UPMC
LIFE CHANGING MEDICINE

UPMC Cytogenetics Laboratory
X High Resolution Microarray

GENES COVERED BY X-HR AND SYNDROMES DETECTED BY X-HR		
Gene	**Location**	**Syndrome**
ABCB7	Xq12-q13	Anemia, sideroblastic, with ataxia
ABCD1	Xq28	Adrenoleukodystrophy
ACSL4	Xq22.3-q23	X-linked mental retardation
AFF2	Xq28	Fragile X E (FRAXE) mental retardation
AGTR2	Xq22-q23	Mental retardation, X-linked
ALAS2	Xp11.21	Anemia, sideroblastic, X-linked
AP1S2	Xp22.2	Mental retardation, X-linked
AR	Xq11.2-q12	Androgen insensitivity syndrome
ARHGEF6	Xq26.3	Mental retardation, X-linked
ARHGEF9	Xq11.1	X-linked mental retardation (XLMR)//Hyperekplexia and epilepsy
ARSE	Xp22.3	Chondrodysplasia punctata, X-linked recessive (CDPX)
ARX	Xp21	Infantile spasm syndrome, X-linked (ISSX), West, Proud, XLAG, Partington, multifocal epilepsy //Early infantile epileptic encephalopathy-1 (EIEE1)(X-linked infantile spasm syndrome-1-ISSX1)
ATP6AP2	Xp11.4	Mental retardation, X-linked, with epilepsy (XMRE)
ATP7A	Xq13.2-q13.3	Menkes disease (MNK) and occipital horn syndrome
ATRX	Xq13.1-q21.1	ATRX, XLMR-Hypotonic facies syndrome, ATR-X, and others
AVPR2	Xq28	Nephrogenic syndrome of inappropriate antidiuresis; NSI; Diabetes insipidus, nephrogenic
BCOR	Xp21.2-p11.4	Microphthalmia syndromic (MCOPS2)
BMP15	Xp11.2	Ovarian dysgenesis 2
BRWD3	Xq21.1	Mental retardation, X-linked 93
BTK	Xq21.33-q22	X-linked agammaglobulinemia
CACNA1F	Xp11.23	Aland Island eye disease
CASK	Xp11.4	X-linked mental retardation with microcephaly & disproportionate pontine and cerebellar hypoplasia (MICPCH) syndrome
CD40LG	Xq26	Immunodeficiency, X-linked, with hyper-IgM
CDKL5	Xp22	Early infantile epileptic encephalopathy-2 (EIEE2)
CHM	Xq21.2	Choroidoremia
CLCN5	Xp11.23-p11.22	Dent disease//Candidate, chloride channel
COL4A5	Xq22.2	Alport syndrome, X-linked
COL4A5, COL4A6	Xq22.3	X-linked Alport plus diffuse leiomyomatosis (ATS-DL)
CSF2RA	Xp22.33	Surfactant metabolism dysfunction, pulmonary, 4
CUL4B	Xq23	CUL4B, Cabezas syndrome, Mental retardation X-L syndromic 15

CYBB	Xp21.1	Chronic granulomatous disease, X-linked
DCX	Xq22.3-q23	X-linked lissencephaly-1 (LISX1)
DIAPH2	Xq21.33	Premature ovarian failure (POF2A)
DKC1	Xq28	Dyskeratosis congenita
DLG3	Xq13.1	Discs large, Drosophila homologue 3/Mental retardation X-linked 90
DMD	Xp21.2	Duchenne muscular dystrophy/ Becker muscular dystrophy
EBP	Xp11.23	Chondrodysplasia punctata, X-linked dominant
ED1	Xq12-q13.1	Hypohydrotic ectodermal dysplasia (EDA)
EFHC2	Xp11.3	Juvenile myoclonic epilepsy(JME)
EFNB1	Xq12	Craniofrontonasal dysplasia
EMD	Xq28	Emery-Dreifuss muscular dystrophy
F8	Xq28	Hemophilia A
F9	Xq27.1-q27.2	Hemophilia B
FAM123B/WTX	Xq11.2	Osteopathia striata with cranial sclerosis
FAM58A	Xq28	STAR syndrome
FANCB	Xp22.2	Fanconi anemia, complementation group B
FGD1	Xp11.21	Faciogenital dysplasia
FHL1	Xq26	Cardiac Involvement, Scapuloperoneal myopathy (X-linked dominant)
FLNA	Xq28	Heterotopia, periventricular, X-linked dominant/ Otopalatodigital syndrome type 1 (OPD)/multifocal epilepsy in females//Melnick-Needles syndrome; BPNH; OPD; Multifocal epilepsy in females
FMR1	Xq27.3	Fragile-X mental retardation syndrome (FMR1)//Fragile X-Syndrome (epilepsy)
FOXP3	Xp11.23	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
FRMD7	Xq26.2	Nystagmus 1, congenital, X-linked
G6PD	Xq28	G6PD deficiency
GATA1	Xp11.23	Dyserythropoietic anemia with thrombocytopenia
GDI1	Xq28	X-linked mental retardation (XLMR)
GJB1	Xq13.1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1
GK	Xp21.3	Glycerol kinase deficiency (GKD)
GLA	Xq22.1	Fabry disease
GPC3	Xq26.1	Simpson-Golabi-Behmel syndrome type 1;(SGBS1)
GPC4	Xq26.1	Simpson-Golabi-Behmel syndrome type 1; Wilms'tumor
GPR143	Xp22.2	Ocular albinism, type I, Nettleship-Falls type

GRIA3	Xq25-q26	X-linked mental retardation (XLMR) (MRX94)//Candidate, glutamate receptor
GRPR	Xp22.2-p22.13	Autism
HSD17B10	Xp11.22	17-beta-hydroxysteroid dehydrogenase X deficiency
HUWE1	Xp11.22	Mental Retardation, X-linked
IDS	Xq28	Mucopolysaccharidosis type II (MPS2)/Hunter syndrome
IGBP1	Xq13.1-q13.3	Cardiac Involvement, Agenesis of the corpus callosum with MR and ocular coloboma
IKBKG	Xq28	Ectodermal dysplasia, hypohidrotic, with immune deficiency
IL1RAPL1	Xp22.1-p21.3	Autism
IL2RG	Xq13.1	Severe combined immunodeficiency, X-linked
KAL1	Xp22.32	Kallmann syndrome 1
KDM5C	Xp11.22-p11.21	Mental retardation, X-linked, syndromic, JARID1C-related
KDM6A	Xp11.3	Kabuki syndrome 2
L1CAM	Xq28	Hydrocephalus
LAMP2	Xq24	Danon disease
MAMLD1	Xq28	Hypospadias 2, X-linked
MAOA	Xp11.3	Brunner syndrome
MBTPS2	Xp22.12	Ichthyosis follicularis, atrichia, and photophobia
MECP2	Xq28	MECP2 male duplication syndrome/Rett syndrome (RTT); MECP2 deletion
MED12	Xq13	Opitz-Kaveggia syndrome
MID1	Xp22	Opitz BBB syndrome
MTM1	Xq28	Myotubular myopathy, X-linked
Multiple	Xp22.2	Microphthalmia 7 with linear skin defects
Multiple	Xp11.22	Xp11.22-linked mental retardation
Multiple	Xp11.3	Xp11.3 Microdeletion
Multiple	Xp11.1-p21.2	Xp11.4-p21.2 Contiguous gene deletion
NDP	Xp11.4	Norrie disease
NDUFA1	Xq24	Mitochondrial complex I deficiency
NHS	Xp22.13	Nance-Horan syndrome
NLGN3	Xq13.1	Autism / Asperger syndrome-1, susceptibility
NLGN4X	Xp22.32-p22.31	Autistic features, X-linked, susceptibility to, 1

NR0B1/DAX1	Xp21.2	Adrenal hypoplasia congenita (AHC)/Dosage sensitive sex reversal
NSDHL	Xq28	CHILD syndrome/CK syndrome
NXF5	Xq22.1	X-Linked Mental Retardation (XLMR)
NYX	Xp11.4	Night blindness, congenital stationary, type 1A
OCRL	Xq25-26	Lowe syndrome/Dent disease
OFD1	Xp22	Oro-facio-digital 1 (OFD1)/Joubert syndrome 10
OPHN1	Xq12	Cerebellar hypoplasia//Syndromic X-linked mental retardation with epilepsy, rostral ventricular enlargement and cerebellar hypoplasia
OPN1LW	Xq28	Colorblindness, protan ; Blue-cone monochromacy,
OPN1MW	Xq28	Colorblindness, deutan ; Blue-cone monochromacy
OTC	Xp21.1	Ornithine transcarbamylase deficiency (OTC)
PAK3	Xq22.3	X-linked mental retardation 30
PCDH19	Xq22.1	Epileptic encephalopathy, early infantile, 9
PDHA1	Xp22.12	Leigh syndrome, X-linked
PGK1	Xq21.1	Phosphoglycerate kinase 1 deficiency
PHEX	Xp22.11	hosphatemic rickets, X-linked dominant
PHF6	Xq26.3	Borjeson-forssman-lehmann syndrome (epilepsy)
PHF8	Xp11.22	Siderius type X-linked mental retardation
PHKA1	Xq12-q13	Muscle glycogenosis
PHKA2	Xp22.2-p22.1	Glycogenosis, X-linked hepatic, type I (3)
PLP1	Xq22.2	Pelizaeus-Merzbacher
PORCN	Xp11.23	Focal dermal hypoplasia/Goltz
POU3F4	Xq21.1	Deafness, X-linked 2 (DFNX2)
PQBP1	Xp11.23	Renpenning syndrome 1 (RENS1)/Sutherland-Haan XLMR syndrome/Golabi-Ito-Hall syndrome
PRPS1	Xq22.3	Deafness, X-linked (DFNX1), Arts syndrome
PTCHD1	Xp22.11	Autism
RP2	Xp11.4-p11.21	X-linked retinitis pigmentosa
RPGR	Xp11.4	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness
RPL10	Xq28	Autism
RPS6KA3	Xp22.2-p22.1	Coffin-Lowry syndrome (CLS)
RS1	Xp22.13	Retinoschisis 1, X-Linked, Juvenile; Rs1

SH2D1A	Xq25-q26	X-linked lymphoproliferative disease (XLP)
SHROOM4	Xp11.22	Stocco dos Santos XLMR syndrome
SLC16A2	Xq13.2	Allan-Herndon-Dudley syndrome
SLC6A8	Xq28	Creatine deficiency syndrome /X-linked mental retardation (XLMR)//X-linked mental retardation with seizures
SLC9A6	Xq26.3	X-linked mental retardation (XLMR), syndromic, Chrystianson type//X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome
SMC1A	Xp11.22-p11.21	Cornelia de Lange syndrome (CDLS), X-linked
SMPX	Xp22.12	Deafness, X-linked 4 (DFNX4)
SMS	Xp22.1	XLMR / Snyder-Robinson syndrome
SOX3	Xq27.1	Mental retardation X-linked with isolated growth hormone deficiency (MRGH)
SRPX2	Xq21.33-q23	Perisylvian polymicrogyria (Polymicrogyria, bilateral perisylvian; BPP//Rolandic epilepsy, mental retardation, and speech dyspraxia, X-linked (RESDX)
STS	Xp22.32	Ichthyosis, X-linked (steroid sulfatase deficiency)
SYN1	Xp11.23	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
SYP	Xp11.23-p11.22	Mental retardation, X-linked, with or without epilepsy
TAZ	Xq28	Barth syndrome
TBX22	Xq21.1	Cleft palate with or without ankyloglossia
TIMM8A	Xq22.1	Mohr-Tranebjaerg syndrome
TRAPPC2	Xp22	Spondyloepiphyseal dysplasia tarda
TSPAN7	Xp11.4	X-linked mental retardation (XLMR) (MRX58)
UBE2A	Xq24-q25	Mental retardation, X-linked syndromic
UBQLN2	Xp11.21	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
UPF3B	Xq25-q26	Mental retardation, X-linked, syndromic 14
VBP1	Xq28	Von Hippel-Lindau syndrome
VCX3A	Xp22	X-linked mental retardation (XLMR)
WAS	Xp11.4-p11.21	Wiskott-Aldrich syndrome
XIAP	Xq25	Lymphoproliferative syndrome, X-linked, 2
XIST	Xq13.2	X-inactivation, familial skewed, XIST deficiency
XK	Xp21.1	McLeod syndrome (3); McLeod syndrome with neuroacanthosis (3)]
YIPF6	Xq12	severe psychomotor retardation, lack of expressive language, and seizures
ZCCHC12	Xq24	X-linked mental retardation (XLMR)
ZDHHC15	Xq13.3	X-linked mental retardation (XLMR) (MRX91)

ZDHHC9	Xq26.1	X-linked mental retardation (XLMR)
ZIC3	Xq26.2	X-linked heterotaxy, ZIC3
ZMYM3	Xq13.1	X-linked mental retardation (XLMR)
ZNF41	Xp11.23	X-Linked Mental Retardation (XLMR) (MRX89)
ZNF674	Xp11.2-p11.3	Xp11.3 deletion with mental retardation(XLMR)
ZNF81	Xp11.23	X-linked mental retardation (XLMR) (MRX45)