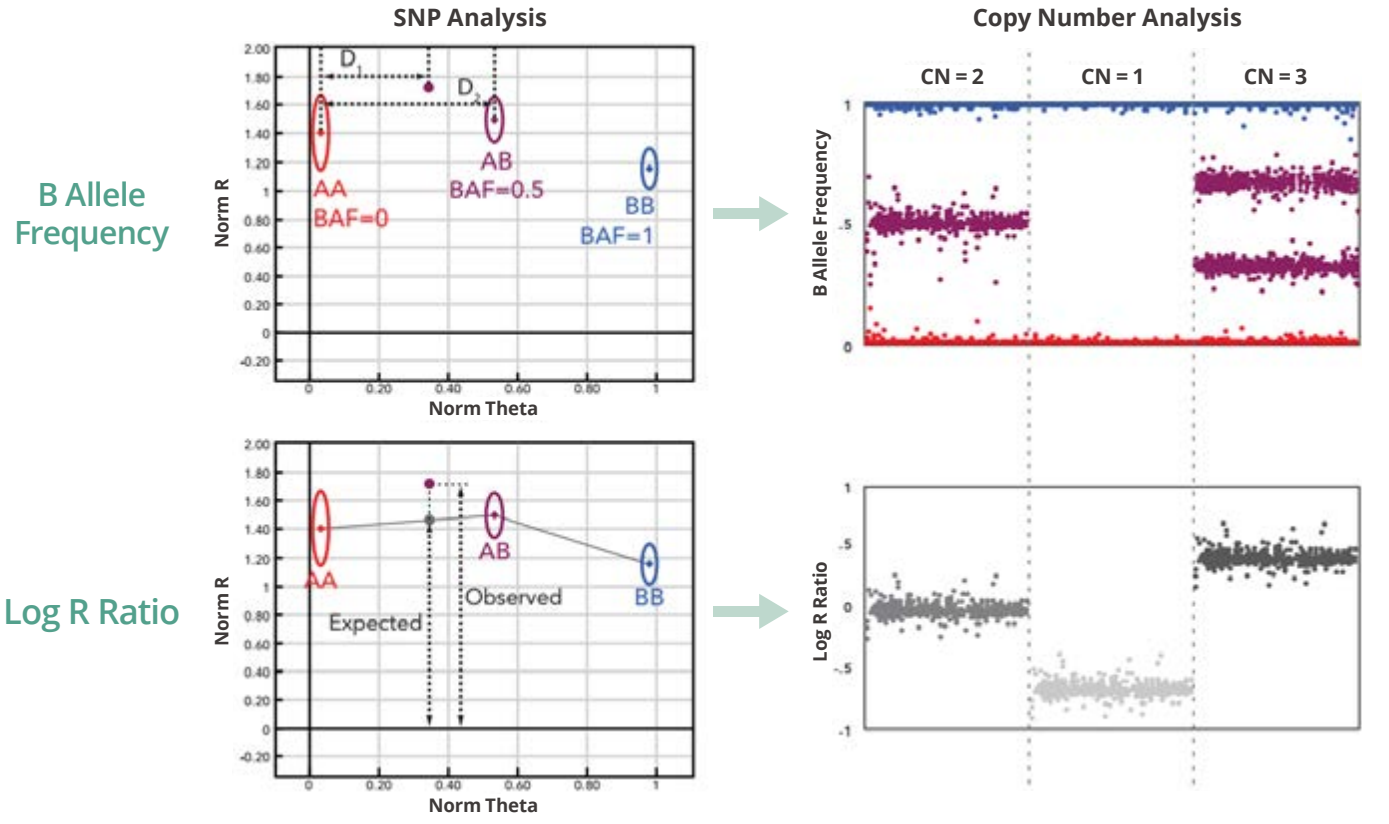
BEBEGENE
'Newborn Copy Number Variation Screening'

Chromosomal microarray analysis (CMA) can detect difficult-to-identify diseases in approximately 15-20% of individuals with unexplained developmental delay, autism spectrum disorder and multiple congenital anomalies. The CMA with SNP probes can detect CNVs, copy neutral changes (i.e., uniparental disomy) as well as identify ploidy changes[1-3].

EDGC's BEBEGENE has two advantages. First, EDGC analyzes CNV analysis with two kinds of data: B allele frequency (BAF) and Log2 R ratio (LRR)[4] [figure 1]. In addition to CNV analysis according to intensity, analysis of Loss of heterogeneity (LOH) is possible. Second, 'genome waves' that hinder accurate analysis were offset by machine learning. Six types were identified by clustering with 5,399 samples, and the corresponding reference was applied through classification. This prevents false detection, enabling more accurate CNV detection.



[Figure 1]. How to calculate BAF and LRR. The allelic copy ratio in terms of BAF is calculated from the θ value of a sample and the expected cluster positions (ellipses, left top panel). The allele frequency is determined as a linear interpolation in the θ-dimension related to the allele frequency of each cluster (0.0, 0.5, and 1.0). In this example, a data point (purple dot) falling approximately 2/3 of the distance from the AA to the AB cluster (D1/D2 = 0.66) has an allele frequency of 0.33 (0.66 * 0.5). The $\log_2$ R ratio is calculated as the ratio between observed normalized intensity of the experimental sample to the expected intensity (left bottom panel). The expected intensity is determined as a linear interpolation as a function of the sample θ (grey line) of the expected cluster positions (ellipses). These two transformed parameters, B allele frequency and $\log_2$ R ratio, are then plotted along the entire genome for all SNPs on the array (right panel). The plot of these two parameters exhibits diagnostic signature profiles of copy number (example copy numbers 2, 1, and 3 shown) and specific classes of structural variation. [Adapted from illumina's Technical Note]

BEBEGENE's disease

Regions that were associated with disease were selected by OMIM (https://www.omim.org/) or decipher (https://www.deciphergenomics.org/) [Figure 2]. BEBEGENE includes monosomy (deletion), trisomy (duplication), and tetrasomy (triplication) according to CN, and is divided into whole chromosome deletion (duplication) and micro-deletion (micro-duplication) according to the length of the region.

Table 1. Number of 239 CNV-related chromosomal disorders.
[See appendix for a detailed list of diseases.]

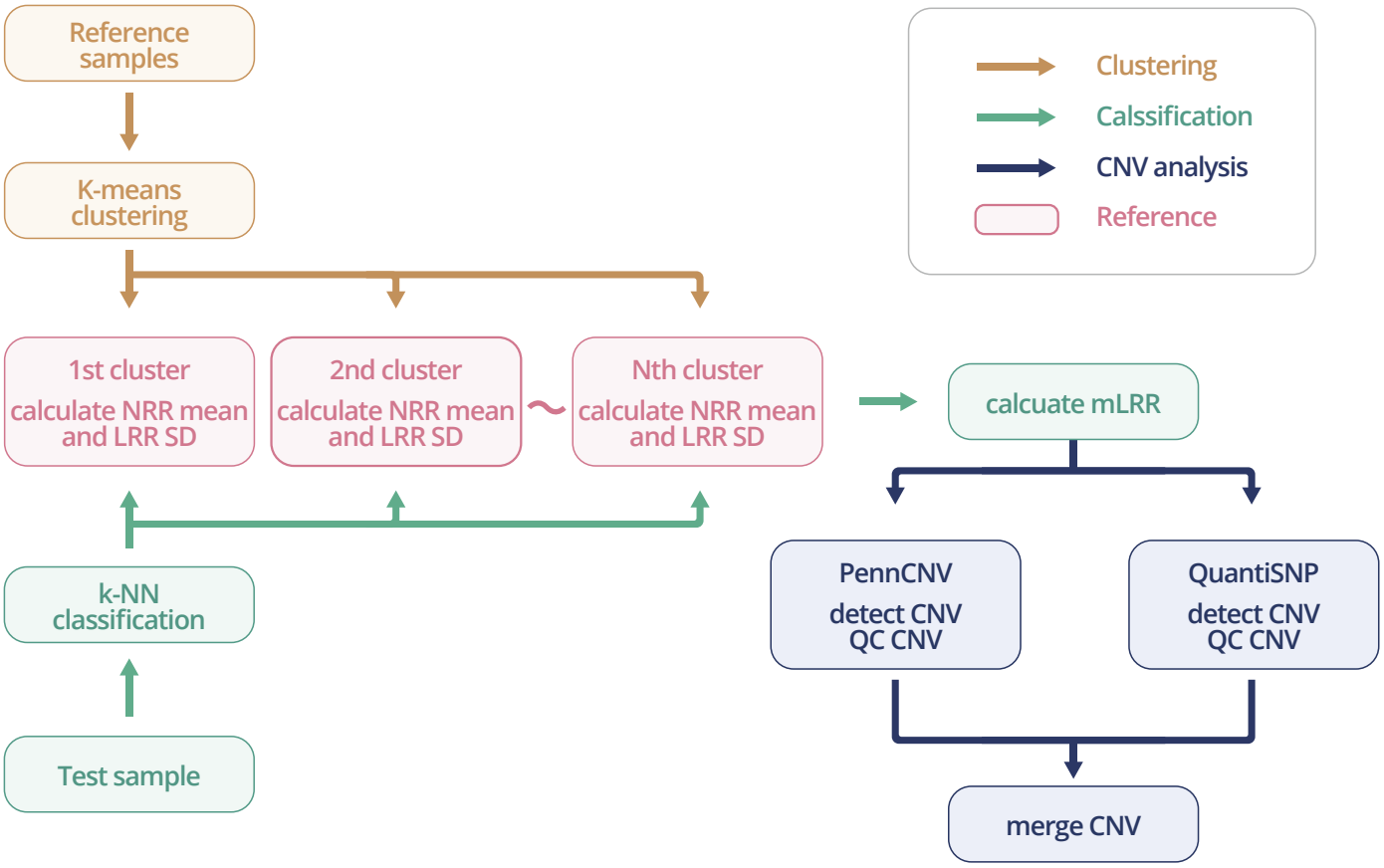
Chromosome	Number of Disease	Deletion	Duplication	Both	Whole chromosome
1	12	9	3	-	-
2	21	14	7	-	-
3	9	6	3	-	-
4	7	4	3	-	-
5	11	8	3	-	-
6	11	7	3	1	-
7	14	7	6	1	-
8	14	10	4	-	-
9	9	8	1	-	-
10	9	5	4	-	-
11	12	9	2	1	-
12	5	4	1	-	-
13	7	6	1	-	1 (Dup)
14	8	6	2	-	-
15	12	7	5	-	-
16	17	12	5	-	-
17	20	9	10	1	-
18	4	2	2	-	1 (Dup)
19	4	2	1	1	-
20	5	5	-	-	-
21	2	1	1	-	1 (Dup)
22	10	5	5	-	-
X	16	8	7	1	3 (1 Del, 2 Dup)
total	**239**	**154**	**79**	**6**	**6 (1 Del, 5 Dup)**

BEBEGENE analysis

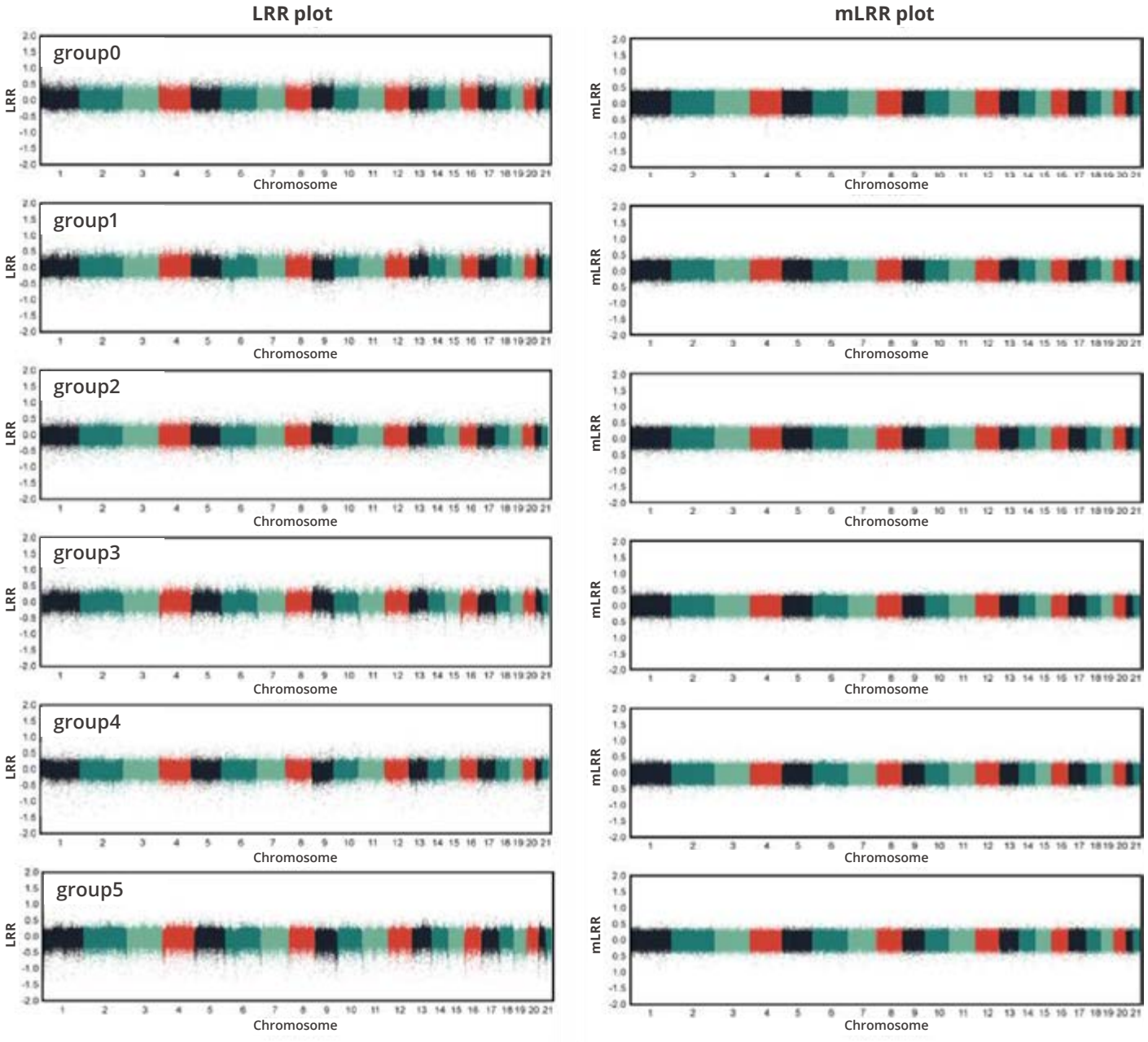
The genomic wave pattern in the Illumina's Global Screening Array (GSA) chip was confirmed using 5,399 clinical samples. Autosomal chromosomes were divided into 1 Mb bin, and LRR mean and standard deviation (SD) of markers within the region were calculated. Bin with no markers or LRR SD θ 0.05 were excluded from the analysis because small SDs result in even distribution and are not very useful for analysis.

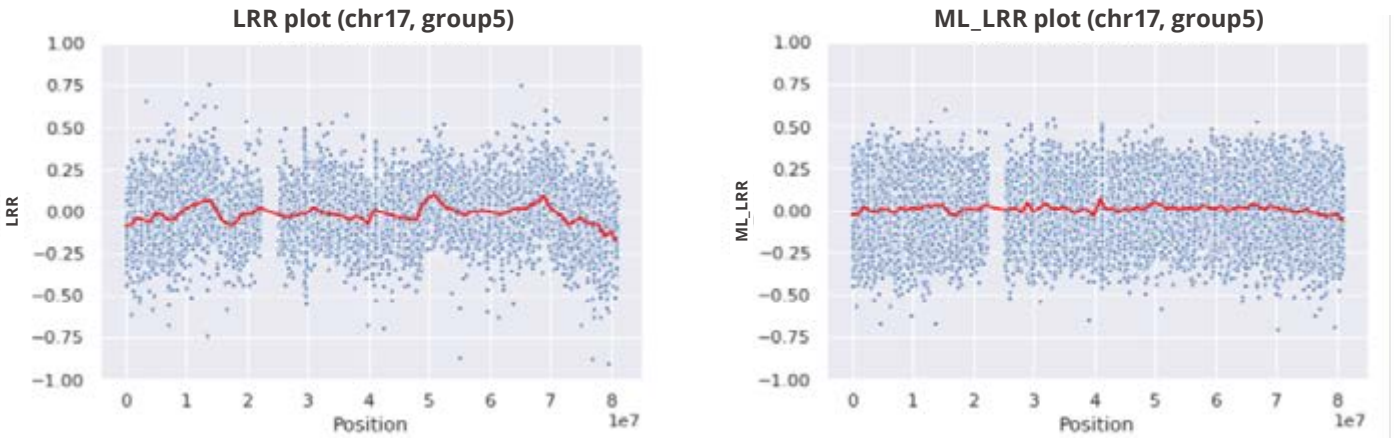
5,399 samples were clustered using the k-means method [Figure 3, 4]. The cluster name was indicated on the classified samples, and the mean was calculated for the marker LRR included in the bin for samples in the cluster. The samples in each cluster were plotted with the mean, and cluster patterns were confirmed. For the sample feature to be used for classification, the LRR mean for the 238 regions used for k-means[5-7] analysis was calculated and classified using k-Nearest Neighbor (k-NN)[8-10]. The matched cluster sample LRR data were used to normalize into the Z-score.

Since the range of normalized values is different from the original LRR, adjustment to the original range is necessary. The original LRR SD and Z-score SD were resized to a similar value to create a new LRR called modified LRR (mLRR).



[Figure 3]. Pipeline of BEBEGENE with Genomic Wave offset. The preprocessing used machine learning to offset the wave. CNV analysis was performed with two tools using offset data. CNV results were merged to reduce false negatives.



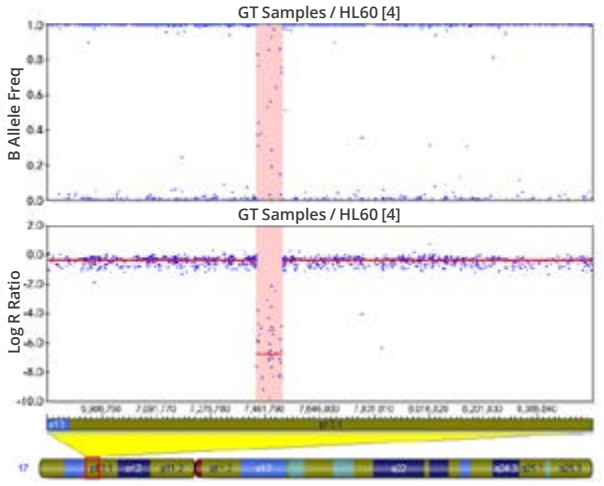


[Figure 4]. Results of waves offset. In the results of group5, both ends of the chromosomes were offset in the modified LRR for all chromosomes.

Interpretation of BEBEGENE analysis result

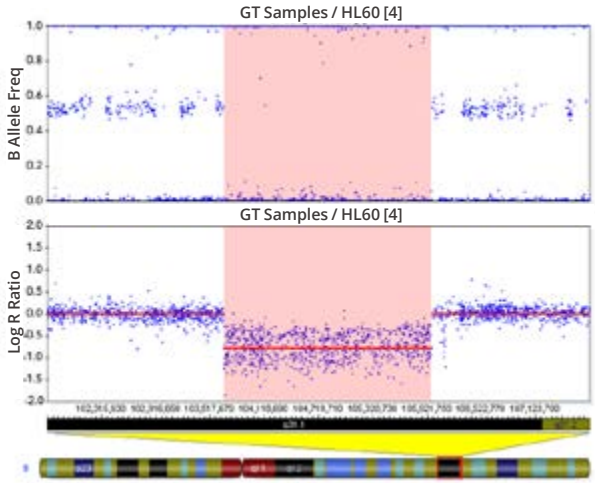
In the BEBEGENE result, the CNV is determined according to the intensity of the LRR value [Figure 5]. In addition, it can be confirmed by the distribution of BAF values. LOH can be confirmed with BAF, and mosaic can also be confirmed.

A)



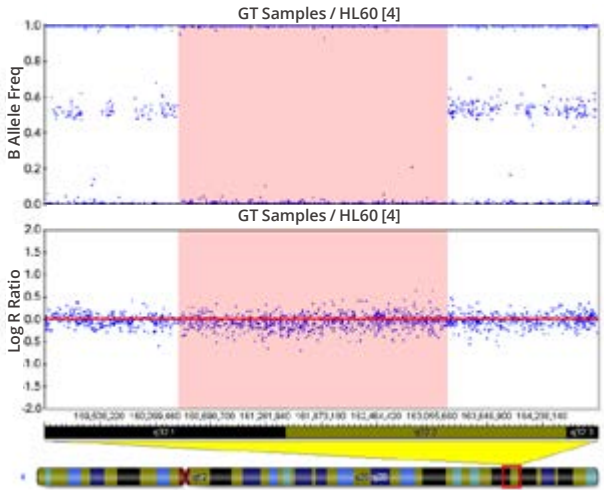
A region of homozygous is where both copies of the chromosome have been lost (shaded). In this case, there are no SNPs present, so the genotyping data (B Allele Freq plot) appears like a "waterfall" as s result of noise in the absence of signal. The log R ratio in thin region is the log2 of ~0/2, which is a highly negative value and is shown in the Log R Ratio plot as a large deflection downward.

B)



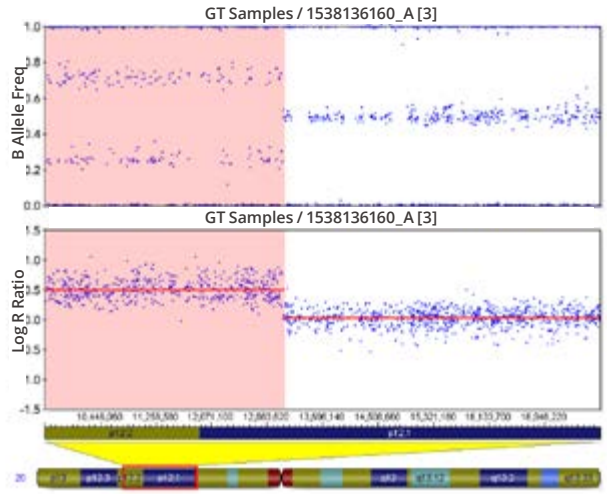
A hemizygous deletion (loss of one copy) is depicted in the ICB as a loss of heterozygotes in the B Allele Freq plot (top) and a loss of signal intensity in the Log R Ratio plot (bottom). In the region of the deletion (shaded), the log R ratio is log2 of 1/2, or -1.

C)



A region of copy-neutral LOH (shaded) is depicted by a loss of heterozygotes in the B allele frequency dada but no change in the log R ratio (physical copy number).

D)



Regions of deletion (not shaded) are depicted by loss of signal intensity in the Log R Ratio plot to -0.5. An overlapping duplication (shaded) is depicted in the middle of the window by an increase in the Log R Ratio plot.

[Figure 5]. Distribution of LRR and BAF by CN. The region marked in pink is the corresponding CNV, and another region is the normal. A) homozygous deletion CN = 0, B) hemizygous deletion CN = 1, C) Loss of heterozygotes CN = 2, D) duplication CN = 3, E) Mean and SD of LRR and BAF values according to copy number. [Adapted from illumina's Technical Note]

E)

Copy Number	Genotype	LRR Mean	LRR SD	BAF Mean	BAF SD
0	DD	-5	2	NA	NA
1	A	-0.45	0.18	0	0.03
	B	-0.45	0.18	1	0.03
2	AA	0	0.18	0	0.03
	AB	0	0.18	0.5	0.03
	BB	0	0.18	1	0.03
3	AAA	0.3	0.18	0	0.03
	AAB	0.3	0.w18	0.33	0.03
	ABB	0.3	0.18	0.66	0.03
	BBB	0.3	0.18	1	0.03
4	AAAA	0.75	0.18	0	0.03
	AAAB	0.75	0.18	0.25	0.03
	AABB	0.75	0.18	0.5	0.03
	ABBB	0.75	0.18	0.75	0.03
	BBBB	0.75	0.18	1	0.03

BEBEGENE Results Statistics for Newborns

Between June 2021 and October 2022, total 24,816 newborns samples were collected for genetic analysis from clinical centers in South Korea. Our genome-wide SNP array chip targeting CNV-related chromosomal disorders was able to identify 464 cases (1.870%) of 92 CNV-associated chromosomal disorders from a total of 24,816 newborns. Table 2 summarizes the CNV-related chromosomal disorders involved in each case.

Table 2. Screening results for CNV-related chromosomal disorders in Korean population.

Diseases	Cytoband	Count	%
16p13.11 duplication syndrome	16p13.11	49	0.197
17p13.3 telomeric duplication syndrome	17p13.3	33	0.133
22q11.2 duplication syndrome	22q11.2	33	0.133
Klinefelter syndrome	chrX	24	0.097
Duchenne muscular dystrophy (DMD)	Xp21.2-p21.1	19	0.077
Triple-X syndrome	chrX	16	0.064
Xp22.31 microdeletion syndrome	Xp22.31	15	0.06
1q21.1 deletion (distal) syndrome	1q21.1	14	0.056
16p13.11 deletion syndrome	16p13.11	14	0.056
16p11.2 duplication (distal) syndrome	16p11.2	13	0.052
16p11.2 duplication (proximal) syndrome	16p11.2	13	0.052
1q21.1 duplication (distal) syndrome	1q21.1	12	0.048
Down syndrome	chr21	11	0.044
16p11.2 deletion (distal) syndrome	16p11.2	11	0.044
1q21.1 deletion (proximal) syndrome	1q21.1	11	0.044
17p12 deletion syndrome	17p12	9	0.036
16p11.2 deletion (proximal) syndrome	16p11.2	8	0.032
16p12.2 deletion (proximal) syndrome	16p12.2	7	0.028
Charcot-Marie-Tooth disease, type 1A (CMT1A)	17p12	7	0.028
22q11.2 deletion syndrome (LCR22 B/C-D)	22q11.21	7	0.028
DiGeorge/Velocardiofacial syndrome (CATCH22)	22q11.21	7	0.028
2q23.1 microdeletion syndrome	2q23.1	7	0.028
15q13.3 deletion syndrome (BP4 to BP5)	15q13.3	6	0.024
15q13.3 duplication syndrome	15q13.3	6	0.024
1q21.1 duplication (proximal) syndrome	1q21.1	6	0.024
2q13 deletion syndrome	2q13	6	0.024
17q12 duplication syndrome	17q12	5	0.02
2p16.3 deletion syndrome	2p16.3	4	0.016
Prader-Willi/Angelman Syndrome (BP1-BP3)	15q11.2-q13.1	4	0.016
18p deletion syndrome	18p	4	0.016
2q11.2 deletion syndrome	2q11.2	3	0.012
Turner syndrome	chrX	3	0.012
Trisomy 9p syndrome	9p	2	0.008
Trisomy 12p syndrome	12p	2	0.008
12p13.33 microdeletion syndrome	12p13.33	2	0.008
17q12 deletion syndrome	17q12	2	0.008
Hypertrichosis terminalis, generalized, with or without gingival hyperplasia (HTC3)	17q24.2-q24.3	2	0.008

Diseases	Cytoband	Count	%
22q11.2 deletion syndrome (distal, D-E/F)	22q11.21-q11.22	2	0.008
2q13 duplication syndrome	2q13	2	0.008
2q21.1 deletion syndrome	2q21.1	2	0.008
2q23.1 microduplication syndrome	2q23.1	2	0.008
5p13 duplication syndrome	5p13	2	0.008
Spinal muscular atrophy, type 1 (SMA1)	5q13	2	0.008
Axenfeld-Rieger syndrome, type 3 (RIEG3)	6p25.3	2	0.008
7p22.1 microduplication syndrome	7p22.1	2	0.008
Monosomy 7	7q	2	0.008
7q36.3 duplication syndrome	7q36.3	2	0.008
8q22.1 duplication syndrome	8q22.1	2	0.008
Xq28 duplication syndrome	Xq28	2	0.008
Patau syndrome	chr13	1	0.004
Monosomy 10p syndrome	10p	1	0.004
DiGeorge syndrome/velocardiofacial syndrome complex 2 (DGS2)	10p14-p13	1	0.004
10q22.3-q23.2 duplication syndrome	10q22.3-q23.2	1	0.004
10q26 deletion syndrome	10q26	1	0.004
Partial Trisomy 11q syndrome	11q	1	0.004
11q22.2-q22.3 microdeletion syndrome	11q22.2-q22.3	1	0.004
Partial Monosomy 13q syndrome	13q	1	0.004
Congenital microcoria	13q32	1	0.004
Alpha-thalassemia/mental retardation syndrome, chromosome 16-related (ATR-16 syndrome)	16pter-p13.3	1	0.004
Smith-Magenis syndrome	17p11.2	1	0.004
17q11.2 duplication syndrome	17q11.2	1	0.004
18q deletion syndrome	18q	1	0.004
1q41-q42 deletion syndrome	1q41-q42	1	0.004
2q35 duplication syndrome	2q35	1	0.004
2q37 deletion syndrome	2q37	1	0.004
3q13.31 deletion syndrome	3q13.31	1	0.004
3q29 duplication syndrome	3q29	1	0.004
Wolf-Hirshhorn syndrome (WHS)	4p16.3	1	0.004
4q deletion syndrome	4q	1	0.004
Trisomy 5p syndrome	5p	1	0.004
5q14.3 deletion (distal) syndrome	5q14.3-q15	1	0.004
5q35 duplicationsyndrome	5q35	1	0.004
Sotos syndrome	5q35.3	1	0.004
Chordoma	6q27	1	0.004
7q11.23 duplication syndrome	7q11.23	1	0.004
7q11.23 deletion (distal) syndrome	7q11.23	1	0.004
Split-hand/foot malformation 1 (SHFM1)	7q21.2-q21.3	1	0.004
Monosomy 8p syndrome	8p	1	0.004
Trisomy 8p syndrome	8p	1	0.004
8p23.1 duplication syndrome	8p23.1	1	0.004
Joubert syndrome 6 (JBTS6)	8q22.1	1	0.004
9q31.1-q31.3 microdeletion syndrome	9q31.1-q31.3	1	0.004

Diseases	Cytoband	Count	%
Xq27.3-q28 duplication syndrome	Xq27.3-q28	1	0.004
2q12.2-q13_DEL	2q12.2-q13	1	0.004
12q24.31-q24.32_DEL	12q24.31-q24.32	1	0.004
15q11.2_DEL	15q11.2	1	0.004
19q13.33-q13.43_DUP	19q13.33-q13.43	1	0.004
22q13.2_DEL	22q13.2	1	0.004
Xq27.1-q28_DUP	Xq27.1-q28	1	0.004
Xq28_DUP	Xq28	1	0.004
Xp22.33-p21.1_DEL	Xp22.33-p21.1	1	0.004
Tetrasomy X	chrX	1	0.004
		464	**1.87**

Reference

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Appendix.

Table. BEBEGENE disease list

Chromosome	Cytoband	info ID
1	1p21.3	1p21.3 microdeletion syndrome
1	1p32-p31	1p32-p31 deletion syndrome
1	1p35.2	1p35.2 microdeletion syndrome
1	1p36	1p36 deletion syndrome
1	1p36	1p36 duplication syndrome
1	1q21.1	1q21.1 deletion (distal) syndrome
1	1q21.1	1q21.1 duplication (distal) syndrome
1	1q21.1	1q21.1 deletion (proximal) syndrome
1	1q21.1	1q21.1 duplication (proximal) syndrome
1	1q41-q42	1q41-q42 deletion syndrome
1	1q43-q44	1q43-q44 deletion syndrome
1	1q44	1q44 microdeletion syndrome
2	2p12-p11.2	2p12-p11.2 deletion syndrome
2	2p16.1-p15	2p15-p16.1 deletion syndrome
2	2p16.1-p15	2p15-p16.1 duplication syndrome
2	2p16.3	2p16.3 deletion syndrome
2	2p21	2p21 microdeletion syndrome
2	2p24.3	2p24.3 duplication syndrome
2	2q11.2	2q11.2 deletion syndrome
2	2q11.2	2q11.2 duplication syndrome
2	2q13	2q13 deletion syndrome
2	2q13	2q13 duplication syndrome
2	2q21.1	2q21.1 deletion syndrome
2	2q22	Mowat-Wilson syndrome (MOWS)
2	2q23.1	2q23.1 microdeletion syndrome
2	2q23.1	2q23.1 microduplication syndrome
2	2q24	2q24 microdeletion syndrome
2	2q31.1	2q31.1 microdeletion syndrome
2	2q31.1	2q31.1 duplication syndrome
2	2q31.2	2q31.2 deletion syndrome
2	2q32-q33	2q32-q33 microdeletion syndrome
2	2q34-q36	2q35 duplication syndrome
2	2q37	BDMR syndrome
3	3p25.3	3p25.3 deletion syndrome
3	3pter-p25	3pter-p25 deletion syndrome
3	3q13.31	3q13.31 deletion syndrome
3	3q21-qter	3q21 duplication syndrome
3	3q22-q24	Dandy-Walker syndrome (DWS)
3	3q26	3q26 microduplication syndrome
3	3q26-q27	3q26-q27 microdeletion syndrome
3	3q29	3q29 deletion syndrome
3	3q29	3q29 duplication syndrome
4	4p	4p duplication syndrome
4	4p16.3	Wolf-Hirshhorn syndrome (WHS)
4	4p16.3	4p16.3 duplication syndrome
4	4q	4q deletion syndrome
4	4q	Distal trisomy 4q syndrome
4	4q21	4q21 deletion syndrome
4	4q25	Axenfeld-Rieger syndrome, type 1 (RIEG1)
5	5p	Trisomy 5p syndrome
5	5p13	5p13 duplication syndrome
5	5p15	Cri du chat syndrome
5	5q12	5q12 deletion syndrome
5	5q13	Spinal muscular atrophy, type 1 (SMA1)
5	5q14.3	5q14.3 deletion (proximal) syndrome
5	5q14.3-q15	5q14.3 deletion (distal) syndrome
5	5q22.2	Familial adenomatous polyposis
5	5q31.3	5q31.3 microdeletion syndrome
5	5q35	Sotos syndrome
5	5q35	5q35 duplicationsyndrome
6	6p22	6p22 microdeletion syndrome
6	6p25.3	Axenfeld-Rieger syndrome, type 3 (RIEG3)
6	6pter-p24	6pter-p24 deletion syndrome
6	6q	Partial Trisomy 6q syndrome
6	6q11-q14	6q11-q14 deletion syndrome
6	6q16	6q16 microdeletion syndrome
6	6q24	6q24 duplication syndrome
6	6q24-q25	6q24-q25 deletion syndrome
6	6q25	6q25 microdeletion syndrome
6	6q25.3	Coffin-Siris syndrome 1 (CSS1)
6	6q27	Chordoma
7	7p	7p deletion syndrome
7	7p11.2-p13	Silver-Russell syndrome
7	7p14.1	Greig cephalopolysyndactyly syndrome (GCPS)
7	7p22.1	7p22.1 microduplication syndrome
7	7q	Monosomy 7
7	7q11.23	Williams-Beuren syndrome (WBS)
7	7q11.23	7q11.23 deletion (distal) syndrome
7	7q11.23	7q11.23 duplication (distal) syndrome
7	7q11.23	7q11.23 duplication syndrome
7	7q21.2-q21.3	Split-hand/foot malformation 1 (SHFM1)
7	7q31	7q31 microdeletion syndrome
7	7q36.3	Currarino syndrome
7	7q36.3	7q36.3 duplication syndrome

Chromosome	Cytoband	info ID
7	7q36.3	Triphalangeal thumb-polysyndactyly (TPT-PS) syndrome
8	8p	Monosomy 8p syndrome
8	8p	Trisomy 8p syndrome
8	8p11.2	8p11.2 deletion syndrome
8	8p23.1	8p23.1 deletion syndrome
8	8p23.1	8p23.1 duplication syndrome
8	8q12	8q12 microduplication syndrome
8	8q13	Mesomelia-synostoses syndrome
8	8q13.1-q13.2	Joubert syndrome 21 (JBTS21)
8	8q21.11	8q21.11 deletion syndrome
8	8q22.1	Joubert syndrome 6 (JBTS6)
8	8q22.1	Nablus mask-like facial syndrome (NMLFS)
8	8q22.1	8q22.1 duplication syndrome
8	8q24.1	Trichorhinophalangeal syndrome type 2 (TRPS2)
8	8q24.3	8q24.3 microdeletion syndrome
9	9p	Trisomy 9p syndrome
9	9p	9p deletion syndrome
9	9p13	9p13 microdeletion syndrome
9	9p24.3	9p24.3 deletion syndrome
9	9q22.3	9q22.3 deletion syndrome
9	9q31.1-q31.3	9q31.1-q31.3 microdeletion syndrome
9	9q33.3-q34.11	9q33.3q34.11 microdeletion syndrome
9	9q34.11	Early infantile epileptic encephalopathy 4 (EIEE4)
9	9q34.3	Kleefstra syndrome 1 (KLEFS1)
10	10p	Monosomy 10p syndrome
10	10p	Trisomy 10p syndrome
10	10p11.21-p12.31	10p11.21-p12.31 microdeletion syndrome
10	10p14-p13	DiGeorge syndrome/velocardiofacial syndrome complex 2 (DGS2)
10	10q	Distal Trisomy 10q syndrome
10	10q22.3-q23.3	10q22.3-q23.2 deletion syndrome
10	10q22.3-q23.3	10q22.3-q23.2 duplication syndrome
10	10q24	Split-hand/foot malformation 3 (SHFM3)
10	10q26	10q26 deletion syndrome
11	11p11.2	Potocki-Shaffer syndrome
11	11p13	11p13 deletion (distal) syndrome
11	11p13	WAGR syndrome
11	11p13	11p13 duplication syndrome
11	11p13-p12	WAGRO syndrome
11	11p15.5	Beckwith-Wiedemann syndrome (BWS)
11	11q	Partial Monosomy 11q syndrome
11	11q	Partial Trisomy 11q syndrome
11	11q13.2-q13.4	11q13.2-q13.4 deletion syndrome
11	11q22.2-q22.3	11q22.2-q22.3 microdeletion syndrome
11	11q23	11q23 deletion syndrome
11	11q23-qter	Jacobsen syndrome (JBS)
12	12p	Trisomy 12p syndrome
12	12p12.1	12p12.1 microdeletion syndrome
12	12p13.33	12p13.33 microdeletion syndrome
12	12q14	12q14 microdeletion syndrome
12	12q15-q21.1	12q15q21.1 microdeletion syndrome
13	13	Patau syndrome
13	13q	Partial Monosomy 13q syndrome
13	13q12.2-q13	Moebius syndrome
13	13q12.3	13q12.3 microdeletion syndrome
13	13q14	13q14 deletion syndrome
13	13q32	Congenital microcoria
13	13q34	Monosomy 13q34 syndrome
14	14q11.2	14q11.2 microdeletion syndrome
14	14q11.2	14q11.2 microduplication syndrome
14	14q11-q22	14q11-q22 deletion syndrome
14	14q12	14q12 microdeletion syndrome
14	14q22	Anophthalmia-hypothalamo pituitary insufficiency syndrome
14	14q22.1-q22.3	Frias syndrome
14	14q24.1-q24.3	14q24.1-q24.3 microdeletion syndrome
14	14q32	14q32 duplication syndrome
15	15q	Distal Trisomy 15q syndrome
15	15q11.2-q13.1	15q11-q13 duplication syndrome (BP1-BP3)
15	15q11.2-q13.1	Prader-Willi/Angelman Syndrome (BP1-BP3)
15	15q13.3	15q13.3 deletion syndrome (BP4 to BP5)
15	15q13.3	15q13.3 duplication syndrome
15	15q14	15q14 microdeletion syndrome
15	15q24	15q24 deletion syndrome (A-D)
15	15q24	15q24 duplication syndrome (A-C)
15	15q25.2	15q25.2 deletion (proximal) syndrome
15	15q25.2-q25.3	15q25.2-q25.3 deletion (distal) syndrome
15	15q26-qter	15q26-qter deletion syndrome
15	15q26-qter	Tetrasomy 15q26 syndrome
16	16p11.2	16p11.2 deletion (distal) syndrome
16	16p11.2	16p11.2 duplication (distal) syndrome
16	16p11.2	16p11.2 deletion (proximal) syndrome
16	16p11.2	16p11.2 duplication (proximal) syndrome
16	16p11.2-p12.2	16p11.2-p12.2 microdeletion syndrome
16	16p11.2-p12.2	16p11.2-p12.2 microduplication syndrome
16	16p12.2	16p12.2 deletion (proximal) syndrome
16	16p13.11	16p13.11 duplication syndrome
16	16p13.11	16p13.11 deletion syndrome
16	16p13.2	16p13.2 deletion syndrome

Chromosome	Cytoband	info ID
16	16p13.3	Polycystic kidney disease, infantile severe, with tuberous sclerosis (PKDTS)
16	16p13.3	Rubinstein-Taybi syndrome
16	16p13.3	16p13.3 duplication syndrome
16	16pter-p13.3	Alpha-thalassemia/mental retardation syndrome, chromosome 16-related (ATR-16 syndrome)
16	16q22	16q22 deletion syndrome
16	16q24.1	16q24.1 microdeletion syndrome
16	16q24.3	KBG syndrome
17	17p	Trisomy 17p syndrome
17	17p11.2	Smith-Magenis syndrome
17	17p11.2	Potocki-Lupski syndrome
17	17p11.2-p12	Yuan-Harel-Lupski syndrome (YUHAL)
17	17p12	17p12 deletion syndrome
17	17p12	Charcot-Marie-Tooth disease, type 1A
17	17p13.1	17p13.1 deletion syndrome
17	17p13.3	Miller-Dieker lissencephaly syndrome (MDLS)
17	17p13.3	17p13.3 duplication syndrome
17	17p13.3-p13.1	17p13.3 telomeric duplication syndrome
17	17q11.2	17q11.2 deletion syndrome
17	17q11.2	17q11.2 duplication syndrome
17	17q12	17q12 deletion syndrome
17	17q12	17q12 duplication syndrome
17	17q21.31	17q21.31 deletion syndrome
17	17q21.31	17q21.31 duplication syndrome
17	17q22	17q22 deletion syndrome
17	17q23.1-q23.2	17q23.1-q23.2 deletion syndrome
17	17q23.1-q23.2	17q23.1-q23.2 duplication syndrome
17	17q24.2-q24.3	Hypertrichosis terminalis, generalized, with or without gingival hyperplasia (HTC3)
18	18	Edward's syndrome
18	18p	Tetrasomy 18p syndrome
18	18p	18p deletion syndrome
18	18q	18q deletion syndrome
19	19p13	19p13 duplication syndrome
19	19p13.12	19p13.12 microdeletion syndrome
19	19p13.13	19p13.13 microdeletion syndrome
19	19q13.11	19q13.11 microdeletion syndrome
20	20p12.2	Alagille syndrome 1 (ALGS1)
20	20p12.3	20p12.3 microdeletion syndrome
20	20p13	20p13 microdeletion syndrome
20	20q11	20q11.2 microdeletion syndrome
20	20q13.33	20q13.33 microdeletion syndrome
21	21	Down syndrome
21	21q22.11-q22.12	21q22.11-q22.12 microdeletion syndrome
22	22q11	22q11.2 deletion syndrome
22	22q11	Emanuel Syndrome
22	22q11.1-q11.21	Cat eye syndrome
22	22q11.21-q11.22	22q11.2 deletion syndrome (distal, D-E/F)
22	22q11.21	DiGeorge/Velocardiofacial syndrome (CATCH22)
22	22q11.21	22q11.2 deletion syndrome (LCR22 B/C-D)
22	22q11.21	22q11.2 duplication syndrome (proximal, A-D)
22	22q11.21-q11.22	22q11.2 duplication (distal) syndrome
22	22q13	22q13 deletion syndrome
22	22q13	22q13 duplication syndrome
X	X	Turner syndrome
X	X	Klinefelter syndrome
X	X	Triple-X syndrome
X	Xp11.22	Xp11.22 duplication syndrome
X	Xp11.22-p11.23	Xp11.22-p11.23 duplication syndrome
X	Xp11.23	Xp11.23 microdeletion syndrome
X	Xp11.3	Xp11.3 deletion syndrome
X	Xp21	Xp21 microdeletion syndrome
X	Xp21.2	Xp21.2 microduplication syndrome
X	Xp21.2-p21.1	Duchenne muscular dystrophy (DMD)
X	Xp22.31	Xp22.31 microdeletion syndrome
X	Xq21	Xq21 microdeletion syndrome
X	Xq22.3	Xq22.3 telomeric deletion syndrome
X	Xq27.3-q28	Xq27.3-q28 duplication syndrome
X	Xq28	Xq28 deletion syndrome
X	Xq28	Xq28 duplication syndrome