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TITLE: KromaTiD dGH SCREEN Whole Genome Analysis Report

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I. PROJECT INFORMATION

Project Quote #	Q23xxxx
Sample Type	Cell type
Sample ID	S01xxxx, [sample name]
Gender	Female
Passage number (or N/A)	N/A
Study Objective	Assessment of genomic integrity
Sample Receipt Date	8/xx/23
Report Date	8/xx/23

I. CHROMOSOME HARVEST

Harvest is performed per SOP-0044.

Condition	Result
Analog incubation time:	**27 hrs**
Colcemid incubation time	**Performed by customer**
Harvest Option	**Standard Protocol: X** **Modified Conditions: N/A**

II.   dGH SCREEN ASSAY

Assay	Standard dGH SCREEN **AS-0002.1 (50 Cells)**
Metaphase and Karyotype Qualification	Samples must be prepared and qualified for dGH analysis prior to running the assay. dGH SCREEN is designed for samples with grossly normal karyotypes and has not been qualified for highly rearranged genome analysis. Spread resolution of 350+ (G-band equivalent) is selected for analysis.
Assay Description	The five-color whole genome assay (5CWG or dGH SCREEN) is a dGH paint combination assay for all 24 human chromosomes. The assay is composed of unique sequence, high-density (HD) dGH chromosome paints in five color panels such that chromosomes painted in the same color can be differentiated by size, shape, and centromere position. Results include per-chromosome attribution of inter- and intra-chromosomal structural events including inversions, translocations, aneuploidy (gain and loss), insertions, centromere abnormalities and complex events across a sample. Prior to analysis, images of dGH SCREEN painted metaphase spreads are qualified, processed and sorted into karyograms for rapid, consistent reading of the assay. Per-cell event assessment is performed in an excel workbook which is built to be leveraged for population-level analysis of events ranging from random to clonal.

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III. RESULTS

Cells Analyzed	50	Total Karyograms	50

Chromosome Aberration Data:

Table 1: Structural Events

Event	Number of Events	Number Of Cells with Events
Gain	0	0
Loss	6	4
Inversion	41	32
SCE	211	49
Insertion	0	0
Size Diff	146	46
Translocations	0	0
Complex	6	5
Total	**410**	**50**

Table 2: Distribution of Events by Chromosome

Count of Events by Chromosome

Chromosome	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	X	Y	Number of Events	Number of Cells w/ Events
Gain	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Loss	0	0	0	0	0	0	0	0	0	0	1	1	0	0	1	0	0	0	0	1	1	1	0	0	6	4
Inversion	0	2	1	1	1	0	2	27	1	2	0	0	2	0	0	0	0	1	0	0	0	0	1	0	41	32
SCE	20	19	15	21	10	15	9	8	11	10	11	8	12	6	3	3	5	6	3	1	1	2	12	0	211	49
Insertion	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Size Diff	16	17	11	12	10	15	9	0	10	5	7	7	1	1	3	3	3	4	3	2	0	0	7	0	146	46
Translocations	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Complex	1	1	1	0	1	1	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	0	0	6	5
Total	37	39	28	34	22	31	20	35	22	17	19	16	15	8	7	6	8	11	6	4	2	3	20	0	410	50

Figure 1: Distribution of Event Rates by Number of Cells

Number of Cells by Event Type

Legend: Gain, Loss, Inversion, SCE, Insertion, Size Diff, Translocations, Complex
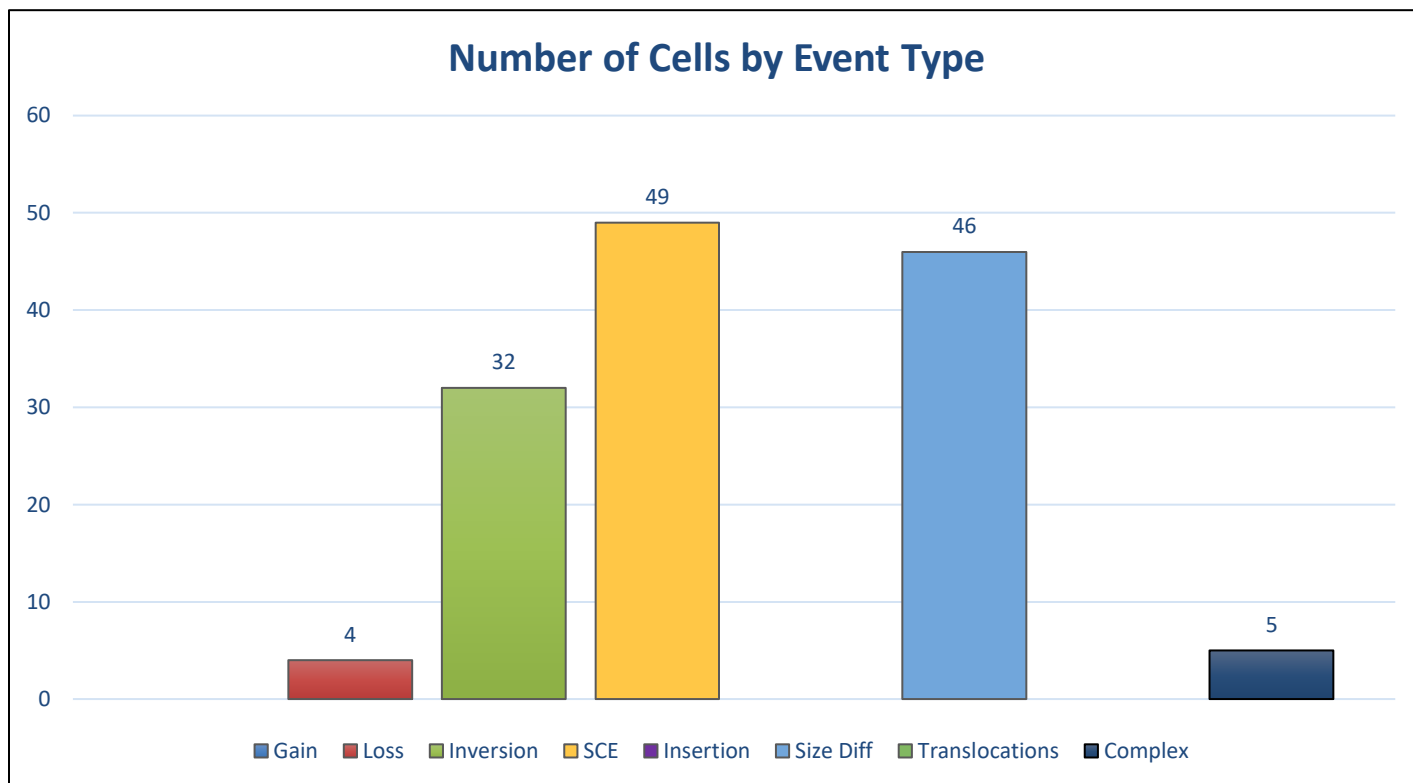
Table 3: Aneuploidy Summary (whole chromosome gain and loss)

	Number of Cells	Percentage of Cells
Gain Event	0	0%
Loss Event	4	8%

Table 4: Aneuploidy detail, by chromosome identity and cell number.

Location	Cell Name	Event	Gain Loss Values
Cell 16 / Chrom-12	Cell 16	Loss	-1
Cell 16 / Chrom-15	Cell 16	Loss	-1
Cell 22 / Chrom-21	Cell 22	Loss	-1
Cell 23 / Chrom-22	Cell 23	Loss	-1
Cell 33 / Chrom-11	Cell 33	Loss	-1
Cell 33 / Chrom-20	Cell 33	Loss	-1

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Table 5: Inversion and Sister Chromatid Exchange Event Summary

Inversion/SCE Event Counts by Chromosome

	P-Arm Inversion Centric Small	P-Arm Inversion Mid-Arm Mid-Size	P-Arm Inversion Mid-Arm Small	P-Arm Exchange Large Sister Chromatid	P-Arm Exchange Mid-Sized Sister Chromatid	P-Arm Exchange Small Sister Chromatid	P-Arm Terminal Inversions	Pericentric Inversion	Q-Arm Inversion Centric Small	Q-Arm Inversion Mid-Arm Mid-Size	Q-Arm Inversion Mid-Arm Small	Q-Arm Exchange Large Sister Chromatid	Q-Arm Exchange Mid-Sized Sister Chromatid	Q-Arm Exchange Small Sister Chromatid	Q-Arm Terminal Inversions	Grand Total
1	0	0	0	3	8	2	0	1	0	0	0	1	5	0	0	20
2	0	0	0	2	6	0	0	0	0	1	1	4	7	0	0	21
3	0	0	0	0	8	1	0	0	0	1	0	2	4	0	0	16
4	0	0	0	0	4	1	0	1	0	1	0	3	12	0	0	22
5	1	0	0	0	5	0	0	1	0	0	0	2	2	0	0	11
6	0	0	0	0	1	1	0	0	0	0	0	2	11	0	0	15
7	0	1	1	0	3	0	0	0	0	0	0	2	4	0	0	11
8	0	0	27	0	0	1	0	0	0	0	0	0	7	0	0	35
9	0	0	1	0	4	0	0	1	0	0	0	0	4	2	0	12
10	0	0	2	0	2	0	0	0	0	0	0	0	7	1	0	12
11	0	0	0	0	3	1	0	0	0	0	0	2	5	0	0	11
12	0	0	0	0	2	0	0	0	0	0	0	1	5	0	0	8
13	0	0	0	0	0	0	0	0	1	1	0	3	9	0	0	14
14	0	0	0	0	0	0	0	0	0	0	0	0	6	0	0	6
15	0	0	0	0	0	0	0	0	0	0	0	0	3	0	0	3
16	0	0	0	0	1	0	0	0	0	0	0	0	2	0	0	3
17	0	0	0	0	1	0	0	0	0	0	0	1	3	0	0	5
18	0	0	0	0	0	0	0	0	0	0	1	1	5	0	0	7
19	0	0	0	0	2	0	0	0	0	0	0	0	1	0	0	3
20	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	1
21	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	1
22	0	0	0	0	0	0	0	0	0	0	0	0	1	1	0	2
X	0	0	1	0	2	2	0	1	0	0	0	2	5	0	0	13
Y	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
Grand Total	1	1	32	5	52	9	0	5	1	4	2	26	110	4	0	**252**
Inversion Total	1	1	32				0		1	4	2				0	41
SCE Total				5	52	9		5				26	110	4		211

Recurrent SCE/Inversion Event Summary

There is a recurrent small inversion on Chr8p present in 54% of cells.
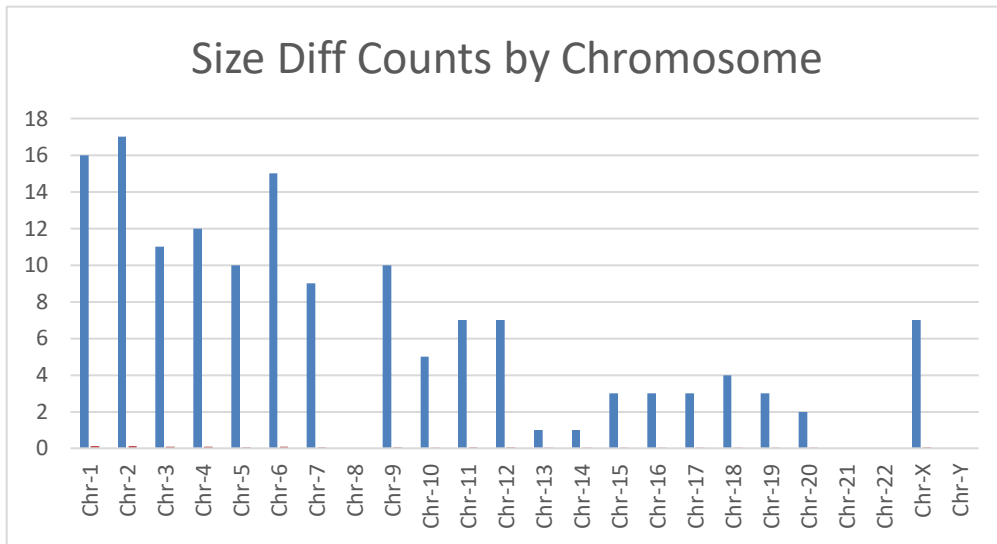
Table 6: Insertion Event Summary

There are no insertion events in the sample.

Table 7: Translocation Event Summary

There are no translocation events in the sample.

Table 8: Size Difference Summary:

Complex Events:

There were four cells that each contained a single complex event, and one cell with two events.

Table 9: Complex Events Summary:

Cell	Chromosome	Complex Event	Code
4	3	Broken chromosome	C4
15	2	Broken chromosome	C4
28	1	Broken chromosome	C4
28	5	Whole arm deletion	C1
34	14	Broken chromosome	C4
43	6	Broken chromosome	C4

Interpretation / Significance:

Sample karyotype is diploid with aneuploidy observed in 8% of cells. Aneuploidy observed was loss of a single chromosome homolog. Random SCE and inversion events (events that do not appear repeatedly in over 30% of cells) were seen at the highest prevalence. The sample had no insertions or translocations. There is a recurrent small inversion on Chr8p present in 54% of the cells. There were six complex events that all involved major structural changes to the impacted chromosome.

Figure 2: Example Karyogram. There are SCE events present on Chr13q and Chr14q. Chromosome 14q has a broken chromatid.
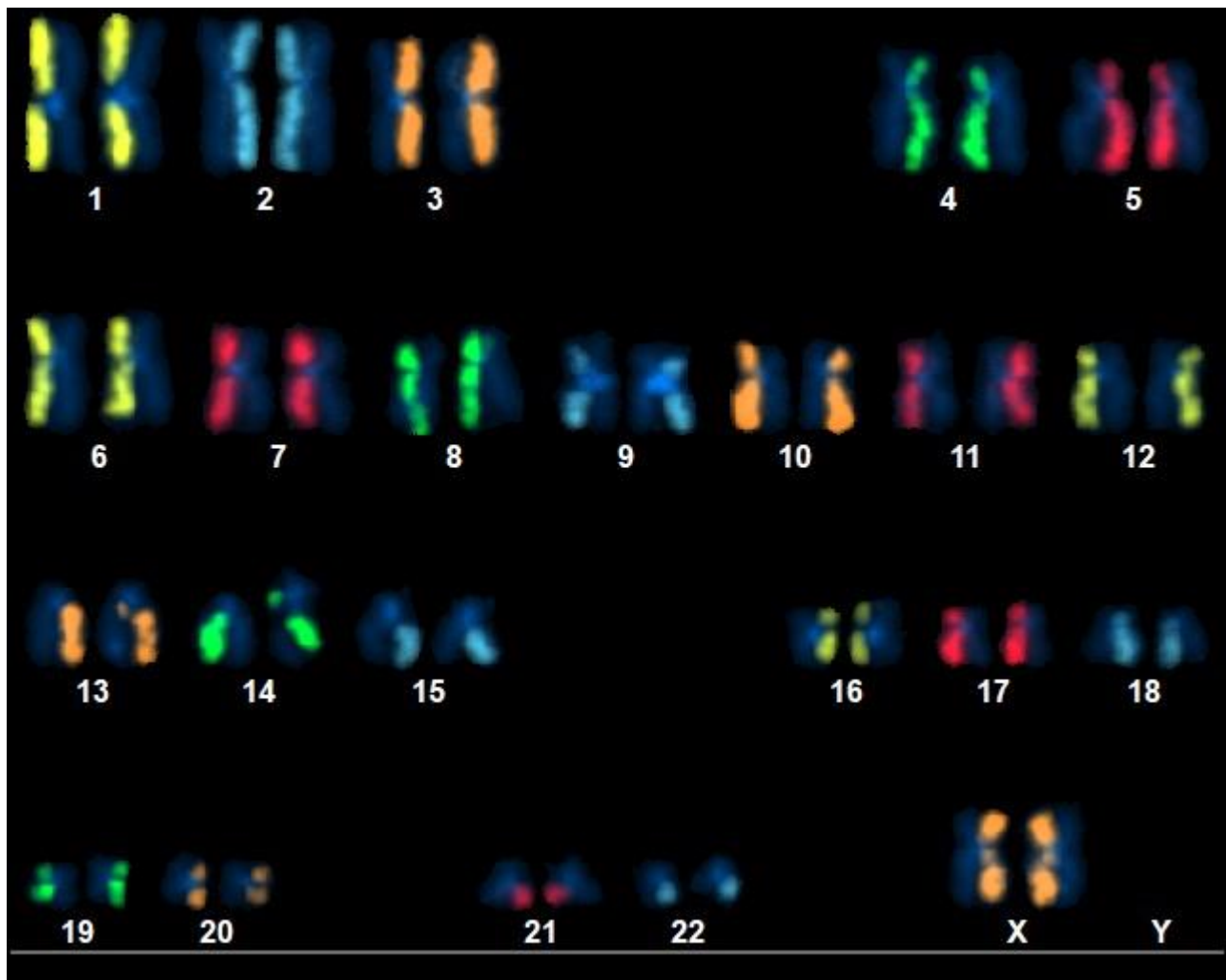
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Figure 3: Example Karyogram. There are SCE events present on Chr3p, Chr7q, Chr12q, and Chr19p. Size difference between homologs is observed for Chr4, Chr9, Chr15, and Chr19. The recurrent small inversion on Chr8p is present. Chr3p has a broken chromatid. Note that the aqua color on Chr5 and the pink on Chr15 are due to an overlap in the metaphase spread.
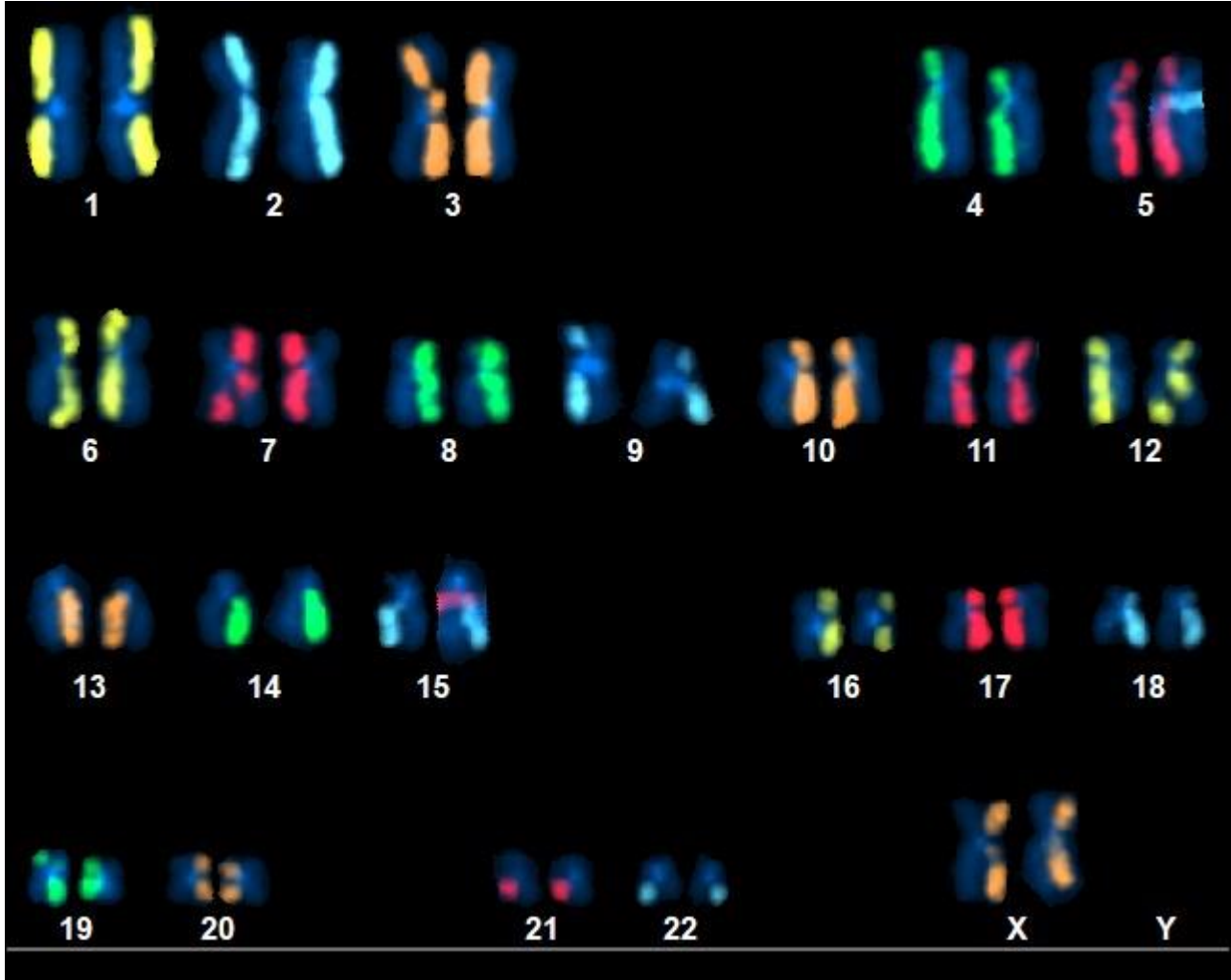
dGH SCREEN 5-Color Whole Genome Assay is for research use only and is not a medical diagnostic test.