

FIG. 1

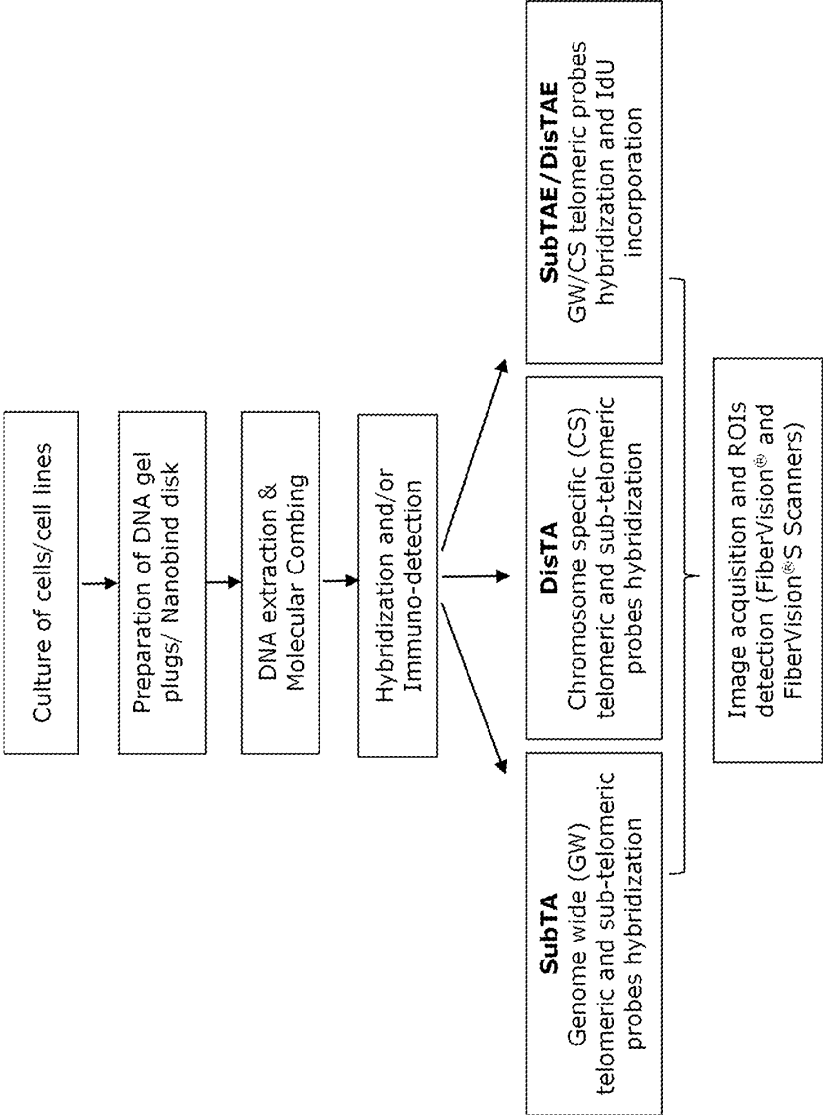


FIG. 2

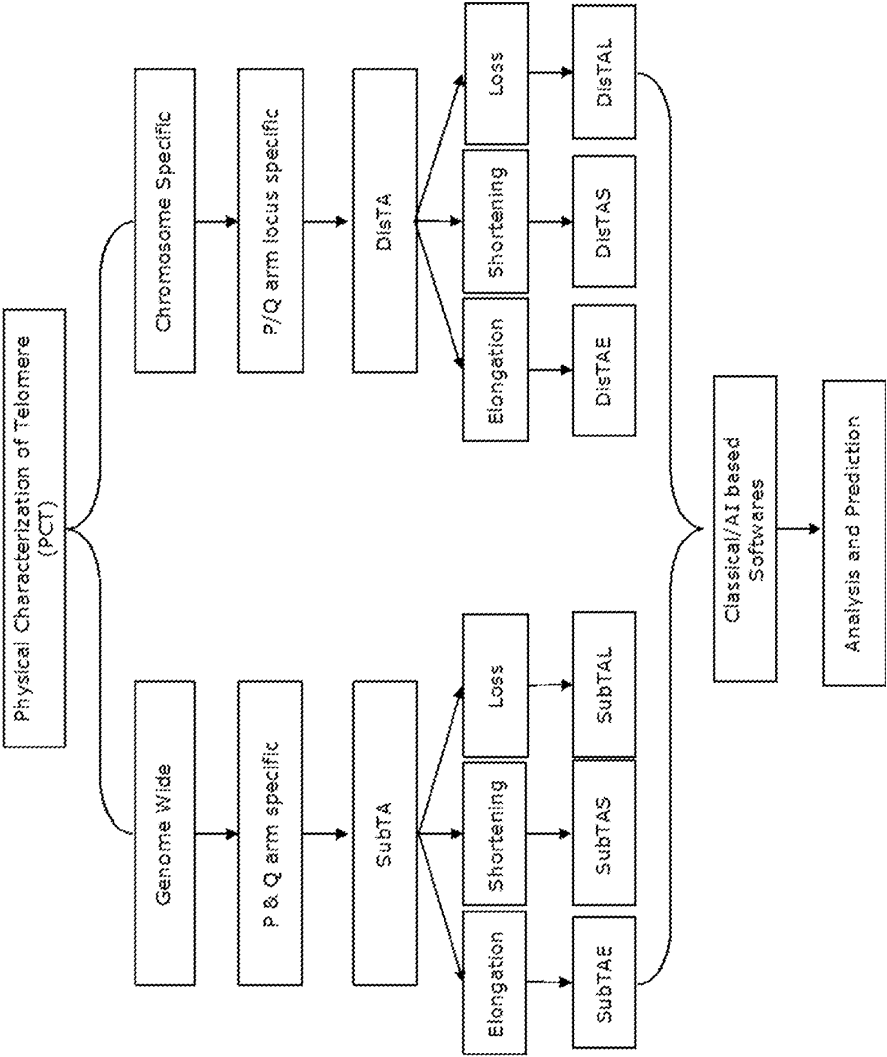


FIG. 3A

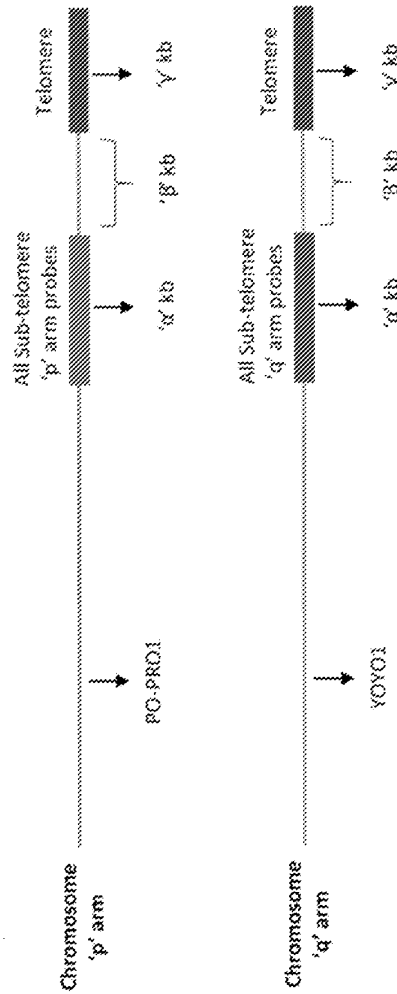


FIG. 3B

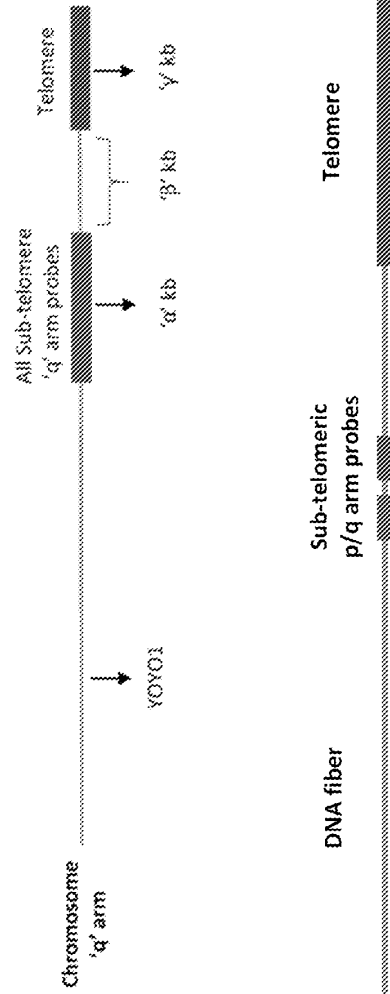


FIG. 4A

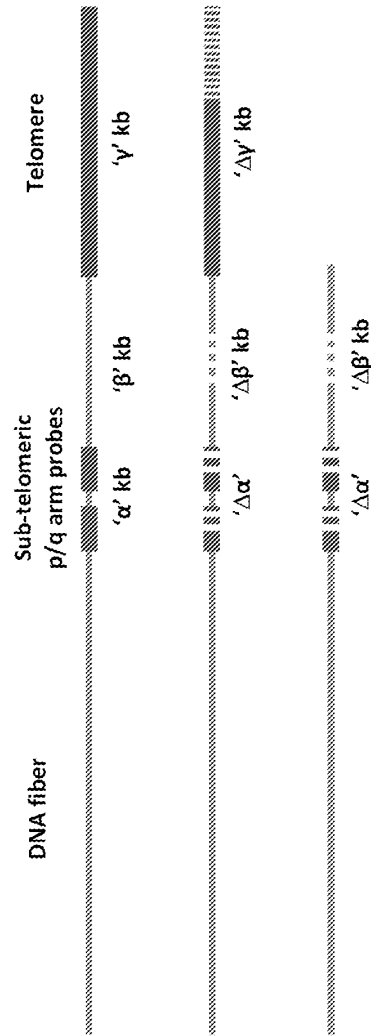


FIG. 4B

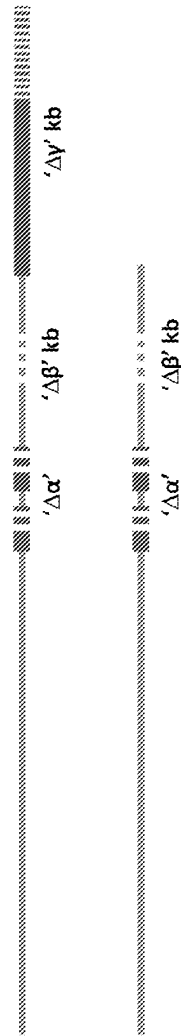
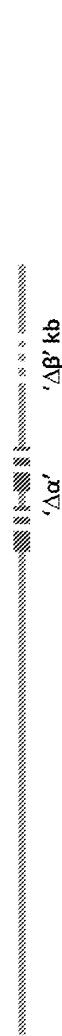


FIG. 4C



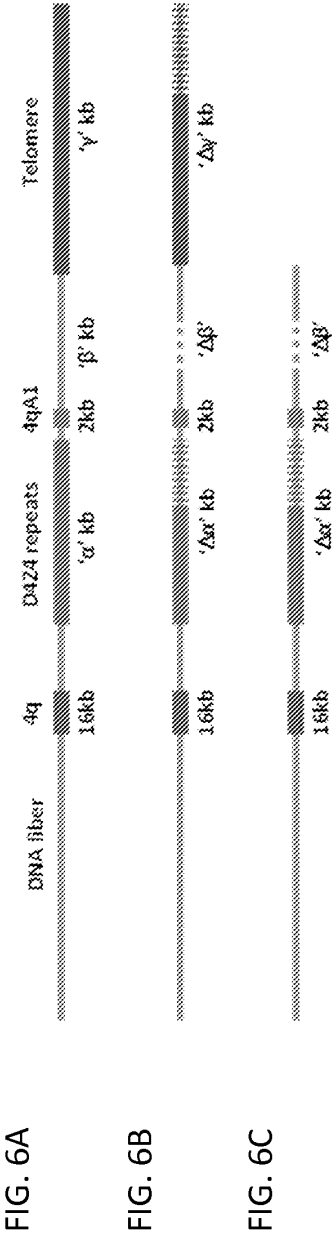
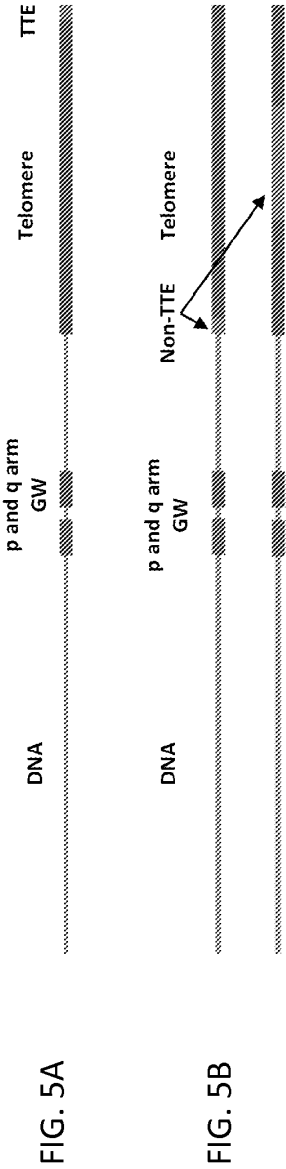


FIG. 7A

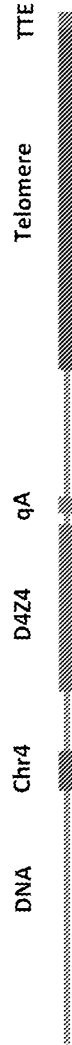


FIG. 7B

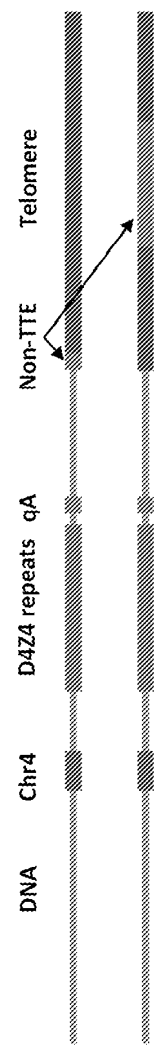


FIG. 8

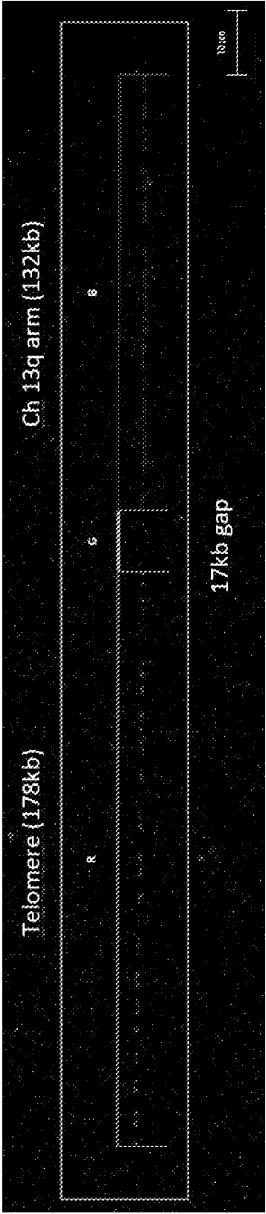


FIG. 9

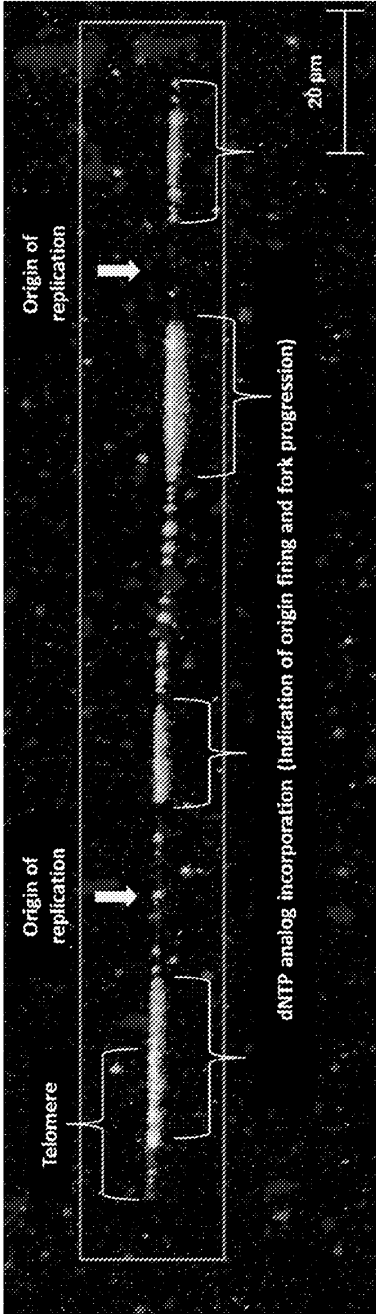
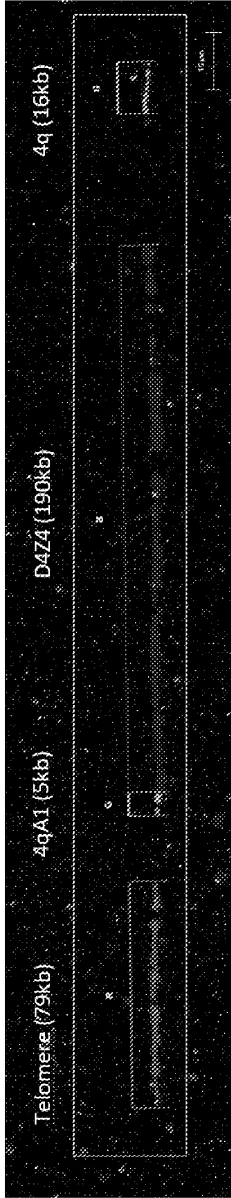


FIG. 10



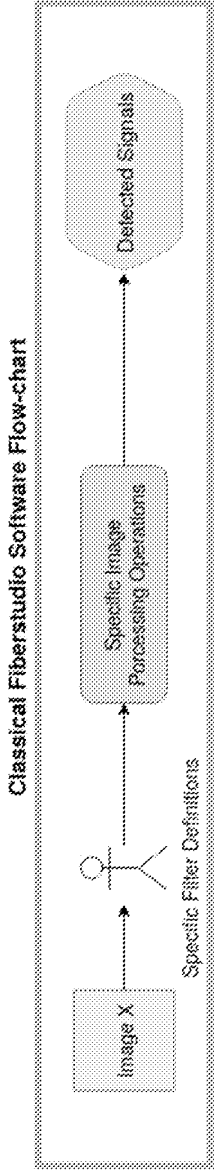


FIG. 11A

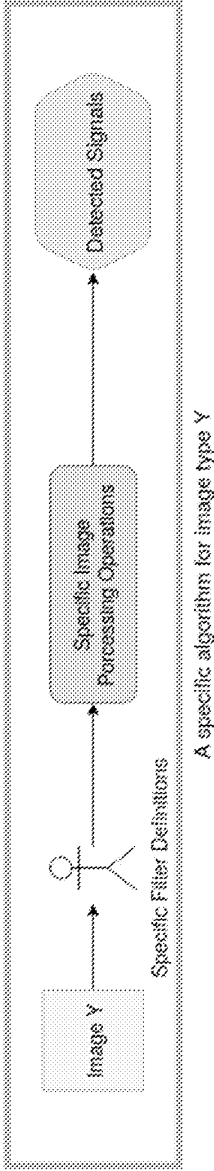


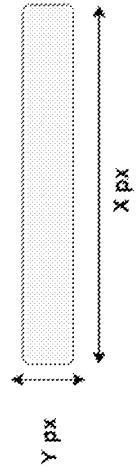
FIG. 11B

FIG. 12A

$$\begin{bmatrix} 1 & -1 & -1 \\ 1 & 8 & -1 \\ 1 & -1 & -1 \end{bmatrix}$$

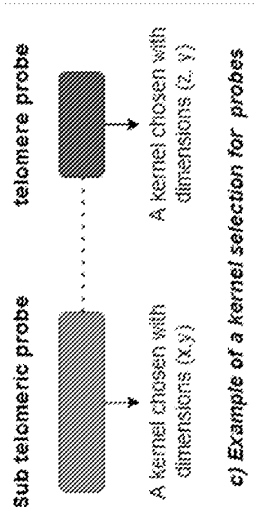
a) A kernel matrix example

FIG. 12B



b) Representation of a kernel with dimensions (x,y)

FIG. 12C



c) Example of a kernel selection for probes

FIG. 13

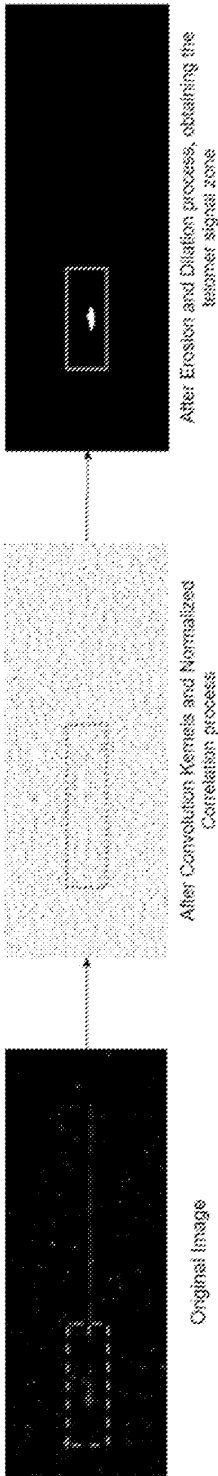


FIG. 14

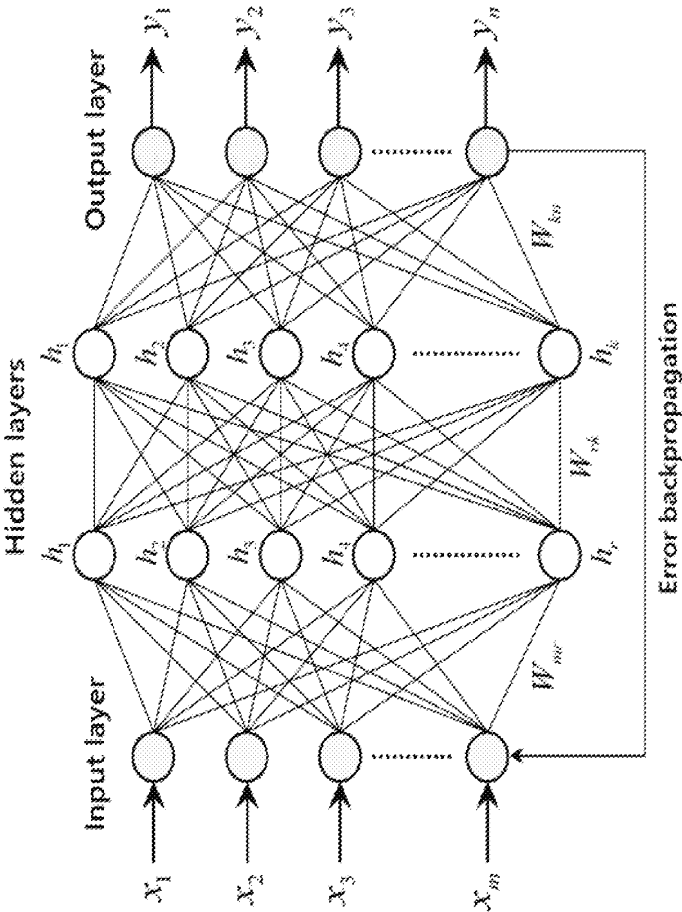


FIG. 15

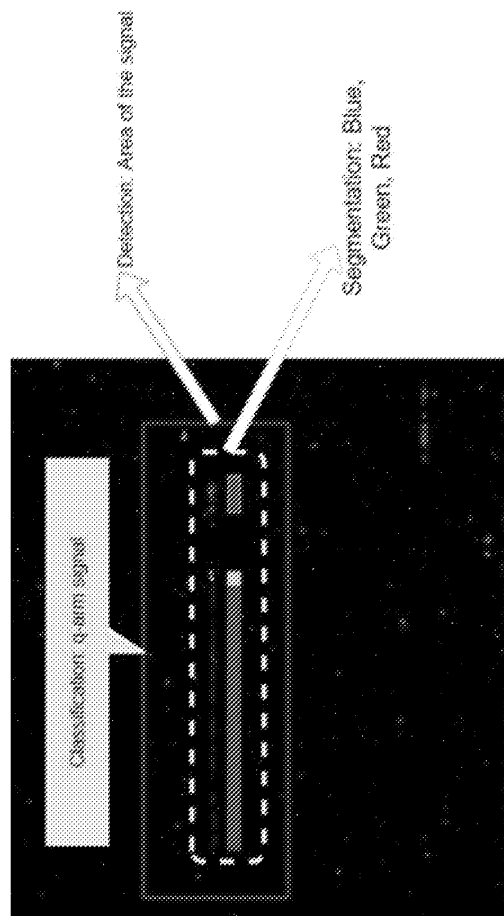


FIG. 16

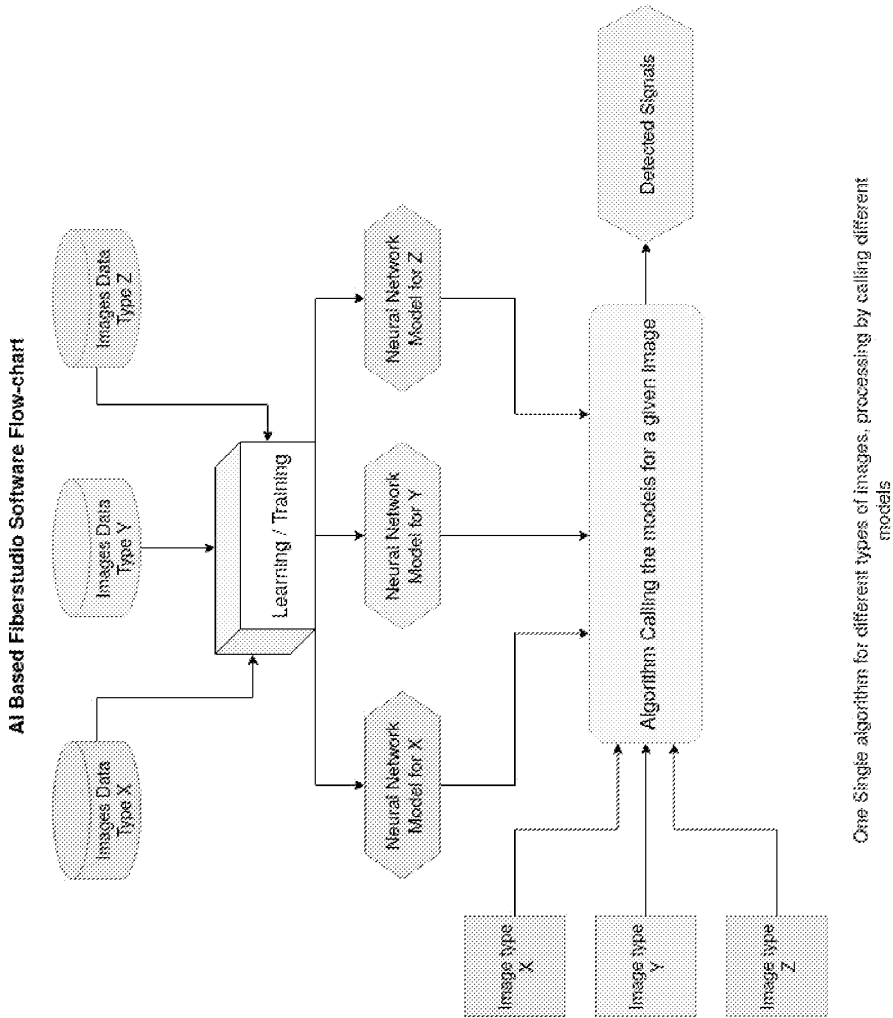


FIG. 17

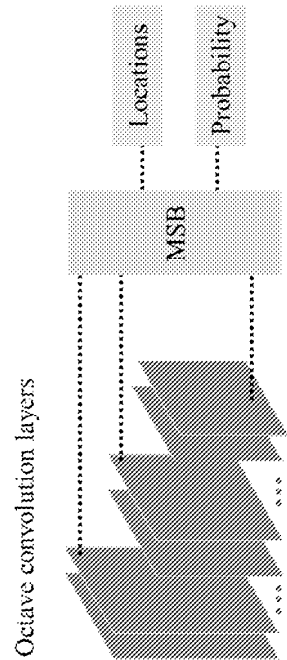


FIG. 18



FIG. 19

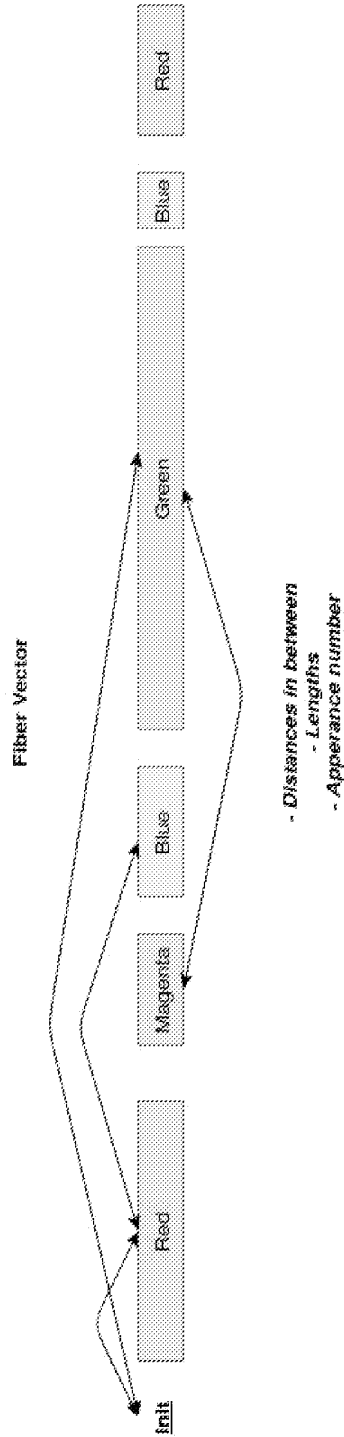


FIG. 20

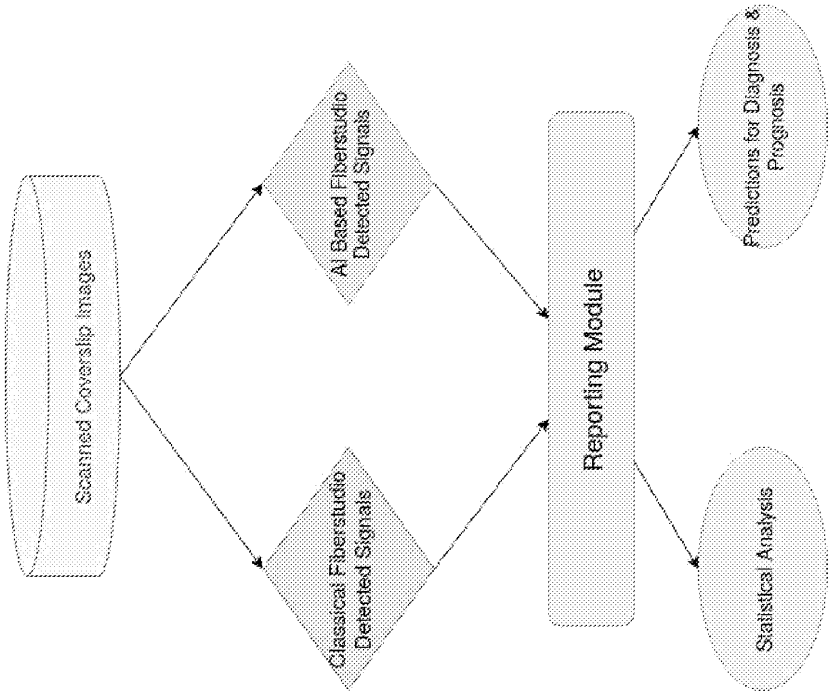


FIG. 21

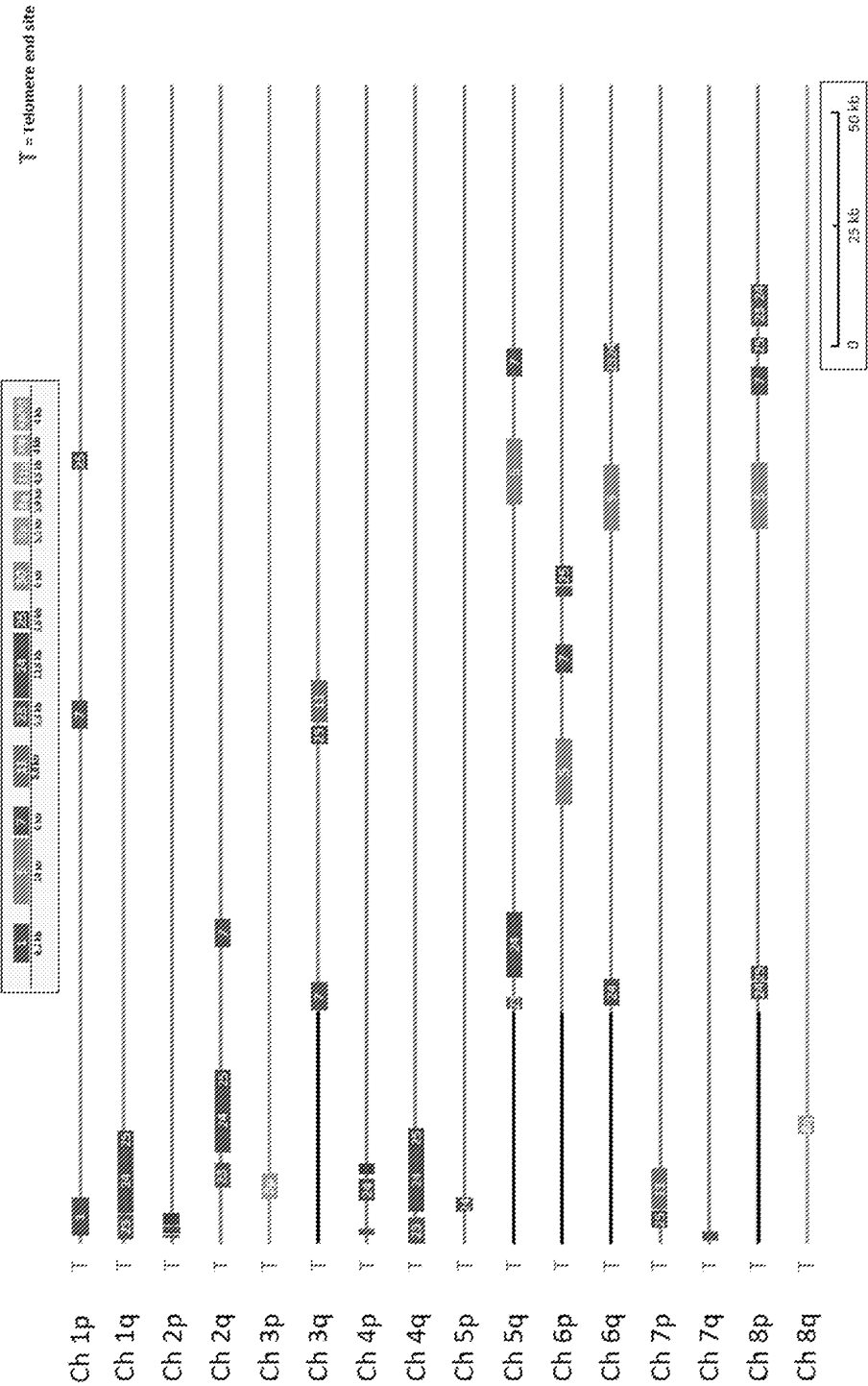


FIG. 22

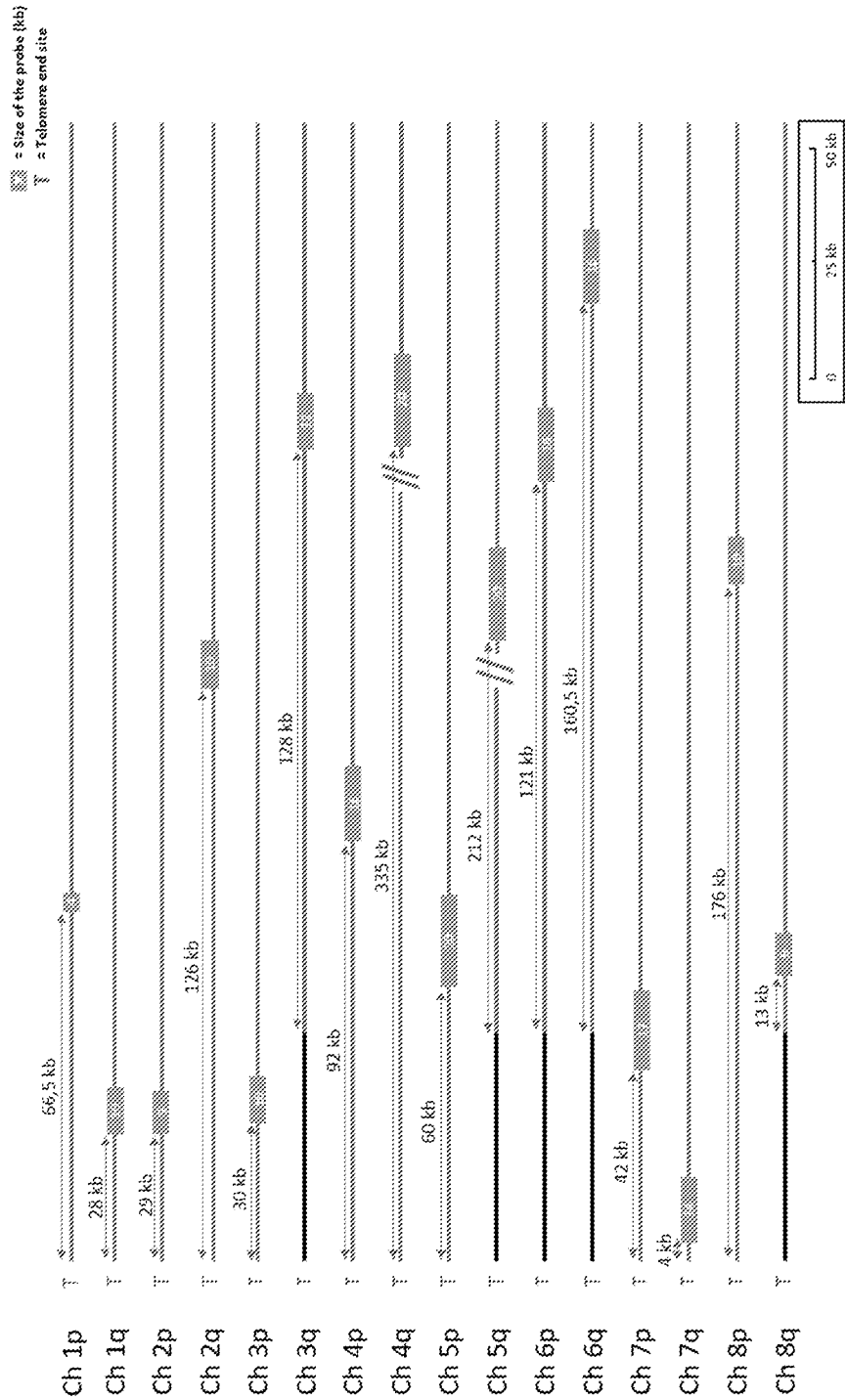


FIG. 23

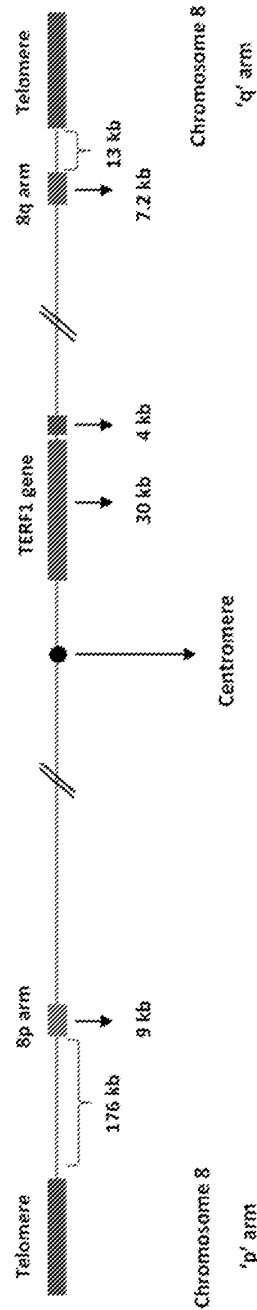


FIG. 24

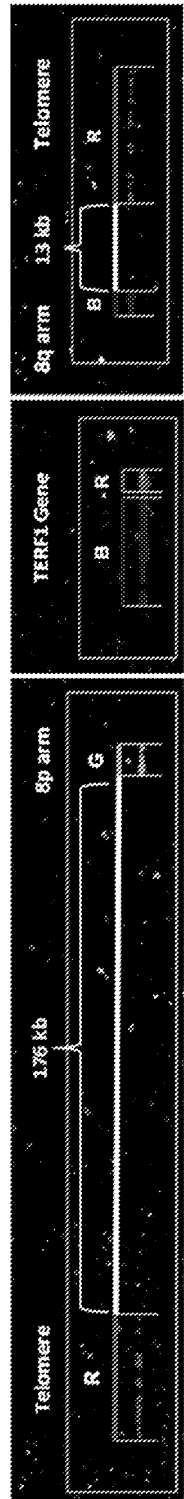


FIG. 25

Genome Reference: GRCh38.p13 (Accession number: GCA_000001405.28)									
Chromosome arms	Block 1 (8,186 bp) Accession number: NC_0000016.10	Block 5 (13,994 bp) Accession number: NC_000008.11	Block 7 (5,943 bp) Accession number: NC_000008.11	Block 11 (8,790 bp) Accession number: NC_000007.14	Block 30 (5,962 bp) Accession number: NC_0000018.10	Block 23 (5,319 p) Accession number: NC_0000022.11	Block 24 (13,840 bp) Accession number: NC_0000022.11	Block 25 (3,594 bp) Accession number: NC_000001.11	Specific probes
Chr1p	Cover: 100% Id 98.5% (1,548,939,941)	X (>200 Mb)	Cover: 100% Id 98.25% (111,839,117,844)	Residual	X	Residual	Residual	Cover: 90% Id 92.7% (166,786,170,939)	X
Chr1q	X	X	Residual	Residual	X	Cover: 99% Id 97.67% (163,819,193,195)	Cover: 97% Id 97.92% (163,170,195,229)	Cover: 100% Id 99.89% (179,934,987,971)	X
Chr2p	X	X	X	X	X	Cover: 100% Id 92.17% (11,822,18,27)	Cover: 11% Id 96.39% (12,336,3,352)	Cover: 84% Id 91.30% (4,243,5,793)	X
Chr2q	X	X	Cover: 100% Id 96.87% (130,704,136,243)	X	X	Cover: 99% Id 97.62% (184,649,187,757)	Cover: 97% Id 96.57% (174,482,189,552)	Cover: 81% Id 94.07% (171,885,178,796)	X
Chr2q	X	X	X	X	X	Residual	Residual	X	5,383 bp Id 99.53% (9,503,14,000) Accession number: NC_000008.12
Chr3q	X	X	Cover: 100% Id 98.04% (154,034,160,938)	Cover: 97% Id 94.94% (183,535,187,711)	X	Residual	Residual	Cover: 90% Id 92.82% (138,013,142,211)	X
Chr4p	X	X	Residual	X	X	Cover: 99% Id 94.53% (2,332,3,076)	Cover: 69% Id 92.23% (9,324,13,842)	Cover: 78% Id 92.98% (15,277,17,155)	X
Chr4q	X	X	X	X	X	Cover: 90% Id 97.66% (184,827,200,000)	Cover: 99% Id 96.50% (181,241,183,167)	Cover: 100% Id 91.90% (178,662,182,565)	X
Chr5p	X	X	Residual	Residual	X	Residual	Cover: 40% Id 94.72% (6,529,9,553)	Residual	X
Chr5q	X	Cover: 100% Id 99.40% (77,255,81,263)	Cover: 100% Id 98% (96,854,93,849)	Residual	X	Cover: 52% Id 98.27% (197,455,199,942)	Cover: 83% Id 95.57% (188,982,193,634)	X	X
Chr6p	X	Cover: 100% Id 98.98% (42,503,51,629)	Cover: 100% Id 96.40% (73,000,76,985)	Residual	X	Cover: 63% Id 93.36% (89,610,91,535)	Residual	Cover: 100% Id 92.94% (91,491,95,184)	X
Chr6q	X	Cover: 100% Id 99.11% (81,856,95,854)	Residual	X	X	Cover: 63% Id 90.99% (59,022,62,159)	Cover: 100% Id 95.40% (192,406,198,409)	X	X
Chr7p	X	Residual	X	Cover: 99% Id 98.25% (5,348,16,344)	X	Residual	Residual	Cover: 90% Id 92.67% (3,084,5,339)	X

FIG. 25 (CON'T)

Genome Reference: GRCh38.p13 (Accession number: GCA_000001405.2)									
Chromosomes arms	Block 1 (8,196 bp) Accession number: NC_0000016.10	Block 5 (13,994 bp) Accession number: NC_000008.11	Block 7 (5,943 bp) Accession number: NC_000008.11	Block 11 (8,790 bp) Accession number: NC_000007.14	Block 30 (5,962 bp) Accession number: NC_0000018.10	Block 23 (5,319 p) Accession number: NC_000002.11	Block 24 (13,844 bp) Accession number: NC_000002.11	Block 25 (3,594 bp) Accession number: NC_000001.11	Specific probes
Chr7q	X	X	Residual	X	X	Cover 42% Id 93.48% (137,616;199,260)	Residual	X	X
Chr9p	X	Cover 100% Id 99.94% (356,796;118,795)	Cover 100% Id 99.95% (133,180;139,181)	X	X	Cover 69% Id 96.38% (147,874;151,987)	Cover 100% Id 94.13% (157,368; 149,478;152,746)	Cover 100% Id 92.14% (2,039;5,989) (181,944;195,158)	X
Chr9q	X	X	Residual	Residual	X	Residual	Residual	X	3,823 bp Id 98.19% (177,603;178,800) Accession number: NC_000008.11
Chr9p	Cover 100% Id 98.72% (1,769;9,954)	X	Cover 100% Id 97.62% (138,660;39,657)	Residual	X	Cover 24% Id 81.58% (366,1,943)	Residual	X	X
Chr9q	X	Cover 100% Id 99.62% (1,769;187,498)	Cover 100% Id 98.04% (157,910;158,091)	Cover 99% Id 96.92% (193,330;103,514)	X	Residual	Residual	Cover 90% Id 92.73% (107,796;105,991)	X
Chr10p	X	X	Residual	Residual	Cover 100% Id 99.22% (1,964;7,362)	Residual	Residual	X	X
Chr10q	X	X	X	X	X	Cover 100% Id 97.62% (14,971;29,757)	Cover 99% Id 94.59% (138,598;192,821)	Cover 100% Id 91.98% (135,731;187,257)	X
Chr11p	X	Cover 100% Id 99.07% (126,360;39,651)	Cover 100% Id 98.23% (154,259;60,048)	Cover 99% Id 96.09% (115,985;113,395)	X	Cover 69% Id 89.13% (140,027;131,799)	Residual	Cover 90% Id 92.79% (111,977;115,859)	X
Chr11q	X	X	Residual	Residual	X	Residual	Residual	X	4,486 bp Id 99.89% (150,001;164,300) Accession number: NC_000001.11
Chr12p	Cover 99% Id 97.82% (1,279;9,876)	X	X	X	X	Residual	Residual	X	X
Chr12q	X	X	Residual	Residual	X	Cover 40% Id 93.65% (137,022;195,889)	Residual	X	X
Chr13p	X	X	Residual	Residual	X	Cover 99% Id 94.98% (134,565;169,709)	Cover 100% Id 94.72% (128,356;196,105)	Residual	X

FIG. 25 (CON'T)

Genome Reference: GRCh38.p13 (Accession number: GCA_000001405.2)									
Chromosomes arms	Block 1 (8,196 bp) Accession number: NC 0000016.10	Block 5 (13,994 bp) Accession number: NC 000008.11	Block 7 (5,943 bp) Accession number: NC 000006.11	Block 11 (8,790 bp) Accession number: NC 000007.14	Block 30 (5,962 bp) Accession number: NC 0000018.10	Block 23 (5,319 p) Accession number: NC 0000022.11	Block 24 (13,844 bp) Accession number: NC 0000022.11	Block 25 (3,594 bp) Accession number: NC 000001.11	Specific probes
Chr14q	X	X	X	X	X	Residual	Cover 25% Id 94.10% (135,446,139,447)	X	X
Chr14p									
Chr15q	Cover 100% Id 98.04% (136,933,136,137) (135,020,97,031)	Cover 100% Id 96.80%	X	X	X	Residual	Residual	X	X
Chr16p	Cover 100% Id 99.38% (1,3293,130)	X	Residual	X	X	Cover 32% Id 83.63% (13,128)	Residual	X	X
Chr16q	X	X	Cover 100% Id 96.37% (150,751,156,752)	Cover 96% Id 95.63% (84,094,83,897)	X	Residual	Cover 41% Id 94.95% (154,848,200,000)	Cover 100% Id 92.73% (94,113,97,994) (153,221,155,177)	X
Chr17p	X	X	X	Cover 87% Id 95.92% (53,094,53,793)	X	Cover 49% Id 96.45% (73,665,76,344)	Residual	Cover 92% Id 92.70% (45,382,53,044)	X
Chr17q	X	X	X		X	Cover 50% Id 96.76% (136,872,199,697)	Cover 96% Id 94.62% (183,938,194,476)	X	X
Chr18p	X	X	X	Residual	Cover 100% Id 96.37% (1,117,113)	Residual	Cover 35% Id 92.47% (85,797,93,551)	Cover 100% Id 92.38% (91,224,95,147)	X
Chr18q	X	X	Residual	X	X	Residual	Cover 35% Id 92.75% (135,753,169,185)	X	X
Chr19p	Cover 17% Id 98.82% (13,1448)	Cover 100% Id 96.94% (104,580,109,284)	Cover 100% Id 98.03% (123,293,126,975)	Residual	X	Cover 100% Id 96.41% (135,965,131,186)	Cover 75% Id 96.07% (139,655,169,137)	Cover 96% Id 92.42% (182,331,185,483)	X
Chr19q	X	X	Residual	Residual	X	Cover 91% Id 96.20% (134,919,139,321)	Cover 100% Id 98.67% (152,996,166,428)	Cover 100% Id 92.85% (173,143,182,733)	X
Chr20p	X	Residual	Residual	Residual	X	Residual	Residual	X	4000 bp Id 99.28% (6,33670,333) Accession number NC_000001.11

FIG. 25 (CON'T)

Genome Reference: GRCh38.p13 (Accession number: GCA_000001405.28)									
Chromosomes arms	Block 1 (8,196 bp) Accession number: NC_0000016.10	Block 5 (13,994 bp) Accession number: NC_000008.11	Block 7 (5,943 bp) Accession number: NC_000006.11	Block 11 (8,790 bp) Accession number: NC_000007.14	Block 30 (5,962 bp) Accession number: NC_000018.10	Block 23 (5,319 p) Accession number: NC_000022.11	Block 24 (13,844 bp) Accession number: NC_000022.11	Block 25 (3,594 bp) Accession number: NC_000001.11	Specific probes
Chr20q	X	X	Cover: 100% Id 96.48% (162,111;162,104)	X	X	Cover: 74% Id 88.73% (147,853;151,615)	Cover: 23% Id 83.56% (147,759;149,029)	X	X
Chr21q	X	X	X	X	X	Cover: 100% Id 97.73% (154,152;154,042)	Cover: 100% Id 97.57% (181,818;195,663)	Cover: 44% Id 94.45% (180,553;182,147)	X
Chr22q	X	Residual	X	Residual	X	Cover: 100% Id 98.5% (134,030;139,429)	Cover: 100% Id 96.99% (181,241;195,138)	Cover: 100% Id 97.88% (137,874;181,564)	X
Chr7p	X	X	X	X	X	Residual	Residual	X	3,971 bp Id 96.28% (17,901;11,000) Accession number: NC_000022.11
Chr9q	Cover: 100% Id 98.42% (126,327;137,517)	X	Residual	Residual	X	Residual	Residual	X	X
Chr7p	X	X	X	X	X	Residual	Residual	X	3,972 bp Id 96.28% (17,901;11,000) Accession number: NC_000022.11
Chr7q	Cover: 100% Id 98.42% (126,327;137,517)	X	Residual	Residual	X	Residual	Residual	X	X

*Source: Ensembl Rest API - Ensembl REST API Endpoints, [online], [Accessed 31 August 2021]. Available from: <https://rest.ensembl.org/>

Color Coding:	80% and greater coverage of probe
	Below 80% but above 30% coverage of probe
	Below 30% coverage of probe
	Non-unique Contaminant
	Unique probe

FIG. 26

Genes of Interest (GOI) probes														Adjacent probe to GOI			
Chromosome arms	Accession number	Coordinates	Size	Distance from Telomere	Gene	Accession number	Coordinates	Size	Distance from Telomere	Accession number	Coordinates	Size	Distance from GOI				
Chr4p	NC_000001.11	(76,501,80,500)	4,000 bp	66.5 kb													
Chr4q	NC_000001.11	(248,908,423-248,918,422)	10,000 bp	28 kb													
Chr2p	NC_000002.12	(39,001-48,000)	9,000 bp	29 kb													
Chr2q	NC_000002.12	(242,047,530-242,057,529)	10,000 bp	126 kb													
Chr3p	NC_000003.12	(40,001-50,000)	10,000 bp	30 kb													
Chr3q	NC_000003.12	(198,095,560-198,107,559)	12,000 bp	128 kb													
Chr6p	NC_000004.12	(102,001-118,000)	16,000 bp	92 kb													
Chr4q	NC_000004.12	(188,849,554-188,869,553)	20,000 bp	335 kb													
Chr5p	NC_000005.10	(70,001-90,000)	20,000 bp	60 kb													
Chr5q	NC_000005.10	(181,246,260-191,266,259)	20,000 bp	212 kb													
Chr6p	NC_000006.12	(181,001-197,000)	16,000 bp	121 kb													
Chr6q	NC_000006.12	(170,569,480-170,585,479)	16,000 bp	160.5 kb													
Chr7p	NC_000007.14	(52,001-69,000)	17,000 bp	42 kb													
Chr7q	NC_000007.14	(159,317,972-159,331,971)	14,000 bp	4 kb													
Chr8p	NC_000008.11	(236,248,246,242)	10,000 bp	176 kb													
Chr8q	NC_000008.11	(145,056,636-145,065,636)	9,000 bp	13 kb	TERF1	NC_000008.11	(73,008,887-73,046,137)	37,250 bp	72.03 Mb	NC_000008.11	(73,048,624-73,052,623)	4,000 bp	2,487 kb				
Chr9p	NC_000009.12	(51,001-71,000)	20,000 bp	41 kb													
Chr9q	NC_000009.12	(138,110,718-138,124,717)	14,000 bp	210 kb													
Chr10p	NC_000010.11	(130,001-144,000)	14,000 bp	120 kb													
Chr10q	NC_000010.11	(133,611,421-133,621,420)	10,000 bp	166 kb													
Chr11p	NC_000011.10	(193,001-213,000)	20,000 bp	133 kb													
Chr11q	NC_000011.10	(135,044,631-135,064,630)	20,000 bp	12 kb													
Chr12p	NC_000012.12	(80,001-95,550)	15,500 bp	70 kb													
Chr12q	NC_000012.12	(133,225,310-133,237,309)	12,000 bp	28 kb													
Chr13p	NC_000013.11	(114,317,829-114,327,328)	9,500 bp	27 kb													
Chr13q	NC_000013.11	(106,843,719-106,855,718)	12,000 bp	28 kb													
Chr14p	NC_000014.9	(101,843,188-101,857,187)	14,000 bp	124 kb													
Chr14q	NC_000014.9	(55,001-70,000)	15,000 bp	45 kb													
Chr15p	NC_000015.10	(90,035,346-90,053,345)	18,000 bp	175 kb													
Chr16p	NC_000016.10	(161,001-181,000)	20,000 bp	101 kb													
Chr17p	NC_000017.11	(83,184,440-83,204,439)	20,000 bp	43 kb													
Chr17q	NC_000017.11	(115,001-127,000)	12,000 bp	105 kb													
Chr18p	NC_000018.10	(80,240,286-80,255,785)	15,500 bp	7.5 kb													
Chr18q	NC_000018.10	(292,001-314,000)	22,000 bp	232 kb													
Chr19p	NC_000019.10	(58,544,617-58,564,616)	20,000 bp	43 kb													
Chr19q	NC_000019.10	(82,501-97,000)	14,500 bp	22.5 kb													
Chr20p	NC_000020.11	(64,263,668-64,279,167)	16,500 bp	55 kb													
Chr20q	NC_000020.11	(46,645,982-46,664,981)	19,000 bp	35 kb													
Chr21p	NC_000021.9	(50,714,467-50,734,466)	20,000 bp	74 kb													
Chr22p	NC_000022.11	(255,001-275,000)	20,000 bp	245 kb													
Chr2p	NC_000002.12	(155,661,616-155,681,616)	20,000 bp	350 kb													
Chr1p	NC_000001.11	(255,001-275,000)	20,000 bp	245 kb													
Chr1q	NC_000001.11	(155,661,616-155,681,616)	20,000 bp	245 kb													
*Acentric Chromosomes																	
*Source: Ensembl, RefSeq API Endpoints. [online]. [Accessed 31 August 2021]. Available from: https://transfer.protected.secure.ensembl.org/																	