

Lista sindroamelor de microdeleție/duplicație a căror detecție este inclusă în setul de teste NIFTY®Pro

<i>Size</i>	<i>Location</i>	<i>Suspected disease</i>	<i>Reference</i>	<i>Size</i>	<i>Location</i>	<i>Suspected disease</i>	<i>Referințe/Reference</i>
99.2	7q	Chromosome 7q deletion	252270	7.9	16p13.3	Chromosome 16p13.3 deletion syndrome	600273
60.9	18q	Chromosome 18q deletion syndrome	601808	7.8	16p12.2-p11.2	Chromosome 16p11.2-p12.2 microduplication syndrome	<a href="https://decipher.sanger.ac.uk/syndrome/96#overview">https://decipher.sanger.ac.uk/syndrome/96#overview</a>
49.0	9p	Chromosome 9p deletion syndrome	158170	7.8	5q12	Chromosome 5q12 deletion syndrome	615668
40.5	14q11-q22	Chromosome 14q11-q22 deletion syndrome	613457	7.6	1p32-p31	Chromosome 1p32-p31 deletion syndrome	613735
28.0	1p36	Chromosome 1p36 deletion syndrome	607872	7.6	1p31.3	Chromosome 1p31 duplication syndrome	164750
27.0	6q11-q14	Chromosome 6q11-q14 deletion syndrome	613544	7.4	16q22	Chromosome 16q22 deletion syndrome	614541
25.3	8q12.2-q21.2	Chromosome 8q12.1-q21.2 deletion syndrome	600257	7.2	14q22.1-q22.3	Frias syndrome	609640

22.3	Xq21	Chromosome Xq21 deletion syndrome	303110	6.7	15q11	Chromosome 15q11-q13 duplication syndrome	608636
22.1	1q41-q42	Chromosome 1q41-q42 deletion syndrome	612530	6.6	6q27	CHDM	215400
22.0	6q24-q25	Chromosome 6q24-q25 deletion syndrome	612863	6.5	15q14	Chromosome 15q14 deletion syndrome	616898
19.7	3q22-q24	Dandy-Walker syndrome	220200	6.3	17q12	Chromosome 17q12 duplication syndrome	614526
17.2	18p	Chromosome 18p deletion syndrome	146390	6.3	17q12	Chromosome 17q12 deletion syndrome	614527
16.4	10q26	Chromosome 10q26 deletion syndrome	609625	5.7	3q29	Chromosome 3q29 duplication syndrome	611936
16.4	3pter-p25	Chromosome 3pter-p25 deletion Syndrome	613792	5.7	3q29	Chromosome 3q29 deletion syndrome	609425
15.5	2p12-p11.2	Chromosome 2p12-p11.2 deletion syndrome	613564	5.7	8q22.1	Chromosome 8q22.1 duplication syndrome	151200
15.4	5q14.3-q15	Chromosome 5q14.3 deletion syndrome	612881	5.7	8q22.1	Chromosome 8q22.1 deletion syndrome	608156

15.2	13q14	Chromosome 13q14 deletion syndrome	613884	5.7	15q11.2	Prader-Will/Angelman syndrome	176270/105830
15.0	10q23	Chromosome 10q22.3-q23.2 deletion syndrome	612242	5.5	21q22.3	Holoprosencephaly 1	236100
13.4	15q26-qter	Levy-Shanske syndrome	614846	5.4	11p13	WAGR syndrome	194072
13.4	15q26-qter	Chromosome 15q26-qter deletion syndrome	612626	5.3	7q11.23	Chromosome 7q11.23 deletion syndrome	613729
13.4	6pter-p24	Chromosome 6pter-p24 deletion syndrome	612582	5.3	7q11.23	Chromosome 7q11.23 duplication syndrome	609757
13.3	2q31	Split-hand/foot malformation 5	606708	5.3	11p11.2	Potocki-Shaffer syndrome	601224
13.2	Xq27.3-q28	Chromosome Xq27.3-q28 duplication syndrome	300869	5.2	15q26.1	HCD	142340
13.1	2q37.1-q37.3	Holoprosencephaly 6	605934	5.0	Xq22.3	Chromosome Xq22.3 telomeric deletion syndrome	300194
12.7	Xp21	Chromosome Xp21 deletion syndrome	300679	4.5	4p16.3	Wolf-Hirschhorn syndrome	194190
12.5	5p	Cri du Chat syndrome	123450	4.0	17q21.31	Chromosome 17q21.31	613533

						duplication syndrome	
12.5	11p13-p12	WAGRO syndrome	612469	4.0	Xp11.3	Chromosome Xp11.3 deletion syndrome	300578
11.7	4q21	Chromosome 4q21 deletion syndrome	613509	3.8	3q13.31	Chromosome 3q13.31 deletion syndrome	615433
11.5	17p12-p11.2	Yuan-Harel-Lupski syndrome	616652	3.7	8p23.1	Chromosome 8p23.1 deletion syndrome	<a href="https://decipher.sanger.ac.uk/syndrome/39#overview">https://decipher.sanger.ac.uk/syndrome/39#overview</a>
11.2	22q11	Cat-Eye syndrome	115470	3.7	8p23.1	Chromosome 8p23.1 duplication syndrome	<a href="https://decipher.sanger.ac.uk/syndrome/85#overview">https://decipher.sanger.ac.uk/syndrome/85#overview</a>
10.8	11q23	Jacobsen syndrome	147791	3.6	12q14	Chromosome 12q14 microdeletion syndrome	<a href="https://decipher.sanger.ac.uk/syndrome/76#overview">https://decipher.sanger.ac.uk/syndrome/76#overview</a>
10.7	10p14-p13	DiGeorge syndrome 2	601362	3.5	17q23.1-q23.2	Chromosome 17q23.1-q23.2 deletion syndrome	613355
9.6	8q24.11-q24.13	Langer-Giedion syndrome	150230	3.4	17p11.2	Potocki-Lupski syndrome	610883
9.1	2p16.1-p15	Chromosome 2p16.1-p15 deletion syndrome	612513	3.4	17p11.2	Smith-Magenis syndrome	182290
8.9	4q32.1-q32.2	Chromosome 4q32.1-q32.2	613603	3.3	17p13.3	Chromosome 17p13.3	613215

		triplication syndrome				duplication syndrome	
8.7	16p12.2-p11.2	Chromosome 16p12.2-p11.2 deletion syndrome	613604	3.3	17p13.3	Chromosome 17p13.3 deletion syndrome	247200
8.4	Xp11.23-p11.22	Chromosome Xp11.23-p11.22 duplication syndrome	300801	3.1	19q13.11	Chromosome 19q13.11 deletion syndrome	617219
8.3	2q31.1	Chromosome 2q31.1 duplication syndrome	613681	22.0	2q34-q36	Chromosome 2q35 duplication syndrome	185900
8.3	2q33.1	Chromosome 2q33.1 deletion syndrome	612313	10.8	15q25	Chromosome 15q25 deletion syndrome	614294
8.2	Xq28	Chromosome Xq28 deletion syndrome	300845	8.0	22q11.2	Chromosome 22q11.2 deletion syndrome	611867
8.0	22q11.2	Chromosome 22q11.2 duplication syndrome	608363	2.4	22q11.21	DiGeorge syndrome	188400
7.9	16pter-p13.3	Chromosome 16p deletion syndrome	141750				