iGene® NIPT Comprehensive MAX
List of Deletion Syndromes

#	Size	Location	Suspected Condition	Reference
1	99.2	7q	Chromosome 7q deletion	252270
2	60.9	18q	Chromosome 18q deletion syndrome	601808
3	49	9p	Chromosome 9p deletion syndrome	158170
4	40.5	14q11-q22	Chromosome 14q11-q22 deletion syndrome	613457
5	28	1p36	Chromosome 1p36 deletion syndrome	607872
6	27	6q11-q14	Chromosome 6q11-q14 deletion syndrome	613544
7	25.3	8q12.2-q21.2	Chromosome 8q12.1-q21.2 deletion syndrome	600257
8	22.3	Xq21	Chromosome Xq21 deletion syndrome	303110
9	22.1	1q41-q42	Chromosome 1q41-q42 deletion syndrome	612530
10	22	6q24-q25	Chromosome 6q24-q25 deletion syndrome	612863
11	19.7	3q22-q24	Dandy-Walker syndrome	220200
12	17.2	18p	Chromosome 18p deletion syndrome	146390
13	16.4	10q26	Chromosome 10q26 deletion syndrome	609625
14	16.4	3pter-p25	Chromosome 3pter-p25 deletion Syndrome	613792
15	15.5	2p12-p11.2	Chromosome 2p12-p11.2 deletion syndrome	613564
16	15.4	5q14.3-q15	Chromosome 5q14.3 deletion syndrome	612881
17	15.2	13q14	Chromosome 13q14 deletion syndrome	613884
18	15	10q23	Chromosome 10q22.3-q23.2 deletion syndrome	612242
19	13.4	15q26-qter	Levy-Shanske syndrome	614846
20	13.4	15q26-qter	Chromosome 15q26-qter deletion syndrome	612626
21	13.4	6pter-p24	Chromosome 6pter-p24 deletion syndrome	612582
22	13.3	2q31	Split-hand/foot malformation 5	606708
23	13.2	Xq27.3-q28	Chromosome Xq27.3-q28 duplication syndrome	300869
24	13.1	2q37.1-q37.3	Holoprosencephaly 6	605934
25	12.7	Xp21	Chromosome Xp21 deletion syndrome	300679
26	12.5	5p	Cri du Chat syndrome	123450
27	12.5	11p13-p12	WAGRO syndrome	612469
28	11.7	4q21	Chromosome 4q21 deletion syndrome	613509
29	11.5	17p12-p11.2	Yuan-Harel-Lupski syndrome	616652
30	11.2	22q11	Cat-Eye syndrome	115470
31	10.8	11q23	Jacobsen syndrome	147791
32	10.7	10p14-p13	DiGeorge syndrome 2	601362
33	9.6	8q24.11-q24.13	Langer-Giedion syndrome	150230
34	9.1	2p16.1-p15	Chromosome 2p16.1-p15 deletion syndrome	612513
35	8.9	4q32.1-q32.2	Chromosome 4q32.1-q32.2 triplication syndrome	613603
36	8.7	16p12.2-p11.2	Chromosome 16p12.2-p11.2 deletion syndrome	613604
37	8.4	Xp11.23-p11.22	Chromosome Xp11.23-p11.22 duplication syndrome	300801
38	8.3	2q31.1	Chromosome 2q31.1 duplication syndrome	613681
39	8.3	2q33.1	Chromosome 2q33.1 deletion syndrome	612313
40	8.2	Xq28	Chromosome Xq28 deletion syndrome	300845
41	8	22q11.2	Chromosome 22q11.2 duplication syndrome	608363
42	7.9	16pter-p13.3	Chromosome 16p deletion syndrome	141750

#	Size	Location	Suspected Condition	Reference
43	7.9	16p13.3	Chromosome 16p13.3 deletion syndrome	600273
44	7.8	16p12.2-p11.2	Chromosome 16p11.2-p12.2 microduplication syndrome	https://decipher.sanger.ac.uk/syndrome/96#overview
45	7.8	5q12	Chromosome 5q12 deletion syndrome	615668
46	7.6	1p32-p31	Chromosome 1p32-p31 deletion syndrome	613735
47	7.6	1p31.3	Chromosome 1p31 duplication syndrome	164750
48	7.4	16q22	Chromosome 16q22 deletion syndrome	614541
49	7.2	14q22.1-q22.3	Frias syndrome	609640
50	6.7	15q11	Chromosome 15q11-q13 duplication syndrome	608636
51	6.6	6q27	CHDM	215400
52	6.5	15q14	Chromosome 15q14 deletion syndrome	616898
53	6.3	17q12	Chromosome 17q12 duplication syndrome	614526
54	6.3	17q12	Chromosome 17q12 deletion syndrome	614527
55	5.7	3q29	Chromosome 3q29 duplication syndrome	611936
56	5.7	3q29	Chromosome 3q29 deletion syndrome	609425
57	5.7	8q22.1	Chromosome 8q22.1 duplication syndrome	151200
58	5.7	8q22.1	Chromosome 8q22.1 deletion syndrome	608156
59 / 60	5.7	15q11.2	Prader-Willi/Angelman syndrome	176270/105830
61	5.5	21q22.3	Holoprosencephaly 1	236100
62	5.4	11p13	WAGR syndrome	194072
63	5.3	7q11.23	Chromosome 7q11.23 deletion syndrome	613729
64	5.3	7q11.23	Chromosome 7q11.23 duplication syndrome	609757
65	5.3	11p11.2	Potocki-Shaffer syndrome	601224
66	5.2	15q26.1	HCD	142340
67	5	Xq22.3	Chromosome Xq22.3 telomeric deletion syndrome	300194
68	4.5	4p16.3	Wolf-Hirschhorn syndrome	194190
69	4	17q21.31	Chromosome 17q21.31 duplication syndrome	613533
70	4	Xp11.3	Chromosome Xp11.3 deletion syndrome	300578
71	3.8	3q13.31	Chromosome 3q13.31 deletion syndrome	615433
72	3.7	8p23.1	Chromosome 8p23.1 deletion syndrome	https://decipher.sanger.ac.uk/syndrome/39#overview
73	3.7	8p23.1	Chromosome 8p23.1 duplication syndrome	https://decipher.sanger.ac.uk/syndrome/85#overview
74	3.6	12q14	Chromosome 12q14 microdeletion syndrome	https://decipher.sanger.ac.uk/syndrome/76#overview
75	3.5	17q23.1-q23.2	Chromosome 17q23.1-q23.2 deletion syndrome	613355
76	3.4	17p11.2	Potocki-Lupski syndrome	610883
77	3.4	17p11.2	Smith-Magenis syndrome	182290
78	3.3	17p13.3	Chromosome 17p13.3 duplication syndrome	613215
79	3.3	17p13.3	Chromosome 17p13.3 deletion syndrome	247200
80	3.1	19q13.11	Chromosome 19q13.11 deletion syndrome	617219
81	22	2q34-q36	Chromosome 2q35 duplication syndrome	185900
82	10	15q25	Chromosome 15q25 deletion syndorme	614294
83	8	22q11.2	Chromosome 22q11.2 deletion syndrome	611867
84	2.4	22q11.21	DiGeorge syndrome	188400

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