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Supplemental information

Optical genome mapping enables constitutional chromosomal aberration detection

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Supplemental Data Description

Supplemental Data includes 12 figures and 7 tables.

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

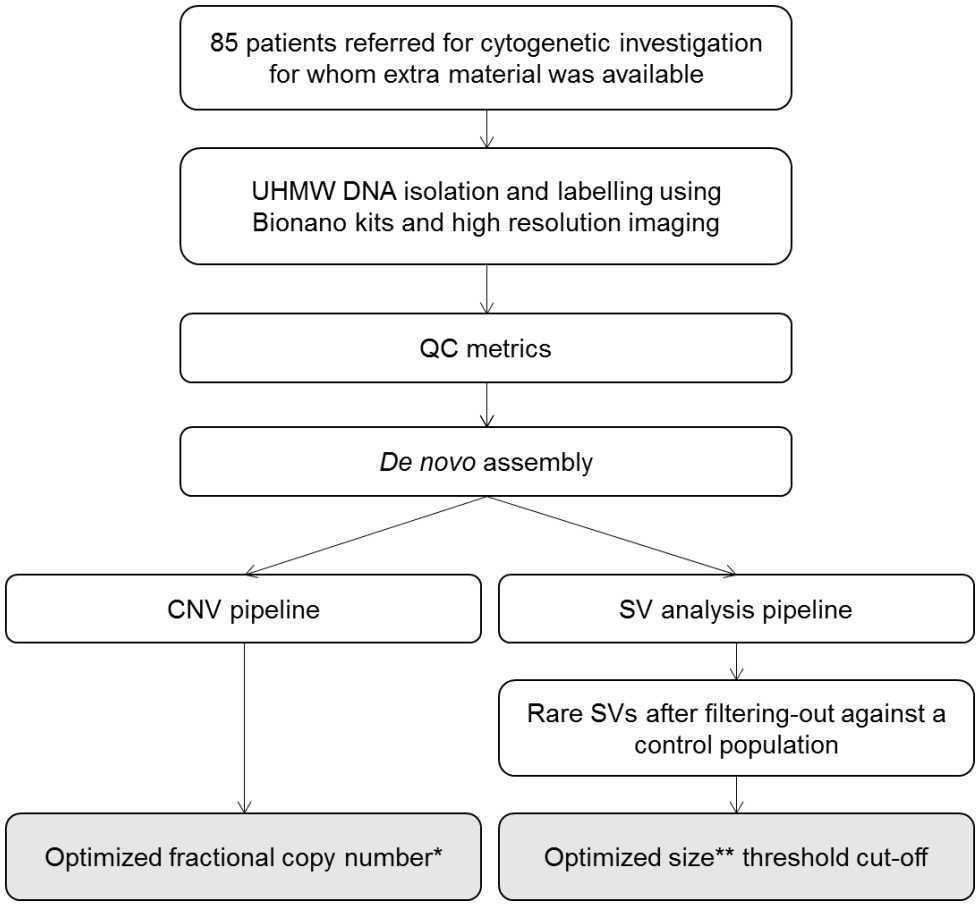


Figure S1. Workflow of optical genome mapping technique.

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 ≤1.2 for losses and ≥2.8 for gains, **recommended SV pipeline size cut-off = 20 Kb

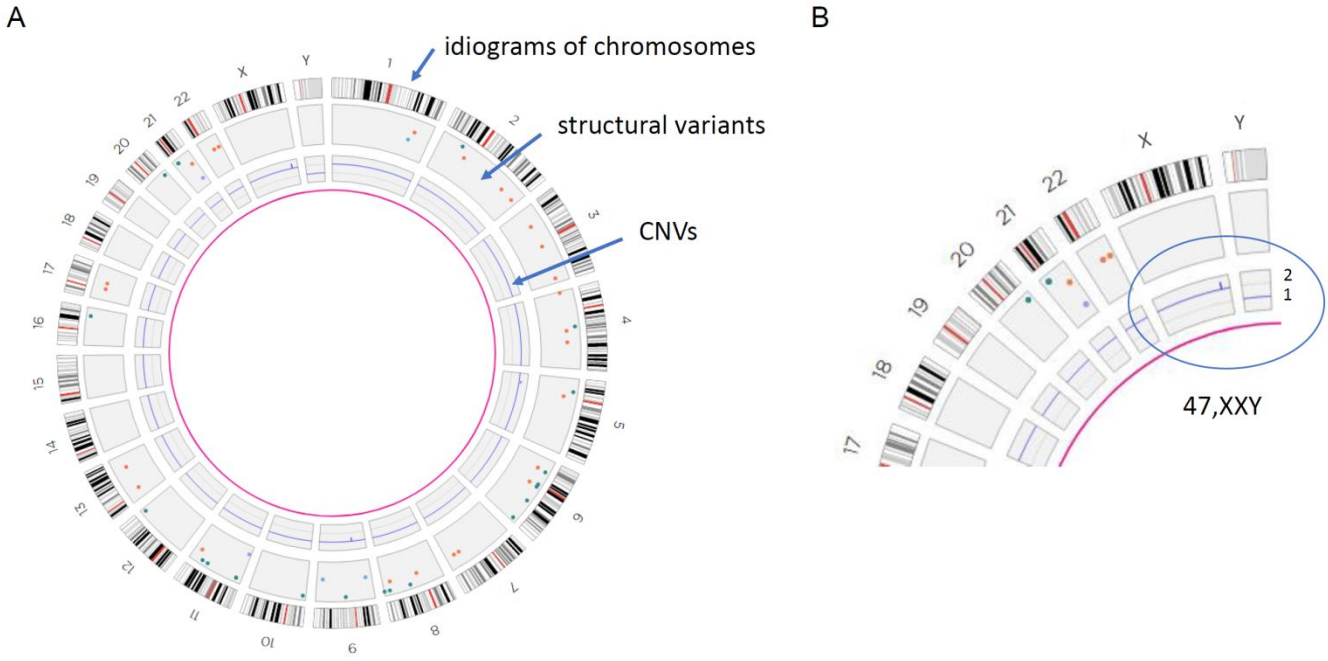
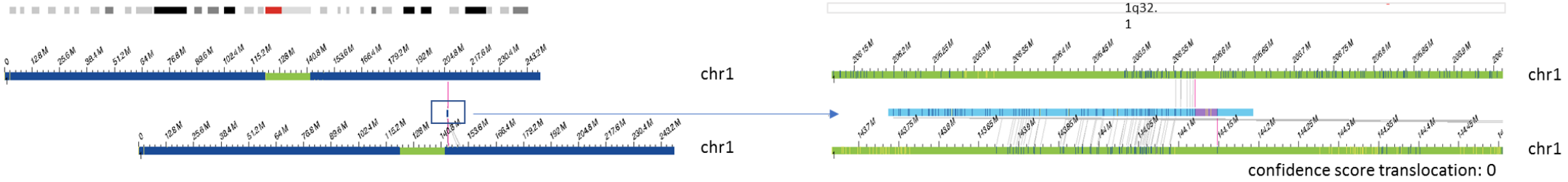


Figure S2. Visual representation of optical genome mapping data.

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).

A) Intrachromosomal rearrangement chr 1 (sample 22)



B) Intrachromosomal rearrangement chr 9 (sample 11)

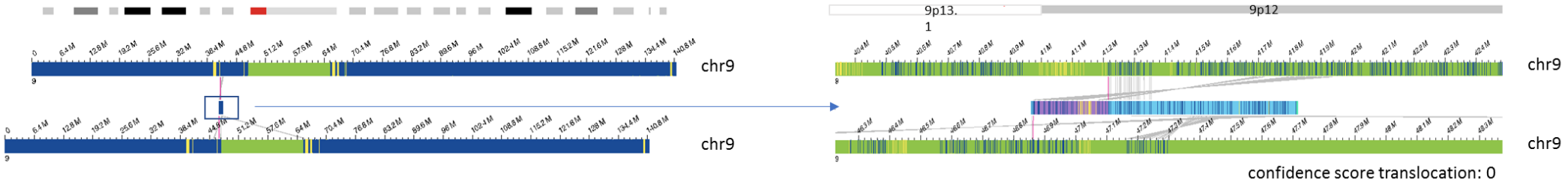


Figure S3 Potential mapping artifacts.

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.

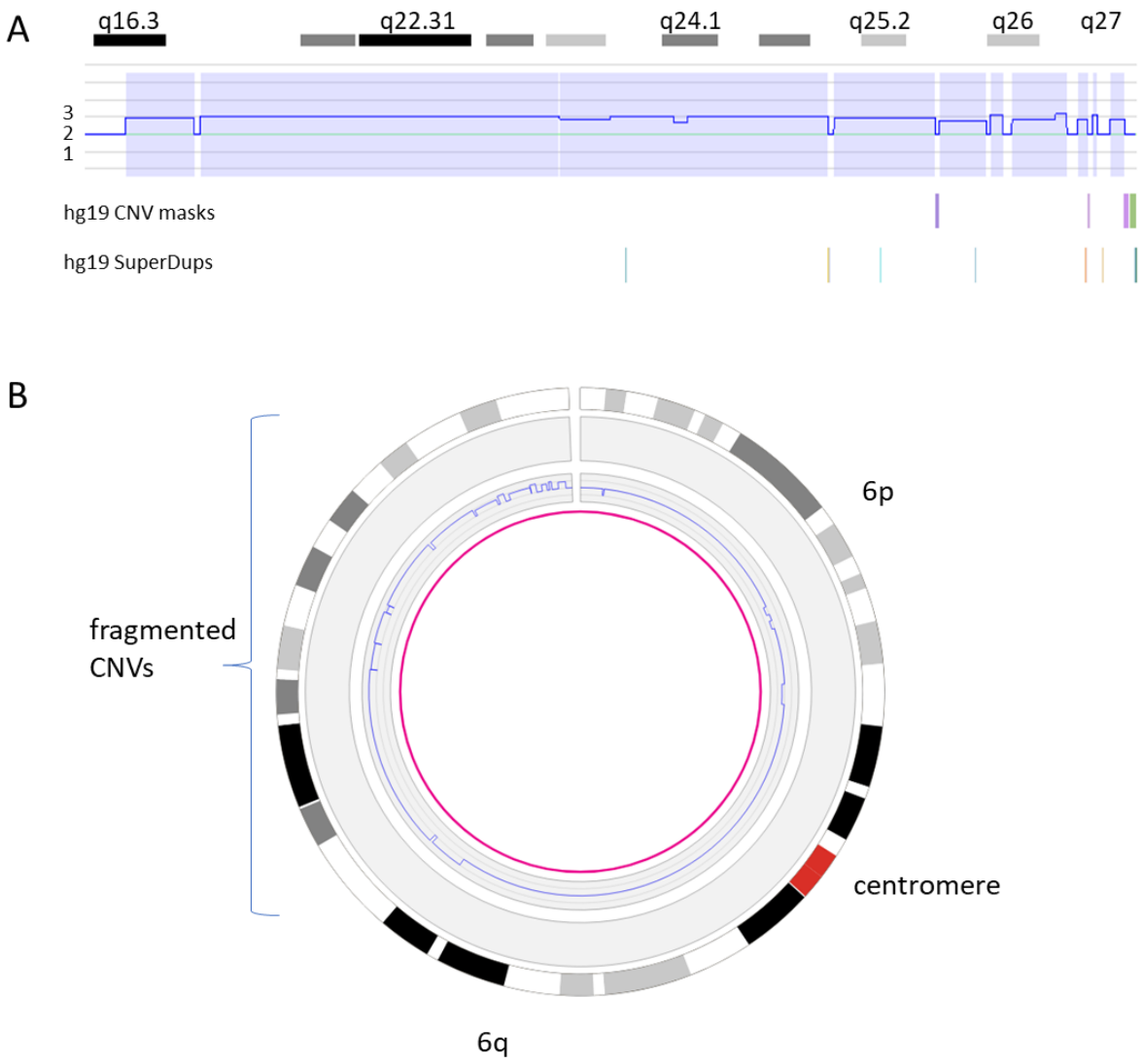
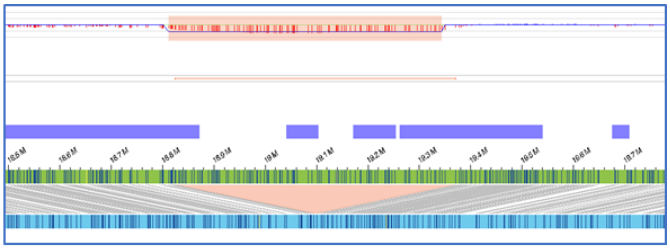


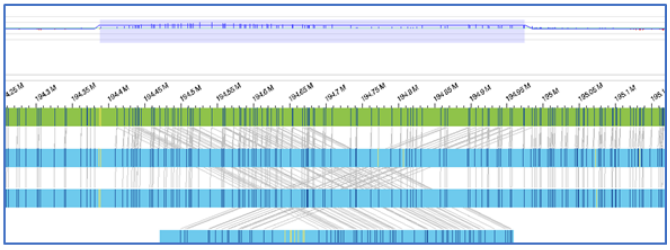
Figure S4. Representative example of fragmented CNVs.

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.

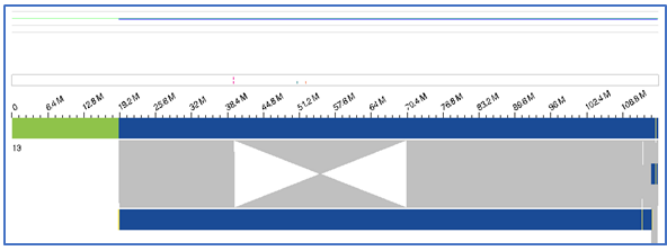
Deletion
Sample 1: 8p22p21.3(18825888_19364764)x1



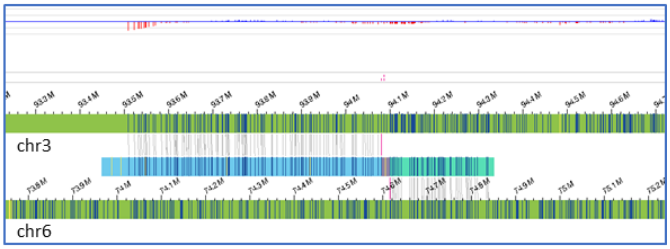
Duplication
Sample 7: 3q29(194349341_194988874)x3



Inversion
Sample 15:
inv(13)(q12q22)



Translocation balanced
Sample 14:
t(3;6)(q11.2;q13)



Translocation unbalanced
Sample 26:
t(12;20)(p13.32;p12.3),
12p13.33p13.32 loss, 20p13p12.3 gain

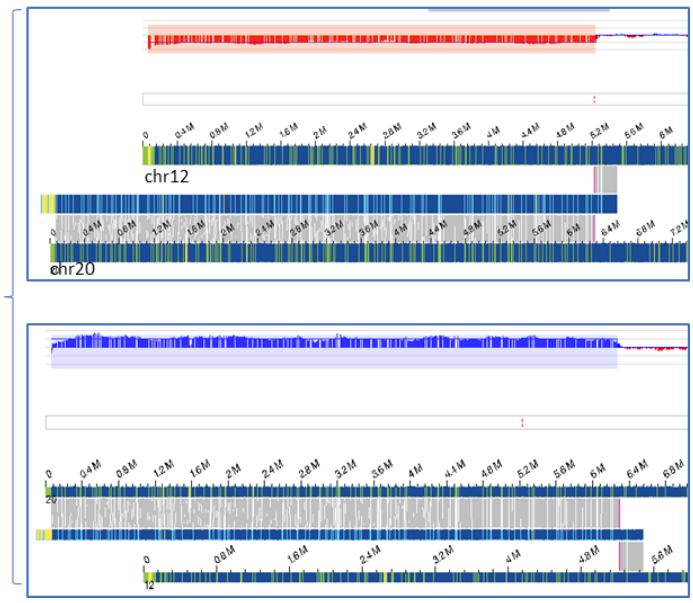
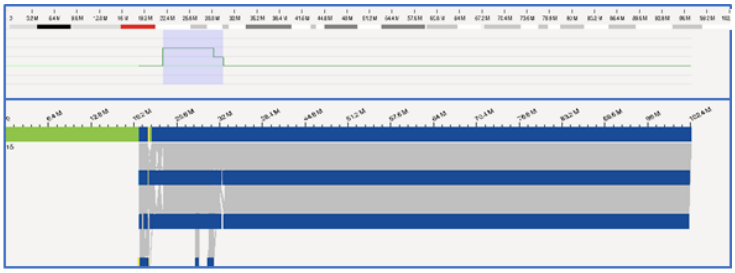
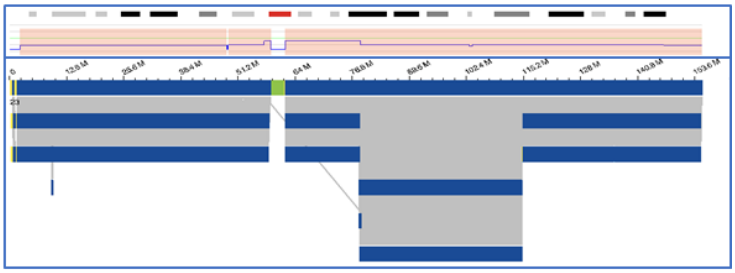


Figure S5 (1/2). Representation of different chromosomal abnormalities.
This figure shows an example of each type of chromosomal aberration that was detected in this study. All aberrations shown are described in detail in Table S1.

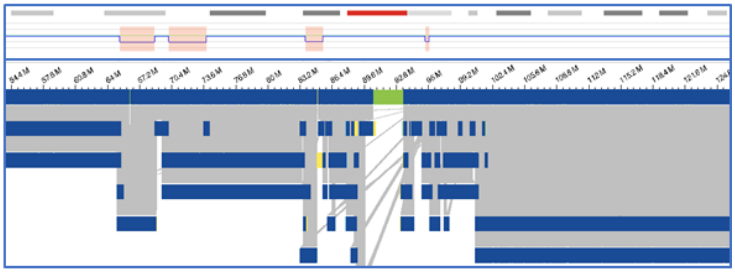
Isochromosome
Sample 77:
15q11.2q13.2 gain (4x)
15q13.3 gain (3x)



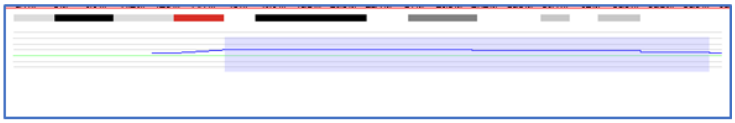
Ring chromosome
Sample 39:
r(X)(p11.21q21.1)



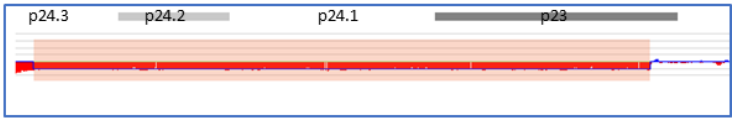
Complex rearrangement
Sample 66:
3p14.1(65238298_68667113)x1
3p13(70127345_73724765)x1
3p12.1(83784489_85467284)x1
3q11.2(97180779_97270083)x1



Aneuploidy
Sample 24:
trisomy 21 via Robertsonian translocation



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



Large duplication
Sample 60:
8p23.1p12 gain

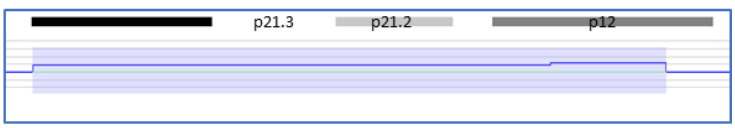


Figure S5 (continued 2/2). Representation of different chromosomal abnormalities.
This figure shows an example of each type of chromosomal aberration that was detected in this study. All aberrations shown are described in detail in Table S1.

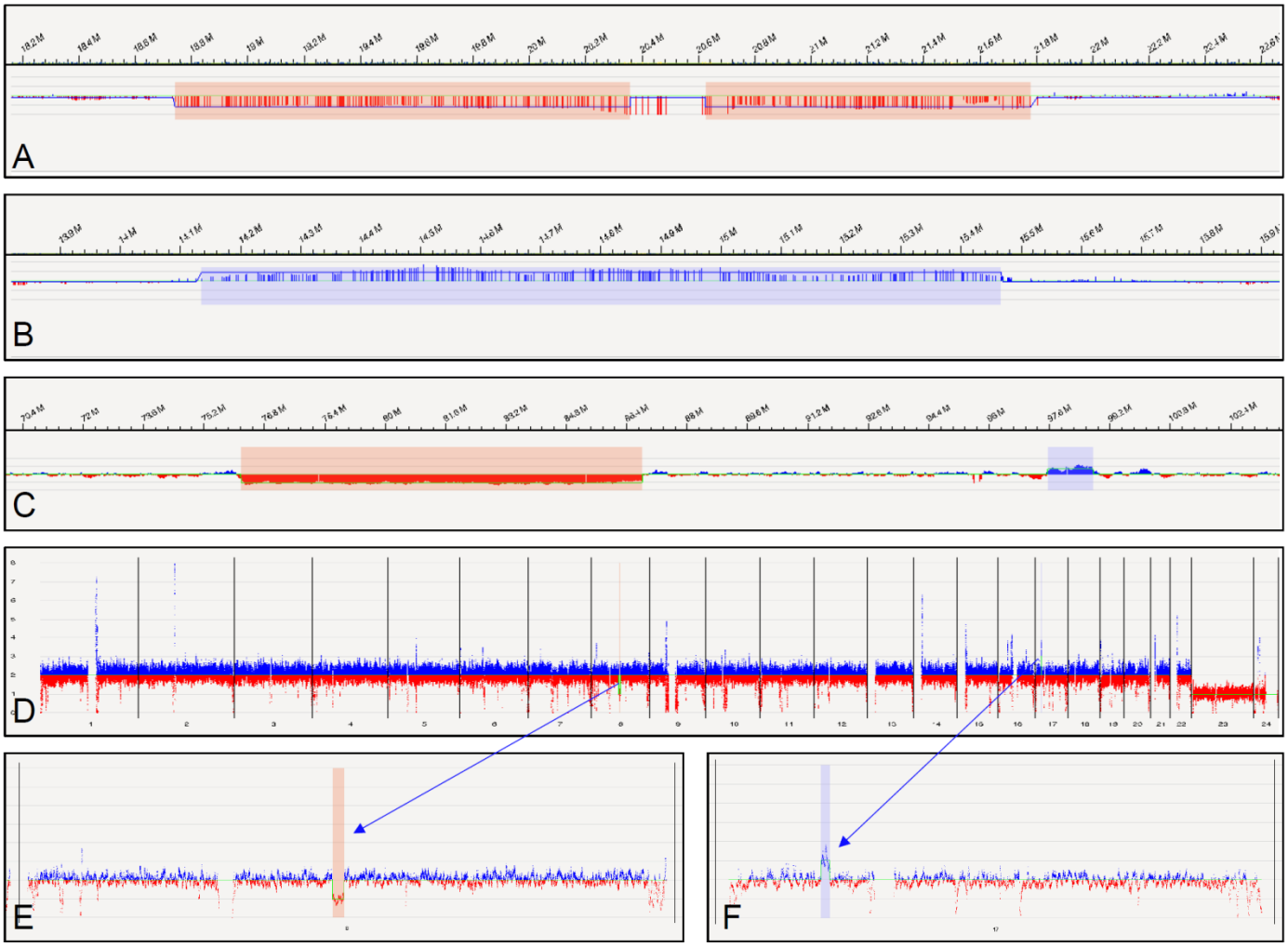


Figure S6. Representative CNV profiles obtained with optical genome mapping, for different samples.

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.

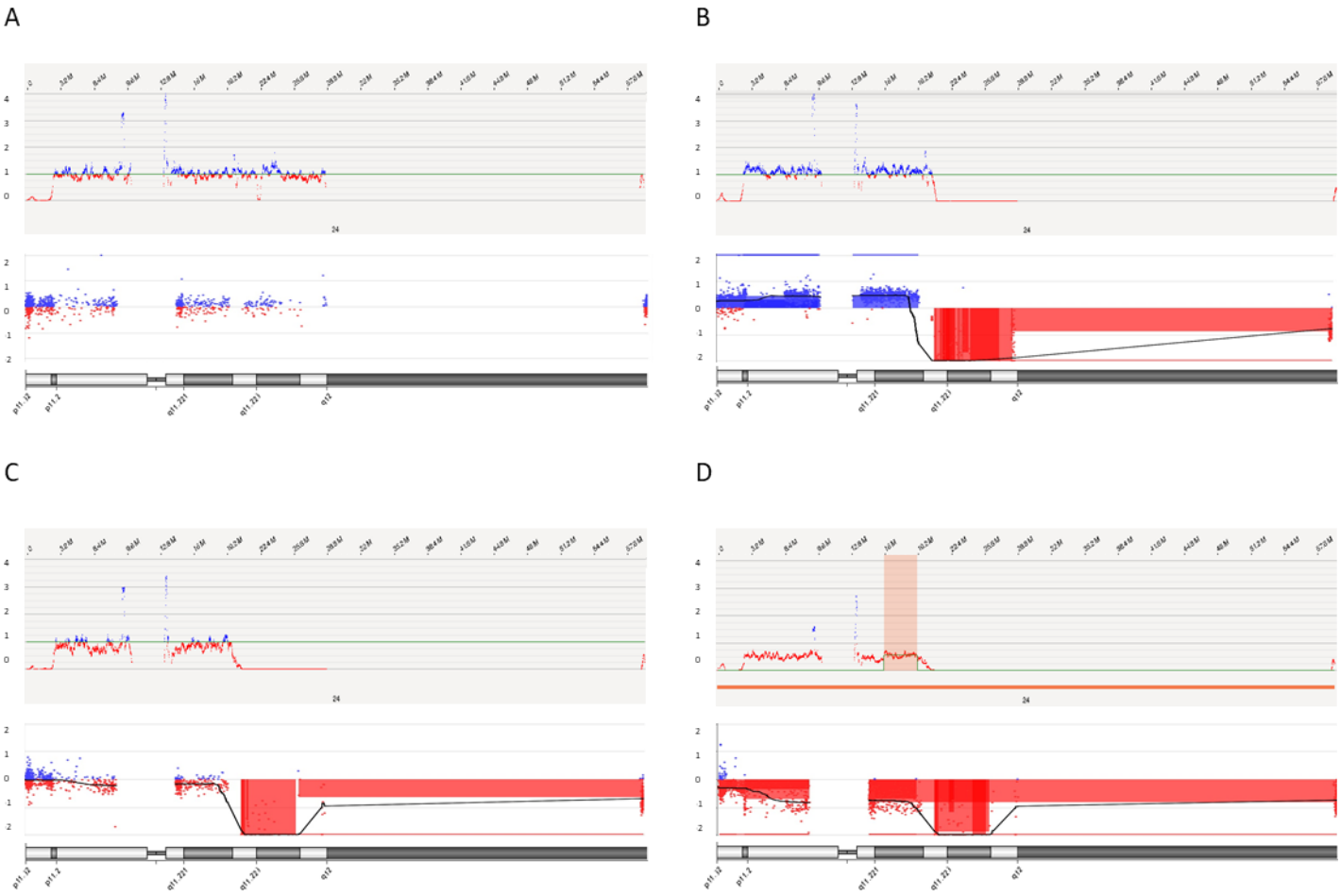


Figure S7: Isodicentric Y-chromosomes.

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.

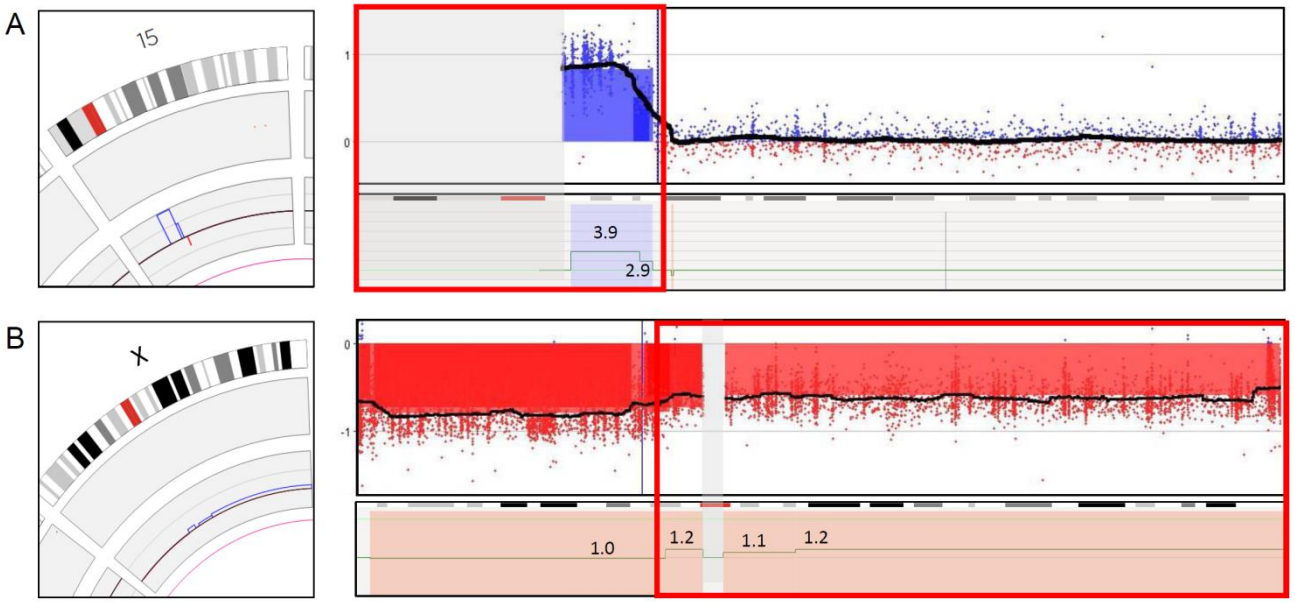


Figure S8. Optical genome mapping and CNV-microarray profiles for isochromosomes 15 and X.

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).

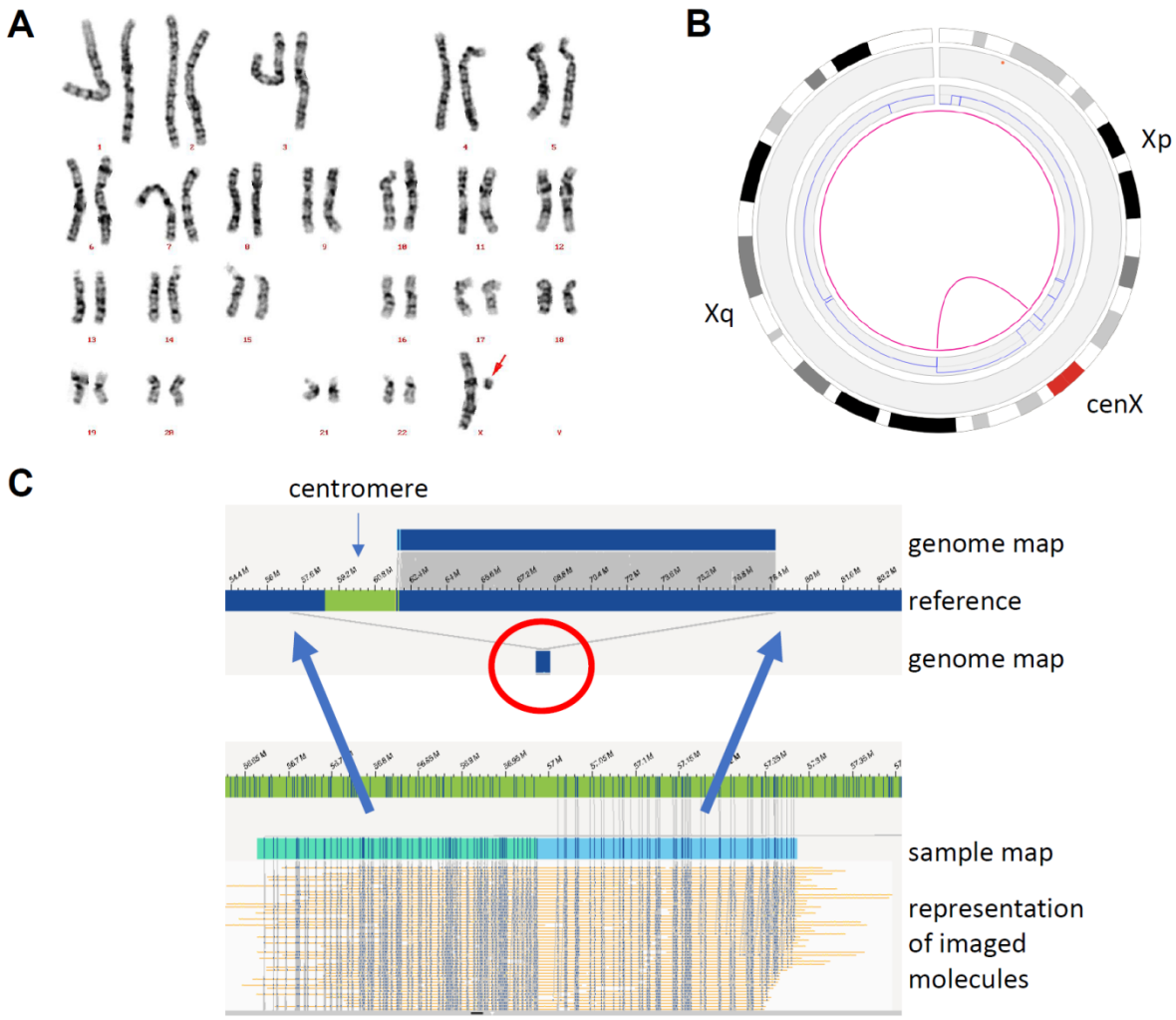


Figure S9. Small ring chromosome X.

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.

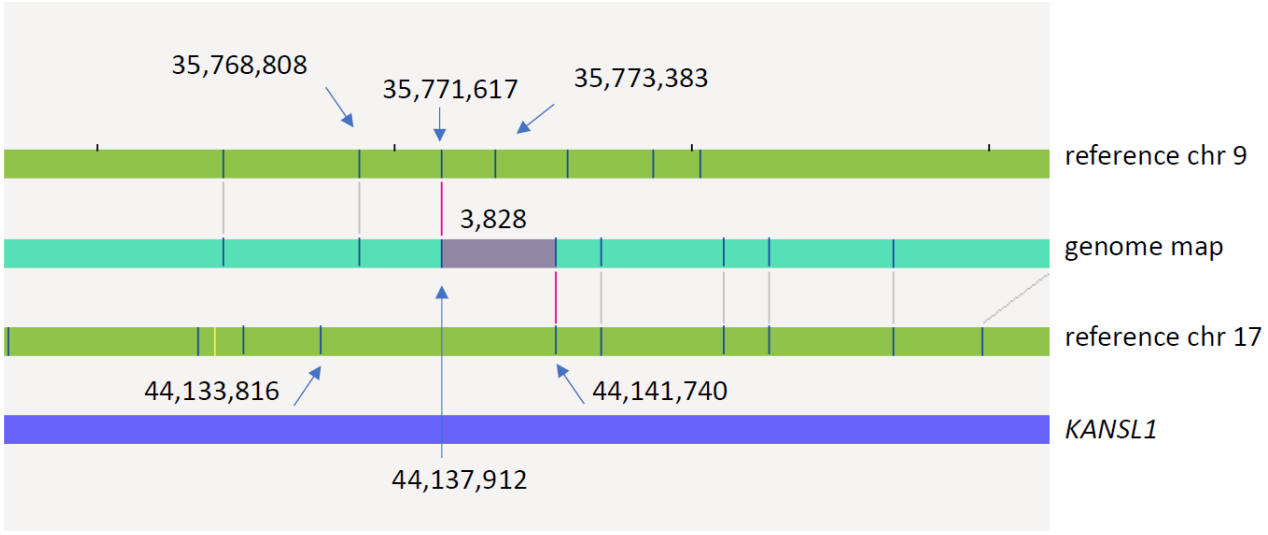


Figure S10. Optical genome mapping breakpoint detection for translocation t(9;17)(p13;q21), disrupting the gene *KANSL1*.

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.

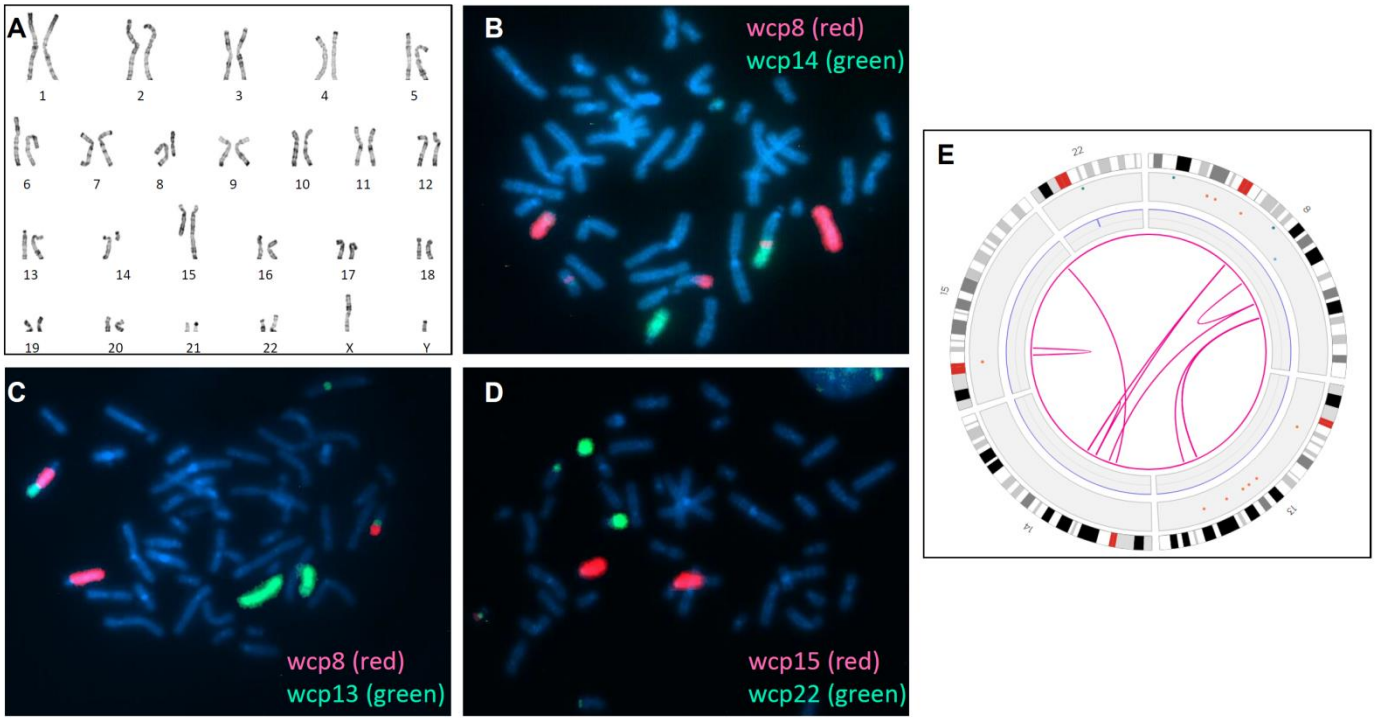


Figure S11. Complex sample 52.

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),der(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.

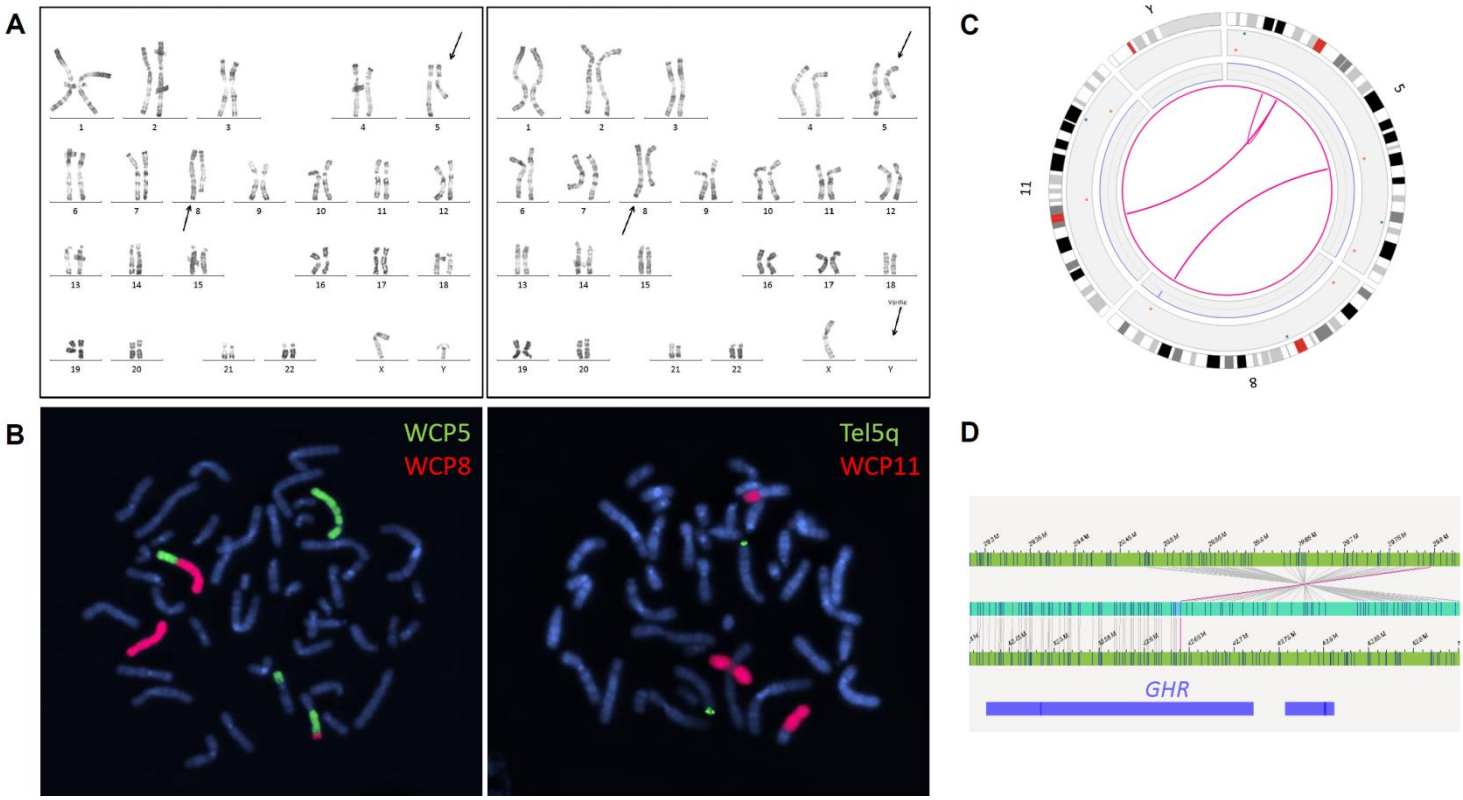


Figure S12: Complex sample 55.

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

Table S2. Technical performance of optical genome mapping.

Sample	Starting material	Total DNA >=150 kbp (Gbp)	Avg N50 >=150kbp (Mbp)	Avg N50 >=20kbp (Mbp)	Map rate (>= 150 kbp)	Effective coverage (x)	Avg label density (per 100kbp)
1	EDTA blood	1 320	0,24	0,12	69%	286	15,69
2	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
4	EDTA blood	1 305	0,24	0,13	58%	243	17,14
5	EDTA blood	1 347	0,26	0,21	87%	368	15,20
6	EDTA blood	811	0,21	0,08	43%	110	15,28
7	EDTA blood	807	0,21	0,10	55%	140	15,34
8	EDTA blood	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	Hep. Blood	813	0,20	0,12	63%	149	13,27
18	Hep. Blood	823	0,20	0,12	64%	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	Amniotic fluid	801	0,30	0,22	85%	214	15,39
28	Amniotic fluid	849	0,26	0,17	78%	186	12,77
29	Chorionic villi	617	0,28	0,21	90%	173	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	Amniotic fluid	817	0,33	0,26	90%	228	14,97
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	Hep. Blood	845	0,30	0,25	89%	233	14,91
37	Hep. Blood	809	0,25	0,10	71%	178	14,74
38	Hep. Blood	857	0,27	0,22	89%	233	14,37
39	Hep. Blood	855	0,25	0,17	86%	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	EDTA blood	499	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	EDTA blood	488	0,23	0,10	70%	107	15,58
48	EDTA blood	486	0,30	0,25	90%	137	14,60
49	Lymphoblastoid cell line	376	0,28	0,20	79%	95	15,79
50	Lymphoblastoid cell line	328	0,28	0,20	70%	73	16,38
51	Lymphoblastoid cell line	484	0,29	0,20	64%	98	15,74
52	Lymphoblastoid cell line	348	0,29	0,20	78%	87	15,71
53	Lymphoblastoid cell line	495	0,28	0,19	73%	115	15,42
54	Lymphoblastoid cell line	345	0,27	0,16	75%	82	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	EDTA blood	324	0,26	0,17	74%	75	15,17
60	EDTA blood	481	0,29	0,22	75%	113	15,27
61	EDTA blood	333	0,22	0,10	68%	71	15,05
62	EDTA blood	492	0,29	0,13	74%	113	15,90
63	EDTA blood	496	0,25	0,16	84%	129	15,04
64	EDTA blood	504	0,28	0,20	90%	143	15,44
65	EDTA blood	346	0,27	0,17	89%	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	EDTA blood	400	0,31	0,31	87%	109	14,47
72	EDTA blood	1 311	0,22	0,12	50%	208	14,88
73	EDTA blood	400	0,32	0,32	84%	105	15,75
74	EDTA blood	911	0,25	0,15	38%	109	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
78	EDTA blood	436	0,27	0,18	91%	122	15,06
79	EDTA blood	483	0,32	0,24	87%	132	14,92
80	EDTA blood	424	0,31	0,25	92%	120	15,06
81	EDTA blood	483	0,31	0,24	93%	140	15,62
82	EDTA blood	449	0,29	0,20	91%	127	15,56
83	EDTA blood	411	0,27	0,17	86%	110	15,94
84	EDTA blood	406	0,27	0,13	88%	110	15,29
85	Lymphoblastoid cell line	481	0,29	0,11	77%	117	15,30
Average		655	0,27	0,18	77%	152	15,13
Max value		1 347	0,36	0,36	93%	368	22,36
Min value		200	0,19	0,08	38%	38	12,77
Standard deviation		270	0,04	0,06	12%	59	1,21

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%
Unbalanced structural aberrations	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 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%
Balanced structural aberrations	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.

Aberration type	Detection of aberration			Localization of aberration		
	Standard of care genome-wide tests		Optical genome mapping	Standard of care genome-wide tests		Optical genome mapping
	CNV-microarray	Karyotyping		CNV-microarray	Karyotyping	
	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	✓	✓	✓	×	✓	✓
Unbalanced Robertsonian translocation	✓	✓	✓	×	✓	×
Balanced Robertsonian translocation	×	✓	×	×	✓	×
Unbalanced reciprocal translocation	✓	✓	✓	×	✓	✓
Balanced reciprocal translocation	×	✓	✓	×	✓	✓
Deletion	✓	✓	✓	×	✓	✓
Insertion	unbalanced only	✓	✓	×	✓	✓
Duplication	✓	✓	✓	×	✓	✓
Inversion	×	✓	✓	×	✓	✓
Isochromosome	✓	✓	✓	×	✓	✓
Ring chromosome	unbalanced only	✓	✓	×	✓	✓

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

Sample ID	Bionano call						CNV-microarray call					Result[a]
	Bionano tool	Chr	Start	End	Size (bp)	Type	Chr	Start	End	Call size or # of probes	LogR	
56	SV	4	161863865	161923311	59447	duplication	4	161869551	161869610	1 probe	0,5	Supported
	SV	10	81438916	81594303	137161	deletion	-	-	-	No probes	-	Inconclusive
	SV	16	34462037	34556454	94418	duplication_inverted	16	34482042	34482101	2 probes	0,5	Supported
	SV	16	34462037	34578881	116845	duplication_inverted	16	34565215	34565274		0,5	Supported
	SV	16	79404969	79430008	20599	deletion	16	79420905	79420964	1 probe	-0,76	Supported
62	SV	1	49908206	50001818	84129	deletion	1	49959780	49959839	1 probe	-0,92	Supported
	SV	1	246313442	246520313	206871	deletion	1	246321447	246501692	180245 bp	mean='-1.12	Confirmed
	SV	2	233239437	233288334	27988	deletion	2	233270613	233270672	1 probe	-0,77	Supported
	SV	5	120421707	120472623	43718	deletion	-	-	-	No probes	-	Inconclusive
	SV	9	71845510	71961535	116026	duplication	9	71846678	71846737	3 probes	0,55	Supported
							9	71917699	71917758		0,39	
							9	71957664	71957723		0,57	
	SV	18	65845905	65907488	49589	deletion	18	65891840	65891899	1 probe	-0,54	Supported
64	CNV	7	76109311	76623844	514533	deletion	7	76139282	76558024	418743 bp	mean=-0.85	Confirmed
	CNV	8	2120483	2349321	228838	deletion	8	2206158	2206217	2 probes	0,16	Not supported
							8	2308926	2308985		-0,07	
	SV	4	138246546	138337696	91151	duplication	4	138259451	138259510	2 probes	0,42	Supported
							4	138324113	138324172		0,59	
	SV	9	32737531,7	32787627,8	36735	deletion	9	32759696	32759755	1 probe	-1,02	Supported
77	CNV	X	6503942	8131734	1627792	duplication	X	6488721	8097511	1608791 bp	mean=0.51	Confirmed
	SV	4	139888286	139919269	23983	deletion	-	-	-	No probes	-	Inconclusive
	SV	11	85146480	85175179	23022	deletion	-	-	-	No probes	-	Inconclusive
	SV	14	86454708	86498388	31877	deletion	-	-	-	No probes	-	Inconclusive
78	SV	1	22308530	22333055	23125	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	100360692	100360751	2 probes	0,54	Supported
							3	100419565	100419624		0,61	
	SV	3	165014055	165083895	42602	deletion	3	165065420	165065479	1 probe	0,17	Not supported
	SV	16	78037966	78081307	36813	deletion	-	-	-	No probes		Inconclusive
79	SV	2	130790519	131181302	390783	deletion	2	130928976	130929035	3 probes	0,08	Not supported
							2	131015497	131015556		-0,03	
							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	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	3 probes	0,5	Supported
							14	106716345	106716404		0,59	
							14	106774613	106774672		0,67	
	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

[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 del), inconclusive = uncovered region (no probes in the microarray at the respective region), Not supported = CNV-microarray and OM results are not matching*