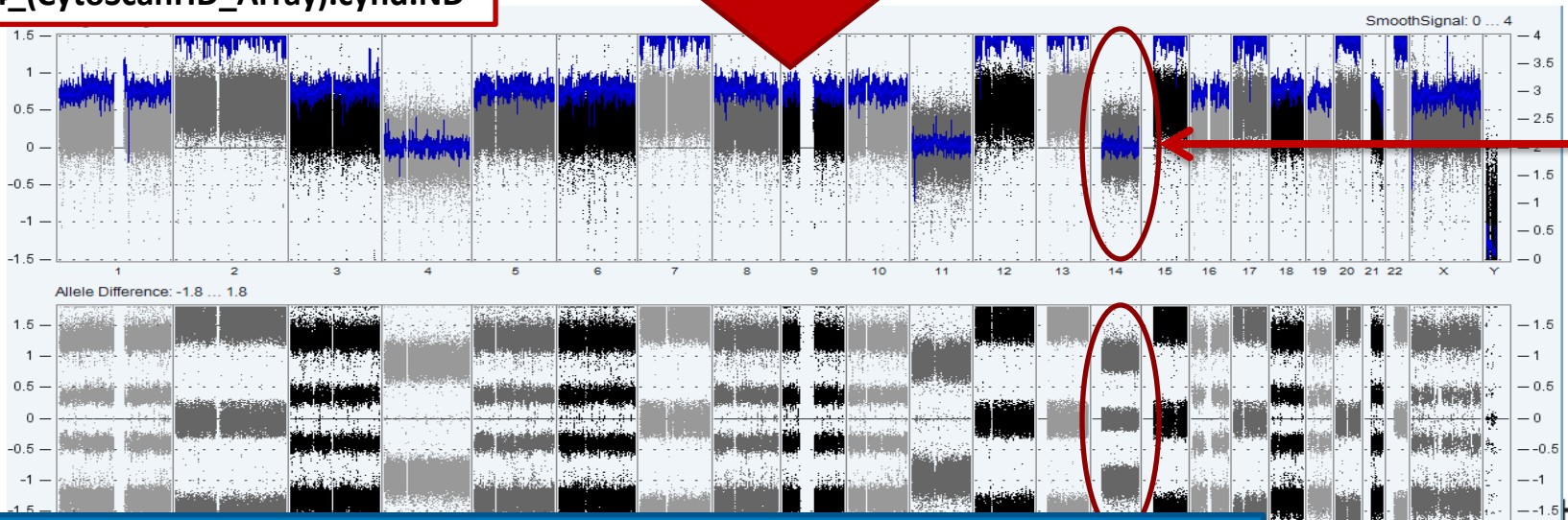
Non-Diploid Normalization

A non-diploid sample processed with the Non-D algorithm

T12_24_(CytoScanHD_Array).cyhd



T12_24_(CytoScanHD_Array).cyhd.ND



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

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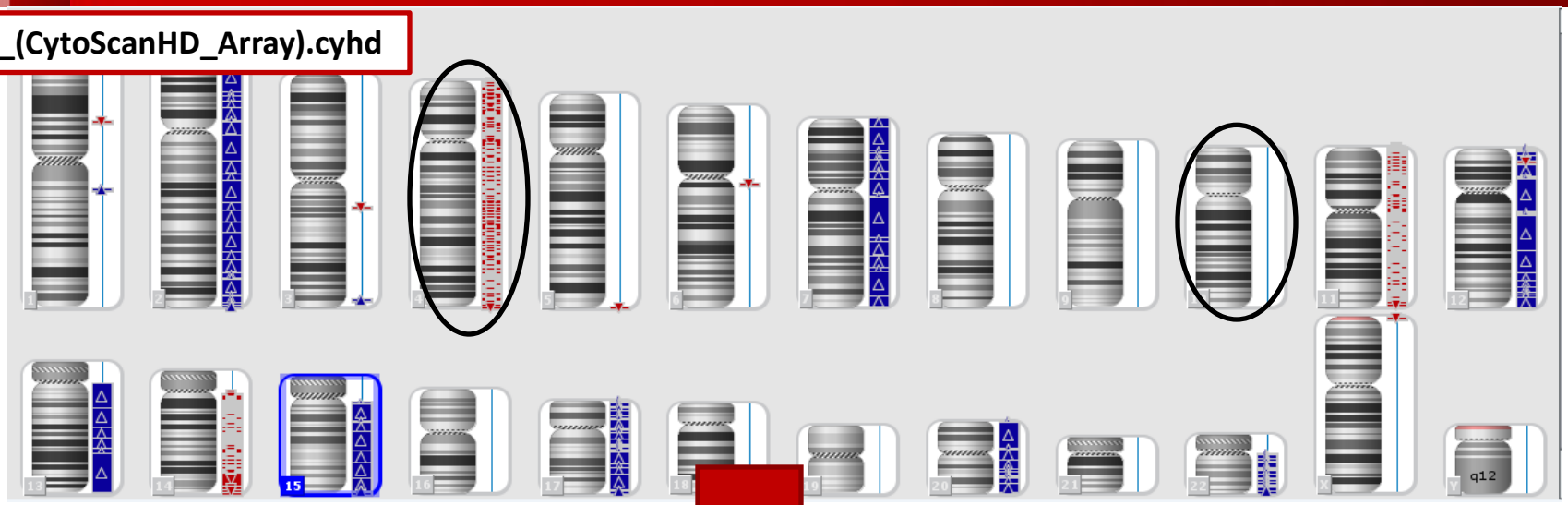
A non-diploid sample processed with the Non-D algorithm – different view

T12_24_(CytoScanHD_Array).cyhd



T12_24_(CytoScanHD_Array).cyhd.ND

- **This sample (T12-24) had several aberrations and hiperdiploid status. The baseline was not set correctly. The ND algorithm successfully corrects the baseline and calls. ~74 chromosomes**

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