



MEDICAL GENETICS LABORATORIES

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Baylor 6.5 44K OLIGO array Disorders

Disorders	Gene(s)/Locus	Location
Monosomy 1p36 syndrome	Multiple	1p36
1q21.2 deletion/duplication (clinical signifi. Unclear)		1q21.2
1q41q42 deletion syndrome		1q41q42
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
8p23 (or8p22) deletion/duplication syndrome		8p22
9p23 deletion syndrome/Trigonocephaly		9p23
9q34.3 deletion syndrome	EHMT1	9q34.3
10q22q23 deletion syndrome		10q22q23
12q24.21 duplication syndrome	Multiple	12q24.21
14q11.2 deletion syndrome		14q11.2
14q12 deletion syndrome	FOXP1B	14q12
15q11 BP1-BP2 interval deletion & duplication		15q11
15q11.2-q12 duplication; reciprocal PWS/AS region		15q11q13
15q13.3 deletion syndrome		15q13.3
15q21 deletion syndrome		15q21.1-q31.2
15q24 microdeletions		15q24
15q26.3 deletion w/ severe intrauterine growth restriction (IUGR)		15q26.3
16p11.2p12.1 deletion/duplication syndrome		16p11.2p12.1
17q21.31 deletion syndrome	MAPT, CRHR1	17q21.31
22q11.2 duplication;reciprocal DGS1 region		22q11.2
22q13.3 Microdeletion syndrome	SHANK3	22q13.33
Xp22.31 duplication;reciprocal STS		Xp22.31
Xp11.3 deletion with mental retardation	ZNF674	Xp11.3
Alagille Syndrome (AGS)	JAG1	20p12.2
Alpha Thalassemia X-Linked Mental Retardation Syndrome	ATRX, SOX8	16p13.3

Alport syndrome, X-linked (ATS)	COL4A5	Xq22.3
Androgen insensitivity syndrome (AIS)	AR	Xq12
Angelman Syndrome (AS)	UBE3A	15q11.2-q12
Aniridia II & WAGR	PAX6	11p13
Autistic features, see 15q11q13 duplication		15q11q13
Autistic features, 16p11.2 deletion/duplication syndrome		16p11.2
Autistic features, X-linked, susceptibility to, 2	NLGN4	Xp22.32
AZFa & AZFb		11q11.22
Bannayan-Riley-Ruvalcaba syndrome (BRRS)	PTEN	10q23.31
Basal cell nevus syndrome (BCNS)	PTCH	9q22.32
Beckwith-Weidemann syndrome (BWS)	IGF2	11p15.5
Bipolar disorder with 8q21.13 deletion	IMPA1	8q21.13
Blepharophimosis, ptosis, epicanthus inversus (BPE)	FOXL2	3q22.3
Brachydactyly	GPC5, GPC6	13q31.3
Brachydactyly, type C (BDC)	GDF5	20q11.2
Brachydactyly-Mental Retardation Syndrome/Albright Hereditary Osteodystrophy-like Syndrome	Multiple	2q37.3
Branchiootorenal dysplasia syndrome (BOR)/Melnick-Frazer Syndrome	EYA1	8q13.3
Bruton agammaglobulinemia; see hypogammaglobulinemia	BTK	Xq22.1
Buschke-Ollendorff Syndrome/Osteopoikilosis, SS and MR	LEMD3	12q14.3
Campomelic dysplasia (CMPD)	SOX9	17q24.3
Cat-eye syndrome (CES);inv dup(22) (q11.2)	Multiple	22q11.21
Charcot-Marie-Tooth disease type 1A (CMT1A)	PMP22	17p12
CHARGE syndrome	CHD7	8q12.2
Chondrodysplasias	COL2A1	12q13.11-q13.2
Chromosome 18q deletion syndrome		18q23
Chromosome 18p deletion syndrome		18p11.31
Chromosome 10q deletion syndrome		10q26
Cleft palate, isolated (CPI)	SATB2	2q32
Cleidocranial dysplasia (CCD)	RUNX2	6p21.1
Congenital adrenal hyperplasia (CAH)		6p21.32
Congenital adrenal hypoplasia (ACH)	NROB1	Xp21.2
Cornelia de Lang syndrome (CDLS)	NIPBL	5p13.2
Cornelia de Lang syndrome 2 (CDLS2)	SMC1L1	Xp11.22-p11.21
Cowden (CD)	PTEN	10q23.31
Craniosynostosis with 11p15.2 disruption	SOX6	11p15.2

Cri-du-Chat syndrome (CMT1A)	Multiple	5p15.2-p13.3
Cutis laxa, X-linked/Occipital horn syndrome	ATP7A	Xq12-q13
Cystinosis, Nephrothic (CTNS)	CTNS	17p13.2-p13.3
Dandy-Walker syndrome (DWS)	ZIC1, ZIC4	3q24
Diaphragmatic hernia, congenital (HCD/DIH1)	NR2F2/CHD2	15q26.1-q26.2
DiGeorge syndrome 1 (DGS1)/Velocardiofacial (VCFS)	TBX1	22q11.21
DiGeorge syndrome 2 (DGS2)	Multiple	10p14
Down syndrome critical region (DSCR)	Multiple	21q22.12
Dyggve-Melchior-Clausen syndrome (DMC)	DYM	18q21.1
DMD Duchenne muscular dystrophy	DMD	Xp21
Feingold syndrome	MYCN	2p24.3
Focal Dermal Hypoplasia/Goltz Syndrome	PORCN	Xp11.23
Forebrain defects; TDGF1	CRIP1	3p21.31
Fragile-X mental retardation syndrome (FMR1)	FMR1	Xq27.3
Generalized epilepsy with febrile seizure plus (GEFS+)	SCN1A	2q24.3
Glycerol kinase deficiency (GKD)	GK	Xp21.2
Greig cephalo-polysyndactyly syndrome (GCPS)	GLI3	7p14.1
Hemophilia A	F8	Xq28
Hereditary Neuropathy with Liability to Pressure Palsies (HNPP)	PMP22	17p11.2
Holoprosencephaly 1 (HPE1)	TMEM1	21q22.3
Holoprosencephaly 2 (HPE2)	SIX3	2p21
Holoprosencephaly 3 (HPE3)	SHH	7q36.3
Holoprosencephaly 4 (HPE4)	TGIF	18p11.31
Holoprosencephaly 5 (HPE5)	ZIC2	13q32.3
Holoprosencephaly 6 (HPE6)	HPE6	2q37.1-q37.3
Holoprosencephaly 7 (HPE7)	PTCH	9q22.32
Holosencephaly 9/ Pituitary anomalies with holoprosencephaly	GLI2	2q14.2
Hypogammaglobulinemia, isolated growth hormone deficiency	BTK	Xq22.1
Hypoparathyroidism, sensorineural deafness, renal (HDR)	GATA3	10p14
Incontinentia pigmenti (IP)	IKBKG (NEMO)	Xq28
Infantile spasm syndrome, X-linked (ISSX)	ARX	Xp21.3
Infantile spasm syndrome, X-linked (ISSX)	CDKL5	Xp21.3
Jacobson Syndrome	Multiple	11q24-q25
Joubert 4	NPHP1	2q13
Kallman syndrome 1 (KAL1)	KAL1	Xp22.31
Langer-Giedion syndrome (LGS)	EXT1, TRPS1	8q23.3-q24.11

Lesch-Nyhan syndrome (LNS)	HPRT1	Xq26
Leukodystrophy with 11q14.2-q14.3		11q14.2-q22.3
Leri-Weill dyschondrosteosis	SHOX, SHOXY	Xp22.33/Yp11
Lissencephaly, X-linked (LISX)	DCX	Xq22.3
Loeys-Dietz syndrome (LDS)	TGFBR1, TGFBR2	9q33-q34,3p22
Marfan syndrome (MFS)	FBN1	15q21.1
Marfan Syndrome, Type II (MFS2)	TGFBR2	3p22
MECP2 male duplication syndrome	MECP2	Xq28
Menkes disease (MNK)	ATP7A	Xq12-q13
Mental Retardation, X-linked 21 (MRX21)	IL1RAPL1	Xp22.1-p21.3
Mental retardation X-linked growth horm. Def (MRGH)	SOX3	Xq26.1
Micophthalmia with linear skin defects	HCCS	Xp22.2
Microcephaly, congenital heart disease	NKX2-5	5q35.1
Microdeletion 2p15-p16.1		2[15-p16.1
Microphthalmia, syn. 3 (MCOPS3)	SOX2	3q26.33
Microphthalmia syn. 6, pituitary hypoplasia (MCOPS6)	SIX6	14q22.2-q22.3
Microphthalmia, syndromic 7 (MCOPS7)/MLS/MIDAS	Multiple	Xp22.2
Miller-Dieker Lissencephaly syndrome (MDLS)	LIS1 and/or YWHAE	17p13.3
Monosomy 9p Syndrome		9p22.3-p23
Mowat-Wilson syndrome	ZFX1B	2q22.3
Mucopolysaccharidosis, type II (MPS2)	IDS	Xq28
Nail-patella syndrome (NPS)	LMX1B	9q33.3
Nephronophthisis 1 (NPH1)/2q13 duplication	NPHP1	2q13
Neurofibromatosis 1 (NF1)	NF1	17q11.2
Neurofibromatosis 2 (NF2)	NF2	22q12.2
Noonan Syndrome (NS1)	PTPN11	12q24.13
Obesity deletion syndrome/Prader-Willi-like phenotype	SIM1	6q16.3
Opitz syndrome	MID1	Xp22.2
Ornithine transcarbamylase deficiency	OTC	Xp11.4
Orofaciodigital syndrome (OFD1)	CXORF5	Xp22.2
Osteogenesis imperfecta congenita	COL1A1	17q21.33
Osteogenesis imperfecta congenita	COL1A2	17q21.3
Pallister-Killian	Multiple	12p
Pelizaeus-Merzbacher disease (PMD)	PLP1	Xq22.2
Polycystic kidney disease 1	PKD1, TSC2	16p13.3
Potocki-Lupski Syndrome/17p11.2 duplication; reciprocal SMS region	RAI1	17p11.2

Potocki-Shaffer	EXT2, ALX4	11p11.2
Prader-Willi syndrome (PWS)	SNRPN, NECDIN, snoRNAs	15q11.2-q12
Prader-Willi-like phenotype;see Obesity deletion syndrome	SIM1	6q16.3
Renal cysts and diabetes (RCAD)	TCF2	17q12
Retinoblastoma (RB1)	RB1	13q14.2
Retinoschisis, X-linked juvenile (RSI)	XLRS1	Xp22.13
Rett syndrome (RTT); MECP2 deletion	MECP2	Xq28
Rieger syndrome, type 1 (RIEG1)	PITX2	4q25
Rubinstein-Taybi syndrome (RSTS)	CREBBP	16p13.3
Sacral/anorectal malformation syndrome		6q25.3
Saethre-Chotzen syndrome (SCS)	TWIST1	7p21.1
Sensorineural deafness and male infertility		15q15.3
Sex reversal, autosomal dominant 2 (SRA2)		9p24.3
Sex reversal, dosage-sensitive (DSS)	NROB1	Xp21.2
Sex reversal, SOX8 related	SOX8	16p13.3
Sex reversal X/Y translocations or deletions	SRY sex determining region of Y	Yp11.31
Sex reversal XY +/- adrenal failure	NR5A1 (SF1)	9q33
Severe myoclonic epilepsy of infancy (SMEI)/Dravet Syndrome	SCN1A	2q24.3
Short stature, pituitary and cerebellar defects, & small sella turcica	LHX4	1q25.2
Short stature, idiopathic, X-linked;see Leri Weill	SHOX, SHOXY	Xp22.33/Yp11.32
Smith-Magenis syndrome (SMS)	RAI1	17p11.2
Sotos syndrome (SOS)	NSD1	5q35.3
Speech delay with FOXP2 deletion	FOXP2	7q31.1
Split-hand/foot malformation-1 (SHFM1)		7q31.1
Split-hand/foot malformation-3 (SHFM3)		10p14
Split-hand/foot malformation-4 (SHFM4)	TP73L	3q28
SRY-Testis determination factor on Y	SRY	Yp11.2
Steroid Sulfatase deficiency (STS)	ARSC2	Xp22.31
STS dup Xp22.31 Unclear		Xp22.31
Stickler syndrome, type II (STL2)	COL11A1	1p21.1
Stocco dos Santos X-Linked Mental Retardation	SHROOM4 (KIAA1202)	Xp11.22-p11.21
Synpolydactyly (SPD1)	HOXD13	2q31-q32
Thrombocytopenia-Absent Radius Syndrome		2q21.1
Treacher Collins-Franceschetti Syndrome (TCOF)	TCOF1	5q32
Trichorhinophalangeal syndrome 1 (TRPS1)	TRPS1	8q23.3

Tuberous Sclerosis 1 (TSC1)	TSC1	9q34
Tuberous Sclerosis 2 (TSC2)	TSC2	16p13.3
Van der Woude syndrome (VWS)	IRF6	1p32.2
Vascular endothelial growth factor (VEGF)	VEGF	6p21.1
Waardenburg syndrome, Type IIA (WS2A)	MITF	3p13
Williams-Beuren syndrome (WBS)	ELN, LIMK1, NCF1	7q11.23
Wilms tumor 1 (WT1) & WAGR	WT1	11p13
Wilms tumor 1 (WT1) & WAGR	PAX6, WT2	11p13
Wolf-Hirschhorn syndrome	Multiple	4p16.3
XIST deficiency; usually ring X chromosome	XIST	Xq13.2 usually ring X chromosome
X-linked heterotaxy (HTX)	ZIC3	Xq26.3
X-linked lymphoproliferative syndrome (XLP)	SH2D1A	Xq25
X-Linked Mental Retardation	CASK	Xp11.4
X-Linked Mental Retardation, 9 (MRX9)	MITF	3p13
All 41 unique subtelomeric regions	multiple	41 sites
All 43 unique pericentromeric regions	multiple	43 sites
Aneuploidy for 24 chromosomes	multiple	24 chromosomes