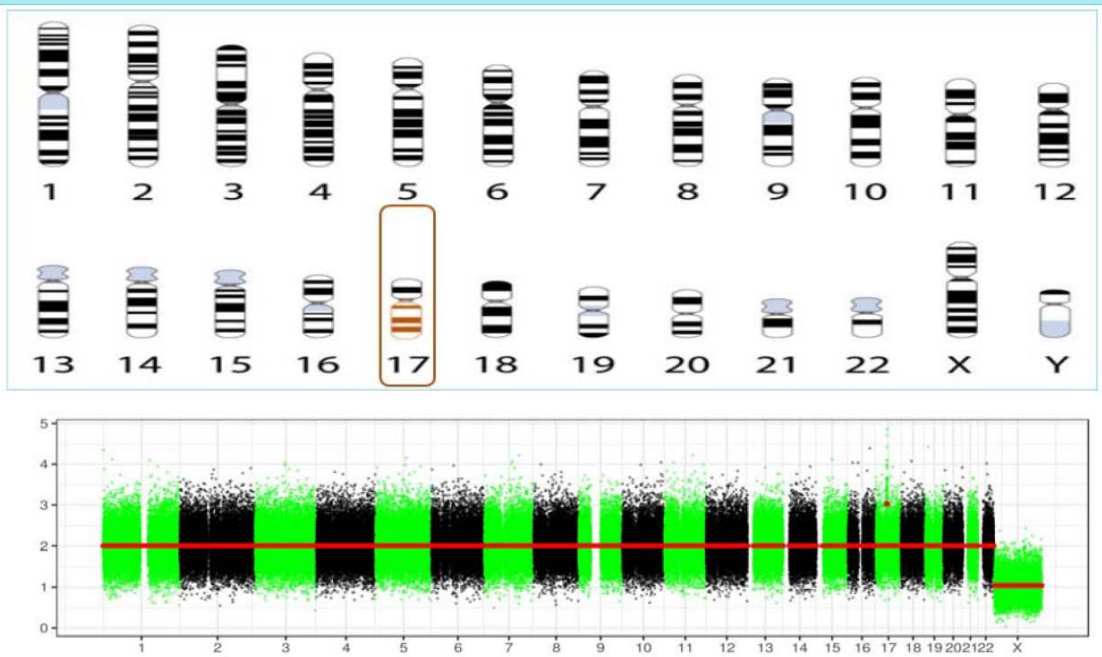
Neonatal Genome Screening Test

Institution		Accession No.	
Name		Age / Sex	
Registration No.		Specimen Type	
Dept./Doctor		Specimen Accepted	

TEST RESULT

Detected - 17q12 duplication

Chromosomal Deletion/Duplication



A . Diagram represents chromosomal deletion/duplication. If there is an abnormality, the chromosome is marked in red.
B . Graph represents results of DNA quantity of each chromosome.

Report Interpretation

Chr17: 34830001-36225001X3
About 1.4Mb duplication on 17q12 has been found. It has major symptoms, intellectual abilities ranging from normal to severe disability. It is often accompanied by speech delay, gross motor delay, seizures (75%), eye or vision problems, cardiac and renal anomalies occur, etc. However, most of cases have been known producing very mild symptoms. Penetrance is estimated about 21%.
(GeneReview :www.ncbi.nlm.nih.gov/books/NBK344340)

* This test examines chromosomal deletion/duplication abnormalities associated with major developmental disorders. The test is a screening test, not diagnostic. Genetic counseling and confirmatory test is needed for accurate diagnostics.

Tested by:Sung-Hee Bang M.T.(33995) / Approved by:Jong-Mun Choi M.D.(924) Eun-Hae Cho M.D.(690)

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DNA Quality	Test Data Quality	Control Material Quality
Good	Good	Good

Test Information

- Test Method : Next Generation Sequencing
- Test Subject : Chromosomal Deletion / Duplication
- Specimen Type : EDTA Whole Blood 0.5ml or 2 Capillary tubes or Cord Blood 0.5ml

Limitations

- This test is a screening test for rare diseases associated with developmental disorders such as Down syndrome, Edwards syndrome, and Patau syndrome. If the result is positive, confirmatory tests such as karyotype analysis, FISH, microarray, etc., are needed for accurate diagnostics.
- Genetic variants(balanced translocation, inversion, point mutation, low-level mosaicism, etc.) other than chromosomal deletion/duplication are not detected.
- It is difficult to rule out the possibility that the disease was caused by chromosomal abnormalities that could not be detected by this test. Chromosomal deletion/duplication that has unclear clinical significance in medical level at the point of reporting is not reported.
- This test is conducted with the consent of the patient and and does not directly aim at the treatment of disease or injury.

Result Details

	Chromosome Number	Deletion/Duplication Syndrome	Chromosome Loci	Test Result		Chromosome Number	Deletion/Duplication Syndrome	Chromosome Loci	Test Result
1	chr1	1p21.3 deletion syndrome	1p21.3	Not Detected	16	chr2	SPD1 syndrome /2q31.1 duplication syndrome	2q31.1	Not Detected
2		1p32-p31 deletion syndrome	1p32-p31	Not Detected	17		2q31.2–q32.3 deletion syndrome	2q31.2-q32.2	Not Detected
3		1p34.1 duplication syndrome	1p34.1	Not Detected	18		2q32–q33 deletion syndrome	2q32-q33	Not Detected
4		1p36 deletion syndrome	1pter-p36.3	Not Detected	19		2q37 deletion syndrome	2q37	Not Detected
5		1q21.1 deletion syndrome	1q21.1	Not Detected	20	chr3	3p14.1–p13 deletion syndrome	3p14.1-p13	Not Detected
6		1q24.3 deletion syndrome	1q24.3	Not Detected	21		3p21.31 deletion syndrome	3p21.31	Not Detected
7		1q24–q25 deletion syndrome	1q24-q25	Not Detected	22		Chromosome 3p deletion syndrome	3pter-p25	Not Detected
8		1q41–42 deletion syndrome	1q41-q42	Not Detected	23		3q deletion syndrome	3q13.11-q13.12	Not Detected
9	chr2	2p15–p16.1deletion syndrome	2p15–p16.1	Not Detected	24		3q13 deletion syndrome	3q13.11-q13.12	Not Detected
10		2p21deletion syndrome	2p21	Not Detected	25		3q27.3–q29 deletion syndrome	3q27.3-q29	Not Detected
11		2q11.2 deletion syndrome	2q11.2	Not Detected	26		3q29 deletion syndrome	3q29	Not Detected
12		2q13 deletion syndrome	2q13	Not Detected	27	chr4	Wolf-Hirschhorn syndrome	4pter-p16.3	Not Detected
13		2q23.1 deletion syndrome	2q23.1	Not Detected	28		4q21 deletion syndrome	4q21	Not Detected
14		2q23.3–q24.1 deletion syndrome	2q23.3-q24.1	Not Detected	29		Rieger syndrome	4q25	Not Detected
15		2q24.3 deletion syndrome	2q24.2-q24.3	Not Detected	30		4q32.1–q32.2 triplication syndrome	4q32.1q32.2	Not Detected

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i-screen NEWBORN GENOME SCREENING

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	Chromosome Number	Deletion/Duplication Syndrome	Chromosome Loci	Test Result		Chromosome Number	Deletion/Duplication Syndrome	Chromosome Loci	Test Result
31	chr5	Cri-du-chat syndrome	5p15.2-p15.33	Not Detected	62	chr16	16p11.2–p12.2 deletion syndrome	16p11.2-p12.2	Not Detected
32		5q14.3–q15 deletion syndrome	5q14.3-q15	Not Detected	63	chr17	Smith-Magenis syndrome	17p11.2	Not Detected
33		Sotos syndrome	5q35.2-q35.3	Not Detected	64		Potocki-Lupski syndrome	17p11.2	Not Detected
34	chr6	6p deletion syndrome	6p25	Not Detected	65		17p13.1 deletion syndrome	17p13.1	Not Detected
35		6q13–14 deletion syndrome	6q13–14	Not Detected	66		Miller-Dieker syndrome	17p13.3	Not Detected
36		6q25 deletion syndrome	6q25.2-q25.3	Not Detected	67		Koolen-de Vries syndrome	17q21.3	Not Detected
37	chr7	Grieg syndrome	7p14.1	Not Detected	68	chr18	Edwards syndrome	18	Not Detected
38		Williams syndrome	7q11.23	Not Detected	69		18q deletion syndrome	18q12.3-q21.1	Not Detected
39		Currarino syndrome	7q36.3	Not Detected	70	chr19	19p13.2 deletion syndrome	19p13.2	Not Detected
40	chr8	Trisomy 8	8	Not Detected	71		19q13.11 deletion syndrome	19q13.11	Not Detected
41		8p23 deletion syndrome	8p23.1	Not Detected	72	chr20	Alagille's syndrome	20p12	Not Detected
42		8q21.11 deletion syndrome	8q21.11	Not Detected	73	chr21	Down syndrome	21q22.13	Not Detected
43		Langer-giedion syndrome	8q24.1	Not Detected	74	chr22	Cat eye syndrome	22p11.1-q11.21	Not Detected
44	chr9	Trisomy 9	9	Not Detected	75		Distal 22q11.2 microdeletion syndrome	22q11.2	Not Detected
45		Tetrasomy 9p	9p	Not Detected	76		DiGeorge syndrome	22q11.21-q11.23	Not Detected
46		Chromosome 9p Deletion Syndrome	9pter-p22.3	Not Detected	77		Phelan-McDermid syndrome	22q13.33	Not Detected
47		9q22.3 deletion syndrome	9q22.3	Not Detected	78	chrX/Y	Turner syndrome	X	Not Detected
48		Kleefstra syndrome	9q34.3	Not Detected	79		Trisomy X	X	Not Detected
49	chr10	DiGeorge syndrome type 2	10p12.31	Not Detected	80		Tetrasomy X	X	Not Detected
50		10q22–q23 deletion synd ro me	10q22-q23	Not Detected	81		Pentasomy X	X	Not Detected
51		10q25–q26 deletion syndrome	10q25-q26	Not Detected	82		Xp11.3 deletion syndrome	Xp11.3	Not Detected
52	chr11	Potocki-Shaffer syndrome	11p11.2	Not Detected	83		Glycerol kinase deficiency	Xp21.2	Not Detected
53		11p deletion syndrome	11p13	Not Detected	84		Nance-Horan syndrome	Xp22.13	Not Detected
54		Jacobsen syndrome	11q23.3-qter	Not Detected	85		Kallmann's syndrome	Xp22.31	Not Detected
55	chr12	Pallister-Killian syndrome	12p	Not Detected	86		Leri-Weill syndrome	Xp22.33	Not Detected
56	chr13	Patau syndrome	13	Not Detected	87		Pelizaeus-Merzbacher syndrome	Xq22.2	Not Detected
57	chr14	14q12 duplication syndrome	14q12	Not Detected	88		Xq22.3–q23 deletion syndrome	Xq22.3-q23	Not Detected
58		14q22–q23 deletion syndrome	14q22-q23	Not Detected	89		MECP2 duplication syndrome	Xq28	Not Detected
59		14q32 deletion syndrome	14q32.2	Not Detected	90		Klinefelter's syndrome	XXY	Not Detected
60	chr15	15q12 duplication syndrome	15q11.2-q13.1	Not Detected	91	Others	Other major chromosomal variants	17q12	Detected
61		Prader-Willi/Angelman syndrome	15q11.2-q13.1	Not Detected					

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