

Localização	Síndrome	Referência
7q	Chromosome 7q deletion	252270
18q	Chromosome 18q deletion syndrome	601808
9p	Chromosome 9p deletion syndrome	158170
14q11-q22	Chromosome 14q11-q22 deletion syndrome	613457
1p36	Chromosome 1p36 deletion syndrome	607872
6q11-q14	Chromosome 6q11-q14 deletion syndrome	613544
8q12.2-q21.2	Chromosome 8q12.1-q21.2 deletion syndrome	600257
Xq21	Chromosome Xq21 deletion syndrome	303110
1q41-q42	Chromosome 1q41-q42 deletion syndrome	612530
6q24-q25	Chromosome 6q24-q25 deletion syndrome	612863
3q22-q24	Dandy-Walker syndrome	220200
18p	Chromosome 18p deletion syndrome	146390
10q26	Chromosome 10q26 deletion syndrome	609625
3pter-p25	Chromosome 3pter-p25 deletion Syndrome	613792
2p12-p11.2	Chromosome 2p12-p11.2 deletion syndrome	613564
5q14.3-q15	Chromosome 5q14.3 deletion syndrome	612881
13q14	Chromosome 13q14 deletion syndrome	613884
10q23	Chromosome 10q22.3-q23.2 deletion syndrome	612242
15q26-qter	Levy-Shanske syndrome	614846
15q26-qter	Chromosome 15q26-qter deletion syndrome	612626
6pter-p24	Chromosome 6pter-p24 deletion syndrome	612582
2q31	Split-hand/foot malformation 5	606708
Xq27.3-q28	Chromosome Xq27.3-q28 duplication syndrome	300869
2q37.1-q37.3	Holoprosencephaly 6	605934
Xp21	Chromosome Xp21 deletion syndrome	300679
5p	Cri du Chat syndrome	123450
11p13-p12	WAGRO syndrome	612469
4q21	Chromosome 4q21 deletion syndrome	613509
17p12-p11.2	Yuan-Harel-Lupski syndrome	616652
22q11	Cat-Eye syndrome	115470
11q23	Jacobsen syndrome	147791
10p14-p13	DiGeorge syndrome 2	601362
8q24.11-q24.13	Langer-Giedion syndrome	150230
2p16.1-p15	Chromosome 2p16.1-p15 deletion syndrome	612513
4q32.1-q32.2	Chromosome 4q32.1-q32.2 triplication syndrome	613603
16p12.2-p11.2	Chromosome 16p12.2-p11.2 deletion syndrome	613604
Xp11.23-p11.22	Chromosome Xp11.23-p11.22 duplication syndrome	300801
2q31.1	Chromosome 2q31.1 duplication syndrome	613681
2q33.1	Chromosome 2q33.1 deletion syndrome	612313
Xq28	Chromosome Xq28 deletion syndrome	300845
22q11.2	Chromosome 22q11.2 duplication syndrome	608363

Referências: OMIM & <http://decipher.sanger.ac.uk>

Localização	Síndrome	Referência
16pter-p13.3	Chromosome 16p syndrome	141750
16p13.3	Chromosome 16p13.3 deletion syndrome	600273
16p12.2-p11.2	Chromosome 16p11.2-p12.2 microduplication syndrome	https://decipher.sanger.ac.uk/syndrome/96#overview
5q12	Chromosome 5q12 deletion syndrome	615668
1p32-p31	Chromosome 1p32-p31 deletion syndrome	613735
1p31.3	Chromosome 1p31 duplication syndrome	164750
16q22	Chromosome 16q22 deletion syndrome	614541
14q22.1-q22.3	Frias syndrome	609640
15q11	Chromosome 15q11-q13 duplication syndrome	608636
6q27	CHDM	215400
15q14	Chromosome 15q14 deletion syndrome	616898
17q12	Chromosome 17q12 duplication syndrome	614526
17q12	Chromosome 17q12 deletion syndrome	614527
3q29	Chromosome 3q29 duplication syndrome	611936
3q29	Chromosome 3q29 deletion syndrome	609425
8q22.1	Chromosome 8q22.1 duplication syndrome	151200
8q22.1	Chromosome 8q22.1 deletion syndrome	608156
15q11.2	Prader-Willi/Angelman syndrome	176270/105830
21q22.3	Holoprosencephaly 1	236100
11p13	WAGR syndrome	194072
7q11.23	Chromosome 7q11.23 deletion syndrome	613729
7q11.23	Chromosome 7q11.23 duplication syndrome	609757
11p11.2	Potocki-Shaffer syndrome	601224
15q26.1	HCD	142340
Xq22.3	Chromosome Xq22.3 telomeric deletion syndrome	300194
4p16.3	Wolf-Hirschhorn syndrome	194190
17q21.31	Chromosome 17q21.31 duplication syndrome	613533
Xp11.3	Chromosome Xp11.3 deletion syndrome	300578
3q13.31	Chromosome 3q13.31 deletion syndrome	615433
8p23.1	Chromosome 8p23.1 deletion syndrome	https://decipher.sanger.ac.uk/syndrome/39#overview
8p23.1	Chromosome 8p23.1 duplication syndrome	https://decipher.sanger.ac.uk/syndrome/85#overview
12q14	Chromosome 12q14 microdeletion syndrome	https://decipher.sanger.ac.uk/syndrome/76#overview
17q23.1-q23.2	Chromosome 17q23.1-q23.2 deletion syndrome	613355
17p11.2	Potocki-Lupski syndrome	610883
17p11.2	Smith-Magenis syndrome	182290
17p13.3	Chromosome 17p13.3 duplication syndrome	613215
17p13.3	Chromosome 17p13.3 deletion syndrome	247200
19q13.11	Chromosome 19q13.11 deletion syndrome	617219
2q34-q36	Chromosome 2q35 duplication syndrome	185900
15q25	Chromosome 15q25 deletion syndrome	614294
22q11.2	Chromosome 22q11.2 deletion syndrome	611867
22q11.21	DiGeorge syndrome	188400