

# Microdeletion / Microduplication Syndromes

## Microdeletion/Microduplication Syndromes

Chromosome	Syndrome	Chromosome	Syndrome	Chromosome	Syndrome
1p36	1p36 deletion	7p14.1	Greig cephalopolysyndactyly syndrome	15q11-q12	Prader-Willi/Angelman Syndrome
2p16.1-p15	2p16.1-p15 deletion	8q23.1-q24.13	Langer-Giedion Syndrome	16p11.2-p12.2	16p11.2-p12.2 deletion
3q29	3q29 deletion	10q26	10q26 deletion	22q11.21	DiGeorge's Syndrome
4p16	Wolf-Hirschhorn Syndrome	11p13	WAGR deletion	22q13.3	Phelan-McDermid Syndrome
5p15.2-p15.33	Cri-du-chat Syndrome	11p23-q25	Jacobsen syndrom	Xp22.3	Kallmann syndrome

## List for other parts

Chromosome	Syndrome	Chromosome	Syndrome	Chromosome	Syndrome	Chromosome	Syndrome
1p36.33	1p36.33 duplication	5p	5p duplication	11p	11p duplication	17q24.2	17q24.2 deletion
1p35.2	1p35.2 deletion	5p	5p deletion	11p	11p deletion	17p11.2	Smith-Magenis Syndrome
1p34.1	1p34.1 duplication	5p13	5p13 duplication	11p15	Silver-Russell syndrome, SRS	17p11.2	Potocki-Lupski Syndrome
1p32-p31	1p32-p31 deletion	5q35	Sotos syndrome	11p15.4	11p15.4 duplication	17p12	Charcot-Marie-Tooth disease type 1A, CMT1A
1p21.3	1p21.3 deletion	5q35	5q35 duplication	11p11.2	Potocki-Shaffer Syndrome(PSS)	17p13.3	Miller-Dieker syndrome
1q21.1	1q21.1 duplication	5q31.3	5q31.3 deletion	11q23-q25	Jacobsen Syndrome	17p21.3	Koolen-de Vries Syndrome
1q21.1	1q21.1 deletion	5q23.3-q31.1	Hougen-Janssen	11q23	11q23 deletion	18p	18p deletion
1q24-q25	1q24-q25 deletion	5q14.3-q15	5q14.3-q15 deletion	11q	11q duplication	18p	18p duplication
1q41-q42	1q41-q42 deletion	6p	6p deletion	11q	11q deletion	18q	18q deletion
1q43-q44	1q43-q44 deletion	6p	6p duplication	12p	12p duplication	19p	19p deletion
1q44	1q44 deletion	6p22	6p22 deletion	12p	12p deletion	19p	19p duplication
2p24.1-p23.3	2p24.1-p23.3 deletion	6p25.3-p25.2	Axenfeld-Rieger Syndrome type 3	12p12.1	12p12.1 deletion	19p13.2	19p13.2 deletion
2q13-q22.3	2q13-q22.3 duplication	6q25.3	ARID1B Syndrome	12q15-q21	12q15-q21 deletion	19p13.13	19p13.13 deletion
2p21	2p21 deletion	6q25.2-q25.3	6q25.2-q25.3 deletion	12q14	12q14 deletion	19p13.12	19p13.12 deletion
2p16.3	2p16.3 deletion	6q25.1	6q25.1 deletion	12q	12q duplication	19p13.3	19p13.3 duplication
2p15-p16.1	2p15-p16.1 deletion	6q21-q22.31	6q21-q22.31 deletion	12q	12q deletion	19q13.11	19q13.11 deletion
2p13.2	2p13.2 deletion	7p21.2-p21.1	Saethre-Chotzen Syndrome	13q	13q duplication	19q	19q duplication
2q13	2q13 deletion	7p22.1	7p22.1 duplication	13q12.3	13q12.3 deletion	20p	20p duplication
2q23.1	2q23.1 deletion	7p15.3-p14.3	Cyclic Vomiting Syndrome	13q14	13q14 deletion	20p	20p deletion
2q23.1	2q23.1 duplication	7q11.23	Williams Syndrome	13q34	13q34 deletion	20p12	Alagille Syndrome
2q24	2q24 deletion	7q11.23	7q11.23 duplication	13q	13q deletion	20p12.3-p12.1	Congenital Myasthenic Syndrome type 18
2q11.2	2q11.2 deletion	7q31	7q31 deletion	14q11-q22	14q11-q22 deletion	20p13	Okhiro syndrome
2q31.1	2q31.1 deletion	7q36.3	Currarino Syndrome	14q11.2	14q11.2 deletion	20q13.33	20q13.33 deletion
2q32-q33	2q32-q33 deletion	7q36.3	Megalencephaly-Capillary Malformation Syndrome	14q11.2	14q11.2 duplication	20q11.2	20q11.2 duplication
2q31.2-q32.3	2q31.2-q32.3 deletion	8p	8p duplication	14q12	14q12 duplication	20q11.2	20q11.2 deletion
2q33.1	2q33.1 deletion	8p23.1	8p23.1 deletion	14q22	14q22 deletion	21q	21q deletion
2q36.1-q36.2	Waardenburg syndrome type 1	8p11.2	8p11.2 deletion	14q22-q23	14q22-q23 deletion	21q22.11-q22.12	21q22.11-q22.12 deletion
2q37	2q37 deletion	8q12	8q12 duplication	14q24.1-q24.3	14q24.1-q24.3 deletion	21q22.13-q22.2	DYRK1A-related intellectual disability syndrome
3q22-q24	Dandy Walker Syndrome	8q13	Mesomelia-synostoses Syndrome	14q31-q32	14q31-q32 deletion	22p	22p duplication
3p25.3	3p25.3 deletion	8q21.11	8q21.11 deletion	15p13.3	15p13.3 deletion	22p	22p deletion
3p25	SETD5 Syndrome	8q22.1	8q22.1 deletion	15q11.2	15q11.2 deletion	22q11	Cat Eye Syndrome
3p14.1-p13	3p14.1-p13 deletion	8q24.3	8q24.3 deletion	15q11.2-13.1	15q11.2-13.1 duplication	22q11.2	22q11.2 duplication
3p21.31	3p21.31 deletion	9p	9p duplication	15q13.3	15q13.3 deletion	22q11.2	Velocardiofacial Syndrome
3q13	3q13 deletion	9p	9p deletion	15q14	15q14 deletion	22q	22q duplication
3q13.31	3q13.31 deletion	9p13	9p13 deletion	15q24	15q24 deletion	22q	22q deletion
3q23	3q23 deletion	9q22.3	9q22.3 deletion	15q	15q duplication	Xp22	Xp22.13-p22.2 duplication
3q26	3q26 duplication	9q31.1-q31.3	9q31.1-q31.3 deletion	15q	15q deletion	Xp21	Xp21 deletion
3q26-q28	3q26-q28 deletion	9q34.3	Kleefstra Syndrome	16p	16p duplication	Xp11.22-p11.23	Xp11.22-p11.23 duplication
3q26.33-q27.2	anophthalmia/Microphthalmos/Esophageal Atresia Syndrome	9q22.2-31.1	Gorlin syndrome	16p	16p deletion	Xq21	Xq21 deletion
3q28-q29	3q28-q29 deletion	10p	10p duplication	16p13.11	16p13.11 duplication	Xq22.3	X-linked Alport Syndrome
4p	4p deletion	10p	10p deletion	16p13.11	16p13.11 deletion	Xq25	Xq25 duplication
4p	4p duplication	10p15.3-15.1	10p15.3 microdeletion syndrome	16p13.3	16p13.3 deletion	Xq27.3-q28	Xq27.3-q28 duplication
4p16.3	4p16.3 duplication	10p12.31	Di George's Syndrome type 2	16p13.3	16p13.3 duplication	Xq28	Xq28 duplication
4q25	Axenfeld-Rieger Syndrome	10q22.3-q23.3	10q22.3-q23.3 duplication	16p13.2	16p13.2 deletion	Yq11	Non-obstructive Azoospermia
4q21	4q21 deletion	10q22.3-q23.3	10q22.3-q23.3 deletion	16p11.2	16p11.2 deletion		
4q32.1-q32.2	4q32.1-q32.2 duplication	10q23	10q23 deletion	17q	17q duplication		
4q	4q deletion	10q24	10q24 duplication	17q24.2-q24.3	17q24.2-q24.3 deletion		