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- 웹 주소: www.orpha.net

ORPHA No.	Disease	Synonym(s)
236	Trisomy 9p	Duplication 9p
280	Wolf-Hirschhorn syndrome	Distal monosomy 4p
281	Monosomy 5p	Cri du chat syndrome
502	Trichorhinophalangeal syndrome type 2	Monosomy 8q24.1
531	Miller-Dieker syndrome	Monosomy 17p13.3
567	22q11.2 deletion syndrome	DiGeorge syndrome
574	Monosomy 21	Partial 21q monosomy
893	WAGR syndrome	Monosomy 11p13
904	Williams syndrome	Monosomy 7q11.23
1001	2q37 microdeletion syndrome	Monosomy 2q37qter
1435	Xq21 microdeletion syndrome	Monosomy Xq21
1587	Monosomy 13q14	Deletion 13q14
1598	Monosomy 18p	De Grouchy syndrome
1600	Monosomy 18q	18q deletion syndrome
1606	1p36 deletion syndrome	Monosomy 1pter
1617	2q24 microdeletion syndrome	Monosomy 2q24
1621	3q13 microdeletion syndrome	Monosomy 3q13
1627	Deletion 5q35	Monosomy 5q35
1699	Trisomy 12p	Duplication 12p
1715	Trisomy 18p	Duplication 18p
1727	22q11.2 duplication syndrome	22q11.2 microduplication syndrome
1738	Trisomy 4p	Duplication 4p
1742	Trisomy 5p	Duplication 5p
1752	Trisomy 8q	Duplication 8q
1762	Proximal Xq28 duplication syndrome	MECP2 duplication syndrome
2308	Jacobsen syndrome	Distal monosomy 11q
2496	Mesomelia-synostoses syndrome	Monosomy 8q13
48652	Monosomy 22q13.3	Phelan-McDermid syndrome
65286	3q29 microdeletion syndrome	Monosomy 3qter
77301	Monosomy 9q22.3	Microdeletion 9q22.3
85332	X-linked intellectual disability-retinitis pigmentosa syndrome	Retinitis pigmentosa and intellectual disability due to monosomy Xp11.3
94063	12q14 microdeletion syndrome	Monosomy 12q14
94065	15q24 microdeletion syndrome	Monosomy 15q24
96072	4p16.3 microduplication syndrome	Telomeric duplication 4p
96078	16p13.3 microduplication syndrome	Telomeric duplication 16p
96095	3q26 microduplication syndrome	Trisomy 3q26

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96095	3q26 microduplication syndrome	Trisomy 3q26
96121	7q11.23 microduplication syndrome	Trisomy 7q11.23
96123	Monosomy 22	Deletion 22
96147	Kleefstra syndrome due to 9q34 microdeletion	Kleefstra syndrome due to monosomy 9q34
96168	Monosomy 13q34	Distal deletion 13q34
97685	17q11 microdeletion syndrome	Monosomy 17q11
98794	Angelman syndrome due to maternal 15q11q13 deletion	Angelman syndrome due to maternal monosomy 15q11q13
139474	17q11.2 microduplication syndrome	Grisart-Destrée syndrome
163693	2p21 microdeletion syndrome	Monosomy 2p21
171829	6q16 microdeletion syndrome	Monosomy 6q16
171929	Trisomy 10p	-
178303	8q22.1 microdeletion syndrome	Monosomy 8q22.1
199318	15q13.3 microdeletion syndrome	Monosomy 15q13.3
217340	17q21.31 microduplication syndrome	Trisomy 17q21.31
217346	19q13.11 microdeletion syndrome	Monosomy 19q13.11
217377	Microduplication Xp11.22p11.23 syndrome	Trisomy Xp11.22p11.23
217385	17p13.3 microduplication syndrome	Trisomy 17p13.3
228384	5q14.3 microdeletion syndrome	Monosomy 5q14.3
228399	8q12 microduplication syndrome	Trisomy 8q12
228402	2q23.1 microdeletion syndrome	Monosomy 2q23.1
228415	5q35 microduplication syndrome	Trisomy 5q35
231137	Silver-Russell syndrome due to 7p11.2p13 microduplication	Silver-Russell syndrome due to trisomy 7p11.2-p13
238446	15q11q13 microduplication syndrome	Trisomy 15q11q13
238750	4q21 microdeletion syndrome	Monosomy 4q21
238769	1q44 microdeletion syndrome	Monosomy 1q44
250994	1q21.1 microduplication syndrome	Trisomy 1q21.1
250999	1q41q42 microdeletion syndrome	Monosomy 1q41q42
251014	2q31.1 microdeletion syndrome	Monosomy 2q31.1
251019	2q32q33 microdeletion syndrome	Monosomy 2q32q33
251028	2q33.1 microdeletion syndrome	Monosomy 2q33.1
251038	3q29 microduplication syndrome	Trisomy 3q29
251046	6p22 microdeletion syndrome	Monosomy 6p22
251056	6q25 microdeletion syndrome	Monosomy 6q25
251061	7q31 microdeletion syndrome	Monosomy 7q31
251066	8p11.2 deletion syndrome	Monosomy 8p11.2
251071	8p23.1 microdeletion syndrome	Monosomy 8p23.1

ORPHA No.	Disease	Synonym(s)
251076	8p23.1 duplication syndrome	Trisomy 8p23.1
254346	19p13.12 microdeletion syndrome	Monosomy 19p13.12
254528	Kagami-Ogata syndrome due to maternal 14q32.2 microdeletion	Maternal monosomy 14q32.2
261112	Monosomy 9p	Alfi syndrome
261120	14q11.2 microdeletion syndrome	Monosomy 14q11.2
261144	14q12 microdeletion syndrome	Monosomy 14q12
261183	15q11.2 microdeletion syndrome	Monosomy 15q11.2
261190	15q14 microdeletion syndrome	Monosomy 15q14
261197	Proximal 16p11.2 microdeletion syndrome	Proximal monosomy 16p11.2
261204	16p11.2p12.2 microduplication syndrome	Trisomy 16p11.2p12.2
261211	16p11.2p12.2 microdeletion syndrome	Monosomy 16p11.2p12.2
261229	14q11.2 microduplication syndrome	Trisomy 14q11.2
261236	16p13.11 microdeletion syndrome	Monosomy 16p13.11
261243	16p13.11 microduplication syndrome	Trisomy 16p13.11
261250	16q24.3 microdeletion syndrome	Monosomy 16q24.3
261265	17q12 microdeletion syndrome	Monosomy 17q12
261272	17q12 microduplication syndrome	Trisomy 17q12
261279	17q23.1q23.2 microdeletion syndrome	Monosomy 17q23.1q23.2
261290	Trisomy 17p	Dup(17p)
261295	20p12.3 microdeletion syndrome	Monosomy 20p12.3
261304	Paternal 20q13.2q13.3 microdeletion syndrome	Paternal monosomy 20q13.2q13.3
261311	20q13.33 microdeletion syndrome	Monosomy 20q13.33
261318	Trisomy 20p	Duplication of 20p
261323	21q22.11q22.12 microdeletion syndrome	Monosomy 21q22.11q22.12
261344	Trisomy 1q	Duplication 1q
261349	2p15p16.1 microdeletion syndrome	Monosomy 2p15p16.1
261476	Xp21 microdeletion syndrome	Monosomy Xp21
261483	Xq27.3q28 duplication syndrome	Trisomy Xq27.3-q28
261537	Mowat-Wilson syndrome due to monosomy 2q22	Hirschsprung disease and intellectual disability due to monosomy 2q22
261584	Familial adenomatous polyposis due to 5q22.2 microdeletion	Familial polyposis coli due to monosomy 5q22.2; Familial adenomatous polyposis due to monosomy 5q22.2; Colorectal adenomatous polyposis due to monosomy 5q22.2
261600	Alagille syndrome due to 20p12 microdeletion	Alagille-Watson syndrome due to monosomy 20p12; Arteriohepatic dysplasia due to monosomy 20p12; Syndromic bile duct paucity due to monosomy 20p12
261638	Okhiro syndrome due to 20q13 microdeletion	Duane-radial ray syndrome due to monosomy 20q13
264200	14q22q23 microdeletion syndrome	Monosomy 14q22-q23
264450	Trisomy 8p	Duplication 8p
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	Monosomy 21q22.13q22.2
276413	10q22.3q23.3 microdeletion syndrome	Monosomy 10q22.3q23.3
276422	10q22.3q23.3 microduplication syndrome	Trisomy 10q22.3q23.3
284160	8q21.11 microdeletion syndrome	Monosomy 8q21.11
284169	Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion	Monosomy 10p11.21p12.31

ORPHA No.	Disease	Synonym(s)
289513	12q15q21.1 microdeletion syndrome	Monosomy 12q15q21.1
293948	1p21.3 microdeletion syndrome	Monosomy 1p21.3
294026	Syndactyly-nystagmus syndrome due to 2q31.1 microduplication	Syndactyly-nystagmus syndrome due to trisomy 2q31.1
300305	11p15.4 microduplication syndrome	Trisomy 11p15.4
313781	20p13 microdeletion syndrome	Monosomy 20p13
313884	12p12.1 microdeletion syndrome	Monosomy 12p12.1
313947	2q23.1 microduplication syndrome	Trisomy 2q23.1
314034	7p22.1 microduplication syndrome	Trisomy 7p22.1
314655	Severe neonatal hypotonia-seizures-encephalopathy syndrome due to 5q31.3 microdeletion	Monosomy 5q31.3
324313	9p13 microdeletion syndrome	Monosomy 9p13
329802	5p13 microduplication syndrome	Trisomy 5p13
352629	16q24.1 microdeletion syndrome	Monosomy 16q24.1
356947	3q26q27 microdeletion syndrome	Monosomy 3q26q27
363958	17q21.31 microdeletion syndrome	Monosomy 17q21.31
401923	9q31.1q31.3 microdeletion syndrome	Monosomy 9q31.1q31.3
401935	14q24.1q24.3 microdeletion syndrome	Monosomy 14q24.1q24.3
401986	1p31p32 microdeletion syndrome	Monosomy 1p31p32
412035	13q12.3 microdeletion syndrome	Monosomy 13q12.3
435638	3p25.3 microdeletion syndrome	Monosomy 3p25.3
444002	11q22.2q22.3 microdeletion syndrome	Monosomy 11q22.2q22.3
444051	20q11.2 microdeletion syndrome	Monosomy 20q11
456298	1p35.2 microdeletion syndrome	Monosomy 1p35.2
456328	X-linked myotubular myopathy-abnormal genitalia syndrome	Xq28 contiguous gene deletion syndrome
477817	PMP22-RAI1 contiguous gene duplication syndrome	Trisomy 17p11.2-p12
488280	14q32 duplication syndrome	Trisomy 14q32
495818	9q33.3q34.11 microdeletion syndrome	Monosomy 9q33.3q34.11
500055	16p13.2 microdeletion syndrome	Monosomy 16p13.2
502437	4q25 proximal deletion syndrome	Proximal monosomy 4q25
508488	8q24.3 microdeletion syndrome	Monosomy 8q24.3

