105K OLIGO ARRAY

Disorder List

BCM
Baylor College of Medicine

MGL
MEDICAL GENETICS LABORATORIES

One Baylor Plaza, Nab 2015, Houston, Texas 77030

MEDICAL GENETICS LABORATORIES

Chromosomal Microarray Analysis (CMA) using Array Comparative Genomic Hybridization (aCGH) is available through the Baylor College of Medicine Medical Genetics Laboratories. Baylor was one of the first labs to offer CMA for clinical application, and we remain a leader in the implementation of new technology for CMA. With a single test, CMA will detect almost all of the disorders detected by chromosome analysis and multiple FISH tests. Included in CMA are probes for all the known microdeletion/duplication syndromes, as well as the pericentromeric and subtelomeric regions. Subtelomeric evaluation is enhanced with CMA by using multiple oligos, covering approximately 10 Mb at each subtelomere. CMA utilizes array-based comparative genomic hybridization with approximately 105,000 oligos covering the whole genome at an average resolution of 30 Kb, with increased coverage at disease loci. Chromosomal Microarray Analysis is a major advance in genetic testing, assisting the clinician in the diagnosis of patients in which a genetic condition is suspected.

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Perisylvian polymicrogyria (Polymicrogyria, bilateral perisylvian; BPP)	SRPX2	Xq22.1
10q22q23 deletion	NRG3, GRID1	10q22q23 deletion
11q11–q13.3 duplication/multiple craniosynostoses, congenital heart defect	FGF3, FGF4	11q13.3 duplication
12q14.1q15 microdeletion	GRIP1	12q14.3 deletion
12q24.21-q24.23 microduplication	THRAP2, NOS1, RFC5	12q24.21-q24.23 duplication
13q33q34 deletion/ genital malformation in males/Microcephaly/MR	EFNB2, ARHGEF7	13q33.3q34 deletion
14q11.2 deletion syndrome	SUPT16H, CHD8	14q11.2 deletion
14q12 deletion syndrome	FOXG1B	14q12 deletion
15q21 microdeletion syndrome		15q21.2 deletion
15q24 deletion	HCN4	15q24.1
15q24 microdeletion	PML	15q24.1 deletion
15q26.3 deletion, Severe IUGR, developmental delay, postnatal growth retardation	IGF1R linsulin-like growth factor -1 receptor	15q26.3 deletion
16p11.2p12 deletion		16p12.1 deletion
17q21.31 microdeletion/microduplication syndrome	MAPT, CRHR1	17q21.31 deletion
1q21.2 deletion/duplication		1q21.2 deletion/ duplication
1q41q42 deletion	DISP1	1q41 deletion
20q13.33 deletion/Autosomal-dominant nocturnal frontal lobe epilepsy,Benign familial neonatal convulsions (type 1), hypotrichosis–lymphedema–telangiectasia	ARFGAP1, CHRNA4, KCNQ2, SOX18	20q13.33 deletion
22q11.2 duplication syndrome reciprocal to DGS deletion	TBX1 T-box 1 ?	22q11.2 duplication
22q13.3 deletion/Autism	SHANK3	22q13.33 deletion
2p15-p16.1 microdeletion		2p15-p16.1 deletion
2q22.3 deletion		2q22.3 deletion

Phenotype	Genes	Cytogenetics
2q22.3q23.3	MBD5, KIF5C	2q23.1
2q35	IHH	2q35
2q37 deletion/brachydactyly-MR/obesity/Albright hereditary osteodystrophy	CENTG2, GPC1, GPR35, ATSV/ KIF1A, STK25	2q37.3 deletion
3p25 deletion	CNTN4	3p25.3 deletion
3q29 deletion syndrome/Autism	PAK2, DLG1	3q29 deletion
5q14.3 deletion	MEF2C	5q14.3
5q21q31 deletion syndrome		5q21q31 deletion
6q24.3q25.1(Diabetes mellitus, insulin-dependent, 5; IDDM5)	SUMO4	6q25.1
7q11.23 duplication (WBS region)		7q11.23 duplication
7q21.13q22.1 deletion/Ectrodactyly/Deafness	CUTL1 (CUX1), FZD1	7q22.13q22.1 deletion
8p23.1 Inverted duplication/deletion		8p22p23 inversion
9q34.3 deletion syndrome	NOTCH1, EHMT1	9q34.3 deletion
Acheiropody/ and preaxial polydactyly	LMBR1	7q36.3 deletion
Action myoclonus-renal failure syndrome (AMRF)	SCARB2	4q21.1 deletion
Adrenal hyperplasia, congenital (CAH) due to 21-alpha hydroxylase deficiency	CYP21A2	6p21.32 deletion
Adrenal hypoplasia congenital (AHC)	NROB1 nuclear receptor family 0 B1 (DAX1)	Xp21.2 deletion
Adrenoleukodystrophy; (ALD)	ABCD1	Xq28 deletion
Alagille syndrome (AGS)	JAG1 jagged 1	20p12.2 deletion
P henotype	Genes	Cytogenetics
Albinism, ocular type 1	GPR143/OA1	Xp22.2 deletion
All 41 unique subtelomeric regions	Multiple	41 sites
All 43 unique pericentromeric regions	Multiple	43 sites
Allan-Herndon-Dudley syndrome, X-linked mental retardation (XLMR)	SLC16A2	Xq13.2 deletion
Alpha thalassemia-MR syndrome	HBA2 & HBA1	16p13.3 deletion

Phenotype	Genes	Cytogenetics
Alport syndrome, X-linked (ATS)	COL4A5	Xq22.3 deletion
Andersen syndrome	KCNJ2	17q24.3 deletion
Androgen insensitivity syndrome (AIS)	AR	Xq12 deletion
Aneuploidy for 24 chromosomes	Multiple	24 chromosomes
Angelman syndrome (AS)	UBE3A ubiquitin ligase 3A, AS-SRO,	15q11.2-q12 deletion
Angelman syndrome (AS)	AS imprinting center	15q11.2 deletion
Aniridia II (AN2)	PAX6 paired box gene 6	11p13 deletion
Anterior segment mesenchymal dysgenesis (ASMD)/ Cataract	PITX3	10q24.32 deletion
Arthropathy, progressive pseudorheumatoid, of childhood; (PPAC)	WISP3	6q21 deletion
Ataxia	ITPR1	3p26.2 deletion
Ataxia teleangiectasia	ATR	3q23 deletion
Atrial septal defect	GATA4	8p23.1 deletion
ATRX, XLMR-Hypotonic facies syndrome, ATR-X, and others	ATRX	Xq21.1 deletion/ duplication
Autism		4q32.1 deletion
Autism	DLX5	7q21.3 deletion
Autism	JMJD1C, TRIP8, REEP3	10q21.3 deletion
Autism	NUFIP1	13q14.12 deletion
Autism	NRXN3	14q31.1 deletion
Autism	A2BP1	16p13.2 deletion
Autism	DLG4	17p13.1 deletion
Autism	NUFIP2	17q11.2 deletion
Autism	CNTNAP1	17q21.31 deletion
Autism	ASMT (ASMTL)	Xp22.33 deletion
Autism	DPP10	2q14.1 deletion
Autism	DPP6	7q36.2 deletion

One Baylor Plaza, Nab 2015, Houston, Texas 77030

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Autism	PCDH9	13q21.32 deletion
Autism	SYBL1	Xq28 deletion
Autism	NRXN1	2p16.3 deletion
Autism	CNTNAP3	9p13.1 deletion
Autism	BDNF	11p14.1 deletion
Autism	NRXN2	11q13.1 deletion
Autism / Asperger syndrome-1, X-linked	NLGN3	Xq13.1 deletion
Autism/Schizophrenia 9 (SCZD9)	DISC1	1q42.2 deletion
Autistic Features 15q11.2-q12	Uncertain	15q11.2-q13 maternal duplication
Autistic features, X-linked, susceptibility to, AUTSX2 (XLMR)	NLGN4	Xp22.32 deletion
Azoospermia factor (AZFa, AZFb)		Yq11.21 deletion
Bartter syndrome type 2	KCNJ1	11q24.3 deletion
Bartter syndrome type 3	CLCNKB	1p36.13 deletion
Bartter syndrome type 4 (Bartter syndrome type 3)?	CLCNKA, CLCNKB, BSND	1p36.13-p32.3
Bartter syndrome, antenatal type 1	SLC12A1	15q21.1 deletion
Bartter with autosomal dominant	CASR	3q21.1 deletion
Basal cell nevus syndrome (BCNS) / Gorlin syndrome (GS)	PTCH1 patched Drosophila homolog	9q22.32 deletion
Beckwith-Wiedemann syndrome (BWS)	IGF2 insulin-like growth factor II, CDKN1C cyclin-dependent kinase inhibitor 1C H19, KCNQ1, p57 (CDKN1C)	11p15.4/15.5 deletion/duplication
Bilateral frontoparietal polymicrogyria/focal or multifocal epilepsy	GPR56	16q13 deletion
Bipolar disorder	IMPA1	8q21.13 deletion
Birk-Barel syndrome	KCNK9	8q24.3

Phenotype	Genes	Cytogenetics
Blepharophimosis, ptosis, and epicanthus inversus (BPES)	FOXL2	3q22.3 deletion
Blepharophimosis/vormian hypolasia/exotropia/DD	POFUT2	21q22.3 deletion
Börjeson-Forssman-Lehmann syndrome	PHF6	Xq26.2 deletion
BP1/BP2 breakpoint region in 15q11.2		15q11.2 deletion/duplication
Brachydactyly and other skeletal anomalies	GPC5, GPC6	13q31.3 deletion
Brachydactyly, type C (BDC)	GDF5	20q11.22 deletion
Branchiootic syndrome-3 (BOS3)	SIX1	14q23.1 deletion
Branchiootorenal dysplasia syndrome (BOR)/ Otofaciocervical (OFC)/Melnick-Fraser	EYA1	8q13.3 deletion
Brunner syndrome/Monoamine oxidase-A deficiency/Antisocial behavior following childhood maltreatment	MAOA	Xp11.3 deletion
Bruton agammaglobulinemia tyrosine kinase	BTK Bruton agammaglobulinemia tyrosine kinase	Xq22.1 deletion
Buschke-Ollendorff syndrome / Osteopoikilosis, short stature and mental retardation	LEMD3	12q14.3 deletion
CADASIL	NOTCH3	19p13.12 deletion
CALC1, behavioral problems and autistic spectrum disorder	CALCA, CALCB	11p15.2 deletion
Campomelic dysplasia	SOX9 SRY- box 9	17q24.3 deletion
Canavan disease	ASPA	17p13.3 deletion
Cardiomyopathy, familial hypertrophic/Congenital heart defect	ACTC	15q14 deletion
Cat eye syndrome (CES)	CECR1, CECR5, CECR6	inv dup(22)(q11.2)
Cerebellar hypoplasia	OPHN1	Xq12 deletion
Cerebral cavernous malformation	CCM2	7p13 deletion
Cerebral cavernous malformations 2	CCM2	7p13 deletion
Ceroid lipofuscinosis, neuronal 8, northern epilepsy variant	CLN8	8p23.3 deletion

Phenotype	Genes	Cytogenetics
Char syndrome	TFAP2B	6p12.3 deletion
Charcot-Marie-Tooth disease type 1A (CMT1A)	PMP22 peripheral myelin protein 22	17p12 duplication
CHARGE syndrome	CHD7 Chromodomain helicase DNA-binding 7	8q12.2 deletion
Chondrodysplasia punctata, X-linked recessive	CDPX1 (ARSE)	Xp22.33 deletion
Chondrodysplasia, grebe tybe	CDMP1 (GDF5)	20q11.22 deletion
Chondrodysplasias (Stickler syndrome/Spondyloepiphyseal dysplasia)	COL2A1	12q13.11 deletion
Choroideremia	CHM	Xq21.2 deletion
Choroideremia	CHM	Xq21.2
Chromosome 10q deletion syndrome		10q26 deletion
Chromosome 18p deletion syndrome		18p11.3 deletion
Chromosome 18q deletion syndrome		18q23 deletion
Chromosome Xp11.3 deletion syndrome/Retinitis pigmantosa X-linked RP2/ XLMR	RP2	Xp11.3 deletion
Chronic pancreatitis	SPINK1	5q32 deletion
Cleft lip/palate	ESR1	6q25.1 deletion
Cleft lip/palate	FGFR2	10q26.13 deletion
Cleft palate, isolated (CPI)	SATB2	2q33.1 deletion
Cleft plate, congenital heart defect	GREM1, GJD2 (CX36), MEIS2	15q13.3 deletion
Cleidocranial dysplasia (CCD)	RUNX2 runt-related transcription factor 2	6p12.3 deletion
Coffin-Lowry syndrome	RPS6KA3/RSK2	Xp22.12 deletion
Coloboma	SNAP25	20p12.2 deletion
Corneal dystrophies	TGFBI	5q31.2
Cornelia de Lange syndrome (CDLS)	NIPBL nipped-B-like	5p13.2 deletion
Cornelia de Lange syndrome (CDLS), X-linked	SMC1A/SMC1L1	Xp11.22 deletion

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Cowden (CD) & Bannayan-Riley-Ruvalcaba syndrome (BRRS)	PTEN	10q23.31 deletion
Craniofrontal dysplasia	CFND	Xq11.2q13.1
Craniometaphyseal dysplasia autosomal dominant (CMDD)	ANKH	5p15.2 deletion
Craniosynostosis	SOX6	11p15.2 deletion
Craniosynostosis (CRS2)	MSX2	5q35.2 deletion
Creatine deficiency syndrome / X-linked mental retardation (XLMR)	SLC6A8	Xq28 deletion
Cri-du-Chat syndrome (cry/speech/face critical regions)	Multiple (TERT, EGR1)	5p15.2 -p13.3 deletion
Currarino syndrome (Sacral agenesis)	HLXB9	7q36.3 deletion
Cystinosis, Nephropathic (CTNS)	CTNS	17p13.2-p13.3 deletion
Dandy-Walker syndrome (DWS)	ZIC1, ZIC4 zinc finger protein of the cerebellum 1, 4	3q24 deletion
Danon disease	LAMP2	Xq24 deletion
Deafness, X-linked 2 (DNFX2)	POU3F4	Xq21.1 deletion
Dent disease	CLCN5	Xp11.22 deletion
Diabetes insipidus, nephrogenic, X-linked	AVPR2	Xq28 deletion
Diabetes mellitus, transient neonatal, IUGR	ZAC (PLAGL1)	6q24.2 paternal deletion
Diaphragmatic hernia	SOX7	8p23.1 deletion
Diaphragmatic hernia, congenital	NR2F2	15q26.1-q26.2 deletion
DiGeorge syndrome 2 (DGS2)		10p14 deletion
DiGeorge/velocardiofacial syndrome DGS1/ VCFS / Autism	GNB1L TBX1 T-box 1	22q11.2 deletion
Dosage sensitive sex reversal (DSS)	NROB1 nuclear receptor family 0 B1 (DAX1)	Xp21.2 duplication
Down syndrome critical regions	DSCR1,DSCR2, DSCR3,DSCR4, DSCR5, DSCR8, DSCR9,DSCR10,	21q22.-2-q22.13/q22.2?

Phenotype	Genes	Cytogenetics
Dyggve-Melchior-Clausen syndrome (DMC) (autosomal recessive)	DYM	18q21.1 deletion/ duplication
Dyskeratosis congenita, X-linked	DKC1	Xq28 deletion
Ectodermal dysplasia, anhidrotic	EDAR	2q13 deletion
Ectodermal dysplasia, X-linked hypohidrotic	EDA	Xq13.1 deletion
Epilepsy	KCND2	7q31.31 deletion
Epilepsy	EFHC2	Xp11.3 deletion
Epilepsy idiopatic generalized/Migraine/Spinocerebellar ataxia 6	CACNA1A	19q21.2 deletion
Epilepsy, Benign neonatal (EBN2)	KCNQ3	8q24.22 deletion
Epilepsy, idiopathic generalized (EIG6)	CACNA1H	16p13.3 deletion
Epilepsy, juvenile absence	GRIK1 (kainate ionotropic)	21q21.3 deletion
Epilepsy, juvenile absence (JAE)	GRIK1	21q21.3 deletion
Epilepsy, juvenile absence (JAE) juvenile myoclonic (EJM1)	EFHC1, c6orf33, LMPB1	6p12.2 deletion
Epilepsy, juvenile myoclonic	CACNB4	2q22-q23 deletion
Epilepsy, juvenile myoclonic	CLCN2	3q27.1 deletion
Epilepsy, juvenile myoclonic	CHRNA7	15q13.3 deletion
Epilepsy, lateral temporal lobe, autosomal dominant (ADLTE)	LGI1	10q23.33 deletion
Epilepsy, nocturnal frontal lobe (type 3)	CHRNB2	1q21.3 deletion
Epilepsy, nocturnal frontal lobe (type 4)	CHRNA2	8p21.2 deletion
Epilepsy, nocturnal frontal lobe, type2/ Autosomal-dominant nocturnal frontal lobe epilepsy (ADNFLE)	CHRNA3, CHRNB4, CHRNA5,	15q25.1 deletion
Epilepsy, progressive myoclonnus	NHLRC1	6p22.3 deletion
Epilepsy, X-linked	SYN1	Xp11.23/11.3 deletion
Episodic ataxia (EA1), partial epilepsy	KCNA1	12p13.32 deletion
Fabry disease	GLA	Xq22.1 deletion

Phenotype	Genes	Cytogenetics
Faciogenital dysplasia	FGD1	Xp11.22 deletion
Faciogenital dysplasia/Aarskog-Scott syndrome	FGD1	Xp11.22 deletion
Feingold syndrome	MYCN	2p24.3 deletion
FGFR3 (Achondroplasia; ACH)	FGFR3	4p16.3
FGS2		Xq28 deletion
Focal dermal hypoplasia (Goltz syndrome), XLMR	PORCN	Xp11.23 deletion
Forebrain defects	TDGF1 (CRIPTO)	3p21.31 deletion
Fragile-X mental retardation syndrome (FMR1)	FMR1	Xq27.3 deletion
Generalized epilepsy with febrile seizures plus (GEFS+)/ Epilepsy, childhood absence (CAE)	GABRG2	5q34 deletion
Generalized epilepsy with febrile seizures-3 plus (type 1)	SCN1B	19q13.11 deletion
Glucose transport defect / Epilepsy, autosomal dominant	GLUT1 (SLC2A1)	17p13.1/1p34.2 deletion
Gonadal dysgenesis, XY female type/Sex reversal X/Y translocations, (Sex determination male)	SRY sex-determining region Y	Yp11.31 translocation/deletion
Granulomatous disease, chronic X-linked	CYBB	Xp11.4 deletion
Greig cephalo-polysyndactyly syndrome (GCPS)	GLI3 GLI-Kruppel family 3	7p14.1 deletion
Hand-foot-uterus syndromes	HOXA13	7p15.2 deletion
Hemophilia A, (Factor 8, hemophilia)	F8	Xq28 deletion
Hereditary neuropathy with pressure palsies (HNPP)	PMP22 peripheral myelin protein 22	17p12 deletion
Heterotaxy	ACVR2B	3p22.2 deletion
Heterotaxy	EBAF (LEFTY2)	1q42.12 deletion
Heterotaxy, visceral 2	CFC1	2q21.1 deletion
Heterotopia, periventricular, X-linked dominant/ Otopalatodigital syndrome type 1 (OPD)/multifocal epilepsy in females	FLNA	Xq28 deletion
Hirschsprung disease	RET	10q12.11 deletion
Hirschsprung disease (HSCR2)	EDNRB	13q22.3 deletion

One Baylor Plaza, Nab 2015, Houston, Texas 77030

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Holoprosencephaly 1 (HPE1)	TMEM1 transmembrane protein1,LSS	21q22.3 deletion
Holoprosencephaly 1 (HPE1)	SIM2	21q22.13 deletion
holoprosencephaly 2(HPE2)	SIX3 sine oculis homolog 3	2p21 deletion
Holoprosencephaly 3 (HPE3)	SHH sonic hedgehog	7q36.3 deletion
Holoprosencephaly 4 (HPE4)	TGIF transforming growth factor-ß induced factor	18p11.31 deletion
Holoprosencephaly 5 (HPE5)	ZIC2 zinc finger protein cerebellum 2	13q32.3 deletion
Holoprosencephaly 6 (HPE6)		2q37.1q37.3 deletion
Holoprosencephaly 7 (HPE7)	PTCH1 patched Drosophila homolog	Missense gain-of-fuction mutations / duplication? duplications?
Holoprosencephaly 9 (HPE9)/Pituitary anomalies with holoprosencephaly	GLI2	2q14.2 deletion
Holt-Oram syndrome	TBX5	12q24.21 deletion
Hunter/Mucopolysaccharidosis, type II (MPS2)	IDS	Xq28 deletion
Hyperekplexia and epilepsy/X-linked mental retardation (XLMR)	ARHGEF9	Xq11.1 deletion
Hyperglycerolemia (Glycerol kinase deficiency)	GK glycerol kinase	Xp21.2 deletion
Hypertension with CHD	BMPR2	2q33.1 deletion
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR)	GATA3 GATA-binding protein 3	10p14 deletion duplication
Hypophosphatemic rickets, X-linked dominant	PHEX	Xp22.11 deletion
Hypotonia-Cystinuria syndrome/ with mitochondrial disease	PPM1B, SLC3A1, PREPL	2p21 deletion
Ichthyosis, X-linked (steroid sulfatase deficiency)	STS steroid sulfatase deficincy	Xp22.31 deletion

Phenotype	Genes	Cytogenetics
Immunodeficiency-centromeric Instability-Facial anomalies syndrome	DNMT3B	20q11.21 deletion
Incontinentia pigmenti (IP)	IKBKG (NEMO)	Xq28 deletion
Infantile spasm syndrome, X-linked (ISSX), West, Proud, XLAG, Partington, multifocal epilepsy	ARX	Xp21.3 deletion
Iridogoniodysgenesis anomaly, Axenfeld-Rieger syndrome, 6p25.3 deletion syndrome	FKHL7 (FOXC1)	6p25.3 deletion
Jacobsen syndrome		11q24-q25 deletion
Jacobsen syndrome (JBS)/ Trigonocephaly 11q deletion		11q24.1 deletion
Johanson-Blizzard syndrome (JBS)	UBR1	15q15.2 deletion
Juvenile myoclonic epilepsy	GABRA1	5q34 deletion
Kallmann syndrome 1 (KAL1)	KAL1 Kallmann syndrome 1	Xp22.31 deletion
Kartagener syndrome	DNAI1	9p13.3 deletion
Kartagener syndrome	DNAH5	5p15.2 deletion
Langer-Giedion syndrome (LGS)/trichorhinophalangeal syndrome type II (TRPS2)	TRPS1 zinc finger transcription TRPS1 & EXT1 exostosin 1	8q23.3-q24.11 deletion
Leri-Weill dyschondrosteosis (LWD) / short stature	SHOX/SHOXY short stature homeobox	Xp22.33 / Yp11.32
Lesch-Nyhan syndrome (LNS)	HPRT1 (HPRT)	Xq26.2 deletion
Leukodystrophy due to mitochondrial complex 1 deficiency	NDUFV1	11q13.2 deletion
Leukodystrophy with microdeletion 11q14.3		11q14.2q14.3 deletion
Leukodystrophy, adult onset autosomal dominant (ADLD)	LMNB1	5q23.2 deletion
Lissencephaly/Autism /multifocal epilepsy	RELN	7q22.1 deletion
Loeys-Dietz syndrome (LDS)	TGFBR1	9q22.33 deletion
Loeys-Dietz syndrome, type 2B (LDS2B)/Marfan syndrome, type II	TGFBR2	3p24.1 deletion
Lowe oculocerebrorenal syndrome	OCRL	Xq25 deletion

Phenotype	Genes	Cytogenetics
LUBS (MRXSL) / MECP2 duplication	MECP2 methyl-CpG-binding protein-2	Xq28 duplication
Lymphedema hereditary, II	FOXC2	16q24.1 deletion
Macrocephaly/seizures	SYT14	1q32.2 deletion
Marfan syndrome (MFS)	FBN1	15q21.1 deletion
MASA syndrome, X-linked mental retardation (XLMR)	L1CAM	Xq28 deletion
McCune-Albright syndrome	GNAS	20q13.32 deletion
McKusick-Kaufman syndrome (MKKS)	MKKS	20p12.2 deletion
Menkes disease (MNK), Cutis laxa / Occipital horn syndrome (OHS)	ATP7A	Xq21.1 deletion
Mental retardation	ARHGAP11A, CHRM5	15q13.3 deletion
Mental retardation X-linked with isolate growth hormone deficiency (MRGH)	SOX3 SRY- box 3	Xq27.1 deletion or duplication
Mental retardation, X-linked 21 (MRX21)	IL1RAPL1	Xp21.3 deletion
Mental retardation, X-linked, syndromic, Turner type/X-linked mental retardation (XLMR)	HUWE1	Xp11.22 deletion
Mental retardation, X-linked, with Epilepsy (XMRE)	ATP6AP2	Xp11.4 deletion
Metachromatic leukodystrophy	ARSA	22q13.33 deletion
Micophthalmia, syndromic 7 (MCOPS7)/Microphthalmia with linear skin defects (MLS)	HCCS	Xp22.2 deletion
Microcephaly, congenital heart disease	NPM1	5q35.1 deletion
Microcephaly/agenesis corpus callosum	AKT3	1q44 deletion
Microphthalmia syndromic (MCOPS2)	BCOR	Xp11.4 deletion
Microphthalmia syndromic 3 (MCOPS3)	SOX2	3q26.33 deletion
Microphthalmia syndromiuc 6 (MCOPS6)/ Anophthalmia, pituitary hypoplasia, and ear anomalies	BMP4	14q22.2 deletion
Microtia	DRD5 (CNV)	4p16.1 deletion
Miller-Dieker lissencephaly syndrome (MDLS)	LIS1, YWHAE	17p13.3 deletion
Mohr-Tranebjaerg syndrome	TIMM8A	Xq22.1 deletion

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Monogenic audiogenic seizure susceptibilty	MASS1 (GPR98, VLGR1)	5q14.3 deletion
Monosomy 1p36 (Facial Clefting Anomalies, Generalized epilepsy with febrile seizures-3 plus (type 5), Cranial Suture Closure, Seizures)	DVL1, SKI, GABRD, MMP23, KCNAB2	1p36 deletion and/or duplication/ triplication
Monosomy 9p syndrome		9p22.3p23
Mowat-Wilson syndrome	ZEB2/ZFHX1B	2q22.3 deletion
Multicentric osteolysis, nodulosis, and arthropathy (MONA)	MMP2	16q12.2 deletion
Muscle-eye-brain disease	POMGNT1	1p34.1 deletion
Myoclonic epilepsy of Lafora	EPM2A	6q24.3 deletion
Myoclonic epilepsy, juvenile/juvenile absence epilepsy	BRD2	6p21.32 deletion
Myoclonic epilepsy, neonatal, with suppression-burst pattern	SLC25A22	11p15.5 deletion
Nail-patella syndrome (NPS)	LMX1B LIM-homeo box factor 1ß	9q33.3 deletion
Nance-Horan	NHS	Xp22.13 deletion
Nebulette	NEBL	10p12.31 deletion
Nephronophthisis 1/Related Joubert Syndrome (NPHP1)	NPHP1, nephrocystin	2q13 homozygous deletion
Neurofibromatosis I (NF1)	NF1 neurofibromin 1	17q11.2 deletion
Neurofibromatosis II (NF2)	NF2 neurofibromin 2	22q12.2 deletion
Neuromotor development delay, cerebellar ataxia, and epilepsy	AF9/MLLT3	9p21.3 deletion
Nievergelt syndrome	LAF4/AFF3	2q11.2
Noonan syndrome (NS1)	PTPN11	12q24.13 duplication
Noonan syndrome (NS4)	SOS1	2p22.1 deletion
Noonan syndrome 5 (NS5)	RAF1	3p25.1 deletion
Norrie disease	NDP	Xp11.3 deletion
Oculopharyngeal muscular dystrophy	PABPN1	14q11.2
Opitz BBB syndrome	MID1	Xp22.2 deletion

Phenotype	Genes	Cytogenetics
Ornithine transcarbamylase deficiency	OTC	Xp11.4 deletion
Orofacial cleft	SUMO1	2q32.2-q33 deletion
Orofaciodigital syndrome (OFD1)	CXORF5 (OFD1)	Xp22.2 deletion
Osler-Rendu-Weber syndrome (Hereditary hemorrhagic teleangiectasia)	ACVRL1	12q13.13 deletion
Osler-Rendu-Weber syndrome 2 (ORW2)	ALK1 (ACVRL1)	12q13.13 deletion
Osteogenesis imperfecta type IV	COL1A1	17q21.33 deletion
Osteogenesis imperfecta congenital	COL1A2	17q21.33 deletion
Oto-dental syndrome (dental and inner-ear disease/ocular coloboma)	FGF3/FADD	11q13.3 deletion
Overgrowth	GPR51/GABBR2	9q22.33 deletion
Ovotestes and male-to female sex-reversal XY	NR5A1 (SF1)	11q13.1 (9q33 deletion)?
Pallister-Killian syndrome		12p triplication
Pancreatic agenesis, congenital	IPF1 (PDX1)	13q12.2 deletion
Papillorenal syndrome (Renal-coloboma syndrome)	PAX2	10q24.31 deletion
Partial anodontia, orofacial clefting/Witkop syndrome	MSX1	4p16.2 deletion
Pelizaeus-Merzbacher disease (PMD)	PLP1 proteolipid protein 1	Xq22.2 duplication or deletion
Persistent Mullerian duct syndrome (PMDS) / Sex ambiguity	AMH/MIS	19p13.3 deletion
Phosphoglycerate kinase deficiency	PGK1	Xq21.1 deletion
Phosphoribosylpyrophosphate synthetase superactivity	PRPS1	Xq22.3 deletion
Pitt-Hopkins syndrome	TCF4	18q21.2 deletion
Pituitary hypoplasia, Oculoauriculovertebral spectrum	SIX6	14q23.1(14q22.--q22.3 del)?

Phenotype	Genes	Cytogenetics
Potocki-Lupski syndrome (PTLS)	RAI1 retinoic acid-induced gene 1	17p11.2 duplication
Potocki-Shaffer syndrome	ALX4 Aristaless-like-4 and/or EXT2 exostosin 2	11p11.2 deletion
Prader –Willi syndrome (PWS)	PWS-SRO, SNRPN, HBII-85 C/D box, NDN necdin, all snoRNAs , snoRNA cluster	15q11.2-q12 deletion
Prader –Willi syndrome (PWS) imprinting center	PWS imprinting center	15q11.2 deletion
Prader-Willi syndrome-like/Obesity	SIM1	6q16.3 deletion
Pseudoachondroplasia (PSACH)	COMP	19p13.11 deletion
Pseudoxanthoma elasticum, forme fruste	ABCC6	16p13.11 deletion
Pyruvate dehydrogenase deficiency	PDHA1	Xp22.12 deletion
Renal cysts and diabetes (RCAD)/epilepsy	TCF2	17q12 deletion/ duplication
Renpenning syndrome 1 (RENS1)/Sutherland-Haan XLMR syndrome/Golabi-Ito-Hall syndrome	PQBP1	Xp11.23 deletion
Retinoblastoma (RB1)	RB1 retinoblastoma 1	13q14.2 deletion
Retinoschisis, X-juvenile, juvenile (RS1)	RS1	Xp22.13 deletion
Retinoschisis, X-linked juvenile (RS1)	XLRS1	Xp22.13 deletion
Rett syndrome (RTT)	MECP2 methyl-CpG-binding protein-2	Xq28 deletion
Rett-like, Infantile spasm syndrome, X-linked (ISSX)	CDKL5	Xp21.3 deletion
Rieger syndrome, type 1 (RIEG1)	PITX2	4q25 deletion
Robinow syndrome/brachydactyly type B1 (BDB1)	ROR2	9q22.31 deletion
Rubinstein-Taybi Syndrome	EP300	22q13.2

One Baylor Plaza, Nab 2015, Houston, Texas 77030

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
Rubinstein-Taybi syndrome (RSTS)	TRAP1, DNASE1, CREBBP, CREB_ binding protein?	16p13.3 deletion
Sacral/anorectal malformation syndrome		6q25.3 deletion
Saethre-Chotzen syndrome (SCS)	TWIST1	7p21.1 deletion
Schizencephaly / multifocal epilepsy	EMX2	10q26.11 deletion
Schizophrenia & epilepsy	CNTNAP2	7q36.1 deletion
Schwartz-Jampel syndrome, type1 (SJS1)	HSPG2	1p36.12 deletion
Seizures	DGKD	2q37.1 deletion
Seizures, benign familial neonatal-infantile	SCN2A	2q23-q24.3 deletion
Severe myoclonic epilepsy of infancy (SMEI) or Dravet syndrome; Generalized epilepsy with febrile seizures plus; GEFS+	SCN1A	2q24.3 deletion
Sex reversal, autosomal dominant 2 (SRA2)		9p24.3 deletion
Shah-Waardenburg syndrome	EDNRB	13q22.3 deletion
Shah-Waardenburg syndrome	EDN3	20q13.32 deletion
Short stature, pituitary and cerebellar defects, & small sella turcica	LHX4	1q25.2 deletion
Siderius type X-linked mental retardation / Cleft lip/palate	PHF8	Xp11.22 deletion
Simpson-Golabi-Behmel syndrome type 1;(SGBS1)	GPC3	Xq26.2 deletion
Smith-Lemli-Opitz syndrome (SLOS)	DHCR7	11q13.4 deletion
Smith-Magenis syndrome (SMS)	RAI1 retinoic acid-induced gene 1	17p11.2 deletion
Sotos syndrome	NSD1 nuclear receptor binding Su-var	5q35.3 deletion
Speech delay/Autism	“FOXP2	
“	7q31.1 deletion	
Split hand/foot malformation 1 with hearing loss		7q21.3 deletion
Split hand/foot malformation 4 (SHFM4)	TP73L (TP63)	3q28 deletion

Phenotype	Genes	Cytogenetics
Split-hand/foot malformation (SHFM5)	DLX1/DLX2	2q31.1 deletion
Split-hand/foot malformation-3 (SHFM3)		10p14 duplication
Split-hand/split-foot malformation 1 (SHFM1)	SHFM1	7q21.3
Split-hand/split-foot malformation 3 (SHFM3)	SHFM3/FBXW4	10q24.32 deletion
Spondylocostal dysostosis autosomal recessive; (SCDO1)	DLL3	19q13.2 deletion
Stickler syndrome, type II (STL2)	COL11A1	1p21.1 deletion
Stickler syndrome, type II (STL2)	COL11A1	
Stocco dos Santos XLMR	SHROOM4 (KIAA1202)	Xp11.22 deletion
Susceptibility to adolescent-onset idiopathic generalized epilepsy	ME2	18q21.2 deletion
Symphalangisms proximal, Multiple synostoses syndrome 1, Stapes ankylosis with broad thumb and toes	NOG	17q22 deletion
Synpolydactyly (SPD1); HOXD deletion syndrome	HOXD13	2q31-q32 deletion
Telangiectasia, hereditary hemorrhagic, of rendu, osler, and weber; (HHT)	ENG	9q34.11 deletion
Tetralogy of Fallot / ASD secundum with atrioventricular conduction defects Microcephaly	NKX2-5 (CSX)	5q35.2 duplication
Tetralogy of Fallot/diaphragmatic hernia	ZFPM2/FOG2	8q23.1 deletion
Thrombocytopenia–Absent Radius Syndrome		1q21.1 deletion
Thrombocytopenia-absent radius syndrome (TAR)		1q21.1 deletion
Timothy syndrome	CACNA1C	12p13.33 deletion
Tooth agenesis, selective 3 (STHAG3) Hypodontia/Oligodontia 3	PAX9	14q13.3 deletion
Townes-Brocks syndrome	SALL1	16q12.1 deletion
Treacher-Collins-Franceschetti syndrome (TCOF)	TCOF1	5q33.1 deletion
Trichorhinophalangeal syndrome I (TRPS1)	TRPS1 zinc finger transcription TRPS1	8q23.3 deletion

Phenotype	Genes	Cytogenetics
Trigonocephaly (9p deletion)		9p23 deletion
Triphalangeal thumb Syndrome (Polydactyly, preaxial II; PPD2)	ZRS regulator of SHH	7q36.3
Ulnar-mammary syndrome	TBX3	12q24.21 deletion/duplication
Van Buchem disease	SOST	17q21.31
Van Buchem disease(Hyperostosis corticalis generalisata)	SOST and downstream	17q21.31
Van der Woude syndrome (VWS)	IRF6	1q32.2 deletion
Vascular endothelial growth factor (VEGF)	VEGFA (VEGF)	6p21.1 deletion
Von Hippel-Lindau syndrome	VBP1	Xq28 deletion
Von Hippel-Lindau syndrome (VHL)	VHL	3p25.3 deletion
Waardenburg syndrome, type 1	PAX3	2q36.1 deletion
Waardenburg syndrome, type IIA (WS2A)	MITF (Microphthalmia-associated transcription factor)	3p14.1 deletion
Waardenburg-Shah syndrome	SOX10	22q13.1 deletion
Walker-Warburg syndrome	POMT1	9q34.13 deletion
Williams-Beuren syndrome (WBS)	ELN elastin, LIMK1 LIM kinase 1, RFC2, CYLN2	7q11.23 deletion
Wilms tumor 1 (WT1)	WT1 Wilms tumor 1 gene	11p13 deletion
Wilms tumor -aniridia genitourinary anomalies--mental retardation syndrome (WAGR)	PAX6 & WT1	11p13 deletion
Wolff-Parkinson-White	BMP2 Bone morphogenetic proteins2	20p12.3 deletion
Wolf-Hirschhorn syndrome (WHS)	Multiple (LETM1, WFS1 WHCR1/2)	4p16.3 deletion

MEDICAL GENETICS LABORATORIES

Phenotype	Genes	Cytogenetics
X-inactivation, familial skewed, XIST deficiency	XIST	Xq13.2 usually ring X chromosome
X-linked heterotaxy, ZIC3	ZIC3 zinc finger protein cerebellum 3	Xq26.3 deletion
X-linked lymphoproliferative syndrome (XLP1)	SH2D1A	Xq25 deletion
X-linked mental retardation (XLMR)	CASK	Xp11.4 deletion
X-linked mental retardation (XLMR)	HSD17B10/HADH2	Xp11.22 deletion
X-linked mental retardation (XLMR)	ELK1	Xp11.23 deletion
X-linked mental retardation (XLMR)	SLC38A5	Xp11.23 deletion
X-linked mental retardation (XLMR)	ZNF41	Xp11.3 deletion
X-linked mental retardation (XLMR)	VCX3A	Xp22.31 deletion
X-linked mental retardation (XLMR)	KLF8	Xp11.21 deletion
X-linked mental retardation (XLMR)	GDI1	Xq28 deletion

Phenotype	Genes	Cytogenetics
X-linked mental retardation (XLMR)	NXF5	Xq22.1 deletion
X-linked mental retardation (XLMR)	ZNF261 (ZMYM3)	Xq13.3 deletion
X-linked mental retardation (XLMR)	KIAA2022	Xq13.3 deletion
X-linked mental retardation (XLMR)	ACSL4	Xq22.3 deletion
X-linked mental retardation (XLMR)	PAK3	Xq22.3 deletion
X-linked mental retardation (XLMR)	AGTR2	Xq23 deletion
X-linked mental retardation (XLMR)	ZDHHC9	Xq25 deletion
X-linked mental retardation (XLMR)	FMR2 (AFF2)	Xq28 deletion
X-linked mental retardation (XLMR) (MRX45)	ZNF81	Xp11.23 deletion
X-linked mental retardation (XLMR) (MRX46)	ARHGEF6	Xq26.3 deletion
X-linked mental retardation (XLMR) (MRX46)	ARHGEF6	Xq28 deletion
X-linked mental retardation (XLMR) (MRX58)	TSPAN7 (TM4SF2)	Xp11.4 deletion
X-linked mental retardation (XLMR) (MRX59)	AP1S2	Xp22.2 deletion
X-linked mental retardation (XLMR) (MRX9)	FTSJ1	Xp11.23 deletion
X-linked mental retardation (XLMR) (MRX90)/ Autism	DLG3	Xq13.1 deletion

Phenotype	Genes	Cytogenetics
X-linked mental retardation (XLMR) (MRX91)	ZDHHC15	Xq13.3 deletion
X-linked mental retardation (XLMR) (MRX94)	GRIA3	Xq25 deletion
X-linked mental retardation (XLMR) with short stature, small testes muscle wasting, and tremor	CUL4B	Xq24 deletion
X-linked mental retardation (XLMR), syndromic, Chrystianson type	SLC9A6	Xq26.3 deletion
X-lissencephaly/multifocal spasms (LISX)	DCX	Xq22.3 deletion
XLMR / Snyder-Robinson syndrome	SMS spermine synthase gene	Xp22.11 deletion
XLMR/Autism	JARID1C/JARIDID	Xp11.2/Y11.222 deletion
Xp11.3 deletion with mental retardation(XLMR)	ZNF674	Xp11.3 deletion
Xq/Yq pseudoautosomal		Xq28 deletion

One Baylor Plaza, Nab 2015, Houston, Texas 77030

MEDICAL GENETICS LABORATORIES

SPECIMEN REQUIREMENTS AND TURN AROUND TIME

Blood in both EDTA (purple top) and Na Heparin (Green top) tubes:

- Adult: 10 cc per tube
- Child: 4 cc per tube
- Infant: 2 cc per tube
- Turn around time: 7 to 10 days

SHIPPING:

Medical Genetics Laboratories
Grand Blvd. Receiving Dock
2450 Holcombe Blvd.
Houston, Tx 77021

Prepaid shipping kits are available upon request: Please call MGL at 1-800-411-gene (4363).

MEDICAL GENETICS LABORATORIES

One Baylor Plaza, Nab 2015, Houston, Texas 77030
713-798-6555 •1-800-411-gene (4363)
www.bcmgeneticlabs.org

10-2008

BCM
Baylor College *of* Medicine