UPMC
LIFECHANGINGMEDICINE

PITTSBURGH CYTOGENETICS LABORATORY

GENES COVERED BY X-HR AND SYNDROMES DETECTED BY X-HR

#	GENES	LOCI	DISORDERS
1	ABCB7	Xq12-q13	Anemia, sideroblastic, with ataxia
2	ABCD1	Xq28	Adrenoleukodystrophy
3	ACSL4	Xq22.3-q23	X-linked mental retardation
4	AFF2	Xq28	Fragile X E (FRAXE) mental retardation
5	AGTR2	Xq22-q23	Mental retardation, X-linked
6	ALAS2	Xp11.21	Anemia, sideroblastic, X-linked
7	AP1S2	Xp22.2	Mental retardation, X-linked
8	AR	Xq11.2-q12	Androgen insensitivity syndrome
9	ARHGEF6	Xq26.3	Mental retardation, X-linked
10	ARHGEF9	Xq11.1	X-linked mental retardation (XLMR)//Hyperekplexia and epilepsy
11	ARSE	Xp22.3	Chondrodysplasia punctata, X-linked recessive (CDPX)
12	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)
13	ATP6AP2	Xp11.4	Mental retardation, X-linked, with epilepsy (XMRE)
14	ATP7A	Xq13.2-q13.3	Menkes disease (MNK) and occipital horn syndrome
15	ATRX	Xq13.1-q21.1	ATRX, XLMR-Hypotonic facies syndrome, ATR-X, and others
16	AVPR2	Xq28	Nephrogenic syndrome of inappropriate antidiuresis; NSI; Diabetes insipidus, nephrogenic
17	BCOR	Xp21.2-p11.4	Microphthalmia syndromic (MCOPS2)
18	BMP15	Xp11.2	Ovarian dysgenesis 2
19	BRWD3	Xq21.1	Mental retardation, X-linked 93
20	BTK	Xq21.33-q22	X-linked agammaglobulinemia
21	CACNA1F	Xp11.23	Aland Island eye disease
22	CASK	Xp11.4	X-linked mental retardation with microcephaly & disproportionate pontine and cerebellar hypoplasia (MICPCH) syndrome
23	CD40LG	Xq26	Immunodeficiency, X-linked, with hyper-IgM
24	CDKL5	Xp22	Early infantile epileptic encephalopathy-2 (EIEE2)
25	CHM	Xq21.2	Choroidoremia
26	CLCN5	Xp11.23-p11.22	Dent disease//Candidate, chloride channel
27	COL4A5	Xq22.2	Alport syndrome, X-linked
28	COL4A5, COL4A6	Xq22.3	X-linked Alport plus diffuse leiomyomatosis (ATS-DL)
29	CSF2RA	Xp22.33	Surfactant metabolism dysfunction, pulmonary, 4
30	CUL4B	Xq23	CUL4B, Cabezas syndrome, Mental retardation X-L syndromic 15
31	CYBB	Xp21.1	Chronic granulomatous disease, X-linked

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32	DCX	Xq22.3-q23	X-linked lissencephaly-1 (LISX1)
33	DIAPH2	Xq21.33	Premature ovarian failure (POF2A)
34	DKC1	Xq28	Dyskeratosis congenita
35	DLG3	Xq13.1	Discs large, Drosophila homologue 3/Mental retardation X-linked 90
36	DMD	Xp21.2	Duchenne muscular dystrophy/ Becker muscular dystrophy
37	EBP	Xp11.23	Chondrodysplasia punctata, X-linked dominant
38	ED1	Xq12-q13.1	Hypohydrotic ectodermal dysplasia (EDA)
39	EFHC2	Xp11.3	Juvenile myoclonic epilepsy(JME)
40	EFNB1	Xq12	Craniofrontonasal dysplasia
41	EMD	Xq28	Emery-Dreifuss muscular dystrophy
42	F8	Xq28	Hemophilia A
43	F9	Xq27.1-q27.2	Hemophilia B
44	FAM123B/WTX	Xq11.2	Osteopathia striata with cranial sclerosis
45	FAM58A	Xq28	STAR syndrome
46	FANCB	Xp22.2	Fanconi anemia, complementation group B
47	FGD1	Xp11.21	Faciogenital dysplasia
48	FHL1	Xq26	Cardiac Involvement, Scapuloperoneal myopathy (X-linked dominant)
49	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
50	FMR1	Xq27.3	Fragile-X mental retardation syndrome (FMR1)//Fragile X-Syndrome (epilepsy)
51	FOXP3	Xp11.23	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked
52	FRMD7	Xq26.2	Nystagmus 1, congenital, X-linked
53	G6PD	Xq28	G6PD deficiency
54	GATA1	Xp11.23	Dyserythropoietic anemia with thrombocytopenia
55	GDI1	Xq28	X-linked mental retardation (XLMR)
56	GJB1	Xq13.1	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1
57	GK	Xp21.3	Glycerol kinase deficiency (GKD)
58	GLA	Xq22.1	Fabry disease
59	GPC3	Xq26.1	Simpson-Golabi-Behmel syndrome type 1;(SGBS1)
60	GPC4	Xq26.1	Simpson-Golabi-Behmel syndrome type 1; Wilms'tumor
61	GPR143	Xp22.2	Ocular albinism, type I, Nettleship-Falls type
62	GRIA3	Xq25-q26	X-linked mental retardation (XLMR) (MRX94)//Candidate, glutamate receptor
63	GRPR	Xp22.2-p22.13	Autism

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64	HSD17B10	Xp11.22	17-beta-hydroxysteroid dehydrogenase X deficiency
65	HUWE1	Xp11.22	Mental Retardation, X-linked
66	IDS	Xq28	Mucopolysaccharidosis type II (MPS2)/Hunter syndrome
67	IGBP1	Xq13.1-q13.3	Cardiac Involvement, Agenesis of the corpus callosum with MR and ocular coloboma
68	IKBKG	Xq28	Ectodermal dysplasia, hypohidrotic, with immune deficiency
69	IL1RAPL1	Xp22.1-p21.3	Autism
70	IL2RG	Xq13.1	Severe combined immunodeficiency, X-linked
71	KAL1	Xp22.32	Kallmann syndrome 1
72	KDM5C	Xp11.22-p11.21	Mental retardation, X-linked, syndromic, JARID1C-related
73	KDM6A	Xp11.3	Kabuki syndrome 2
74	L1CAM	Xq28	Hydrocephalus
75	LAMP2	Xq24	Danon disease
76	MAMLD1	Xq28	Hypospadias 2, X-linked
77	MAOA	Xp11.3	Brunner syndrome
78	MBTPS2	Xp22.12	Ichthyosis follicularis, atrichia, and photophobia
79	MECP2	Xq28	MECP2 male duplication syndrome/Rett syndrome (RTT); MECP2 deletion
80	MED12	Xq13	Opitz-Kaveggia syndrome
81	MID1	Xp22	Opitz BBB syndrome
82	MTM1	Xq28	Myotubular myopathy, X-linked
83	Multiple	Xp22.2	Microphthalmia 7 with linear skin defects
84	Multiple	Xp11.22	Xp11.22-linked mental retardation
85	Multiple	Xp11.3	Xp11.3 Microdeletion
86	Multiple	Xp11.1-p21.2	Xp11.4-p21.2 Contiguous gene deletion
87	NDP	Xp11.4	Norrie disease
88	NDUFA1	Xq24	Mitochondrial complex I deficiency
89	NHS	Xp22.13	Nance-Horan syndrome
90	NLGN3	Xq13.1	Autism / Asperger syndrome-1, susceptibility
91	NLGN4X	Xp22.32-p22.31	Autistic features, X-linked, susceptibility to, 1
92	NR0B1/DAX1	Xp21.2	Adrenal hypoplasia congenita (AHC)/Dosage sensitive sex reversal
93	NSDHL	Xq28	CHILD syndrome/CK syndrome
94	NXF5	Xq22.1	X-Linked Mental Retardation (XLMR)
95	NYX	Xp11.4	Night blindness, congenital stationary, type 1A
96	OCRL	Xq25-26	Lowe syndrome/Dent disease
97	OFD1	Xp22	Oro-facio-digital 1 (OFD1)/Joubert syndrome 10

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98	OPHN1	Xq12	Cerebellar hypoplasia//Syndromic X-linked mental retardation with epilepsy, rostral ventricular enlargement and cerebellar hypoplasia
99	OPN1LW	Xq28	Colorblindness, protan ; Blue-cone monochromacy,
100	OPN1MW	Xq28	Colorblindness, deutan ; Blue-cone monochromacy
101	OTC	Xp21.1	Ornithine transcarbamylase deficiency (OTC)
102	PAK3	Xq22.3	X-linked mental retardation 30
103	PCDH19	Xq22.1	Epileptic encephalopathy, early infantile, 9
104	PDHA1	Xp22.12	Leigh syndrome, X-linked
105	PGK1	Xq21.1	Phosphoglycerate kinase 1 deficiency
106	PHEX	Xp22.11	hosphatemic rickets, X-linked dominant
107	PHF6	Xq26.3	Borjeson-forssman-lehmann syndrome (epilepsy)
108	PHF8	Xp11.22	Siderius type X-linked mental retardation
109	PHKA1	Xq12-q13	Muscle glycogenosis
110	PHKA2	Xp22.2-p22.1	Glycogenosis, X-linked hepatic, type I (3)
111	PLP1	Xq22.2	Pelizaeus-Merzbacher
112	PORCN	Xp11.23	Focal dermal hypoplasia/Goltz
113	POU3F4	Xq21.1	Deafness, X-linked 2 (DFNX2)
114	PQBP1	Xp11.23	Renpenning syndrome 1 (RENS1)/Sutherland-Haan XLMR syndrome/Golabi-Ito-Hall syndrome
115	PRPS1	Xq22.3	Deafness, X-linked (DFNX1), Arts syndrome
116	PTCHD1	Xp22.11	Autism
117	RP2	Xp11.4-p11.21	X-linked retinitis pigmentosa
118	RPGR	Xp11.4	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness
119	RPL10	Xq28	Autism
120	RPS6KA3	Xp22.2-p22.1	Coffin-Lowry syndrome (CLS)
121	RS1	Xp22.13	Retinoschisis 1, X-Linked, Juvenile; Rs1
122	SH2D1A	Xq25-q26	X-linked lymphoproliferative disease (XLP)
123	SHROOM4	Xp11.22	Stocco dos Santos XLMR syndrome
124	SLC16A2	Xq13.2	Allan-Herndon-Dudley syndrome
125	SLC6A8	Xq28	Creatine deficiency syndrome /X-linked mental retardation (XLMR)//X-linked mental retardation with seizures
126	SLC9A6	Xq26.3	X-linked mental retardation (XLMR), syndromic, Chrystianson type//X-linked mental retardation, microcephaly, epilepsy, and ataxia, a phenotype mimicking Angelman syndrome
127	SMC1A	Xp11.22-p11.21	Cornelia de Lange syndrome (CDLS), X-linked
128	SMPX	Xp22.12	Deafness, X-linked 4 (DFNX4)

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129	SMS	Xp22.1	XLMR / Snyder-Robinson syndrome
130	SOX3	Xq27.1	Mental retardation X-linked with isolated growth hormone deficiency (MRGH)
131	SRPX2	Xq21.33-q23	Perisylvian polymicrogyria (Polymicrogyria, bilateral perisylvian; BPP//Rolandic epilepsy, mental retardation, and speech dyspraxia, X-linked (RESDX)
132	STS	Xp22.32	Ichthyosis, X-linked (steroid sulfatase deficiency)
133	SYN1	Xp11.23	Epilepsy, X-linked, with variable learning disabilities and behavior disorders
134	SYP	Xp11.23-p11.22	Mental retardation, X-linked, with or without epilepsy
135	TAZ	Xq28	Barth syndrome
136	TBX22	Xq21.1	Cleft palate with or without ankyloglossia
137	TIMM8A	Xq22.1	Mohr-Tranebjaerg syndrome
138	TRAPPC2	Xp22	Spondyloepiphyseal dysplasia tarda
139	TSPAN7	Xp11.4	X-linked mental retardation (XLMR) (MRX58)
140	UBE2A	Xq24-q25	Mental retardation, X-linked syndromic
141	UBQLN2	Xp11.21	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia
142	UPF3B	Xq25-q26	Mental retardation, X-linked, syndromic 14
143	VBP1	Xq28	Von Hippel-Lindau syndrome
144	VCX3A	Xp22	X-linked mental retardation (XLMR)
145	WAS	Xp11.4-p11.21	Wiskott-Aldrich syndrome
146	XIAP	Xq25	Lymphoproliferative syndrome, X-linked, 2
147	XIST	Xq13.2	X-inactivation, familial skewed, XIST deficiency
148	XK	Xp21.1	McLeod syndrome (3); McLeod syndrome with neuroacanthosis (3)]
149	YIPF6	Xq12	severe psychomotor retardation, lack of expressive language, and seizures
150	ZCCHC12	Xq24	X-linked mental retardation (XLMR)
151	ZDHHC15	Xq13.3	X-linked mental retardation (XLMR) (MRX91)
152	ZDHHC9	Xq26.1	X-linked mental retardation (XLMR)
153	ZIC3	Xq26.2	X-linked heterotaxy, ZIC3
154	ZMYM3	Xq13.1	X-linked mental retardation (XLMR)
155	ZNF41	Xp11.23	X-Linked Mental Retardation (XLMR) (MRX89)
156	ZNF674	Xp11.2-p11.3	Xp11.3 deletion with mental retardation(XLMR)
157	ZNF81	Xp11.23	X-linked mental retardation (XLMR) (MRX45)

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