NICC® Extended - Supplementary Sheet

NICC® Extended provides comprehensive screening for chromosomes 21, 18, 13, 9, 16, 22, X & Y, 80+ microdeletion/duplication syndromes and other numerical chromosomal aneuploidies in the 23chromosomes.The 80+ microdeletions/duplication syndromes tested by **NICC® Extended** are listed in the table below. Should any of these conditions is detected it will be reported under: **Other findings include microdeletion/duplication syndromes and incidental finding.**

Note: The test is powered by NIFTY® Pro.

List of Microdeletion/duplication Syndromes tested by NICC® Extended

Size	Location	Suspected Disease	Reference
99.2	7q	Chromosome 7q deletion	252270
60.9	18q	Chromosome 18q deletion syndrome	601808
49.0	9p	Chromosome 9p deletion syndrome	158170
40.5	14q11-q22	Chromosome 14q11-q22 deletion syndrome	613457
28.0	1p36	Chromosome 1p36 deletion syndrome	607872
27.0	6q11-q14	Chromosome 6q11-q14 deletion syndrome	613544
25.3	8q12.2-q21.2	Chromosome 8q12.1-q21.2 deletion syndrome	600257
22.3	Xq21	Chromosome Xq21 deletion syndrome	303110
22.1	1q41-q42	Chromosome 1q41-q42 deletion syndrome	612530
22.0	6q24-q25	Chromosome 6q24-q25 deletion syndrome	612863
19.7	3q22-q24	Dandy-Walker syndrome	220200
17.2	18p	Chromosome 18p deletion syndrome	146390
16.4	10q26	Chromosome 10q26 deletion syndrome	609625
16.4	3pter-p25	Chromosome 3pter-p25 deletion syndrome	613792
15.5	2p12-p11.2	Chromosome 2p12-p11.2 deletion syndrome	613564
15.4	5q14.3-q15	Chromosome 5q14.3 deletion syndrome	612881
15.2	13q14	Chromosome 13q14 deletion syndrome	613884
15.0	10q23	Chromosome 10q22.3-q23.2 deletion syndrome	612242
13.4	15q26-qter	Levy-Shanske syndrome	614846
13.4	15q26-qter	Chromosome 15q26-qter deletion syndrome	612626
13.3	2q31	Split-hand/foot malformation 5	606708
13.4	6pter-p24	Chromosome 6pter-p24 deletion syndrome	612582
13.2	Xq27.3-q28	Chromosome Xq27.3-q28 duplication syndrome	300869
13.1	2q37.1-q37.3	Holoprosencephaly 6	605934
12.7	Xp21	Chromosome Xp21 deletion syndrome	300679
12.5	5p	Cri du Chat syndrome	123450
12.5	11p13-p12	WAGRO syndrome	612469
11.7	4q21	Chromosome 4q21 deletion syndrome	613509
11.5	17p12-p11.2	Yuan-Harel-Lupski syndrome	616652
11.2	22q11	Cat-Eye syndrome	115470
10.8	11q23	Jacobsen syndrome	147791
10.7	10p14-p13	DiGeorge syndrome 2	601362
9.6	8q24.11-q24.13	Langer-Giedion syndrome	150230
9.1	2p16.1-p15	Chromosome 2p16.1-p15 deletion syndrome	612513
8.9	4q32.1-q32.2	Chromosome 4q32.1-q32.2 triplication syndrome	613603
8.7	16p12.2-p11.2	Chromosome 16p12.2-p11.2 deletion syndrome	613604
8.4	Xp11.23-p11.22	Chromosome Xp11.23-p11.22 duplication syndrome	300801
8.3	2q31.1	Chromosome 2q31.1 duplication syndrome	613681
8.3	2q33.1	Chromosome 2q33.1 deletion syndrome	612313
8.2	Xq28	Chromosome Xq28 deletion syndrome	300845

Size	Location	Suspected Disease	Reference
8.0	22q11.2	Chromosome 22q11.2 duplication syndrome	608363
7.9	16pter-p13.3	Chromosome 16p deletion syndrome	141750
7.9	16p13.3	Chromosome 16p13.3 deletion syndrome	600273
7.8	16p12.2-p11.2	Chromosome 16p11.2-p12.2 microduplication syndrome	https://decipher.sanger.ac.uk/syndrome/96#overview
7.8	5q12	Chromosome 5q12 deletion syndrome	615668
7.6	1p32-p31	Chromosome 1p32-p31 deletion syndrome	613735
7.6	1p31.3	Chromosome 1p31 duplication syndrome	164750
7.4	16q22	Chromosome 16q22 deletion syndrome	614541
7.2	14q22.1-q22.3	Frias syndrome	609640
6.7	15q11	Chromosome 15q11-q13 duplication syndrome	608636
6.6	6q27	CHDM	215400
6.5	15q14	Chromosome 15q14 deletion syndrome	616898
6.3	17q12	Chromosome 17q12 duplication syndrome	614526
6.3	17q12	Chromosome 17q12 deletion syndrome	614527
5.7	3q29	Chromosome 3q29 duplication syndrome	611936
5.7	3q29	Chromosome 3q29 deletion syndrome	609425
5.7	8q22.1	Chromosome 8q22.1 duplication syndrome	151200
5.7	8q22.1	Chromosome 8q22.1 deletion syndrome	608156
5.7	15q11.2	Prader-Will/Angelman syndrome	176270 /105830
5.5	21q22.3	Holoprosencephaly 1	236100
5.4	11p13	WAGR syndrome	194072
5.3	7q11.23	Chromosome 7q11.23 deletion syndrome	613729
5.3	7q11.23	Chromosome 7q11.23 duplication syndrome	609757
5.3	11p11.2	Potocki-Shaffer syndrome	601224
5.2	15q26.1	HCD	142340
5.0	Xq22.3	Chromosome Xq22.3 telomeric deletion syndrome	300194
4.5	4p16.3	Wolf-Hirschhorn syndrome	194190
4.0	17q21.31	Chromosome 17q21.31 duplication syndrome	613533
4.0	Xp11.3	Chromosome Xp11.3 deletion syndrome	300578
3.8	3q13.31	Chromosome 3q13.31 deletion syndrome	615433
3.7	8p23.1	Chromosome 8p23.1 deletion syndrome	https://deciphersanger.ac.uk/syndrome/39#overview
3.7	8p23.1	Chromosome 8p23.1 duplication syndrome	https://decipher.sanger.ac.uk/syndrome/85#overview
3.6	12q14	Chromosome 12q14 microdeletion syndrome	https://decipher.sanger.ac.uk/syndrome/76#overview
3.5	17q23.1-q23.2	Chromosome 17q23.1-q23.2 deletion syndrome	613355
3.4	17p11.2	Potocki-Lupski syndrome	610883
3.4	17p11.2	Smith-Magenis syndrome	182290
3.3	17p13.3	Chromosome 17p13.3 duplication syndrome	613215
3.3	17p13.3	Chromosome 17p13.3 deletion syndrome	247200
3.1	19q13.11	Chromosome 19q13.11 deletion syndrome	617219
22.	2q34-q36	Chromosome 2q35 duplication syndrome	185900
10.	15q25	Chromosome 15q25 deletion syndrome	614294
8.0	22q11.2	Chromosome 22q11.2 deletion syndrome	611867
2.4	22q11.21	DiGeorge syndrome	188400