Appendix 1. Clinically Significant Copy Number Variants Detected by Microarray, According to Nuchal Translucency Thickness

CMA; chromosomal microarray analysis; CNV, copy number variant; NIPT, noninvasive prenatal testing; OMIM, *Online Mendelian Inheritance in Man*, a continuously updated catalog of human genes and genetic disorders and traits. OMIM-morbid – gene associated with clinical disorders.

Case	CMA result (ISCN*) array GRCh37/hg19	CNV size [bp]		Syndromes/affected genes	Category*	Karyotype Detectable	NIPT detectable
Nuchal translucency 3.0 mm (n=198)							
1	21q11.2q22.3(15,016,486-48,093,361)x3	33,076,876		Trisomy 21	Pathogenic	Yes	Yes
2	22q11.21(18,894,865-20,402,655)x3 mat	1,507,791		22q11.21 duplication syndrome OMIM #608363	Pathogenic	No	No
3	Xq21.21-q21.33(88147270-97054063)x2	8,906,794		47 genes – 6 OMIM, 1 OMIM-morbid	LP	No	No
Nuchal translucency 3.1 mm (n=158)							
4	1q21.1q21.2(146,106,723-147,830,830)x3mat	1,724,108		54 genes – 10 OMIM, 2 OMIM-morbid	Pathogenic	No	No
5	4p16.1(8,210,011-10,542,148)x3,4p15.33p15.32(13,420,803-17,009,363)x3	2,332,138 + 3,588,561		75 genes – 10 OMIM, 3 OMIM-morbid; 36 genes – 12 OMIM, 4 OMIM-morbid	LP	No	No
6	21q11.2q22.3(15,016,486-48,093,361)x2-3			Trisomy 21 mosaicism	Pathogenic	Yes	Yes
7-9	21q11.2q22.3(15,016,486-48,093,361)x3			Trisomy 21 (3 cases)	Pathogenic	Yes	Yes
Nuchal translucency 3.2 mm (n=108)							
10	10p15.3q26.3(100,047-135,426,386)x2-3/ 18p11.32q23(136,227-78,013,728)x2-3 40%	135,326,340 + 77,877,502		Trisomy 10 and 18 mosaicism	Pathogenic	Yes	Yes
11	13q21.33q31.1(69,020,199-87,381,357)x1dn	18,361,159		132 genes – 16 OMIM, 9 OMIM-morbid	Pathogenic	Yes	No
12	16p11.2(29631159-30281111)x3	649,953		16p11.2 duplication syndrome (OMIM #614671)	Pathogenic	No	No

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13	18p11.32q23(136,227-78,013,728)x3	77,877,502		Trisomy 18	Pathogenic	Yes	Yes
14-15	21q11.2q22.3(15,016,486-48,093,361)x3	33,076,876		Trisomy 21 (2 cases)	Pathogenic	Yes	Yes
Nuchal translucency 3.3 mm (n=78)							
16	7p22.3q36.3(43,376-159,119,707)x2-3	159,076,332		2227 genes – 63 OMIM-morbid	Pathogenic	Yes	No
17	8p23.1(8094406-11898209)x1	3,803,804		8p23.1 deletion syndrome (ORPHA:251071)	Pathogenic	No	No
18	18p11.32q23(136,227-78,013,728)x3	77,877,502		Trisomy 18	Pathogenic	Yes	Yes
19-22	21q11.2q22.3(15,016,486-48,093,361)x3	33,076,876		Trisomy 21 (4 cases)	Pathogenic	Yes	Yes
23	Xp22.33q28(1-155,270,560)x3	155,270,560		47,XXX syndrome	Pathogenic	Yes	Yes
Nuchal translucency 3.4 mm (n=77)							
24	2p25.3q37.3(12,770-242,782,258)x2-3	242,769,489		Trisomy 2 mosaicism	Pathogenic	Yes	No
25	16p11.2(29503993-30190017)x1	686,025		16p11.2 deletion syndrome (OMIM #611913)	Pathogenic	No	No
26	16p11.2(29,580,020-30,176,508)x3	596,489		16p11.2 duplication syndrome (OMIM #614671)	Pathogenic	No	No
27	21q11.2q22.3(15,016,486-48,093,361)x3	33,076,876		Trisomy 21	Pathogenic	Yes	Yes
28	22q11.21(18916842-21800471)x1	2,883,630		DiGeorge syndrome (OMIM #188400)	Pathogenic	No	No
29	Xp22.33q28(1-155,270,560)x2	155,270,560		Klinefelter syndrome	Pathogenic	Yes	Yes

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Appendix 2. Findings of Questionable Significance Detected by Microarray in 619 Pregnancies With Nuchal Translucency of 3-3.4 mm

CMA; chromosomal microarray analysis; CNV, copy number variant; NIPT, noninvasive prenatal testing; OMIM, *Online Mendelian Inheritance in Man*, a continuously updated catalog of human genes and genetic disorders and traits. OMIM-morbid – gene associated with clinical disorders.

Case	CMA result (ISCN*) array GRCh37/hg19	CNV size [bp]	Syndromes/affected genes	Nuchal translucency	Inheritance
Variants of unknown significance					
1	6q14.3q15(85,958,544-88,235,246)x3	2,276,703	46 genes – 4 OMIM-morbid	3 mm	De novo
2	14q31.3(85,077,027-92,504,745)x3	7,427,719	69 genes – 10 OMIM-morbid	3 mm	Not reported
3	9p13.2(36,516,705-37,665,085)x3	1,148,381	21 genes – 2 OMIM-morbid	3.1 mm	Not reported
4	6q24.1(140,996,521-142,666,633)x1	1,670,113	10 genes – 1 OMIM-morbid	3.1 mm	Not reported
5	3p12.1(86,704,171-87,479,589)x1	775,419	8 genes – 2 OMIM-morbid	3.3 mm	Not reported
High-frequency low-penetrant findings					
1	15q13.2q13.3(30,936,285-32,620,127)x3	1,683,843	22 genes – 2 OMIM-morbid	3.1 mm	Maternal
2	15q13.3(32,003,537-32,439,251)x3	435,715	3 genes – 0 OMIM-morbid	3.3 mm	Paternal
3	15q13.2q13.3(31,104,220-32,915,723)x3	1,811,504	33 genes – 2 OMIM-morbid	3.4 mm	Paternal

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