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KaryoStat™ Report

Client Name: Stanford
Quote No: CB16307.3-R
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Summary of services

Project Summary:

- Stanford (Client) is interested in services provided by the Life Technologies Corporation in the analysis of three (3) client-provided samples using the KaryoStat™ assay.

Service Description:

- The KaryoStat™ assay allows for digital visualization of chromosome aberrations with a resolution similar to g-banding karyotyping. The size of structural aberration that can be detected is > 2 Mb for chromosomal gains and > 1 Mb for chromosomal losses (the resolution depends on the location of the aberration in the chromosome. Due to a lower probe density on the telomere ends and centromeres, the resolution in those locations may be closer to 5Mb). The KaryoStat™ array is optimized for balanced whole-genome coverage with a low-resolution DNA copy number analysis, the assay covers all 36,000 RefSeq genes, including 14,000 OMIM® targets. The assay enables the detection of aneuploidies, submicroscopic aberrations, and mosaic events.

Materials & Methods:

Genomic DNA purification

Cells were prepared according to the Genomic DNA Purification Kit (Catalog #: K0512) and quantified using the Qubit™ dsDNA BR Assay Kit (Catalog #: Q32850)

GeneChip® Preparation

250 ng total gDNA was used to prepare the GeneChip® for KaryoStat™ according to the manual, and is an array that looks for copy number variants and single nucleotide polymorphisms across the genome.

KaryoStat™ Results: Sample information

#	Sample ID	Status
KS-7847	#991	Complete
KS-7848	#477	Complete
KS-7849	#2021	Complete

Table 1. Customer-provided sample information

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KaryoStat™ Results: KS-7847

1. KaryoStat™ analysis of KS-7847 revealed the sample originated from a male individual
2. No chromosomal aberrations were found when comparing against the reference dataset (Figure 1)
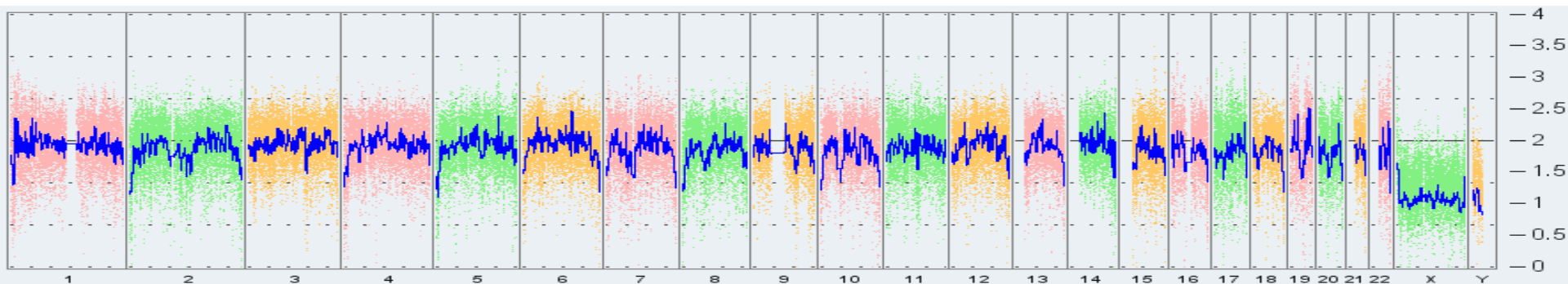
Copy Number State
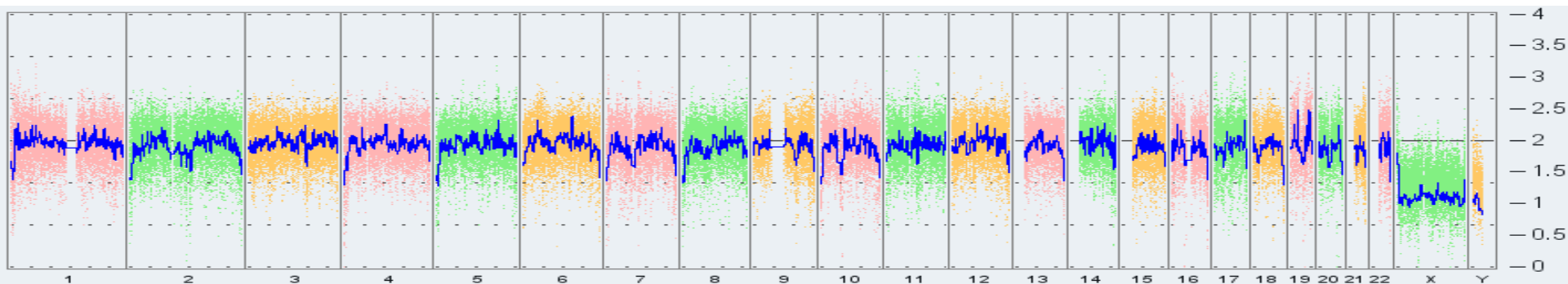
Figure 1: Whole genome view. The whole genome view displays all somatic and sex chromosomes in one frame with high level copy number. The smooth signal plot (right y-axis) is the smoothing of the log2 ratios which depict the signal intensities of probes on the microarray. A value of 2 represents a normal copy number state (CN = 2). A value of 3 represents chromosomal gain (CN = 3). A value of 1 represents a chromosomal loss (CN = 1). The pink, green and yellow colors indicate the raw signal for each individual chromosome probe, while the blue signal represents the normalized probe signal which is used to identify copy number and aberrations (if any).*

Disclaimer: This assay was conducted solely for the listed investigator/institution. The results of this assay are for research use only.

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1. KaryoStat™ analysis of KS-7848 revealed the sample originated from a male individual

2. No chromosomal aberrations were found when comparing against the reference dataset (Figure 2)
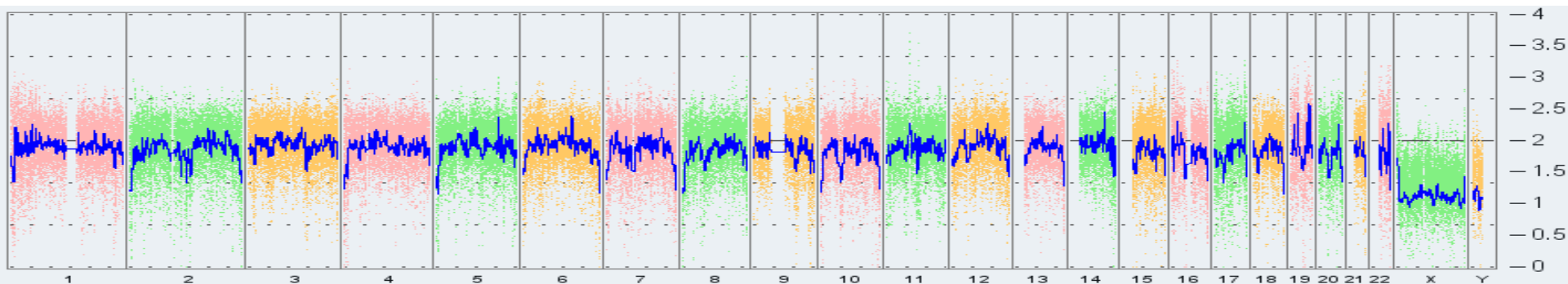
Copy Number State

Figure 2: Whole genome view. The whole genome view displays all somatic and sex chromosomes in one frame with high level copy number. The smooth signal plot (right y-axis) is the smoothing of the log2 ratios which depict the signal intensities of probes on the microarray. A value of 2 represents a normal copy number state (CN = 2). A value of 3 represents chromosomal gain (CN = 3). A value of 1 represents a chromosomal loss (CN = 1). The pink, green and yellow colors indicate the raw signal for each individual chromosome probe, while the blue signal represents the normalized probe signal which is used to identify copy number and aberrations (if any).*

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KaryoStat™ Results: KS-7849

1. KaryoStat™ analysis of KS-7849 revealed the sample originated from a male individual
2. No chromosomal aberrations were found when comparing against the reference dataset (Figure 3)

Copy Number State

Figure 3: Whole genome view. The whole genome view displays all somatic and sex chromosomes in one frame with high level copy number. The smooth signal plot (right y-axis) is the smoothing of the log2 ratios which depict the signal intensities of probes on the microarray. A value of 2 represents a normal copy number state (CN = 2). A value of 3 represents chromosomal gain (CN = 3). A value of 1 represents a chromosomal loss (CN = 1). The pink, green and yellow colors indicate the raw signal for each individual chromosome probe, while the blue signal represents the normalized probe signal which is used to identify copy number and aberrations (if any).*

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