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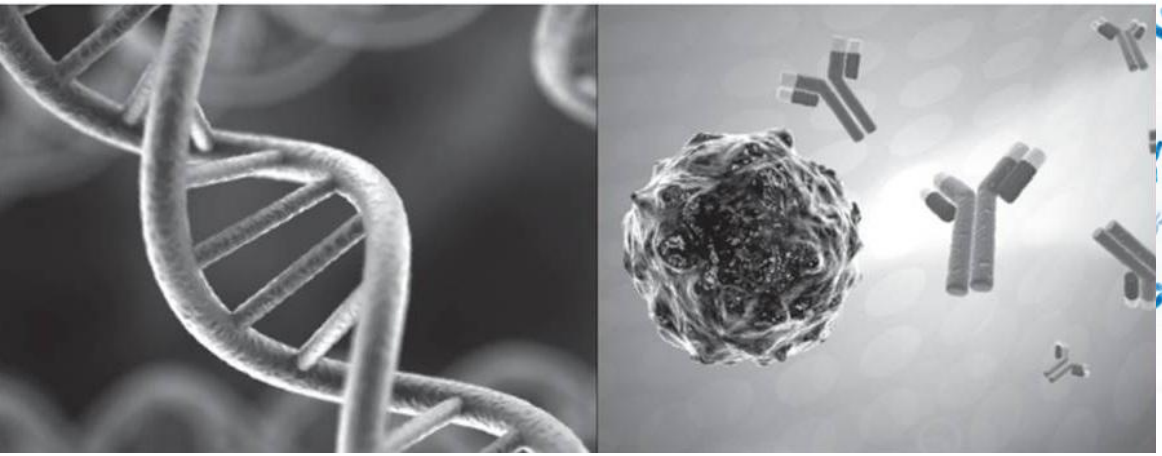

CytoScan® Cytogenetics Suite

CytoScan Optima

Educational Examples of Different Aberration types

eBioscience GeneChip Panomics USB

Summary

- A set of different samples analyzed with CytoScan Optima Suite is provided for educational proposes.

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

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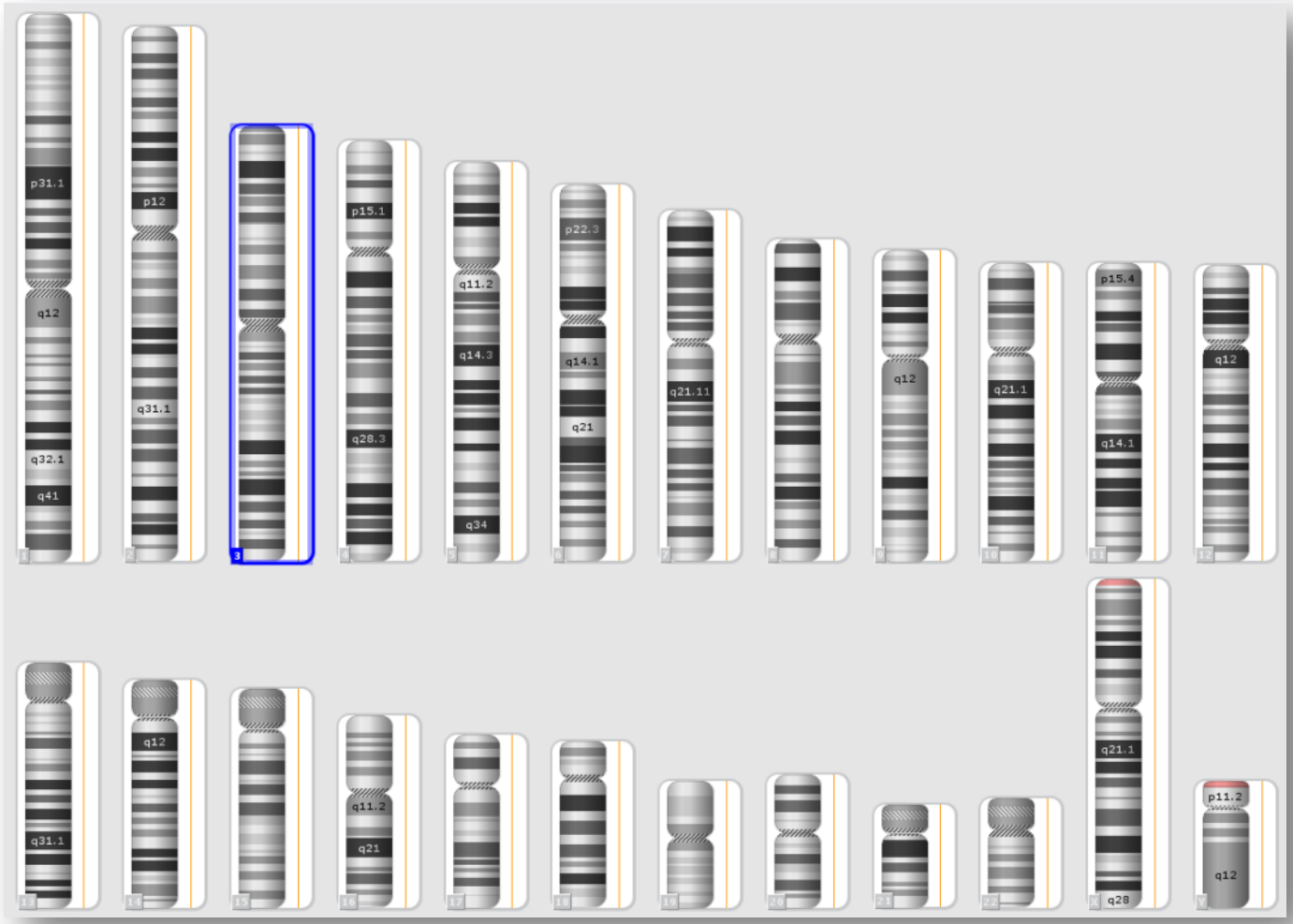
A0014084: Normal Sample



- **This example provides an illustration of a normal sample.**
- **No aberrations are detected.**

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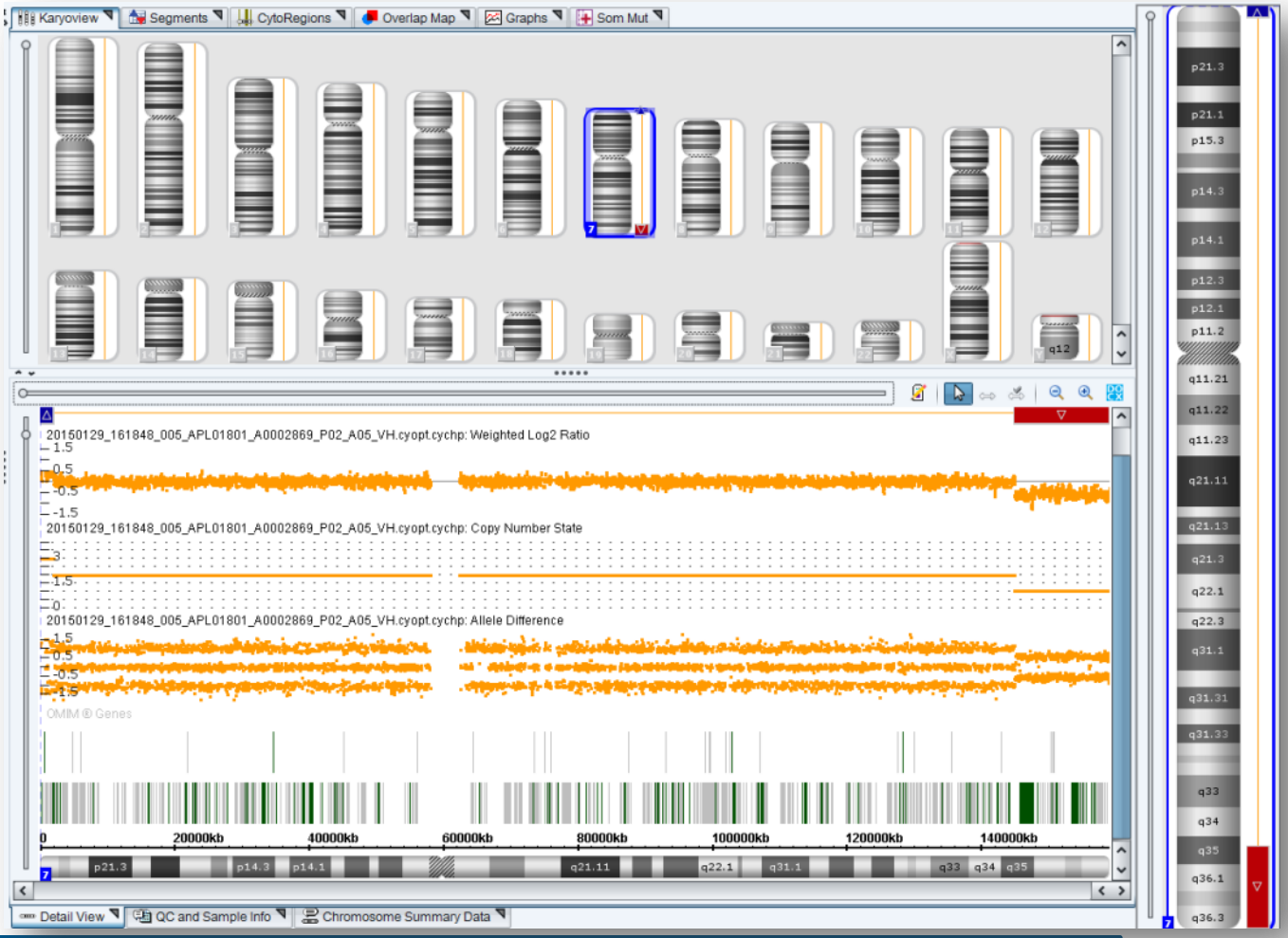
A0002869: Hemizygous loss and gain on Chromosome 7



- **This example illustrates copy number = 1, 2, and 3 on Chromosome 7:**
  - **A subtelomeric gain on the p-arm and a subtelomeric loss on the q arm.**

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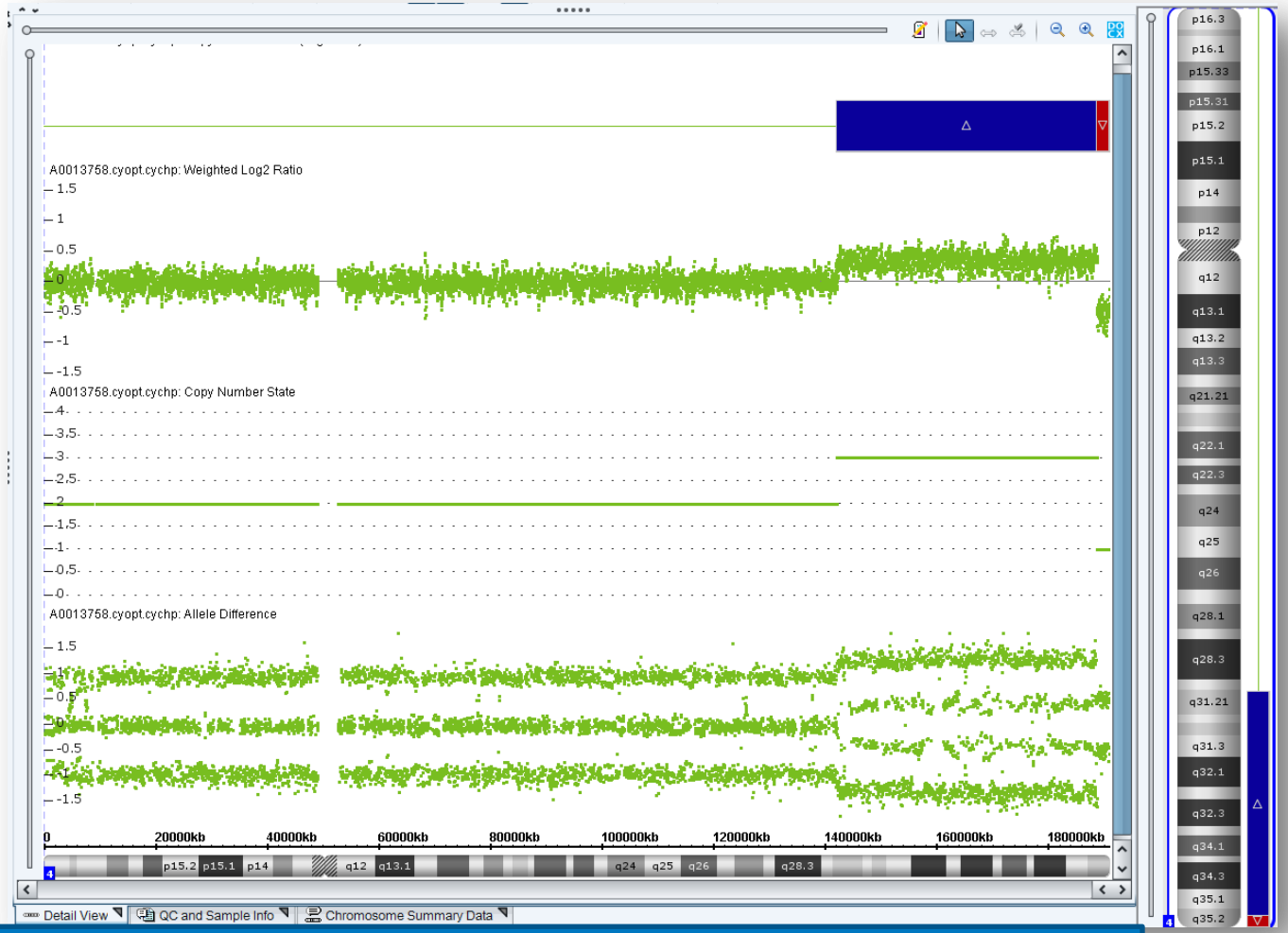
A0013758:Hemizygous loss and gain on Chromosome 4 on a POC sample



- **This example also illustrates copy number = 1, 2, and 3 on Chromosome 4:**
  - **A 46 MB gain is followed by a 2.2 MB loss**

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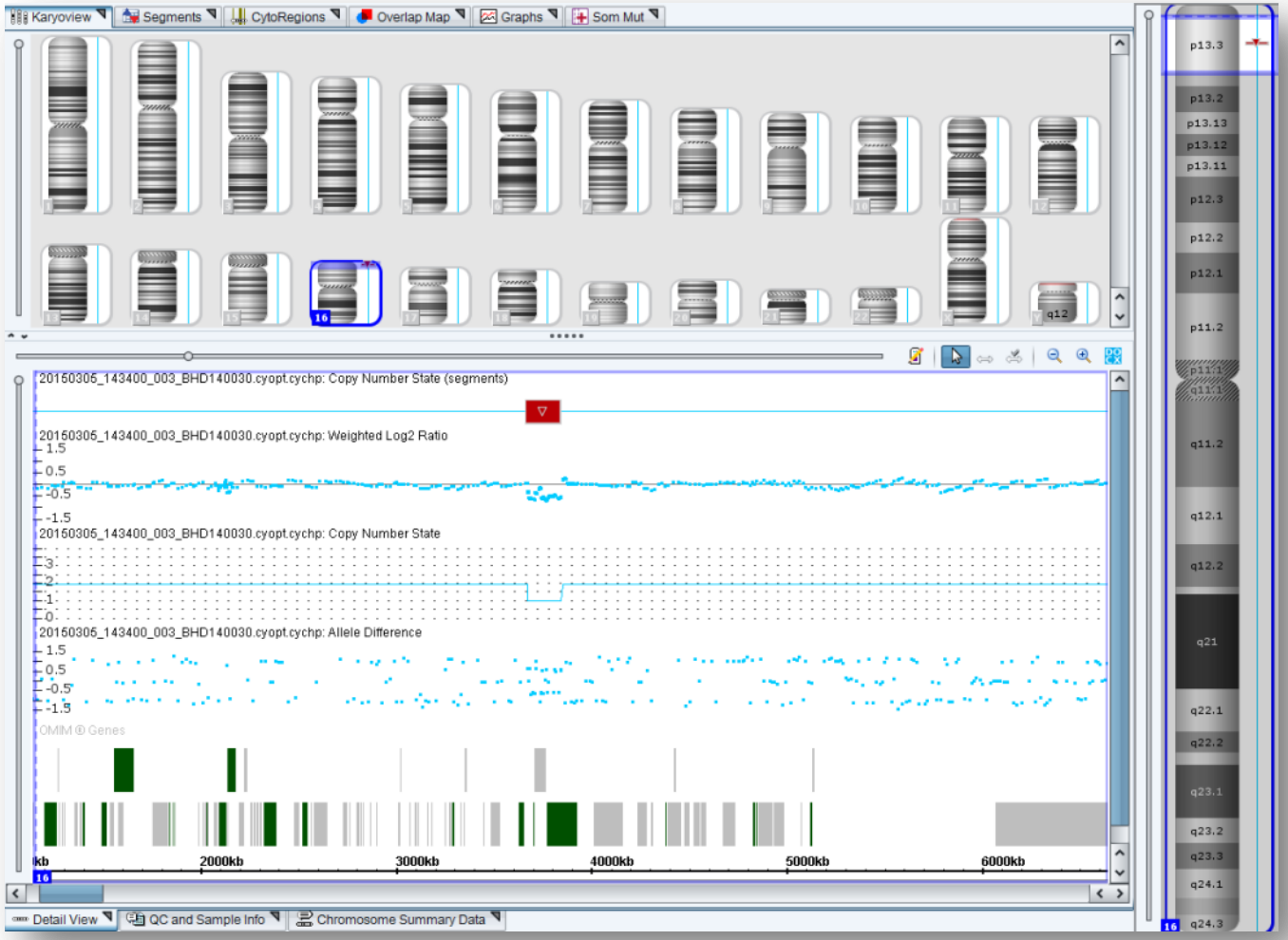
143400: 170kb del in target gene (CREBBP)



- **This example also illustrates a small copy number loss on one of the 396 regions with increased coverage**

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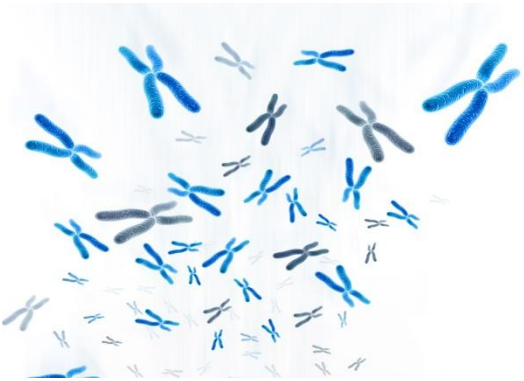

Mosaic Segments

130086: Mosaic Gain on chromosome 1

- This sample represents a mosaic gain in the same region as the previous sample
- 50% mosaic is visible on the smooth signal and allele peaks tracks.

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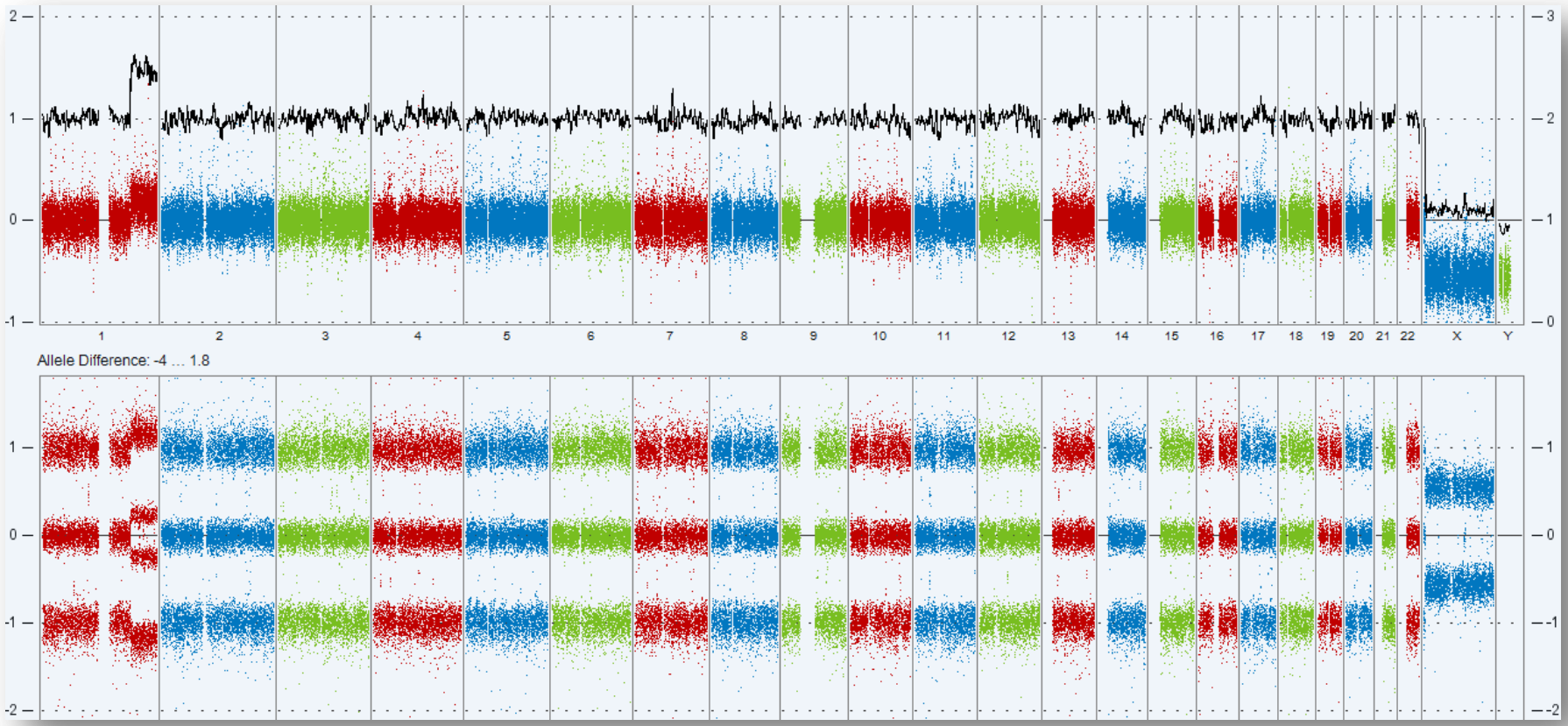
130086: Mosaic Gain on chromosome 1



- The same sample visualized in whole-genome view
- 50% mosaic is clearly visible on the allele difference track and smooth signal

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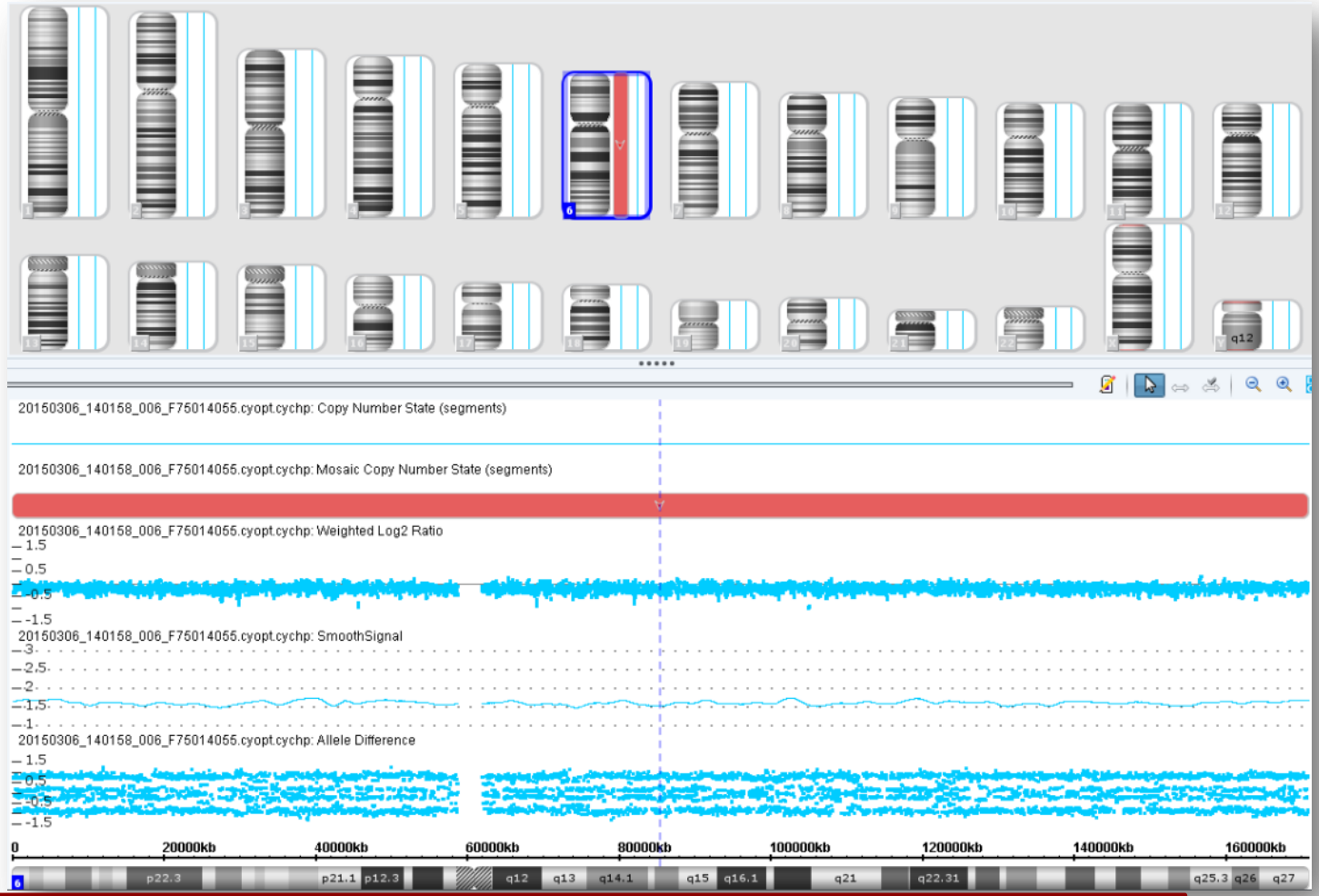
14158: Mosaic monosomy 6 (~35%) on a POC sample



- **This POC sample represents a mosaic loss example**
- **35% mosaic is visible on the smooth signal and allele peaks tracks.**

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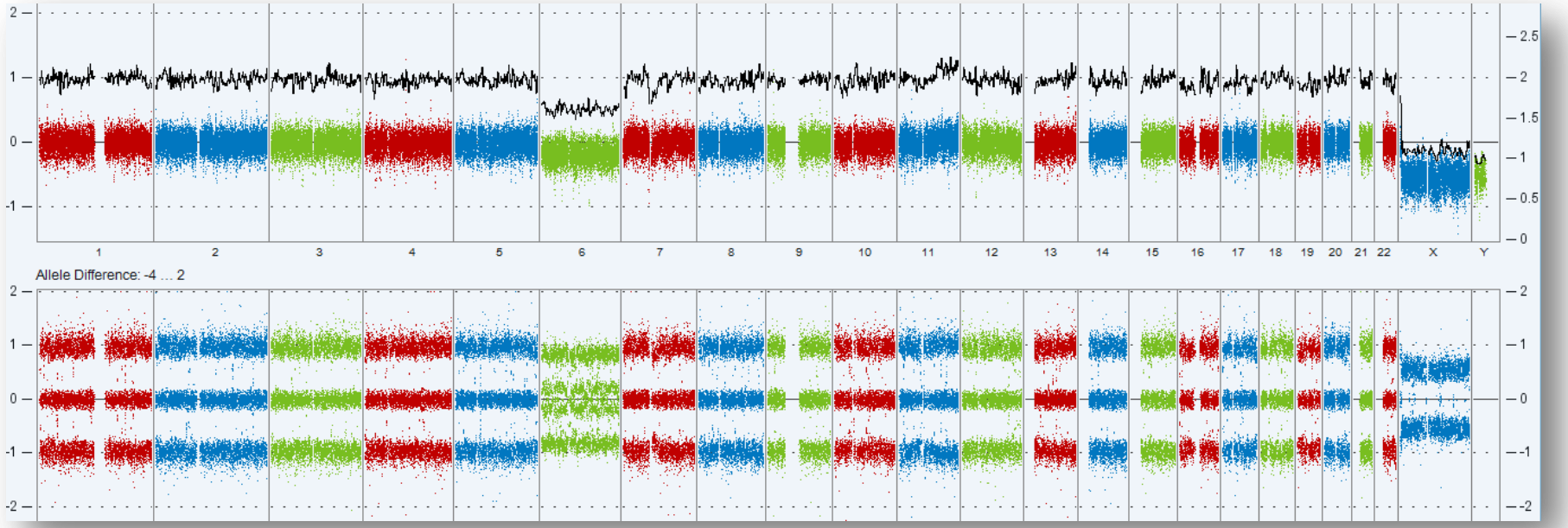
14158: Mosaic monosomy 6 (~35%) on a POC sample



- **Whole-genome view aids in the identification of the mosaic monosomy 6**
- **35% mosaic is clearly visible on the allele difference track and smooth signal**

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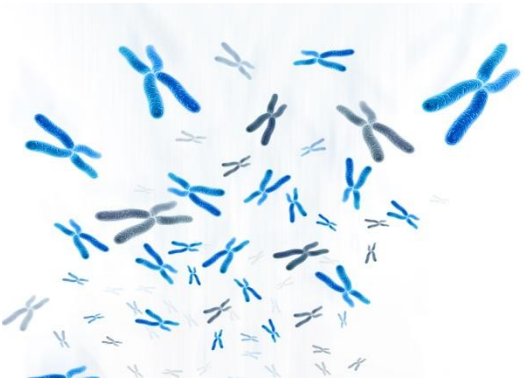
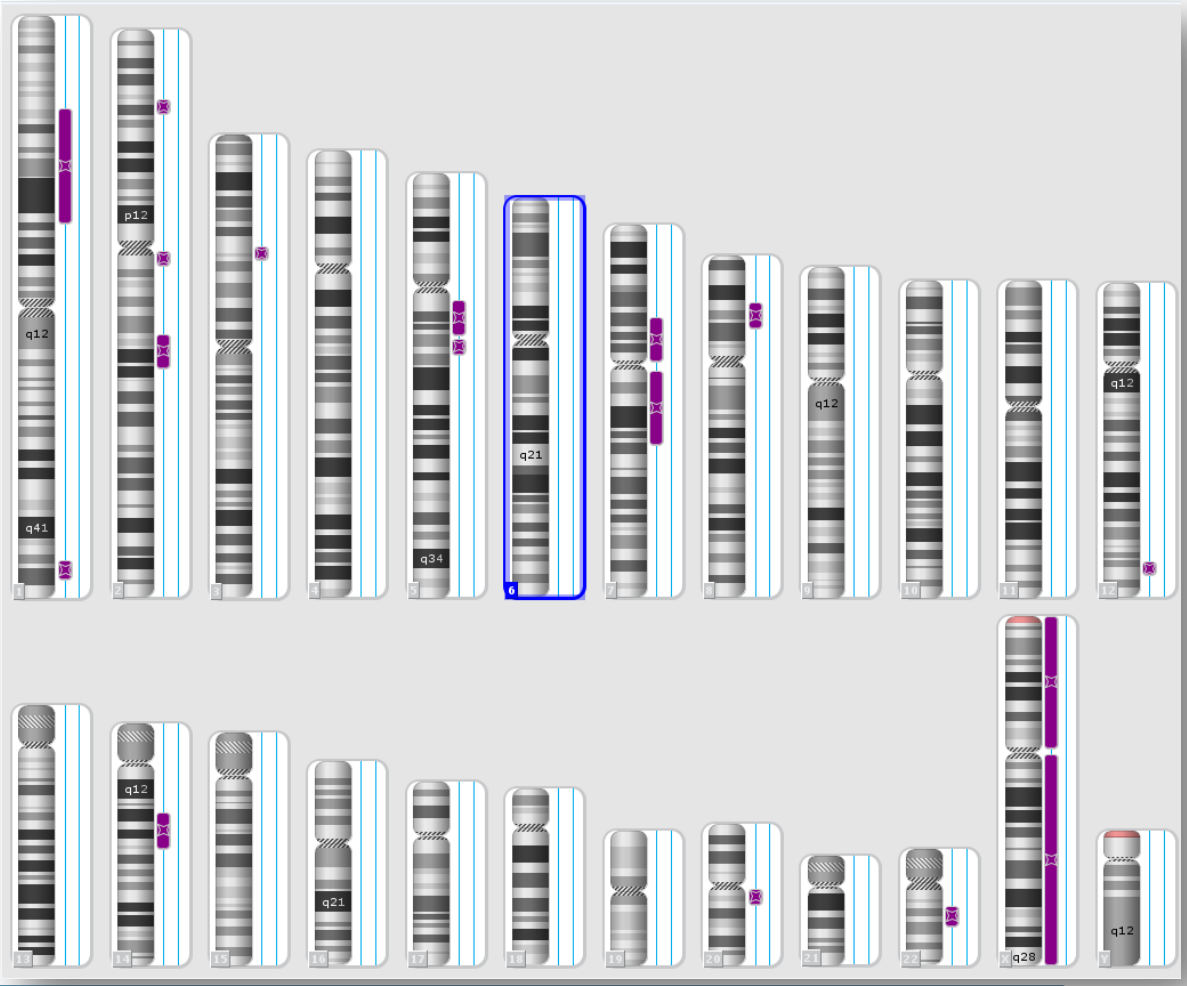


ROH/AOH/LOH Examples

130080: Regions identical by descent Genomic profile of consanguinity



This example illustrates blocks of LOH >5 Mb across a majority of the Chromosomes.

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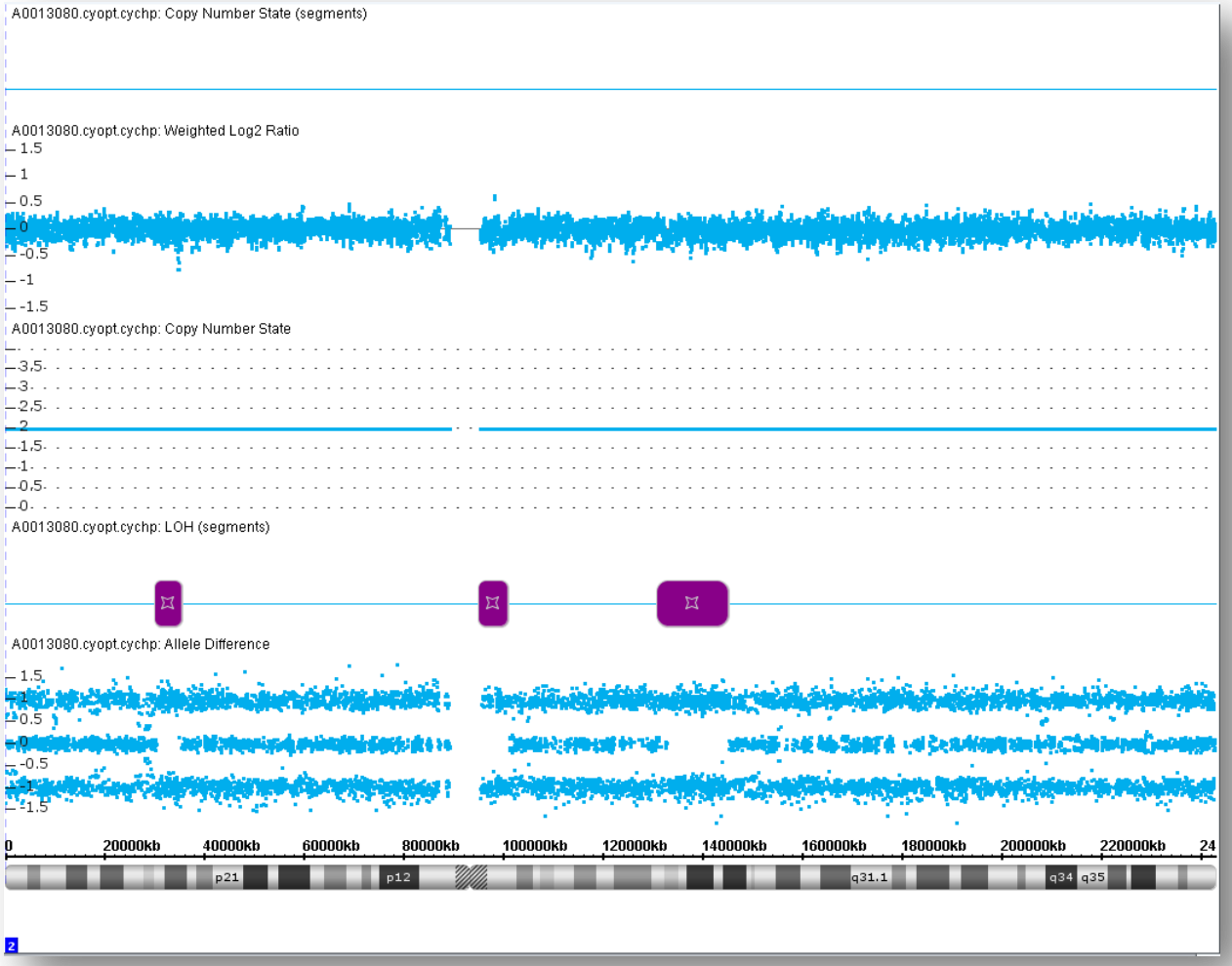
Regions identical by descent
Detailed view of chromosome 2



- **This is an example of two blocks of LOH >5 Mb on Chromosome 2.**

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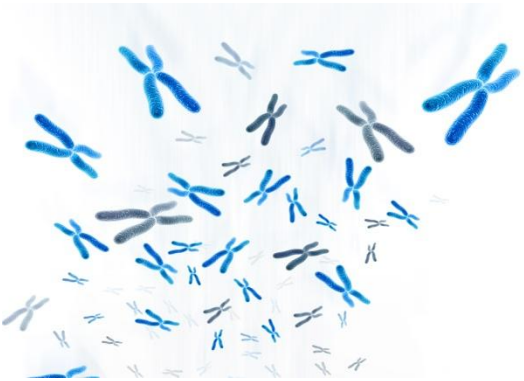

Genotypes - Trio

Mendelian Consistency Checking

#%SetName=RelatednessTest									
#%Columns=5									
#%Rows=3									
AnalysisTy	Reference	FamilialSa	RoleValid	RoleIndexScore					
0	0	2	1	32529.94					
1	0	1	1	23872.11					
2	0	2	1	24169.28					
#%SetName=MIE									
#%Columns=9									
#%Rows=23									
Chromosc	Display	MarkerCo	MIE-Trio	MIE-Mat	MIE-Pat	Percent-T	Percent-M	Percent-Pat-MIE	
1	1	11542	47	12	7	0.41	0.1	0.06	
2	2	12783	51	14	12	0.4	0.11	0.09	
3	3	10603	52	16	12	0.49	0.15	0.11	
4	4	10109	47	14	6	0.46	0.14	0.06	
5	5	9539	35	6	10	0.37	0.06	0.1	
6	6	9108	40	15	6	0.44	0.16	0.07	
7	7	8094	49	8	13	0.61	0.1	0.16	
8	8	7737	19	3	9	0.25	0.04	0.12	
9	9	5941	25	6	5	0.42	0.1	0.08	
10	10	7001	26	6	10	0.37	0.09	0.14	
11	11	6958	28	8	11	0.4	0.11	0.16	
12	12	6986	28	2	6	0.4	0.03	0.09	
13	13	5337	13	3	5	0.24	0.06	0.09	
14	14	4698	23	6	8	0.49	0.13	0.17	
15	15	4303	22	8	3	0.51	0.19	0.07	
16	16	3967	27	8	7	0.68	0.2	0.18	
17	17	3843	14	5	5	0.36	0.13	0.13	
18	18	4129	12	3	6	0.29	0.07	0.15	
19	19	2278	22	4	6	0.97	0.18	0.26	
20	20	3306	13	8	2	0.39	0.24	0.06	
21	21	1895	4	0	0	0.21	0	0	
22	22	1698	11	1	2	0.65	0.06	0.12	
24	X	6595	39	5	11	0.59	0.08	0.17	

- **This set of samples represents a trio. The Mendelian Error Check Function was used to check for relatedness.**
- **The trio is consistent!**

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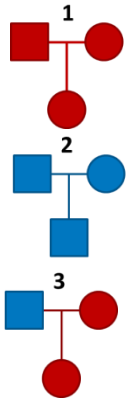
Trio Tool - Optima

- ✓ CytoScan Optima has enough genotypable SNPs across the genome to perform trio/duo analysis (1 & 2)
- ✓ Trio Tool is able to detect inconsistencies using CytoScan Optima Suite (3)



Sample ID	Ref #	Trio #	Relation	Family
A-0015275	NA10847	1	mother	CEPH 1334
A-0012037	NA12239	1	mat grandmother	CEPH 1334
A-0012038	NA12146	1	mat grandfather	CEPH 1334
A-0015266	NA07029	2	father	CEPH 1340
A-0012011	NA07000	2	pat grandmother	CEPH 1340
A-0012010	NA06994	2	pat grandfather	CEPH 1340
A-0015275	NA10847	1	mother	CEPH 1334
A-0012037	NA12239	1	mat grandmother	CEPH 1334
A-0012010	NA06994	2	pat grandfather	CEPH 1340

	Concordance mat	Concordance Pat	Concordance total
1-FAM1	99.89%	99.89%	99.57%
2-FAM2	99.89%	99.83%	99.49%
3-MIX	99.89%	91.69%	84.91%

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Whole-genome coverage

High-density SNPS with >99% genotype accuracy

AOH/LOH detection

CytoScan®
Cytogenetics Suite

Low-level mosaic sensitivity

Many applications – One platform