



One Baylor Plaza, NAB 2015, Houston, Texas 77030
713-798-6555 • 1-800-411-GENE (4363) • genetictest@bcm.edu
www.bcmgeneticlabs.org

Baylor 7.0 105K OLIGO array Disorders

Disorders	Gene(s)/Locus	Location
1q21.2 deletion/duplication (clinical signifi. unclear)		1q21.2
1q41q42 deletion	<i>DISP1</i>	1q41
2p15p16.1 microdeletion		2p15p16.1
2q22.3 deletion syndrome		2q22.3
3q29 Microdeletion syndrome	Multiple	3q29
5q21.1-q31.2 deletion syndrome		5q21.1q31.2
7q11.23 duplication;reciprocal Williams-Beuren region		7q11.23
8p23p22 deletion/duplication syndrome		8p22
9p23 deletion syndrome/Trigonocephaly		9p23
9q34.3 deletion syndrome	<i>EHMT1</i>	9q34.3
10q22q23 deletion syndrome	<i>GRID1, NRG3</i>	10q23.1q23.2
12q24.21 duplication syndrome	Multiple	12q24.21
14q11.2 syndrome	<i>CHD8, SUPT16H</i>	14q11.2
14q12 deletion syndrome	<i>FOXP1B</i>	14q12
15q11 BP1-BP2 interval deletion & duplication		15q11.2
15q11.2-q12 duplication; reciprocal PWS/AS region		15q11.2q12
15q13.3 microdeletion	<i>CHRNA7</i>	15q13.3
15q21 deletion syndrome		15q21.1q31.2
15q24 microdeletion		15q24
15q26.3 deletion w/ severe intrauterine growth restriction (IUGR)		15q26.3
16p11.2p12 deletion		16p11.2p12
17q21.31 deletion syndrome	<i>MAPT, CRHR1</i>	17q21.31
17q21.31 duplication syndrome; reciprocal 17q21.31 deletion syndrome	<i>MAPT, CRHR1</i>	17q21.31
22q11.2 duplication;reciprocal DGS1 region		22q11.2
22q13.3 Microdeletion syndrome	<i>SHANK3</i>	22q13.33
Aarskog-Scott	<i>FGD1</i>	Xp11.22
ADLD adult-onset, Autosomal Dominant Leukodystrophy	<i>LMNB1</i>	5q23.2
Adrenoleukodystrophy	<i>ABCD1</i>	Xq28
Alagille Syndrome (AGS)	<i>JAG1</i>	20p12.2
Alpha Thalassemia X-Linked Metal Retardation syndrome	<i>ATRX, SOX8</i>	16p13.3
Alport syndrome, X-linked (ATS)	<i>COL4A5</i>	Xq22.3

Androgen insensitivity syndrome (AIS)	<i>AR</i>	Xq12
Angelman syndrome (AS)	<i>UBE3A</i>	15q11.2q12
Angelman syndrome (AS) imprinting center		15q11.2q12
Aniridia II & WAGR	<i>PAX6</i>	11p13
ATR-X, and others	<i>ATRX</i>	Xq21.1
Autistic features, see 15q11q13 duplication		15q11q13
Autistic features, X-linked, susceptibility to, 2	<i>NLGN4</i>	Xp22.31p22.32
AZFa & AZFb		Yq11.22
Bannayan-Riley-Ruvalcaba syndrome (BRRS)	<i>PTEN</i>	10q23.31
Basal cell nevus syndrome (BCNS)	<i>PTCH</i>	9q22.32
Beckwith-Wiedemann syndrome (BWS)	<i>IGF2</i>	11p15.5
Bipolar disorder with 8q21.13 deletion	<i>IMPA1</i>	8q21.13
Blepharophimosis, ptosis, epicanthus inversus (BPE)	<i>FOXL2</i>	3q22.3
Börjeson-Forssman-Lehmann	<i>PHF6</i>	Xq26.2
BPNH, OPD	<i>FLNA</i>	Xq28
Brachydactyly	<i>GPC5, GPC6</i>	13q31.3
Brachydactyly, type C (BDC)	<i>GDF5</i>	20q11.2
Brachydactyly-Mental Retardation syndrome/Albright Hereditary Osteodystrophy-like syndrome	Multiple	2q37.3
Branchioototic syndrome-3	<i>SIX1</i>	14q23.1
Branchiootorenal dysplasia syndrome (BOR)/Melnick-Frazer syndrome	<i>EYA1</i>	8q13.3
Bruton agammaglobulinemia; see hypogammaglobulinemia	<i>BTK</i>	Xq22.1
Buschke-Ollendorff syndrome/Osteopoikilosis, SS and MR	<i>LEMD3</i>	12q14.3
Campomelic dysplasia (CMPD)	<i>SOX9</i>	17q24.3
Cat-eye syndrome (CES);inv dup(22) (q11.2)	Multiple	22q11.21
Cerebellar hypoplasia	<i>OPHN1</i>	Xq12
Charcot-Marie-Tooth disease type 1A (CMT1A)	<i>PMP22</i>	17p12
Charcot-Marie-Tooth disease type 1D (CMT1D), autosomal dominant	<i>EGR2</i>	5q31.2
CHARGE syndrome	<i>CHD7</i>	8q12.2
Chondrodyplasia	<i>COL2A1</i>	12q13.11q13.2
Choroiderma	<i>CHM</i>	Xq21.2
Chromosome 10q deletion syndrome		10q26
Chromosome 18p deletion syndrome		18p11.31
Chromosome 18q deletion syndrome		18q23
Chronic Pancreatitis	<i>SPINK1</i>	5q32
Cleft lip/Cleft palate	<i>PHF8</i>	Xp11.22
Cleft palate, isolated (CPI)	<i>SATB2</i>	2q32
Cleidocranial dysplasia (CCD)	<i>RUNX2</i>	6p21.1
Coffin-Lowry	<i>RPS6KA3</i>	Xp22.12
Congenital adrenal hyperplasia (CAH)		6p21.32

Congenital adrenal hypoplasia (ACH)	<i>NROB1</i>	Xp21.2
Cornelia de Lange syndrome (CDLS)	<i>NIPBL</i>	5p13.2
Cornelia de Lange syndrome 2 (CDLS2)	<i>SMC1L1</i>	Xp11.22p11.21
Cowden disease (CD)	<i>PTEN</i>	10q23.31
Craniosynostosis with 11p15.2 disruption	<i>SOX6</i>	11p15.2
Cri-du-Chat syndrome	Multiple	5p15.2p13.3
Cutis laxa, X-linked/Occipital horn syndrome	<i>ATP7A</i>	Xq12q13
Cystinosis, Nephropathic (CTNS)	<i>CTNS</i>	17p13.2p13.3
Cystinuria with mitochondrial disaeze	<i>SLC3A1</i>	2p21
Dandy-Walker syndrome (DWS)	<i>ZIC1, ZIC4</i>	3q24
Danon disease	<i>LAMP2</i>	Xq24
Dent disease	<i>CLCN5</i>	Xp11.22
Diabetes Mellitus, transient neonatal, 1	<i>ZAC</i>	6q24.2
Diaphragmatic hernia, congenital (HCD/DIH1)	<i>NR2F2/CHD2</i>	15q26.1q26.2
DiGeorge syndrome 1 (DGS1)/Velocardiofacial (VCFS)	<i>TBX1</i>	22q11.2
DiGeorge syndrome 2 (DGS2)	Multiple	10p14
Down syndrome critical region 1 (DSCR)	Multiple	21q22.12
Duchenne/Becker muscular dystrophy (DMD/BMD)	<i>DMD</i>	Xp21.1p21.2
Dyggve-Melchior-Clausen syndrome (DMC)	<i>DYM</i>	18q21.1
Dyskeratosis congenita	<i>DKC1</i>	Xq28
Epilepsy	<i>ATP6AP2</i>	Xp11.4
Epilepsy with febrile seizure plus (GEFS+), generalized	<i>SCN1A</i>	2q24.3
Epilepsy, macrocephaly	<i>SYN1</i>	Xp11.23
Fabry disease	<i>GLA</i>	Xq22.1
Feingold syndrome	<i>MYCN</i>	2p24.3
Focal Dermal Hypoplasia/Goltz syndrome	<i>PORCN</i>	Xp11.23
Forebrain defects; TDGF1	<i>CRIPTO</i>	3p21.31
Fragile-X mental retardation syndrome (FMR1)	<i>FMR1</i>	Xq27.3
Glycerol kinase deficiency (GKD)	<i>GK</i>	Xp21.2
Golabi-Ito-Hall syndrome	<i>PQBP1</i>	Xp11.23
Greig cephalo-polysyndactyly syndrome (GCPS)	<i>GLI3</i>	7p14.1
Growth hormone deficiency	<i>SOX3</i>	Xq27.1
HADH2 deficiency	<i>HADH2</i>	Xp11.22
Hearing Loss, Hereditary	<i>GJB2 (Connexin 26)</i>	13q12.11
Hemophilia A	<i>F8</i>	Xq28
Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)	<i>PMP22</i>	17p11.2
Hereditary pancreatitis	<i>PRSS1</i>	7q34
Heterotaxy 2	<i>CFC1</i>	2q21.1
Hirschsprung	<i>RET</i>	10q11.21
Hirschsprung	<i>EDNRB</i>	13q22.3

Holoprosencephaly 1 (HPE1)	<i>TMEM1</i>	21q22.3
Holoprosencephaly 2 (HPE2)	<i>SIX3</i>	2p21
Holoprosencephaly 3 (HPE3)	<i>SHH</i>	7q36.3
Holoprosencephaly 4 (HPE4)	<i>TGIF</i>	18p11.31
Holoprosencephaly 5 (HPE5)	<i>ZIC2</i>	13q32.3
Holoprosencephaly 6 (HPE6)		2q37.1q37.3
Holoprosencephaly 7 (HPE7)	<i>PTCH</i>	9q22.32
Holoprosencephaly 9 (HPE9)/ Pituitary anomalies with holoprosencephaly	<i>GLI2</i>	2q14.2
Holt-Oram syndrome	<i>TBX5</i>	12q24.21
Hunter	<i>IDS</i>	Xq28
Hypertension with CHD	<i>BMPR2</i>	2q33.1
Hypogammaglobulinemia, isolated growth hormone deficiency	<i>BTK</i>	Xq22.1
Hypoparathyroidism, sensorineural deafness, renal (HDR)	<i>GATA3</i>	10p14
Incontinentia pigmenti (IP)	<i>IKBKG (NEMO)</i>	Xq28
Infantile hyperinsulinism, enteropathy, and deafness	<i>USH1C, ABCC8</i>	11p15.1
Infantile spasm syndrome, X-linked (ISSX)	<i>ARX</i>	Xp21.3
Infantile spasm syndrome, X-linked (ISSX)	<i>CDKL5</i>	Xp21.3
Iridogoniodysgenesis anomaly, Axenfeld-Rieger syndrome	<i>FKHL7 (FOXC1)</i>	6p25.3
Jacobson syndrome	Multiple	11q24q25
Joubert 4	<i>NPHP1</i>	2q13
Kallman syndrome 1 (KAL1)	<i>KAL1</i>	Xp22.31
Langer-Giedion syndrome (LGS)	<i>EXT1, TRPS1</i>	8q23.3q24.11
Lenz microphthalmia	<i>BCOR</i>	Xp11.4
Leri-Weill dyschondrosteosis	<i>SHOX, SHOXY</i>	Xp22.33/Yp11.32
Lesch-Nyhan syndrome (LNS)	<i>HPRT1</i>	Xq26.2
Leukodystrophy with 11q14.2-q14.3		11q14.2q22.3
Lissencephaly, X-linked (LISX)	<i>DCX</i>	Xq22.3q23
Loeys-Dietz syndrome (LDS)	<i>TGFBR1</i>	9q22.33
Loeys-Dietz syndrome (LDS)	<i>TGFBR2</i>	3p24.1
Lowe syndrome	<i>OCRL</i>	Xq25
Marfan syndrome (MFS)	<i>FBN1</i>	15q21.1
Marfan syndrome, Type II (MFS2)	<i>TGFBR2</i>	3p22
MASA, CRASH, HSAS syndromes	<i>LICAM</i>	Xq28
MECP2 male duplication syndrome	<i>MECP2</i>	Xq28
Menkes disease (MNK)	<i>ATP7A</i>	Xq12q13
Mental retardation X-linked growth horm. Def (MRGH)	<i>SOX3</i>	Xq27.1
Mental Retardation, X-linked 21 (MRX21)	<i>IL1RAPL1</i>	Xp22.1p21.3
Micophthalmia with linear skin defects	<i>HCCS</i>	Xp22.2
Microcephaly, congenital heart disease	<i>NKX2-5</i>	5q35.1
Microphthalmia syn. 6, pituitary hypoplasia (MCOPS6)	<i>SIX6</i>	14q22.2q22.3

Microphthalmia, syn. 3 (MCOPS3)	SOX2	3q26.33
Microphthalmia, syndromic 7 (MCOPS7)/MLS/MIDAS	Multiple	Xp22.2
Miller-Dieker Lissencephaly syndrome (MDLS)	<i>LIS1</i> and/or <i>YWHAE</i>	17p13.3
Mohr-Tranebjærg	<i>TIMM8A</i>	Xq22.1
Monoamine oxidase-A deficiency	<i>MAOA</i>	Xp11.3
Monosomy 1p36 syndrome	Multiple	1p36
Monosomy 9p syndrome		9p22.3p23
Mowat-Wilson syndrome	<i>ZFHX1B</i>	2q22.3
Mucopolysaccharidosis, type II (MPS2)	<i>IDS</i>	Xq28
Nail-patella syndrome (NPS)	<i>LMX1B</i>	9q33.3
Nance-Horan	<i>NHS</i>	Xp22.13
Nebulette	<i>NEBL</i>	10p12.31
Nephronophthisis 1 (NPH1)/2q13 deletion	<i>NPHP1</i>	2q13
Neurofibromatosis 1 (NF1)	<i>NF1</i>	17q11.2
Neurofibromatosis 2 (NF2)	<i>NF2</i>	22q12.2
Noonan syndrome	<i>SOS1</i>	2p22.1
Noonan syndrome	<i>RAF1</i>	3p25.1
Noonan syndrome	<i>PTPN11</i>	12q24.13
Norrie disease	<i>NDP</i>	Xp11.3
Opitz syndrome	<i>MID1</i>	Xp22.2
Ornithine transcarbamylase deficiency	<i>OTC</i>	Xp11.4
Orofaciodigital syndrome (OFD1)	<i>OFD1</i>	Xp22.2
Osteogenesis imperfecta congenita	<i>COL1A1</i>	17q21.33
Osteogenesis imperfecta congenita	<i>COL1A2</i>	7q21.3
Pallister-Killian	Multiple	12p
Pelizaeus-Merzbacher disease (PMD)	<i>PLP1</i>	Xq22.2
Phosphoglycerate kinase deficiency	<i>PGK1</i>	Xq21.1
Pitt-Hopkins	<i>TCF4</i>	18q21.2
Polycystic kidney disease 1	<i>PKD1, TSC2</i>	16p13.3
Potocki-Lupski syndrome/17p11.2 duplication; reciprocal SMS region	<i>RAI1</i>	17p11.2
Potocki-Shaffer syndrome	<i>EXT2, ALX4</i>	11p11.2
Prader-Willi syndrome (PWS)	<i>SNRPN, NECDIN, snoRNAs</i>	15q11.2q12
Prader-Willi syndrome (PWS) imprinting center		15q11.2q12
Prader-Willi-like phenotype; see Obesity deletion syndrome	<i>SIM1</i>	6q16.3
PRPP synthetase hyperactivity	<i>PRPS1</i>	Xq22.3
Pyruvate dehydrogenase deficiency	<i>PDHA1</i>	Xp22.12
Renal cysts and diabetes (RCAD)	<i>TCF2</i>	17q12
Renpenning, Sutherland-Haan, Hamel	<i>PQBP1</i>	Xp11.23
Retinoblastoma (RB1)	<i>RB1</i>	13q14.2
Retinoschisis, X-linked juvinille (RSI)	<i>XLRS1</i>	Xp22.13

Rett syndrome (RTT); MECP2 deletion	<i>MECP2</i>	Xq28
Rett-like, infantile spasms	<i>CDKL5</i>	Xp22.13
Rieger syndrome, type 1 (RIEG1)	<i>PITX2</i>	4q25
Rubinstein-Taybi syndrome (RSTS)	<i>CREBBP</i>	16p13.3
Sacral/anorectal malformation syndrome		6q25.3
Saethre-Chotzen syndrome (SCS)	<i>TWIST1</i>	7p21.1
Schizophrenia & epilepsy	<i>CNTNAP2</i>	7q35
Sensorineural deafness and male infertility		15q15.3
Severe myoclonic epilepsy of infancy (SMEI)/Dravet syndrome	<i>SCN1A</i>	2q24.3
Sex Determining Region/Sex Reversal	<i>SRY</i>	Yp11.31
Sex reversal XY +/- adrenal failure	<i>NR5A1 (SF1)</i>	9q33
Sex reversal, autosomal dominant 2 (SRA2)		9p24.3
Sex reversal, dosage-sensitive (DSS)	<i>NROB1</i>	Xp21.2
Sex reversal, SOX8 related	<i>SOX8</i>	16p13.3
Short stature, idiopathic, X-linked; see Leri Weill	<i>SHOX, SHOXY</i>	Xp22.33/Yp11.32
Short stature, pituitary and cerebellar defects, & small sella turcica	<i>LHX4</i>	1q25.2
Simpson-Golabi Behmel	<i>GPC3</i>	Xq26.2
Smith-Magenis syndrome (SMS)	<i>RAI1</i>	17p11.2
Sotos syndrome (SOS)	<i>NSD1</i>	5q35.3
Speech delay with FOXP2 deletion	<i>FOXP2</i>	7q31.1
Spermine synthase deficiency	<i>SMS</i>	Xp22.11
Split-Hand/Foot Malformation 5 (SHFM5)	<i>DLX1, DLX2</i>	2q31
Split-hand/foot malformation-1 (SHFM1)		7q31.1
Split-hand/foot malformation-3 (SHFM3)		10p14
Split-hand/foot malformation-4 (SHFM4)	<i>TP73L</i>	3q28
Steroid Sulfatase deficiency (STS)	<i>STS</i>	Xp22.31
Stickler syndrome, type II (STL2)	<i>COL11A1</i>	1p21.1
Stocco dos Santos X-Linked Mental Retardation	<i>SHROOM4 (KIAA1202)</i>	Xp11.22p11.21
Synpolydactyly (SPD1)	<i>HOXD13</i>	2q31q32
T3 transporter deficiency	<i>SLC16A12</i>	Xq13.2
Tetralogy of Fallot	<i>ZFPM2/FOG2</i>	8q23.1
Thrombocytopenia-Absent Radius syndrome		1q21.1
Timothy syndrome (TS)	<i>CACNA1C</i>	12p13.33
Townes-Brocks syndrome (TBS)	<i>SALL1</i>	16q12.1
Treacher Collins-Franceschetti syndrome (TCOF)	<i>TCOF1</i>	5q32
Trichorhinophalangeal syndrome 1 (TRPS1)	<i>TRPS1</i>	8q23.3
Tuberous Sclerosis 1 (TSC1)	<i>TSC1</i>	9q34
Tuberous Sclerosis 2 (TSC2)	<i>TSC2</i>	16p13.3
Ulnar-mammary syndrome	<i>TBX3</i>	12q24.21
Van der Woude syndrome (VWS)	<i>IRF6</i>	1p32.2

Vascular endothelial growth factor (VEGF)	<i>VEGF</i>	6p21.1
Waardenburg syndrome, Type IIA (WS2A)	<i>MITF</i>	3p13
West, Proud, XLAG, Partington syndromes	<i>ARX</i>	Xp21.3
Williams-Beuren syndrome (WBS)	<i>ELN, LIMK1, NCF1</i>	7q11.23
Wilms tumor 1 (WT1) & WAGR	<i>PAX6, WT2</i>	11p13
Wolf-Hirschhorn syndrome	Multiple	4p16.3
XIST deficiency;usually ring X chromosome	<i>XIST</i>	Xq13.2
X-linked chronic granulomatous disease	<i>CYBB</i>	Xp11.4
X-Linked Cornelia De Lange syndrome	<i>SMC1L1</i>	Xp11.22
X-linked dyskeratosis congenita	<i>DKC1</i>	Xq28
X-linked heterotaxy (HTX)	<i>ZIC3</i>	Xq26.3
X-linked hypohidrotic ectodermal dysplasia	<i>EDA</i>	Xq13.1
X-linked hypophosphatemic rickets	<i>PHEX</i>	Xp22.11
X-linked lymphoproliferative syndrome (XLP)	<i>SH2D1A</i>	Xq25
X-Linked Mental Retardation (XLMR)	<i>CASK</i>	Xp11.4
X-Linked Mental Retardation (XLMR)	<i>NXF5</i>	Xq22.1
X-Linked Mental Retardation (XLMR)	<i>VCX3A</i>	Xp22.31
X-Linked Mental Retardation (XLMR)	<i>AP1S2</i>	Xp22.2
X-Linked Mental Retardation (XLMR)	<i>IL1RAPL1</i>	Xp21.2p21.3
X-Linked Mental Retardation (XLMR)	<i>TM4SF2 (TSPAN7)</i>	Xp11.4
X-Linked Mental Retardation (XLMR)	<i>CASK</i>	Xp11.4
X-Linked Mental Retardation (XLMR)	<i>ZNF674</i>	Xp11.3
X-Linked Mental Retardation (XLMR)	<i>ZNF41</i>	Xp11.3
X-Linked Mental Retardation (XLMR)	<i>ELK1</i>	Xp11.23
X-Linked Mental Retardation (XLMR)	<i>ZNF81</i>	Xp11.23
X-Linked Mental Retardation (XLMR)	<i>SLC38A5</i>	Xp11.23
X-Linked Mental Retardation (XLMR)	<i>FTSJ1</i>	Xp11.23
X-Linked Mental Retardation (XLMR)	<i>JARID1C</i>	Xp11.22
X-Linked Mental Retardation (XLMR)	<i>KLF8</i>	Xp11.21
X-Linked Mental Retardation (XLMR)	<i>ARHGF9</i>	Xq11.1
X-Linked Mental Retardation (XLMR)	<i>DLG3</i>	Xq13.1
X-Linked Mental Retardation (XLMR)	<i>ZNF261 (ZMYM3)</i>	Xq13.3
X-Linked Mental Retardation (XLMR)	<i>KIAA2022</i>	Xq13.3
X-Linked Mental Retardation (XLMR)	<i>ZDHHC15</i>	Xq13.3
X-Linked Mental Retardation (XLMR)	<i>ACSL4</i>	Xq22.3
X-Linked Mental Retardation (XLMR)	<i>PAK3</i>	Xq22.3
X-Linked Mental Retardation (XLMR)	<i>AGTR2</i>	Xq23
X-Linked Mental Retardation (XLMR)	<i>CUL4B</i>	Xq24
X-Linked Mental Retardation (XLMR)	<i>GRIA3</i>	Xq25
X-Linked Mental Retardation (XLMR)	<i>ZDHHC9</i>	Xq25

X-Linked Mental Retardation (XLMR)	<i>ARHGEF6</i>	Xq28
X-Linked Mental Retardation (XLMR)	<i>FMR2 (AFF2)</i>	Xq28
X-Linked Mental Retardation (XLMR)	<i>SLC6A8</i>	Xq28
X-Linked Mental Retardation (XLMR)	<i>GDI1</i>	Xq28
X-Linked Mental Retardation, 9 (MRX9)	<i>MITF</i>	3p13
X-linked nephrogenic diabetes insipidus	<i>AVPR2</i>	Xq28
X-linked recessive chondrodysplasia	<i>CDPX1 (ARSE)</i>	Xp22.33
XLMR retinitis pigmentosa		Xp11.3
Xp11.3 deletion with mental retardation	<i>ZNF674</i>	Xp11.3
Xp22.31 duplication; reciprocal STS	<i>STS</i>	Xp22.31
All 41 unique subtelomeric regions	multiple	41 sites
All 43 unique pericentromeric regions	multiple	43 sites
Aneuploidy for 24 chromosomes	multiple	24 chromosomes