# LISTADO DE SÍNDROMES ESTUDIADOS

| | Condition | OMIM# | Gene/ Locus | Location |
|---|---|---|---|---|
| 1 | 1p36 Microdeletion * | 607872 | Multiple | 1p36 |
| 2 | 1q21.1 Distal microdeletion * | 612474 | Multiple<br>ACP6 candidate<br>GJA5 candidate<br>GJA8 candidate | 1q21.1-q21.2 |
| 3 | 1q21.1 Microdeletion with susceptibility for thrombocytopenia-absent radius (TAR) | 274000 | Multiple | 1q21.1 |
| 4 | 1q41-q42 Microdeletion/Fryns | 612530/229850 | Multiple | 1q41-q42 |
| 5 | 1q44 Microdeletion | 612337 | Multiple<br>AKT3 candidate | 1q44 |
| 6 | 2p15-p16.1 Microdeletion | 612513 | Multiple | 2p15-p16.1 |
| 7 | 2p16.3 Microdeletion/Pitt-Hopkins-like 2 * | 600565 | NRXN1 | 2p16.3 |
| 8 | 2p21 Microdeletion, autosomal recessive | 606407 | Multiple | 2p21 |
| 9 | 2q23.1 Microdeletion | | MBD5<br>EPC2 | 2q23.1 |
| 10 | 2q32.2-q33 Microdeletion | 119540 | Multiple<br>SATB2 candidate | 2q33.1 |
| 11 | 3q29 Microdeletion | 609425 | Multiple | 3q29 |
| 12 | 3q29 Microduplication * | 611936 | Multiple | 3q29 |
| 13 | 5p13.2 Microduplication * | 613174 | NIPBL | 5p13.2 |
| 14 | 5q35.2-q35.3 Microduplication * | | NSD1 | 5q35.2-q35.3 |
| 15 | 6p25.3 Microdeletion | 612582 | Multiple | 6p25.3 |
| 16 | 6q24.3 Microdeletion | 612863 | Multiple | 6q24.3 |

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| 17 | 6q25.2-q25.3 Microdeletion | 612863 | Multiple | 6q25.2-q25.3 |
| 18 | 7q11.23 Microduplication * | 609757 | Multiple | 7q11.23 |
| 19 | 8p23.1 Microdeletion/Congenital diaphragmatic hernia 2 (CDH2) | 222400 | Multiple GATA4 candidate | 8p23.1 |
| 20 | 8p23.1 Microduplication * | | Multiple GATA4 candidate | 8p23.1 |
| 21 | 8q12 Microduplication * | | Multiple CHD7 candidate | 8q12.1-q12.3 |
| 22 | 9q22.32-q22.33 Microdeletion | | Multiple TGFBR1 candidate GABBR2 candidate | 9q22.32-q22.33 |
| 23 | 9q34 Microdeletion/Kleefstra * | 610253 | EHMT1 | 9q34.3 |
| 24 | 10q22.3-q23.31 Microdeletion | | Multiple | 10q23.1-q23.2 |
| 25 | 12q14.1-q15 Microdeletion * | | Multiple LEMD3 candidate GRIP1 candidate HMGA2 candidate | 12q14.3-q15 |
| 26 | 12q14.3-related primordial dwarfism/Russell-Silver-like | | HMGA2 | 12q14.3 |
| 27 | 12q24.21-q24.23 Microduplication * | | Multiple | 12q24.21-q24.23 |
| 28 | 14q11.2 Microdeletion | | Multiple CHD8 candidate SUPT16H candidate | 14q11.2 |
| 29 | 14q12 Microduplication * | 164874 | Multiple FOXG1 candidate | 14q12 |
| 30 | 14q22-q23 Microdeletion | | Multiple | 14q22-q23 |
| 31 | 14q32.2 Microdeletion causing upd(14)mat phenotype | | Multiple DLK1 candidate RTL1 candidate | 14q32.2 |
| 32 | 14q32.2 Microdeletion causing upd(14)pat phenotype | 608149 | Multiple MEG3 candidate MEG8 candidate RTL1 candidate | 14q32.2 |
| 33 | 15q11-q13 Microduplication * | 608636 | Multiple | 15q11-q13 |

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| 34 | 15q13.3 Microdeletion * | 612001 | Multiple<br>CHRNA7<br>candidate | 15q13.3 |
| 35 | 15q24.1-q24.2 Microdeletion | 613406 | Multiple | 15q24.1-q24.2 |
| 36 | 15q24.1-q24.2 Microduplication * | 613406 | Multiple | 15q24.1-q24.2 |
| 37 | 16p11.2 Microdeletion * | 611913 | Multiple | 16p11.2 |
| 38 | 16p11.2-p12.2 Microdeletion * | 613604 | Multiple | 16p11.2-p12.2 |
| 39 | 16p12.1 Microdeletion | | Multiple<br>CDR2<br>candidate<br>EEF2K<br>candidate<br>UQCRC2<br>candidate | 16p12.2 |
| 40 | 16p13.11 Microdeletion * | | Multiple | 16p13.1 |
| 41 | 16p13.3 Microdeletion/Severe Rubinstein-Taybi | 610543 | Multiple<br>CREBBP | 16p13.3 |
| 42 | 16p13.3 Microduplication * | 613458 | CREBBP | 16p13.3 |
| 43 | 16q11.2-q12.2 Microdeletion | | Multiple<br>SALL1<br>candidate<br>ZNF423<br>candidate | 16q11.2-q12.1 |
| 44 | 17p13.1 Microdeletion | 613776 | Multiple<br>TP53 | 17p13.1 |
| 45 | 17p13.3 Distal microdeletion | | YWHAE<br>CRK | 17p13.3 |
| 46 | 17p13.3 Distal microduplication, not including lissencephaly region (PAFAH1B1) * | 613215 | YWHAE<br>CRK | 17p13.3 |
| 47 | 17p13.3 Proximal microduplication, including lissencephaly region (PAFAH1B1) * | 613215 | PAFAH1B1 (LIS1)<br>YWHAE<br>CRK | 17p13.3 |
| 48 | 17q12 Microduplication * | | Multiple<br>HNF1B<br>candidate<br>LHX1<br>candidate | 17q12 |
| 49 | 17q21.31 Microdeletion | 610443 | Multiple<br>MAPT<br>candidate | 17q21.31 |
| 50 | 17q21.31 Microduplication * | 613533 | Multiple | 17q21.31 |
| 51 | 17q23.1-q23.2 Microdeletion * | 613355 | Multiple<br>TBX2<br>candidate<br>TBX4<br>candidate | 17q23.1-q23.2 |

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| 52 | 19q13.11 Microdeletion | 613026 | Multiple<br>LSM14A<br>candidate<br>UBA2<br>candidate | 19q13.11 |
| 53 | 22q11.2 Distal microdeletion * | 611867 | Multiple | 22q11.2 |
| 54 | 22q11.21 Microduplication * | 608363 | Multiple<br>TBX1<br>candidate | 22q11.21 |
| 55 | 22q13.3 Microdeletion * | 606232 | Multiple<br>SHANK3<br>candidate | 22q13.3 |
| 56 | Xp11.22-p11.23 Microduplication * | 300801 | Multiple | Xp11.22-<br>p11.23 |
| 57 | Xp11.22-linked intellectual disability/Mental retardation 17, X-linked/Mental retardation 31, X-linked * | 300705 | Multiple<br>HSD17B10<br>candidate<br>HUWE1<br>candidate | Xp11.22 |
| 58 | Xp11.3 Microdeletion | 300578 | Multiple<br>RP2<br>candidate<br>ZNF674<br>candidate | Xp11.23-<br>p11.3 |
| 59 | Xp11.4-p21.2 Contiguous gene deletion | | Multiple<br>IL1RAPL1<br>OTC | Xp11.4-<br>p21.3 |
| 60 | Xq28 Microduplication * | 300815 | Multiple<br>GDI1<br>candidate<br>IKBKG<br>candidate | Xq28 |
| 61 | Adrenal hypoplasia congenita, X-linked (AHC) | 300200 | NR0B1 | Xp21.2 |
| 62 | Agammaglobulinemia, X-linked/Bruton agammaglobulinemia, X-linked | 300755 | BTK | Xq22.1 |
| 63 | Alagille | 118450 | JAG1 | 20p12.2 |
| 64 | Albright hereditary osteodystrophy-like/Brachydactyly with intellectual disability/2q37 Microdeletion | 600430 | HDAC4 | 2q37.3 |
| 65 | Alpha thalassemia with intellectual disability (AT-ID) | 141750 | Multiple<br>HBA1<br>HBA2<br>SOX8<br>candidate | 16p13.3 |
| 66 | Alport, X-linked (ATS) | 301050 | COL4A5 | Xq22.3 |
| 67 | Alport plus diffuse leiomyomatosis, X-linked (ATS-DL) | 301050 | COL4A5<br>COL4A6 | Xq22.3 |
| 68 | Androgen insensitivity, X-linked | 300068 | AR | Xq12 |
| 69 | Angelman | 105830 | UBE3A | 15q11.2 |
| 70 | Aniridia II | 106210 | PAX6 | 11p13 |

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| 71 | ANKRD11 haploinsufficiency/16q24.3 Microdeletion | | ANKRD11 | 16q24.3 |
| 72 | Atrial septal defect (ASD) with atrioventricular conduction defects | 108900 | NKX2-5 | 5q35.1 |
| 73 | Autism with intellectual disability, SHANK2-related | 613436 | SHANK2 | 11q13.3-q13.4 |
| 74 | Bannayan-Riley-Ruvalcaba (BRRS)/PTEN hamartoma tumor | 153480 | PTEN | 10q23.31 |
| 75 | Basal cell nevus/Gorlin-Goltz | 109400 | PTCH1 | 9q22.32 |
| 76 | Beckwith-Wiedemann, IGF2-related * | 130650 | IGF2 | 11p15.5 |
| 77 | Beckwith-Wiedemann, KCNQ1OT1-related | 130650 | KCNQ1OT1 | 11p15.5 |
| 78 | Benign neonatal epilepsy | 121200 | KCNQ2 | 20q13.33 |
| 79 | Beta thalassemia | 141900 | HBB | 11p15.4 |
| 80 | Bilateral frontoparietal polymicrogyria, autosomal recessive (BFPP) | 606854 | GPR56 | 16q21 |
| 81 | Blepharophimosis, ptosis epicanthus inversus (BPE) | 110100 | FOXL2 | 3q22.3 |
| 82 | Boston-type craniosynostosis/Craniosynostosis type 2 * | 604757 | MSX2 | 5q35.2 |
| 83 | Branchio-oto-renal (BOR)/Melnick-Fraser | 113650 | EYA1 | 8q13.3 |
| 84 | Campomelic dysplasia (CMPD) | 114290 | SOX9 | 17q24.3 |
| 85 | Cat-eye * | 115470 | Multiple | 22q11.1-q11.21 |
| 86 | Cerebellar hypoplasia, VLDLR-related, autosomal recessive/Hutterite dysequilibrium, autosomal recessive | 224050 | VLDLR | 9p24.2 |
| 87 | Cerebral cavernous malformations, type 1 (CCM1) | 116860 | KRIT1 | 7q21.2 |
| 88 | Cerebral cavernous malformations, type 2 (CCM2) | 603284 | CCM2 | 7p13 |
| 89 | Cerebral cavernous malformations, type 3 (CCM3) | 603285 | PDCD10 | 3q26.1 |
| 90 | CHARGE | 214800 | CHD7 | 8q12.1-q12.2 |
| 91 | Chondrodysplasia 1, X-linked (CDPX1) | 302950 | ARSE | Xp22.33 |
| 92 | Choroideremia, X-linked | 303100 | CHM | Xq21.2 |
| 93 | Chronic granulomatous disease, X-linked | 306400 | CYBB | Xp11.4 |
| 94 | Cleidocranial dysplasia (CCD) | 119600 | RUNX2 | 6p21.1 |
| 95 | Cohen, autosomal recessive * | 216550 | VPS13B | 8q22.2 |
| 96 | Congenital diaphragmatic hernia (CDH) | 142340 | Multiple CHD2 candidate NR2F2 candidate | 15q26.1-q26.3 |
| 97 | Congenital hemidysplasia with ichthyosiform erythroderma & limb defects (CHILD) | 308050 | NSDHL | Xq28 |
| 98 | Cornelia de Lange/Brachmann-de Lange | 122470 | NIPBL | 5p13.2 |
| 99 | Cowden/PTEN hamartoma tumor | 158350 | PTEN | 10q23.31 |
| 100 | Craniofrontonasal, X-linked | 304110 | EFNB1 | Xq13.1 |

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| 101 | Cri-du-chat | 123450 | Multiple TERT candidate CTNND2 candidate | 5p15.2-p15.33 |
| 102 | Currarino | 176450 | MNX1 | 7q36.3 |
| 103 | Dandy-Walker malformation (DWM) | 220200 | Multiple ZIC1 candidate ZIC4 candidate | 3q23-q25.1 |
| 104 | Deafness 22, autosomal recessive | 607039 | OTOA | 16p12.2 |
| 105 | Diamond-Blackfan anemia 1 | 105650 | RPS19 | 19q13.2 |
| 106 | DiGeorge/Velocardiofacial (VCF)/22q11.21 Microdeletion | 188400/ | TBX1 | 22q11.21 |
| 107 | DiGeorge 2 | 601362 | Multiple | 10p14 |
| 108 | Dopa-responsive dystonia (DRD)/Segawa | 128230 | GCH1 | 14q22.2 |
| 109 | Dosage-sensitive sex reversal, X-linked * | 300018 | NR0B1 | Xp21.2 |
| 110 | Early-onset ataxia with oculomotor apraxia & hypoalbuminemia, autosomal recessive | 208920 | APTX | 9p21.1 |
| 111 | Emery-Dreifuss muscular dystrophy, X-linked (EDMD) | 181350 | EMD | Xq28 |
| 112 | EPHA7 haploinsufficiency/6q16.1 Microdeletion | | EPHA7 | 6q15-q16.1 |
| 113 | Epilepsy & intellectual disability restricted to females, X-linked/Juberg-Hellman, X-linked/Dravet-like, X-linked | 300088 | PCDH19 | Xq22.1 |
| 114 | Epileptic encephalopathy, SCN2A-related * | 613721 | SCN2A | 2q24.3 |
| 115 | Epileptic encephalopathy, STXBP1-related | 612164 | STXBP1 | 9q34.11 |
| 116 | Faciogenital dysplasia, X-linked/Aarskog-Scott, X-linked | 305400 | FGD1 | Xp11.22 |
| 117 | Familial adenomatous polyposis with intellectual disabilities (FAP-ID)/5q22 Microdeletion | 175100 | APC | 5q22.2 |
| 118 | Feingold | 164280 | MYCN | 2p24.3 |
| 119 | FMR1 microdeletion, X-linked | 300624 | FMR1 | Xq27.3 |
| 120 | Focal dermal hypoplasia, X-linked/Goltz, X-linked | 305600 | PORCN | Xp11.23 |
| 121 | GLUT1 deficiency | 606777 | SLC2A1 | 1p34.2 |
| 122 | Glycerol kinase deficiency, X-linked (GKD) | 300474 | GK | Xp21.2 |
| 123 | Greig cephalopolysyndactyly | 175700 | GLI3 | 7p14.1 |
| 124 | Growth hormone insensitivity, autosomal recessive/Laron, autosomal recessive | 262500 | GHR | 5p12-p13.1 |
| 125 | Hemophilia A, X-linked | 306700 | F8 | Xq28 |
| 126 | Hemophilia B, X-linked | 306900 | F9 | Xq27.1 |
| 127 | Hereditary hemorrhagic telangiectasia, type 1/Osler-Rendu-Weber | 187300 | ENG | 9q34.11 |

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| 128 | Hereditary hemorrhagic telangiectasia, type 2 | 600376 | ACVRL1 | 12q13.13 |
| 129 | Hereditary paraganglioma-pheochromocytoma, SDHB-related | 115310 | SDHB | 1p36.13 |
| 130 | Hereditary paraganglioma-pheochromocytoma, SDHD-related | 168000 | SDHD | 11q23.1 |
| 131 | Heterotaxy, X-linked | 306955 | ZIC3 | Xq26.3 |
| 132 | Hirschsprung, EDNRB-related | 600155 | EDNRB | 13q22.3 |
| 133 | Hirschsprung, RET-related | 142623 | RET | 10q11.21 |
| 134 | Holoprosencephaly 1 | 236100 | Multiple TRAPPC10 candidate PWP2 candidate | 21q22.3 |
| 135 | Holoprosencephaly 2 | 157170 | SIX3 | 2p21 |
| 136 | Holoprosencephaly 3 | 142945 | SHH | 7q36.3 |
| 137 | Holoprosencephaly 4 | 142946 | TGIF1 | 18p11.31 |
| 138 | Holoprosencephaly 5 | 609637 | ZIC2 | 13q32.3 |
| 139 | Holoprosencephaly 8 | 609408 | Multiple | 14q13.1-q13.3 |
| 140 | Holt-Oram * | 142900 | TBX5 | 12q24.21 |
| 141 | Hydrocephalus and nephrogenic diabetes insipidus, X-linked | | L1CAM AVPR2 | Xq28 |
| 142 | Hyper-IgE recurrent infection, autosomal recessive | 243700 | DOCK8 | 9p24.3 |
| 143 | Hypohidrotic ectodermal dysplasia, X-linked (XHED) | 305100 | EDA | Xq13.1 |
| 144 | Hypoparathyroidism, sensorineural deafness, renal disease (HDR) | 146255 | GATA3 | 10p14 |
| 145 | Hypospadius 2, X-linked | 300758 | MAMLD1 | Xq28 |
| 146 | Hypotonia-cystinuria, autosomal recessive | 606407 | SLC3A1 PREPL | 2p21 |
| 147 | Idiopathic short stature, X- & Y-linked (ISSX) | 300582 | SHOX | Xp22.33 & Yp11.32 |
| 148 | Infantile hyperinsulinism with enteropathy & deafness, autosomal recessive | 606528 | USH1C ABCC8 | 11p15.1 |
| 149 | Infantile spasms, CDKL5-related, X-linked/Atypical Rett, CDKL5-related, X-linked | 300672 | CDKL5 | Xp22.13 |
| 150 | Infantile spasms, MAGI2-related | | MAGI2 | 7q21.11 |
| 151 | Intellectual disability with cerebellar hypoplasia & distinctive facial appearance, X-linked/Mental retardation 60, X-linked * | 300486 | OPHN1 | Xq12 |
| 152 | Intellectual disability with isolated growth hormone deficiency, X-linked * | 300123 | SOX3 | Xq27.1 |
| 153 | Intellectual disability with language impairment and autistic features | 613670 | FOXP1 | 3p13 |

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| 154 | Intellectual disability with microcephaly & disproportionate pontine and cerebellar hypoplasia, X-linked | 300749 | CASK | Xp11.4 |
| 155 | Intellectual disability with stereotypical movements, epilepsy, and/or cerebral malformations/5q14.3 Microdeletion | 613443 | MEF2C | 5q14.3 |
| 156 | Jacobsen/11q terminal deletion | 147791 | Multiple | 11q23-q25 |
| 157 | Joubert 4, autosomal recessive | 609583 | NPHP1 | 2q13 |
| 158 | Juvenile polyposis (JPS), BMPR1A-related | 174900 | BMPR1A | 10q23.2 |
| 159 | Juvenile polyposis (JPS), SMAD4-related | 174900 | SMAD4 | 18q21.2 |
| 160 | Kallmann 1, X-linked | 308700 | KAL1 | Xp22.31 |
| 161 | Langer-Giedion/Trichorhinophalangeal type II | 150230 | TRPS1<br>EXT1 | 8q23.3<br>8q24.11 |
| 162 | Langer mesomelic dysplasia, X- & Y-linked (LMD) | 249700 | SHOX | Xp22.33 & Yp11.32 |
| 163 | Leri-Weill dyschondrosteosis, X- & Y-linked (LWD) | 127300 | SHOX | Xp22.33 & Yp11.32 |
| 164 | Lesch-Nyhan, X-linked (LNS) | 300322 | HPRT1 | Xq26.2-q26.3 |
| 165 | Li-Fraumeni 1 (LFS) | 151623 | TP53 | 17p13.1 |
| 166 | Lissencephaly, X-linked | 300067 | DCX | Xq23 |
| 167 | Lissencephaly 1 | 607432 | PAFAH1B1 (LIS1) | 17p13.3 |
| 168 | Lowe, X-linked | 309000 | OCRL | Xq25-q26.1 |
| 169 | Lymphoproliferative, X-linked (XLP) | 308240 | SH2D1A | Xq25 |
| 170 | Marfan 1 (MFS1) | 154700 | FBN1 | 15q21.1 |
| 171 | McLeod neuroacanthocytosis, X-linked | 314850 | XK | Xp21.1 |
| 172 | Menkes, X-linked (MNK) | 309400 | ATP7A | Xq21.1 |
| 173 | Mental retardation 1, autosomal dominant (MRD1) | 156200 | MBD5 | 2q23.1 |
| 174 | Mental retardation 5, autosomal dominant | 612621 | SYNGAP1 | 6p21.32 |
| 175 | Mental retardation 6, autosomal recessive | 611092 | GRIK2 | 6q16.3 |
| 176 | Mental retardation 7, autosomal recessive | 611093 | TUSC3 | 8p22 |
| 177 | Mental retardation 9, X-linked/Mental retardation 44, X-linked | 309549 | FTSJ1 | Xp11.23 |
| 178 | Mental retardation 21, X-linked/Mental retardation 34, X-linked | 300143 | IL1RAPL1 | Xp21.2-p21.3 |
| 179 | Mental retardation 94, X-linked * | 300699 | GRIA3 | Xq25 |
| 180 | Mesomelic dysplasia Kantaputra type * | 156232 | Multiple HOXD gene cluster candidate | 2q31.1 |
| 181 | Metachromatic leukodystrophy, autosomal recessive (MLD)/Arylsulfatase A deficiency | 250100 | ARSA | 22q13.33 |
| 182 | Microphthalmia 3 | 206900 | SOX2 | 3q26.33 |

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| 183 | Microphthalmia 7 with linear skin defects, X-linked | 309801 | HCCS | Xp22.2 |
| 184 | Miller-Dieker | 247200 | PAFAH1B1 (LIS1) | 17p13.3 |
| 185 | Mohr-Tranebjaerg, X-linked/Deafness-dystonia-optic neuropathy, X-linked (DDON) | 304700 | TIMM8A | Xq22.1 |
| 186 | Mowat-Wilson * | 235730 | ZEB2 | 2q22.3 |
| 187 | Myoclonus dystonia | 159900 | SGCE | 7q21.3 |
| 188 | Myotubular myopathy 1, X-linked | 310400 | MTM1 | Xq28 |
| 189 | Nablus mask-like facial | 608156 | Multiple | 8q22.1 |
| 190 | Nail-patella (NPS) | 161200 | LMX1B | 9q33.3 |
| 191 | Neonatal hemolytic anemia associated with hemoglobin beta cluster/Epsilon-gamma-delta-beta-thalassemia | 141900 | Hemoglobin beta gene cluster | 11p15.4 |
| 192 | Nephronophthisis 1, autosomal recessive | 256100 | NPHP1 | 2q13 |
| 193 | Nephropathic cystinosis, autosomal recessive | 219800 | CTNS | 17p13.2 |
| 194 | Neurofibromatosis 1 with intellectual disabilities (NF1-ID) * | 613675 | NF1 | 17q11.2 |
| 195 | Neurofibromatosis 2 (NF2) | 101000 | NF2 | 22q12.2 |
| 196 | Neurosensory deafness, autosomal recessive (DFNB1) | 220290 | GJB6 | 13q12.11 |
| 197 | NFIA haploinsufficiency/1p31.3 Microdeletion | 613735 | NFIA | 1p31.3 |
| 198 | Norrie, X-linked * | 310600 | NDP | Xp11.3 |
| 199 | Oculocutaneous albinism 2, autosomal recessive (OCA2) * | 203200 | OCA2 | 15q12-q13.1 |
| 200 | Oculofaciocardiodental, X-linked/Microphthalmia 2, X-linked | 300166 | BCOR | Xp11.4 |
| 201 | Okihiro/Duane radial ray (DRRS) | 607323 | SALL4 | 20q13.2 |
| 202 | Opitz GBBB, X-linked | 300000 | MID1 | Xp22.2 |
| 203 | Ornithine transcarbamylase deficiency, X-linked (OTC) | 311250 | OTC | Xp11.4 |
| 204 | Osteopathia striata with cranial sclerosis, X-linked | 300373 | FAM123B | Xq11.2 |
| 205 | Oto-dental | 166750 | FGF3 | 11q13.3 |
| 206 | Pallister-Killian * | 601803 | Multiple | 12p |
| 207 | Paris-Trousseau thrombocytopenia | 188025 | FLI1 | 11q24.3 |
| 208 | Pelizaeus-Merzbacher, X-linked * | 312080 | PLP1 | Xq22.2 |
| 209 | Peutz-Jeghers (PJS) | 175200 | STK11 | 19p13.3 |
| 210 | Pitt-Hopkins | 610954 | TCF4 | 18q21.2 |
| 211 | Pitt-Hopkins-like 1, autosomal recessive/Cortical dysplasia-focal epilepsy, autosomal recessive (CDFE) | 610042 | CNTNAP2 | 7q35-q36.1 |
| 212 | Polycystic kidney disease 1 (PKD1) | 173900 | PKD1 | 16p13.3 |

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| 213 | Potocki-Lupski/17p11.2 Microduplication * | 610883 | Multiple RAI1 candidate | 17p11.2 |
| 214 | Potocki-Shaffer | 601224 | EXT2 ALX4 | 11p11.2 |
| 215 | Prader-Willi (PWS) | 176270 | SNORD116 cluster candidate | 15q11.2 |
| 216 | Prader-Willi-like phenotype | 176270 | Multiple SIM1 candidate | 6q16.1-q16.3 |
| 217 | Renal cysts and diabetes (RCAD) | 137920 | HNF1B | 17q12 |
| 218 | Retinoblastoma with intellectual disability | 180200 | RB1 | 13q14.2 |
| 219 | Rett, congenital variant | 613454 | FOXG1 | 14q12 |
| 220 | Rieger 1 (RIEG1)/Axenfeld-Rieger | 180500 | PITX2 | 4q25 |
| 221 | Rubinstein-Taybi (RTS) | 180849 | CREBBP | 16p13.3 |
| 222 | Saethre-Chotzen | 101400 | TWIST1 | 7p21.1 |
| 223 | Severe myoclonic epilepsy of infancy (SMEI)/Dravet | 607208 | SCN1A | 2q24.3 |
| 224 | Sex reversal, autosomal dominant 2 (SRA2) | 154230 | Multiple DMRT1 candidate | 9p24.3 |
| 225 | Short stature, pituitary and cerebellar defects, small sella turcica/Pituitary hormone deficiency, combined, 4 | 262700 | LHX4 | 1q25.2 |
| 226 | Siderius type intellectual disability, X-linked | 300263 | PHF8 | Xp11.22 |
| 227 | Simpson-Golabi-Behmel, X-linked (SGBS) | 312870 | GPC3 | Xq26.2 |
| 228 | Smith-Magenis (SMS) | 182290 | RAI1 | 17p11.2 |
| 229 | Sotos | 117550 | NSD1 | 5q35.2-q35.3 |
| 230 | Speech & language disorder 1 | 602081 | FOXP2 | 7q31.1 |
| 231 | Split-hand/foot malformation 1 (SHFM1)/Ectrodactyly | 183600 | SHFM1 | 7q21.3 |
| 232 | Split-hand/foot malformation 3 (SHFM3)/Ectrodactyly * | 246560 | FBXW4 | 10q24.32 |
| 233 | Split-hand/foot malformation 5 (SHFM5)/Ectrodactyly | 606708 | Multiple EVX2 candidate HOXD gene cluster candidate | 2q31.1 |
| 234 | Steroid sulfatase deficiency, X-linked/Ichthyosis, X-linked | 308100 | STS | Xp22.31 |
| 235 | Synpolydactyly/Syndactyly II | 186000 | HOXD gene cluster | 2q31.1 |
| 236 | Toe syndactyly, telecanthus, anogenital & renal malformations, X-linked (STAR) | 300707 | FAM58A | Xq28 |
| 237 | Townes-Brocks 1 | 107480 | SALL1 | 16q12.1 |

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| 238 | Trichorhinophalangeal 1 | 190350 | TRPS1 | 8q23.3 |
| 239 | Tuberous sclerosis 1 (TSC1) | 191100 | TSC1 | 9q34.13 |
| 240 | Tuberous sclerosis 2 (TSC2) | 613254 | TSC2 | 16p13.3 |
| 241 | Ulnar-mammary | 181450 | TBX3 | 12q24.21 |
| 242 | Usher IIC, autosomal recessive | 605472 | GPR98 | 5q14.3 |
| 243 | Van der Woude | 119300 | IRF6 | 1q32.2 |
| 244 | von Hippel-Lindau | 193300 | VHL | 3p25.3 |
| 245 | Waardenburg I | 193500 | PAX3 | 2q36.1 |
| 246 | Waardenburg IIA | 193510 | MITF | 3p13-p14.1 |
| 247 | Waardenburg IIE | 611584 | SOX10 | 22q13.1 |
| 248 | Waardenburg IVA/Waardenburg-Shah, autosomal recessive | 277580 | EDNRB | 13q22.3 |
| 249 | Waardenburg-Shah, neurologic variant/Peripheral demyelinating neuropathy, central dysmyelination, Waardenburg, and Hirschsprung (PCWH)/Waardenburg IVC | 609136/ 613266 | SOX10 | 22q13.1 |
| 250 | Walker-Warburg, LARGE-related, autosomal recessive/Muscular dystrophy-dystroglycanopathy A1, autosomal recessive | 236670 | LARGE | 22q12.3 |
| 251 | Williams-Beuren (WBS) | 194050 | ELN | 7q11.23 |
| 252 | Wilms Tumor 1 (WT1) | 194070 | WT1 | 11p13 |
| 253 | Wilms tumor-aniridia-genital anomalies-retardation (WAGR) | 194072 | PAX6 WT1 | 11p13 |
| 254 | Wolf-Hirschhorn | 194190 | Multiple | 4p16.3 |
| 255 | XX male/SRY dosage abnormalities | 278850 | SRY | Yp11.31 |
| 256 | XY gonadal dysgenesis/SRY dosage abnormalities | 400044 | SRY | Yp11.31 |
| 257 | XY sex-reversal, +/- adrenal failure/46,XY sex reversal 3/Adrenocortical insufficiency | 612965 | NR5A1 | 9q33.3 |
| 258 | All 43 unique pericentromeric regions | | Multiple | 43 sites |
| 259 | All 41 unique subtelomeric regions | | Multiple | 41 sites |
| 260 | Aneuploidy for 24 chromosomes | | Multiple | 24 chromosomes |