

Table 1: List of Investigated Microdeletion/Microduplication Syndromes

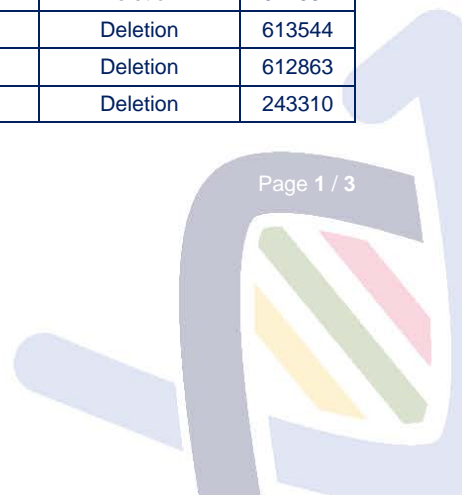
Chromosomal Locus	Microdeletion/Microduplication Syndrome	Type of Chromosomal Anomaly	OMIM
1p36	1p36 Deletion Syndrome (distal)	Deletion	607872
1p36	1p36 Deletion Syndrome (proximal)	Deletion	619343
1p35	1p35 Deletion Syndrome	Deletion	617930
1p32-p31	1p32-p31 Deletion Syndrome	Deletion	613735
1p31.3	1p31 Deletion Syndrome	Deletion	164750
1q21.1	1q21.1 Microdeletion Syndrome	Deletion	612474
1q21.1	1q21.1 Microduplication Syndrome	Duplication	612475
1q41-q42	1q41-q42 Deletion Syndrome	Deletion	612530
1q43-q44	1q43-q44 Deletion Syndrome	Deletion	612337
2p25.3	2p25.3 Deletion Syndrome	Deletion	616521
2p25.3	2p25.3 Duplication Syndrome	Duplication	616521
2p16.1-p15	2p16.1-p15 Deletion Syndrome	Deletion	612513
2p12-p11.2	2p12-p11.2 Deletion Syndrome	Deletion	613564
2q23.1	2q23.1 Microdeletion Syndrome	Deletion	156200
2q23.1	2q23.1 Microduplication Syndrome	Duplication	156200
2q31	Split-Hand/Foot Malformation 5	Deletion	606708
2q31.1	2q31.1 Duplication Syndrome	Duplication	613681
2q31.2	2q31.2 Deletion Syndrome	Deletion	612345
2q32-q33	Glass Syndrome (2q32-q33 Deletion Syndrome)	Deletion	612313
2q33.1	2q33.1 Deletion Syndrome	Deletion	612313
2q34-q36	2q35 Duplication Syndrome	Duplication	185900
2q37	2q37 Deletion Syndrome (Brachydactyly-Intellectual Deficit Syndrome)	Deletion	600430
2q37.1-q37.3	Holoprosencephaly Type 6	Deletion	605934
3pter-p25	3pter-p25 Deletion Syndrome	Deletion	613792
3q13.31	3q13.31 Deletion Syndrome	Deletion	615433
3q22-q24	Dandy-Walker Syndrome	Deletion	220200
3q29	3q29 Duplication Syndrome	Duplication	611936
3q29	3q29 Deletion Syndrome	Deletion	609425
4p16.3	Wolf-Hirschhorn Syndrome (4p16.3 Deletion Syndrome)	Deletion	194190
4q21	4q21 Deletion Syndrome	Deletion	613509
4q32.1-q32.2	4q32.1-q32.2 Triplication Syndrome	Duplication	613603
5p15.3	Cri du Chat Syndrome (5p Deletion Syndrome)	Deletion	123450
5q12	5q12 Deletion Syndrome	Deletion	615668
5q14.3	5q14.3 Deletion Syndrome (proximal)	Deletion	613443
5q14.3-q15	5q14.3 Deletion Syndrome	Deletion	612881
5q35.3	Sotos Syndrome (5q35 Deletion Syndrome)	Deletion	117550
6pter-p24	6pter-p24 Deletion Syndrome	Deletion	612582
6q11-q14	6q11-q14 Deletion Syndrome	Deletion	613544
6q24-q25	6q24-q25 Deletion Syndrome	Deletion	612863
7p22	7p22 Deletion Syndrome (Baraitser-Winter Syndrome)	Deletion	243310

GENOMICA srl
REGISTERED OFFICE

Rome: Via Arduino 38 - 00162
 Pec: info@pec.genomicalab.it
 VAT no.: 14554101007
 REA: RM-1530210

LABORATORIES

Rome: Via Arduino 38 - 00162 - Tel.: 06.21115020
 E-mail: info@genomicalab.it
 Website: www.genomicalab.it



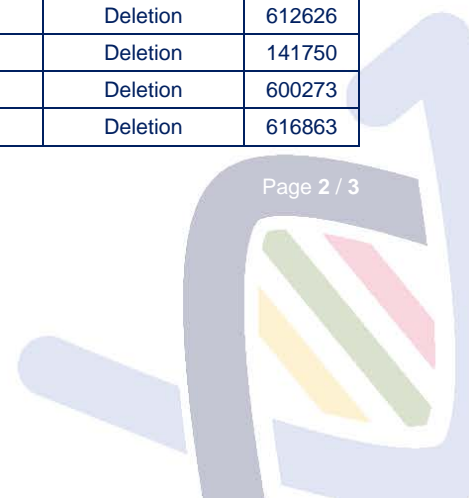
7q	Monosomy 7q	Deletion	252270
7q11.23	7q11.23 Deletion Syndrome (distal, 1.2 Mb)	Deletion	613729
7q11.23	7q11.23 Duplication Syndrome	Duplication	609757
7q11.23	Williams-Beuren Syndrome	Deletion	194050
8pter-p23.1	Recombinant Chromosome 8 Syndrome (8pter-p23.1 Deletion)	Deletion	179613
8p23.1	8p23.1 Deletion Syndrome	Deletion	146390
8p23.1	8p23.1 Duplication Syndrome	Duplication	614290
8q12.2-q21.2	8q12.2-q21.2 Deletion Syndrome	Deletion	600257
8q21.11	8q21.11 Deletion Syndrome	Deletion	614230
8q22.1	8q22.1 Duplication Syndrome	Duplication	151200
8q22.1	8q22.1 Deletion Syndrome	Deletion	608156
8q22.1-qter	Recombinant Chromosome 8 Syndrome (8q22.1-qter Duplication)	Duplication	179613
8q24.11-q24.13	Langer-Giedion Syndrome (8q24.1 Deletion Syndrome)	Deletion	150230
8q24.3	8q24.3 Deletion Syndrome (Verheij Syndrome)	Deletion	615583
9p	Monosomy 9p	Deletion	158170
9q34.3	9q34.3 Deletion Syndrome (Kleefstra Syndrome)	Deletion	610253
10p14-p13	DiGeorge Syndrome Type 2	Deletion	601362
10p12-p11	10p12-p11 Deletion Syndrome (DeSanto-Shinawi Syndrome)	Deletion	616708
10q22.3-q23.2	10q22.3-q23.2 Deletion Syndrome	Deletion	612242
10q26	10q26 Deletion Syndrome	Deletion	609625
11p13	WAGR Syndrome (11p13 Deletion Syndrome)	Deletion	194072
11p13-p12	WAGRO Syndrome (11p13-p12 Deletion Syndrome)	Deletion	612469
11p11.2	Potocki-Shaffer Syndrome (11p11.2 Deletion Syndrome)	Deletion	601224
11q23	Jacobsen Syndrome (11q Deletion Syndrome)	Deletion	147791
12q14	12q14 Microdeletion Syndrome	Deletion	166700
12q15	12q15 Deletion Syndrome	Deletion	618608
12q21.33	12q21.33 Deletion Syndrome	Deletion	612868
13q14	13q14 Deletion Syndrome	Deletion	613884
13q33-q34	13q33-q34 Deletion Syndrome	Deletion	619148
14q11-q22	14q11-q22 Deletion Syndrome	Deletion	613457
14q22.1-q22.3	Frias Syndrome (14q22 Deletion Syndrome)	Deletion	609640
14q32	14q32 Deletion Syndrome	Deletion	618147
15q11-q13	15q11-q13 Duplication Syndrome	Duplication	608636
15q11.2	Prader-Willi Syndrome	Deletion	176270
15q11.2	Angelman Syndrome	Deletion	105830
15q13.3	15q13.3 Microdeletion Syndrome	Deletion	612001
15q14	15q14 Deletion Syndrome	Deletion	616898
15q24	Witteveen-Kolk Syndrome (15q24 Deletion Syndrome)	Deletion	613406
15q24	Witteveen-Kolk Syndrome (15q24 Duplication Syndrome)	Duplication	613406
15q25	15q25 Deletion Syndrome	Deletion	614294
15q26.1	Congenital Diaphragmatic Hernia	Deletion	142340
15q26-qter	Levy-Shanske Syndrome (Tetrasomy 15q26)	Duplication	614846
15q26-qter	Distal 15q Monosomy (15q26-qter Deletion Syndrome)	Deletion	612626
16pter-p13.3	16p Deletion Syndrome	Deletion	141750
16p13.3	16p13.3 Deletion Syndrome	Deletion	600273
16p13.2	16p13.2 Deletion Syndrome (Hao-Fountain Syndrome)	Deletion	616863

GENOMICA srl
REGISTERED OFFICE

Rome: Via Arduino 38 - 00162
Pec: info@pec.genomicalab.it
VAT no.: 14554101007
REA: RM-1530210

LABORATORIES

Rome: Via Arduino 38 - 00162 - Tel.: 06.21115020
E-mail: info@genomicalab.it
Website: www.genomicalab.it



16p13.11	16p13.11 Microdeletion Syndrome	Deletion	Decipher
16p13.11	16p13.11 Microduplication Syndrome	Duplication	Decipher
16p12.2-p11.2	16p12.2-p11.2 Deletion Syndrome	Deletion	613604
16p12.2-p11.2	16p11.2-p12.2 Microduplication Syndrome	Duplication	136570
16q22	16q22 Deletion Syndrome	Deletion	614541
17p13.3	17p13.3 Duplication Syndrome	Duplication	613215
17p13.3	Miller-Dieker Lissencephaly Syndrome (17p13.3 Deletion Syndrome)	Deletion	247200
17p13.1	17p13.1 Deletion Syndrome	Deletion	613776
17p12	Charcot-Marie-Tooth Type 1A (CMT1A)	Duplication	118220
17p12	Hereditary Liability to Pressure Palsies (HNPP)	Deletion	162500
17p12-p11.2	Yuan-Harel-Lupski Syndrome	Duplication	616652
17p11.2	Potocki-Lupski Syndrome	Duplication	610883
17p11.2	Smith-Magenis Syndrome (17p11.2 Deletion Syndrome)	Deletion	182290
17q11.2	NF1 Microdeletion Syndrome (17q11.2 Deletion Syndrome)	Deletion	613675
17q11.2	17q11.2 Duplication Syndrome (NF1 Microduplication Syndrome)	Duplication	618874
17q12	17q12 Duplication Syndrome	Duplication	614526
17q12	17q12 Deletion Syndrome	Deletion	614527
17q12	Renal Cysts and Diabetes Syndrome	Deletion	137920
17q21.31	17q21.31 Duplication Syndrome	Duplication	613533
17q23.1-q23.2	17q23.1-q23.2 Deletion Syndrome	Deletion	613355
17q23.1-q23.2	17q23.1-q23.2 Duplication Syndrome	Duplication	613618
18q	18q Monosomy	Deletion	601808
18p	De Grouchy Syndrome (18p Deletion Syndrome)	Deletion	146390
19p13.13	19p13.13 Deletion Syndrome	Deletion	613638
19p13.13	19p13.13 Duplication Syndrome	Duplication	613638
19p13	19p13 Duplication Syndrome	Duplication	615830
19q13.11	19q13.11 Deletion Syndrome	Deletion	617219
19q13.11	19q13.11 Deletion Syndrome (distal)	Deletion	613026
20q11-q12	20q11-q12 Deletion Syndrome	Deletion	614257
21q22.3	Type 1 Holoprosencephaly	Deletion	236100
22q11	Cat Eye Syndrome	Duplication	115470
22q11.2	DiGeorge Syndrome (22q11.2 Deletion Syndrome)	Deletion	188400
22q11.2	22q11.2 Duplication Syndrome	Duplication	608363
22q11.2 Distal	22q11.2 Deletion Syndrome (distal)	Deletion	611867
22q13	Phelan-McDermid Syndrome (22q13.3 Deletion Syndrome)	Deletion	606232
Xp22.31	Steroid Sulphatase Deficiency (X-linked Ichthyosis)	Deletion	308100
Xp21	Xp21 Deletion Syndrome	Deletion	300679
Xp11.3	Xp11.3 Deletion Syndrome	Deletion	300578
Xp11.23-p11.22	Xp11.23-p11.22 Duplication Syndrome	Duplication	300801
Xq13.2-q13.3	Xq13 Duplication Syndrome	Duplication	301069
Xq21	Xq21 Deletion Syndrome	Deletion	303110
Xq22.3	Xq22.3 Telomeric Deletion Syndrome	Deletion	300194
Xq27.3-q28	Xq27.3-q28 Duplication Syndrome	Duplication	300869
Xq28	Xq28 Deletion Syndrome	Deletion	300845
Xq28	Xq28 Chromosome Duplication Syndrome	Duplication	300815

GENOMICA srl
REGISTERED OFFICE

Rome: Via Arduino 38 - 00162
 Pec: info@pec.genomicalab.it
 VAT no.: 14554101007
 REA: RM-1530210

LABORATORIES

Rome: Via Arduino 38 - 00162 - Tel.: 06.21115020
 E-mail: info@genomicalab.it
 Website: www.genomicalab.it

