The American Journal of Human Genetics, Volume 108

Supplemental information

Optical genome mapping enables constitutional

chromosomal aberration detection

Tuomo Mantere, Kornelia Neveling, Céline Pebrel-Richard, Marion Benoist, Guillaume van der Zande, Ellen Kater-Baats, Imane Baatout, Ronald van Beek, Tony Yammine, Michiel Oorsprong, Faten Hsoumi, Daniel Olde-Weghuis, Wed Majdali, Susan Vermeulen, Marc Pauper, Aziza Lebbar, Marian Stevens-Kroef, Damien Sanlaville, Jean Michel Dupont, Dominique Smeets, Alexander Hoischen, Caroline Schluth-Bolard, and Laïla El Khattabi

Supplemental Data Description

Supplemental Data includes 12 figures and 7 tables.

Figure S1. Workflow of optical genome mapping technique.

Figure S2. Visual representation of optical genome mapping data.

Figure S3. Potential mapping artifacts.

Figure S4. Representative example of fragmented CNVs.

Figure S5. Representation of different chromosomal abnormalities.

Figure S6. Representative CNV profiles obtained with optical genome mapping, for different samples.

Figure S7. Isodicentric Y-chromosomes.

Figure S8. Optical genome mapping and CNV-microarray profiles for isochromosomes 15 and X.

Figure S9. Small ring chromosome X.

Figure S10. Optical genome mapping breakpoint detection for translocation t(9;17)(p13;q21), disrupting the gene *KANSL1*.

Figure S11. Complex sample 52.

Figure S12. Complex sample 55.

 Table S1. Comparison of previous diagnostic findings with optical genome mapping results.

Table S2. Technical performance of optical genome mapping.

 Table S3. Overall numbers of variants per sample.

Table S4. SV and CNV statistics per size range.

Table S5. Detection rate of chromosomal aberrations, by each pipeline, according to their type.

Table S6. Comparison of capabilities and limitations of optical genome mapping versus conventional cytogenetic methods, karyotyping and CNV-microarray, in detecting different types of chromosomal aberrations.

 Table S7. Concordance of all filtered SVs and CNVs between optical genome mapping and CNV-microarray



Figure S1. Workflow of optical genome mapping technique.

For this study, 85 samples for whom extra material was available were included. Ultra-high molecular weight DNA was extracted using the Bionano solution phase DNA isolation method. Labeling was done using the DLE-1 chemistry. High resolution imaging of DNA molecules was done on Bionano Saphyr instruments. As different centers were included, different amount of data was produced (~800Gbp for Radboud UMC, ~300Gbp for the French centers), and samples were analyzed using different software versions (3.4.1 and 3.5). A *de novo* assembly was performed, and both SVs and CNVs were called.

Grey boxes are optional steps to reduce the number of calls without losing sensitivity for clinically relevant aberrations. *FCN \leq 1.2 for losses and \geq 2.8 for gains, **recommended SV pipeline size cut-off = 20 Kb



Figure S2. Visual representation of optical genome mapping data.

A) Genome-wide circos plot showing all 24 chromosomes in a circular way. For each chromosome, the idiograms are shown at the outside of the circosplot, with ideogram-style chromosomal banding and the centromeres in red. Different colored dots in the boxes underneath represent different called SVs. The blue line in the box underneath represents the CNV profile, with each peak representing a CNV call. B) Part of a circos plot, showing the sex chromosomes. The blue CNV line shows two copies of chromosome X, as for autosomes, and one copy of chromosome Y consistent with a sex chromosome aneuploidy (47,XXY, resulting in Klinefelter syndrome).



Figure S3 Potential mapping artifacts.

This figure shows two different rearrangements called in two different samples that likely derive from mapping difficulties. A) Intrachromosomal rearrangement in chr1, B) Intrachromosomal rearrangement in chr9. In both samples, the rearrangement breakpoints occur in highly repetitive regions, often in close proximity to the centromere. The left side of each subfigure shows the whole chromosome view of the respective translocation, whereas the right side shows the zoom-in of the corresponding molecule map. The many different connecting lines between the labels in the reference genome map and the samples' genome map let assume that mapping of highly repetitive regions leads to false positive calls.



Figure S4. Representative example of fragmented CNVs.

This figure shows an example of a large CNV which is a consequence of a Robertsonian translocation. However, instead of having one call of a large CNV, several smaller CNVs are called. A) CNV coverage plot of 6q16.3q27 region. B) Circos plot of the entire chromosome 6.



Figure S5 (1/2). Representation of different chromosomal abnormalities.

This figure shows an example of each type of chromosomal aberration that was detected in this study. All aberrations shown are described in detail in Table S1.

Isochromosome

Sample 77: 15q11.2q13.2 gain (4x) 15q13.3 gain (3x)

Ring chromosome

Sample 39: r(X)(p11.21q21.1)





Complex rearrangement

Sample 66: 3p14.1(65238298_68667113)x1 3p13(70127345_73724765)x1 3p12.1(83784489_85467284)x1 3q11.2(97180779_97270083)x1





Sample 24: trisomy 21 via Robertsonian translocation



Large terminal deletion Sample 41: del(9)(p23pter)

Large duplication		p21.3	p21.2	_	p12	
Sample 60:						1
8p23.1p12 gain						

Figure S5 (continued 2/2). Representation of different chromosomal abnormalities.

This figure shows an example of each type of chromosomal aberration that was detected in this study. All aberrations shown are described in detail in Table S1.



Figure S6. Representative CNV profiles obtained with optical genome mapping, for different samples.

A) Sample 2. Loss of 22q11.21(18645354_21465660). B) Sample 8. Gain of 17p12(14087934_15436895). C) Sample 70. Loss of 6q14.1q14.3(76385698_86884355), and gain of 6q16.1(97661978_98726638). D) Genome-wide CNV view (available in Bionano Solve v1.5) of sample 73 with E) chromosome 8 highlighted (showing a deletion) and F) chromosome 17 highlighted (showing a duplication). Blue: gains, Red: losses.



Figure S7: Isodicentric Y-chromosomes.

Optical genome mapping CNV profile (top) and CNV-microarray CNV profile (bottom) of Y-chromosomes of samples 80 (normal chrY, A), sample 55 (B), sample 57 (C) and sample 79 (D). The chromosomal position is given on top, the corresponding chromosome banding below each CNV-microarray profile. Sample 27 is not shown here because it was analyzed with a different CNV-microarray platform.



Figure S8. Optical genome mapping and CNV-microarray profiles for isochromosomes 15 and X.

A) Sample 77 (ish idic(15)(D15Z1+,SNRPN++,D15Z1+)). Left: Circos plot showing an abnormal CNV profile on chromosome 15. Top right: CNV-microarray data showing a gain on chr15. Bottom right: optical mapping data, showing a CNV profile that is nearly identical to the CNV-microarray profile. Numbers present fractional copy numbers. B) Sample 78 (46,X,idic(X)(p11.21)). Left: Circos plot showing a CNV baseline suggesting one copy of chromosome X (compared to the CNV line of chr22 partially shown on the left side). Additionally, the CNV profile shows a mosaic "gain" (compared to the baseline) on part of the chrX p-arm and the whole q-arm. Top right: CNV-microarray data showing a global loss on chrX (compared to a 46,XX control sample). However, the degree of loss varies within the chromosome consistent with a mosaic 45,X/46,X,idic(X)(p11.21) karyotype. Bottom right: optical mapping data showing a CNV profile that is nearly identical to the CNV-microarray profile. Numbers present fractional copy numbers. Red box shows parts of the chromosome 15 and X respectively that make up the iso-chromosomes. Grey box indicates the centromere (15 and X) and/or acrocentric p-arm (15).



Figure S9. Small ring chromosome X.

A) Karyogram of sample 39. The red arrow is pointing towards the small X ring chromosome. B) Circos-plot (of chromosome X only) of sample 39. The pink line in the center of the circosplot is indicating the presence of the ring chromosome (called as an intrachromosomal translocation). C) Different genome maps (dark blue bars on top and below the reference) indicating the presence of the ring chromosome. The individual molecules for the genome map below the reference (highlighted by a red circle) are shown at the bottom of this figure. The left part of these molecules (light green bar) map to a region upstream of the centromere, whereas the right part of the same molecules (light blue bar) map to a region downstream of the centromere.



Figure S10. Optical genome mapping breakpoint detection for translocation t(9;17)(p13;q21), disrupting the gene *KANSL1*.

The two green bars represent the references of chromosomes 9 and 17, respectively. The mint bar in between represents the genome map of the translocation. The blue bar underneath represents the *KANSL1* gene. Small vertical black lines represent identified labels, and the red vertical lines indicate the translocation breakpoints, with an uncertain region of 3,828 bp in between shown in purple. The breakpoints are located between basepair-positions 35,771,617 and 35,773,383 on chromosome 9, and between 44,137,912 and 44,141,740 on chromosome 17.



Figure S11. Complex sample 52.

A) Karyotype of sample 52, interpreted as 46,XY,der(8)t(8;22)(q12;q12),der(13)t(8;13)(q31;q23),der(14)t(14;15)(q11.2;q25),der(15)t(14;15)(q21;q24),d er(22)t(13;22)(q31.1;p11.2). B) FISH of sample 52, using FISH probes wcp8 (red), wcp14 (green). C) FISH of sample 52, using FISH probes wcp8 (green), wcp13 (red). D) FISH of sample 52, using FISH probes wcp15 (green), wcp22 (red). E) Optical mapping circos plot, showing different translocations t(8;13), t(8;14), t(14,15), and intrachromosomal translocations on chr 8 and chr 15.



Figure S12: Complex sample 55.

A) Karyotype of sample 55, interpreted as 46,X,idic(Y)(q11.22),t(5;8)(q23;q24),t(5;11)(p12;p13)[32/50] /45,X,t(5;8)(q23;q24),t(5;11)(p12;p13)[10/50]/47,XY,idic(Y)(q11.22),t(5;8)(q23;q24),t(5;11)(p12;p13) [8/50]. B) FISH of sample 55, showing the translocations t(5;8) (left) and t(5;11) (right). C) Optical mapping circos plot of sample 55, showing the translocations t(5;8), t(5;11) and an intrachromosomal translocations 5. D) Bionano genome maps, showing the intrachromosomal translocation on chromosome 5, which is disrupting the gene *GHR*.

 Table S1. Comparison of previous diagnostic findings with optical genome mapping results.

Provided in Excel sheet format

Sample Starting material Total DNA Avg N50 Avg N50 Map rate Effective Avg label density =150 kbp (Gbp) >=150kbp (Mbp) >=20kbp (Mbp) (>= 150 kbp) coverage (x) (per 100kbp) EDTA blood 1 320 0,24 0,12 69% 286 15.69 1 EDTA blood 1 332 0,26 0.15 78% 323 15,19 3 EDTA blood 1 302 0.26 0.13 57% 233 17.88 0,13 58% 243 17,14 4 EDTA blood 1 305 0,24 1 347 0,21 87% 5 EDTA blood 0,26 368 15,20 6 EDTA blood 811 0,21 0,08 43% 110 15,28 EDTA blood 807 0,21 0,10 55% 140 15,34 EDTA blood 8 867 0,24 0,12 60% 163 15,19 9 EDTA blood 816 0,25 0,17 67% 167 15,38 10 EDTA blood 824 0,22 0,11 68% 175 15,25 11 EDTA blood 840 0,24 0,13 67% 176 15,40 12 EDTA blood 534 0,33 0,33 40% 69 16,16 13 EDTA blood 814 0,24 0,15 79% 200 15,19 14 Hep. Blood 801 0,24 0.14 66% 162 15,25 15 Hep. Blood 655 0.24 0.15 63% 124 14.40 16 Hep. Blood 821 0,22 0,14 72% 175 13,76 17 0,12 63% 149 Hep. Blood 813 0,20 13,27 18 0,12 64% Hep. Blood 823 0,20 157 14,28 19 Hep. Blood 825 0,22 0,13 69% 169 14,21 20 Hep. Blood 817 0,19 0,12 65% 150 12,97 21 Hep. Blood 832 0,21 0,13 67% 162 13,83 22 Amniotic fluid 200 0,30 0,30 60% 38 15,40 23 Amniotic fluid 812 0,27 0,21 75% 177 13,47 24 Chorionic villi 826 0,25 0,18 73% 176 13,74 25 Amniotic fluid 850 0,26 0,20 72% 177 13,26 26 Amniotic fluid 802 0.24 0.18 88% 220 15.33 27 0.22 85% 214 Amniotic fluid 801 0.30 15.39 28 0,17 78% 849 186 Amniotic fluid 0,26 12,77 29 617 0,21 90% 173 Chorionic villi 0,28 14,96 30 Amniotic fluid 842 0,33 0,27 89% 242 14,72 31 Chorionic villi 820 0,27 0,21 88% 223 14,40 32 0,33 0,26 90% 228 14,97 Amniotic fluid 817 33 Chorionic villi 802 0,29 0,20 84% 219 14,97 34 Lymphoblastoid cell line 839 0,32 0,21 86% 224 15,15 35 Hep. Blood 814 0.27 0.21 87% 218 14.37 36 37 Hep. Blood 845 0.30 0.25 89% 233 14.91 Hep. Blood 809 0.25 0.10 71% 178 14.74 38 Hep. Blood 857 0.27 0.22 89% 233 14.37 39 0.17 86% Hep. Blood 855 0.25 228 14.01 40 Hep. Blood 810 0,24 0,17 77% 189 12,92 41 Hep. Blood 811 0,21 0,13 73% 184 14,23 42 Hep. Blood 823 0,24 0,18 82% 208 14,19 43 499 EDTA blood 0,26 0,17 87% 135 14,50 44 EDTA blood 506 0,26 0,12 82% 130 15,11 45 EDTA blood 503 0,25 0,17 85% 134 15,27 46 EDTA blood 495 0,24 0.16 78% 122 14,96 47 48 EDTA blood 488 0.23 0.10 70% 107 15.58 486 0,30 0,25 90% 137 EDTA blood 14,60 49 Lymphoblastoid cell line 376 0,28 0,20 79% 95 15,79 50 0,20 70% 328 0,28 73 Lymphoblastoid cell line 16,38 484 0,20 64% 98 51 15,74 Lymphoblastoid cell line 0,29 348 0,20 78% 87 52 Lymphoblastoid cell line 0,29 15,71 53 Lymphoblastoid cell line 495 0,28 0,19 73% 115 15,42 54 Lymphoblastoid cell line 345 0,16 75% 82 0,27 15,25 55 EDTA blood 541 0,29 0,20 85% 144 14,47 56 EDTA blood 519 0,26 0,17 88% 142 14,51 57 EDTA blood 324 0,22 0.13 75% 75 14,78 58 EDTA blood 328 0.23 0.14 84% 85 15,10 59 0,17 74% 75 EDTA blood 324 0,26 15,17 60 0,22 75% EDTA blood 481 0,29 113 15,27 61 EDTA blood 333 0,22 0,10 68% 71 15,05 EDTA blood 0,13 74% 113 62 492 0,29 15,90 63 EDTA blood 496 0,25 0,16 84% 129 15,04 504 0,28 143 64 EDTA blood 0,20 90% 15,44 65 0,17 89% EDTA blood 346 0,27 95 15,13 66 EDTA blood 380 0,30 0,19 83% 102 16,28 67 EDTA blood 353 0,31 0,25 90% 100 16,83 68 EDTA blood 371 0,35 0,27 81% 96 17,17 69 EDTA blood 480 0,32 0,25 85% 129 16,46 70 EDTA blood 400 0.36 0.36 89% 111 14.24 71 400 0,31 0,31 87% 14,47 EDTA blood 109 72 0,12 50% EDTA blood 1 311 0,22 208 14,88 EDTA blood 0,32 84% 73 400 0,32 105 15,75 74 EDTA blood 911 0,15 38% 109 0,25 22,36 75 EDTA blood 400 0,28 0,28 86% 108 15,45 76 EDTA blood 400 0,30 0,30 81% 102 15,78 77 EDTA blood 488 0,34 0,28 87% 134 14,82

Table S2. Technical performance of optical genome mapping.

			- /	-, -			
85	Lymphoblastoid cell line	481	0,29	0,11	77%	117	15,30
Average		655	0,27	0,18	77%	152	15,13
Max valu	ie	1 347	0,36	0,36	93%	368	22,36
Min valu	e	200	0,19	0,08	38%	38	12,77
Standard	d deviation	270	0,04	0,06	12%	59	1,21

0,27

0,32

0.31

0.31

0.29

0,27

0,27

0,18 91%

0,24 87%

0.25 92%

0.24 93%

0.20 91%

0,17 86%

0,13 88%

122

132

120

140

127

110

110

15,06

14,92

15.06

15.62

15.56

15,94

15,29

436

483

424

483

449

411

406

78

79

80

81

82

83

84

EDTA blood

EDTA blood

EDTA blood

EDTA blood

EDTA blood

EDTA blood EDTA blood
 Table S3. Overall numbers of variants per sample.

Provided in Excel sheet format

Table S4. SV and CNV statistics per size range.

a. Size distribution of SV calls for the cohort

		Number of rare SVs in each size category							
	Total rare SVs	0-20kb	20-100kb	100-500kb	>500kb				
All chromosomes	6785 (100%)	5996 (88.4%)	423 (6.2%)	160 (2.4%)	206 (3.0%)				

b. CNV size distribution in the total cohort (initial filter-setups: non-masked CNVs only and excluding those with CN=2)

		Number of CNV calls in each size category							
	Total	< 0.5Mb	0.5-1.0Mb	1-5Mb	>5Mb				
All chromosomes	942 (100%)	471/942 (50%)	246/942 (26%)	174/942 (18%)	51/942 (6%)				
Autosomes only	895 (100%)	463/895 (52%)	241/895 (27%)	162/895 (18%)	29/895 (3%)				

c. CNV size distribution in the total cohort (additional filtering: excluding deletions with fractional CN >1.2 and duplications with CN < 2.8)

		Number of CNV calls in each size category								
	Total	< 0.5Mb	0.5-1.0Mb	1-5Mb	>5Mb					
All chromosomes	366 (100%)	224/366 (61.2%)	61/366 (16.7%)	37/366 (10.1%)	44/366 (12%)					
Autosomes only	324 (100%)	218/324 (67.3%)	57/324 (17.6%)	25/324 (7.7%)	24/324 (7.4%)					

Table S5. Detection rate of chromosomal aberrations, by each pipeline, according to their type.

Aberration type	SV tool call	CNV tool call	Only called by one tool	Called by both tools	Global rate of detection	
Aneuploidies (n=7)		NA	7/7 (100%)	7/7 (100%)	NA	100%
	Microdeletions and duplications (n=34)	27/34 (79%)	27/34 (79%)	14/34 (41%)	20/34 (59%)	100%
	Large CNV losses and gains (n=5)*	2/5 (40%)	5/5 (100%)	3/5 (60%)	2/5 (40%)	100%
	Unbalanced translocation (n=7)	4/7 (57%)	5/7 (51%)	3/7 (43%)	4/7 (57%)	100%
Unbalanced structural aberrations	Unbalanced insertions (n=2)	2/2 (100%)	2/2 (100%)	0/2 (0%)	2/2 (100%)	100%
	Ring chromosomes (n=1)	1/1 (100%)	1/1 (100%)	0/1 (0%)	1/1 (100%)	100%
	Isochromosomes (n=6)	0/6 (0%)	6/6 (100%)	6/6 (100%)	0/6 (0%)	100%
	complex chromosomal rearrangements (n=4)	4/4 (100%)	2/4 (50%)	2/4 (50%)	2/4 (50%)	100%
Palancod structural aborrations	Balanced translocation (n=27)	27/27 (100%)	NA	27/27 (100%)	NA	100%
	Inversions (n=6)	6/6 (100%)	NA	6/6 (100%)	NA	100%

*this includes terminal aberrations that are currently not called by the SV pipeline

NA: not applicable

Table S6. Comparison of capabilities and limitations of optical genome mapping versus conventional cytogenetic methods, karyotyping and CNV-microarray, in detecting different types of chromosomal aberrations.

	D	Localization of aberration				
	Standard of care	genome-wide tests	Optical genome	Standard of care §	Optical genome	
Aberration type	CNV-microarray	Karyotyping	mapping	CNV-microarray	Karyotyping	mapping
	aberrations larger than 20-200 kb	aberrations larger than 7-10Mb	aberrations >few kb	aberrations larger than 20-200 kb	aberrations larger than 7-10Mb	aberrations >few kb
Aneuploidy	\checkmark	✓	✓	×	✓	\checkmark
Unbalanced Robertsonian translocation	\checkmark	\checkmark	\checkmark	×	✓	×
Balanced Robertsonian translocation	×	✓	*	×	✓	×
Unbalanced reciprocal translocation	\checkmark	✓	✓	×	✓	\checkmark
Balanced reciprocal translocation	×	\checkmark	✓	×	✓	\checkmark
Deletion	\checkmark	✓	✓	×	✓	\checkmark
Insertion	unbalanced only	\checkmark	✓	×	✓	\checkmark
Duplication	\checkmark	\checkmark	✓	×	✓	\checkmark
Inversion	×	\checkmark	✓	×	✓	\checkmark
Isochromosome	\checkmark	✓	✓	×	✓	\checkmark
Ring chromosome	unbalanced only	✓	✓	×	✓	\checkmark

Binner bed Binner bed Or. Start End	Commits ID				Bionano call		CNV-microarray call				- ua	
N 4 10.8188905 615920311 94947 duplication 4 11.8188905 11.6706 0.5 Seported 56 3V 16 3448207 3453843 34138 duplication juvered 16 3465027 3453843 04138 duplication juvered 16 3465027 3456384 04418 duplication juvered 16 3465027 3456384 04418 duplication juvered 16 3465027 3456384 05 Segported 05 Segported 3V 1.6 79404969 79404989 7940098 19992 deteion 1.6 49595920 199920 1999 04993839 1probe -0.021 Segported 3V 2 2323147 232320824 23820672 1probe -0.077 Segported 3V 3 12041170 20072823 428184 4318 deteion - - No No No Segported 3V 1 165845905 56907488 47385646 7297773 729773 0.37 No Segported </th <th>Sample ID</th> <th>Bionano tool</th> <th>Chr</th> <th>Start</th> <th>End</th> <th>Size (bp) Type</th> <th>Chr</th> <th>Start</th> <th>End</th> <th>Call size or # of probes</th> <th>LogR</th> <th>Result</th>	Sample ID	Bionano tool	Chr	Start	End	Size (bp) Type	Chr	Start	End	Call size or # of probes	LogR	Result
SV 10 81438916 81761 deletion - No probes Inconclusive SV 16 34462037 3357881 116945 3466213 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456215 3456217 3457839 1 probe -0.71 Supported 0.3 Supported 0.3 Supported 0.22 Supported 0.22 Supported 0.22 Supported 0.22 Supported 0.21 0.25 Supported 0.21 0.25 Supported 0.23 Supported 0.24 1.22 2.232 0.25 Supported 0.25 Supported 0.25 Supported 0.25 Supported 0.25 Supported <t< td=""><td></td><td>SV</td><td>4</td><td>161863865</td><td>161923311</td><td>59447 duplication</td><td>4</td><td>161869551</td><td>161869610</td><td>1 probe</td><td>0,5</td><td>Supported</td></t<>		SV	4	161863865	161923311	59447 duplication	4	161869551	161869610	1 probe	0,5	Supported
SV 16 34450207 3453543 94418 duplication_metrical 16 34450207 34545020 3555502 3555274 2 probes 0.5 Supported 5V 16 79404969 7943008 7943008 79420905 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 79420907 7942097		SV	10	81438916	81594303	137161 deletion	-	-	-	No probes	-	Inconclusive
SV 16 34462037 3452881 116843 explication meeter 16 3456213 3452214 2 probes 0.5 Supported 5V 1 40908008 20593 deletion 1 4090205 7482094 1 probe 0.76 Supported 5V 1 40912040 20011818 81129 deletion 1 46324472 26501692 1 probe 0.97 Supported 5V 2 2323294372 233238334 27988 deletion 2 233270471 1 probe 0.77 Supported 5V 3 20242707 120012222 3718 deletion - - - No No 5V 3 654505 5507488 15656 deletion 7 719773 0.57 - - - No No - 0.57 5V 18 6581496 63813984 6381394 1907517 0.55 - - No No Supported - - No No	56	SV	16	34462037	34556454	94418 duplication inverted	16	34482042	34482101	2	0,5	Supported
SV 16 79400069 79430006 79420005 79420905 1 probe 0.76 Supported SV 1.49980206 5000118 80129 deletion 1 4959204 9993891 probe 0.92 Supported SV 1.249312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 246312447 24631247 24537 0.56 0.77 7.61093111 7.6153242 7.5598014 1.4154451 1.60761473 1.57576 1.57576		SV	16	34462037	34578881	116845 duplication inverted	16	34565215	34565274	2 probes	0,5	Supported
SV 1 4999200 9001318 81129 deletion 1 4995200 499389 1 probe -0.92 Supported SV 2 23324917 23328334 27988 deletion 2 23327072 1 probe -0.77 Supported SV 5 120421707 120472623 43718 deletion - - Inconclusive - - Inconclusive - - - Inconclusive -		SV	16	79404969	79430008	20599 deletion	16	79420905	79420964	1 probe	-0.76	Supported
SV 1 24632442 24623043 200871 deletion 1 246321447 24651082 1802425 hpp mean*1.12 Confirmed 5V 2 33239437 2323844 23328434 2332844 233270613 23237067 1 probe		SV	1	49908206	50001818	84129 deletion	1	49959780	49959839	1 probe	-0,92	Supported
SV 2 23323433 23228334 27988 deletion 2 23270612 1 probe -0.77 Supported 62 SV 5 102021707 120072633 43718 deletion - - No probes - Iscondukie 64 SV 9 71845510 7196535 116026 duplication 9 71917583 3 probe -0.55 0.57 0.57 64 ENV 18 65891890 65891890 65891890 65891890 10016 0.54 Supported 64 ENV 7 7612922 2108985 2108915 2206217 21065 0.045 Supported 64 13824526 13823760 9 3273751.7 278726 0.16 Not supported 64 138237661 21382915 2108925 2108925 1 probe -0.07 0.042 Supported 77 SV 4 138237620 2108925 1 probe -0.07 0.07 0.07 0.042		SV	1	246313442	246520313	206871 deletion	1	246321447	246501692	180245 bp	mean='-1.12	Confirmed
SV 5 12042/1707 12047/2623 43718 deletion - - - No probes - Inconclusive SV 9 71845510 7196/578 1160/277 0.55 0.55 SV 18 65848905 65907/88 45898 46180 77917/58 3 probes 0.35 5upported 64 (NV 7 7610/9311 7662/344 51453 deletion 7<701/3282		SV	2	233239437	233288334	27988 deletion	2	233270613	233270672	1 probe	-0,77	Supported
D2 9 71846737 116072 0.55 5V 9 71845510 7191769 7191779 3 probes 0.33 Supported 5V 18 653845905 65907488 49589 deletion 18 65891899 1 probe 0.55 64 (NV 7 76109311 76623841 51433 deletion 77 7613222 76538024 418743 bp mean-0.85 Confirmed 64 (NV 8 2120483 2349321 22883 deletion 8 220611 2 probes -0.07 0.06 Not supported 5V 4 138246546 13837666 91151 duplication 4 138229511 138279510 2 probes -0.07 0.02 Supported -0.07 0.042 Supported -0.07 -0.07 -0.07 -0.06 <t< td=""><td>62</td><td>SV</td><td>5</td><td>120421707</td><td>120472623</td><td>43718 deletion</td><td>-</td><td>-</td><td>-</td><td>No probes</td><td>-</td><td>Inconclusive</td></t<>	62	SV	5	120421707	120472623	43718 deletion	-	-	-	No probes	-	Inconclusive
SV 9 7195153 116026 duplication 9 71917758 3 probes 0.39 Supported 64 57 78109311 76623844 51533 deletion 18 65891899 1 probe -0.54 Supported 64 CNV 8 2120483 2349321 228383 deletion 8 2206152 2206217 2 probes -0.07 Not supported 55 V 4 13824656 138337666 91151 duplication 4 13825910 2 probes -0.07 Not supported 5V 4 13824656 138337666 91151 duplication 4 13825910 2 probes -0.12 Supported 5V 4 13824656 13837666 91351 19827966 32379566 1 probe -1.02 Supported 77 CNV K 6593242 8313731 127792 duplication - No probes - Inconclusive 5V 1 8391262 223022 deletion - - <	62						9	71846678	71846737	•	0,55	
Image: solution of the second secon		SV	9	71845510	71961535	116026 duplication	9	71917699	71917758	3 probes	0,39	Supported
SV 18 6554905 65907488 49589 deletion 18 65891899 1 probe -0.54 Supported 64 CNV 7 76109311 76623844 514533 deletion 7 77139282 76558044 418743 bp mean-0.85 Confirmed 64 18327657 2206158 2206158 2206217 2probes -0.07 57 4 18327662 183229510 2probes -0.02 Supported 58 9 22737531,7 22787627.8 36733 deletion 4 138229755 1 probe -1.02 Supported 77 59 9 22737531,7 2278757 1 probe -1.02 Supported 78 5V 1 39883 deletion - - No probes - Inconclusive 5V 1 2250850 2593305 23215 deletion - - No probes - Inconclusive 5V 2 130234028 131077 deletion - -							9	71957664	71957723		0,57	
CNV 7 76109311 7662844 514533 deletion 7 7712922 76558024 418743 bp mean-0.85 Confirmed 64 CNV 8 2120437 228383 deletion 8 2206158 2206217 2 probes 0.01 Not supported 5V 4 13837696 91151 duplication 4 138329451 13825510 2 probes 0.42 Supported 64 13837531 3278756 127072 4138324112 0.50 0.42 Supported 70 5V 4 13838277 8 373784 6687938 31877 deletion - - No probes - inconclusive 5V 14 86454708 86498388 31877 deletion - - No probes - inconclusive 5V 1 2208530 223125 64212 1307792 geletion - - No probes - inconclusive 5V		SV	18	65845905	65907488	49589 deletion	18	65891840	65891899	1 probe	-0,54	Supported
64 CNV 8 2120483 2349321 228838 deletion 8 2206217 2 probes 0.46 Not supported 5v 4 138246546 13833766 91151 duplication 4 13829451 138229452 2 probes 0.07 Supported 70 5v 9 32737531.7 32787627.8 36735 deletion 9 3273955 1 probe -1.02 Supported 70 5v 4 138824612 809751 1608791 bp mean-0.51 Confirmed 70 5v 1 8545408 8157179 22082 deletion - - No probes - Inconclusive 5v 1 86459388 31877 deletion - - No probes - Inconclusive 5v 2 131031140 131977013 74897 deletion - - No probes - Inconclusive 5v 2 13103140 131977013 74897 deletion 2 - No probes <t< td=""><td></td><td>CNV</td><td>7</td><td>76109311</td><td>76623844</td><td>514533 deletion</td><td>7</td><td>76139282</td><td>76558024</td><td>418743 bp</td><td>mean=-0.85</td><td>Confirmed</td></t<>		CNV	7	76109311	76623844	514533 deletion	7	76139282	76558024	418743 bp	mean=-0.85	Confirmed
64 Ev 6 2100es 2.000 MOS supported 5V 4 138246546 138337696 91151 duplication 4 138224113 138324112 2 probes 0.042 Supported 5V 9 327351.7 32787627.8 36735 deletion 9 327359696 32739755 1 probe -0.02 Supported 77 5V 1 85146480 8313717 220224 8037511 1606791 bp mean-0.51 Confirmed 78 5V 14 8644708 8649388 318177 deletion - - No probes - Inconclusive 5V 14 8644708 8649388 318177 deletion - - No probes - Inconclusive SV 1 2208050 2233055 23125 deletion - - No probes - Inconclusive SV 2 1303140 13197013 74897 deletion - - No probes		CNIV	0	2120492	2240221	228220 deletion	8	2206158	2206217	2 prohos	0,16	Notsupported
Sv 4 138246546 138337696 91151 duplication 4 138224511 138229451 2 probes 0,42 Supported SV 9 32737531,7 32787627,8 36735 delta 383224113 1383241172 2 probes 0,59 T SV 4 138324113 138324113 1383241172 2 probes 0,59 T SV 4 1383241172 1080791 bp mean=0.51 Confirmed SV 14 139832866 13991220 23933 deletion - - No probes - Inconclusive SV 14 86454708 86498388 31877 deletion - - No probes - Inconclusive SV 1 2303030 2333505 23125 deletion - - No probes - Inconclusive SV 2 131031400 131977013 74897 deletion - - No probes - Inconclusive SV 2	64	CINV	٥	2120465	2349321	228838 deletion	8	2308926	2308985	z probes	-0,07	Not supported
N 4 13824010 13824013 13824013 12824012 2100es 0.59 SV 9 32735931 1002 Supported 0.50 0.59 77 SV 4 13988626 13919269 239393 deletion - - No probes - Inconclusive 77 SV 4 139886286 3131734 1627792 2002 deletion - - No probes - Inconclusive SV 14 86454708 86493388 31877 deletion - - No probes - Inconclusive SV 1 2206530 223350204 55948466 27963 duplication - - No probes - Inconclusive SV 2 24651022 24175013 34897 deletion 2 241687451 241687509 1 probe 0.04 Supported SV 2 13031400 131977013 74897 241650751 2416507511	04	sv/	4	120246546	120227606	011E1 duplication	4	138259451	138259510	2 probos	0,42	Supported
SV 9 32737531,7 32737627,8 36735 deletion 9 32759696 32759755 1 probe -1.02 Supported 77 5V 4 139888286 139919269 23983 deletion - - No probes - Inconclusive SV 11 8516480 85175179 23022 deletion - - No probes - Inconclusive SV 14 86454708 86498388 31877 deletion - - No probes - Inconclusive SV 14 86454708 86498388 31877 deletion - - No probes - Inconclusive SV 1 2308530 2233305 23125 deletion - - No probes - Inconclusive SV 2 13101140 131077013 7497 deletion 3 100405671 0,01 Not supported SV 3 100300351 100451628 111278 duplication 3 100419624 2 probes		30	4	138240340	138337090	91191 duplication	4	138324113	138324172	2 probes	0,59	
CNV X 50342 8131734 1627792 duplication X 6488721 8097511 1608791 bp mean=0.51 Confirmed SV 4 139888286 139919269 23983 deletion - - No probes - Inconclusive SV 14 86454708 8649338 31877 deletion - - No probes - Inconclusive SV 14 8649338 31877 deletion - - No probes - Inconclusive SV 2 55920504 55948466 27963 duplication - - No probes - Inconclusive SV 2 241651052 241703013 74897 deletion - - No probes - Inconclusive SV 2 241651052 24105031 100451628 10030751 probes 0.01 Not supported SV 3 165014055 165083895 42602 deletion 3 100306621 100806519 1 probe <td></td> <td>SV</td> <td>9</td> <td>32737531,7</td> <td>32787627,8</td> <td>36735 deletion</td> <td>9</td> <td>32759696</td> <td>32759755</td> <td>1 probe</td> <td>-1,02</td> <td>Supported</td>		SV	9	32737531,7	32787627,8	36735 deletion	9	32759696	32759755	1 probe	-1,02	Supported
SV 4 13988826 139919269 23983 deletion - - No probes - Inconclusive SV 11 85146408 8517317 23022 deletion - - No probes - Inconclusive SV 14 86454708 86498388 31877 deletion - - No probes - Inconclusive SV 1 2230550 23325 deletion - - No probes - Inconclusive SV 2 550204 55944862 27663 duplication - - No probes - Inconclusive SV 2 241651052 241705013 53962 duplication 2 241687509 1 probe 0,42 Supported SV 3 100340551 100451628 111278 duplication 3 100360521 100360751 2 probes 0,61 Supported SV 3 100304055 16008395 42602 deletion 3 1003605479 1 probe 0,17 <td></td> <td>CNV</td> <td>Х</td> <td>6503942</td> <td>8131734</td> <td>1627792 duplication</td> <td>Х</td> <td>6488721</td> <td>8097511</td> <td>1608791 bp</td> <td>mean=0.51</td> <td>Confirmed</td>		CNV	Х	6503942	8131734	1627792 duplication	Х	6488721	8097511	1608791 bp	mean=0.51	Confirmed
SV 11 85146480 85175179 23022 deletion - - No probes - Inconclusive SV 14 8645708 8649888 31877 deletion - - No probes - Inconclusive SV 1 22308530 2233055 23125 deletion - - No probes - Inconclusive SV 2 55920504 559248466 27963 duplication - - No probes - Inconclusive SV 2 241651052 241705013 3497 deletion - - 10 probes mean-0.01 Not supported SV 2 241651052 241705013 53962 duplication 3 10036052 100360751 2 probes 0.61 Supported SV 3 165014055 165083895 42602 deletion 3 160049561 100419565 100360751 0.68 Inconclusive SV 16 78037966 78081307 36813 deletion - <	77	SV	4	139888286	139919269	23983 deletion	-	-	-	No probes	-	Inconclusive
SV 14 86454708 86498388 31877 deletion - - No probes - Inconclusive SV 1 2238530 2233055 23125 deletion - - No probes - Inconclusive SV 2 55920504 55948466 27963 duplication - - No probes - Inconclusive SV 2 13103140 131977013 74897 deletion - - No probes mean=-0.01 Not supported SV 2 241651052 241705013 53962 delpication 2 241687509 1 probe 0,42 Supported SV 3 100340351 100451628 111278 duplication 3 100360622 100360751 2 probes 0,64 Supported Supported 0,64 Supported 0,61 S	//	SV	11	85146480	85175179	23022 deletion	-	-	-	No probes	-	Inconclusive
SV 1 22308530 22132 deletion - - No probes - Inconclusive SV 2 55920504 55948466 27963 duplication - - No probes - Inconclusive SV 2 131031140 131977013 74897 deletion - - - 10 probes mean=-0.01 Not supported SV 2 241651052 241705013 53962 duplication 2 241687451 241687509 1 probe 0.42 Supported SV 3 100340351 100451628 111278 duplication 3 105065420 106905 0.61 Supported SV 3 165014055 165083895 42602 deletion 3 155065479 1 probe 0.17 Not supported SV 16 78037966 78081307 36813 deletion - - No probes - 0.08 SV 2 130790519 131181302 390783 deletion - - No pr		SV	14	86454708	86498388	31877 deletion	-	-	-	No probes	-	Inconclusive
SV 2 55920504 55948466 27963 duplication - - No probes - Inconclusive SV 2 131031140 131977013 74897 deletion - - 10 probes mean=-0.01 Not supported SV 2 241651052 241705013 53962 duplication 2 241687451 241687509 1 probe 0.42 Supported SV 2 100340351 100451628 111278 duplication 3 100419565 100419624 2 probes 0.61 Supported SV 3 165014055 165083895 42602 deletion 3 165065479 1 probe 0.17 Not supported SV 16 78037966 78081307 36813 deletion - - No probes 0.08 SV 16 7803790519 131181302 390783 deletion 7 6146033 6146032 1 probe -0.005 - 0.08 SV 7 6118254 6146732		SV	1	22308530	22333055	23125 deletion	-	-	-	No probes	-	Inconclusive
SV 2 131031140 131977013 74897 deletion - - - 10 probes mean=-0.01 Not supported 78 SV 2 241651052 241705013 53962 duplication 2 241867509 1 probe 0,42 Supported SV 3 100340351 100451628 111278 duplication 3 100360692 100360751 2 probes 0,61 Supported SV 3 165014055 165083895 42602 deletion 3 150565420 165065479 1 probe 0,17 Not supported SV 16 78037966 78081307 36813 deletion - No No probes 0,08 SV 2 130790519 131181302 390783 deletion 2 131015497 131015556 3 probes -0,03 Not supported SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 Not supported SV 10 5		SV	2	55920504	55948466	27963 duplication	-	-	-	No probes	-	Inconclusive
SV 2 241651052 241705013 53962 duplication 2 241687801 241687909 1 probe 0.42 Supported SV 3 100340351 100451628 111278 duplication 3 100360692 1000419624 2 probes 0.64 Supported SV 3 165014055 165083895 42602 deletion 3 165065420 165065479 1 probe 0.17 Not supported SV 16 78037966 78081307 36813 deletion - - No probes 0.08 SV 2 13079519 131181302 390783 deletion 2 13105497 13101556 3 probes -0,03 Not supported SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 Not supported SV 9 10581280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 57		SV	2	131031140	131977013	74897 deletion	-	-	-	10 probes	mean=-0.01	Not supported
SV 3 100340351 100451628 111278 duplication 3 100360692 100360751 2 probes 0,54 Supported SV 3 165014055 165083895 42602 deletion 3 100419654 1 probe 0,17 Not supported SV 16 78037966 78081307 36813 deletion - - No probes 0,68 SV 16 78037966 78081307 36813 deletion - - No probes 0,08 SV 2 130790519 131181302 390783 deletion 2 13101556 3 probes -0,05 SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 Not supported SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 564747 5716793 52047 duplication - - No probes - Inconc	78	SV	2	241651052	241705013	53962 duplication	2	241687451	241687509	1 probe	0,42	Supported
SV 3 100419654 100419624 1 probe 0,61 Chapter SV 3 165014055 165083895 42602 deletion 3 165065420 165065479 1 probe 0,17 Not supported SV 16 78037966 78031307 36813 deletion - - No probes Inconclusive SV 2 130790519 131181302 390783 deletion 2 130928976 130929035 0,08 SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,05 SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 12102683 12140526 37844 duplication - - No probes - Inconclusive SV 10 12102683 12140526 37844 duplication - - No probes - Inconclusive SV 11		SV 3 10034	100340351	100451628	0451628 111278 duplication	3	100360692	100360751	2 probes	0,54	Supported	
SV 3 165083895 42602 deletion 3 165065479 1 probe 0,17 Not supported SV 16 78037966 78081307 36813 deletion - - No probes 0,08 SV 2 130790519 131181302 390783 deletion 2 130929035 0,08 SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684245 1 probe 0,71 Supported SV 10 1202683 12140526 37844 duplication - - No probes - Inconclusive SV 10 1202683 1214052 26589 deletion - - No probes - Inconclusive SV 11 4905539					100.01010		3	100419565	100419624	- p	0,61	
SV 16 78037966 78081307 36813 deletion - - No probes Inconclusive SV 2 130790519 131181302 390783 deletion 2 130928976 130929035 3 probes -0,03 SV 2 130790519 131181302 390783 deletion 7 6146033 6146092 1 probe -0,05 SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 Not supported SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684186 5684245 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion 13 38106904		SV	3	165014055	165083895	42602 deletion	3	165065420	165065479	1 probe	0,17	Not supported
79 Sv 12 130790519 131181302 390783 deletion 2 130028976 130929035 0,08 SV 2 130790519 131181302 390783 deletion 2 131015497 131015556 3 probes -0,03 Not supported SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,05 - Inconclusive SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684186 5684245 1 probe 0,071 Supported SV 10 12102683 12140526 37844 duplication - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion - - No probes - Inconclusive SV 13 38066960 38129134 50		SV	16	78037966	78081307	36813 deletion	-	-	-	No probes		Inconclusive
SV 2 130790519 131181302 390783 deletion 2 131015497 131015556 3 probes -0,03 Not supported SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,05 -0,05 SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 56841265 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion - - No probes - Inconclusive SV 13 38066960 38129134 50292 deletion 13 3810			_				2	130928976	130929035		0,08	
P 2 131084449 131084508 -0,05 SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe -0,06 Not supported SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684186 5684245 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion 13 38106904 1 probe -1,03 Supported SV 13 38066960 38129134 50292 deletion 13 38106904 1 probe -1,03 Suported		SV	2	130790519	131181302	390783 deletion	2	131015497	131015556	3 probes	-0,03	Not supported
SV 7 6118254 6146732 21897 deletion 7 6146033 6146092 1 probe 0,06 Not supported SV 9 105811280 105874071 55238 deletion - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684186 5684245 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion 13 38106845 38106904 1 probe -1,03 Supported SV 13 38066960 38129134 50292 deletion 13 38106904 1 probe -1,03 Supported SV 14 74571941 74597810 </td <td></td> <td></td> <td></td> <td></td> <td></td> <td></td> <td>2</td> <td>131084449</td> <td>131084508</td> <td></td> <td>-0,05</td> <td></td>							2	131084449	131084508		-0,05	
SV 9 105811280 105874071 55238 deletion - - - No probes - Inconclusive SV 10 5664747 5716793 52047 duplication 10 5684186 5684245 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion - - - No probes - Inconclusive SV 13 38066960 38129134 50292 deletion 13 38106845 38106904 1 probe -1,03 Supported SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 106601449 106784215 182767 duplication 14 106665178 0,55 Supported SV 15 86274566 86553109 278544 duplicati		SV	7	6118254	6146732	21897 deletion	7	6146033	6146092	1 probe	-0,06	Not supported
SV 10 5664/47 5/16/93 5/2047 duplication 10 5684186 5684245 1 probe 0,71 Supported SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive SV 11 49055393 49105125 26589 deletion - - - No probes - Inconclusive SV 13 38066960 38129134 50292 deletion 13 38106845 38106904 1 probe -1,03 Supported SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 1066051149 106665178 0,55 0,59 Supported - - - No - - - No - - -		SV	9	105811280	105874071	55238 deletion	-	-	-	No probes	-	Inconclusive
SV 10 12102683 12140526 37844 duplication - - - No probes - Inconclusive 79 SV 11 49055393 49105125 26589 deletion - - - No probes - Inconclusive SV 13 38066960 38129134 50292 deletion 13 38106845 3810904 1 probe -1,03 Supported SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 106601449 106784215 182767 duplication 14 106655178 0,5 Supported SV 14 106601449 106784215 182767 duplication 14 106716345 106716404 3 probes 0,59 Supported SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV 15 86274566 86553109 278544 duplication_split 15 86286466 86542460 253		SV	10	5664747	5/16/93	52047 duplication	10	5684186	5684245	1 probe	0,71	Supported
V9 SV 11 49055393 49105125 26589 deletion - - - No probes - Inconclusive SV 13 38066960 38129134 50292 deletion 13 38106845 38106904 1 probe -1,03 Supported SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 106601449 106784215 182767 duplication 14 106665119 106665178 0,5 SV 14 106601449 106784215 182767 duplication 14 106716345 106716404 3 probes 0,59 Supported SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X </td <td>70</td> <td>SV</td> <td>10</td> <td>12102683</td> <td>12140526</td> <td></td> <td>-</td> <td>-</td> <td>-</td> <td>No probes</td> <td>-</td> <td>Inconclusive</td>	70	SV	10	12102683	12140526		-	-	-	No probes	-	Inconclusive
SV 13 38066960 38129134 50292 deletion 13 38106845 3810904 1 probe -1,03 Supported SV 14 74571941 74597810 25870 duplication - - - No probes - Inconclusive SV 14 106601449 106784215 182767 duplication 14 106665119 106665178 0,59 SV 14 106601449 106784215 182767 duplication 14 106716345 106716404 3 probes 0,59 SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X 23907510 23934260 26751 duplication X 23914694 23914753 1 probe 0.72 Supported	79	SV	11	49055393	49105125		-	-	-	No probes	-	Inconclusive
SV 14 74571941 74597810 25870 duplication - - No probes - Inconclusive SV 14 106601449 106784215 182767 duplication 14 1066651178 0,5 0,5 SV 14 106601449 106784215 182767 duplication 14 106716345 106716404 3 probes 0,59 SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X 23907510 23934260 26751 duplication X 23914694 23914753 1 probes 0.72 Supported		SV	13	38066960	38129134	50292 deletion	13	38106845	38106904	1 probe	-1,03	Supported
SV 14 106601449 106784215 182767 duplication 14 106055119 106055178 0,59 SV 14 106761345 106716345 106716404 3 probes 0,59 SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X 23907510 23934260 26751 duplication X 23914644 23914753 1 probe 0,71		50	14	74571941	74597810	25870 duplication	-	-	-	No probes	-	Inconclusive
SV 14 100/1442 102/10/04213 162/10/04 102/10/0404 5 probes 0,59 Supported SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X 23907510 23934260 26751 duplication X 2301/664 2301/753 1 probe 0,72 Supported		SV	11	106601440	10670/015	182767 duplication	14	106716245	106716404	3 probas	0,5	Supported
SV 15 86274566 86553109 278544 duplication_split 15 86288646 86542460 253814 bp mean=0.55 Confirmed SV X 23907510 23934260 26751 duplication X 23914644 23914753 1 probe 0.72 Supported		30	14	100001449	100784215		14	106774612	106774672	2 highes	0,59	Supported
SV 13 602274300 60333103 276344 duplication_split 13 6026040 60342400 233614 bp Intell=0.53 Colliniting SV X 23907510 23934260 26751 duplication X 2301/604 2301/753 1 probe 0.72 Supported		sv/	15	86274566	86553100	278544 duplication split	14	26288646	265/2/60	253814 hn	0,07	Confirmed
		SV	X 51	23907510	230371360	26751 duplication	× 12	2301/60/	2301/752	233014 UP	0.55 n 72	Supported

Table S7. Concordance of all filtered SVs and CNVs between optical genome mapping and CNV-microarray.

^a confirmed= the event is called by CNV-microarray (>=5 consecutive deviating probes with a LogR>0.25 for duplication and <-0.25 for deletion), supported= less than 5 probes but consistent LogR deviation (threshold>0,25 for dup <-0,25 for dup], inconclusive = uncovered region (no probes in the microarray at the respective region), Not supported = CNV-microarray and OM results are not matching