

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chr10	83,625,077	84,735,341	NRG3	10q22	10q22-23 Deletion	605533	Brain
chr10	87,349,292	88,116,230	GRID1	10q22	10q22-23 Deletion	610659	Brain
chr12	65,029,066	65,359,020	GRIP1	12q14	12q14.3 Deletion	604597	Brain and dysmorphic
chr15	30,110,018	30,248,527	CHRNA7	15q13	15q13.3 MicroDeletion Syndrome	118511	Schizophrenia/Epilepsy
chr2	61,136,239	61,673,319	MicroDeletion Region	2p15	2p15p16.1 MicroDeletion	602559	Multiple
chr3	197,951,312	198,043,751	PAK2	3q29	3q29 MicroDeletion Syndrome	605022	Multiple
chr9	139,725,238	139,850,397	EHMT1	9q34	9q34.3 Syndrome	607001	Multiple
chrX	54,488,612	54,539,324	FGD1	Xp11	Aarskog-Scott Syndrome	300564	Multiple
chrX	152,643,530	152,663,374	ABCD1	Xq28	Adrenoleukodystrophy	300475	Late onset brain disease
chr5	126,140,732	126,200,608	LMNB1	5q23	Adult-onset Autosomal Dominant Leukodystrophy	150340	Late onset brain disease
chr20	56,838,189	56,929,641	GNAS	20q13	Albright Hereditary Osteodystrophy	139320	Mild DD and DF
chr15	20,556,790	20,585,849	NIPA2	15q11	Angelman / Prader Willi	608146	Mental retardation
chr15	20,594,720	20,637,877	NIPA1	15q11	Angelman / Prader Willi	608146	Mental retardation
chr15	21,361,547	21,407,800	MKRN3	15q11	Angelman / Prader Willi	603586	Mental retardation
chr15	21,429,787	21,452,081	MAGEL2	15q11	Angelman / Prader Willi	605283	Mental retardation
chr15	22,619,887	22,774,822	SNRPN	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,713,304	22,720,548	PWS_ICRegion	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,741,227	22,784,821	SNURF	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,747,536	22,751,866	PWS_ICRegion	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,751,228	22,774,822	SNRPN	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,768,233	22,793,232	SNORD107/64/108	15q11	Angelman / Prader Willi	182279	Mental retardation
chr15	22,835,000	23,010,100	SnoRNA	15q11	Angelman / Prader Willi	601491	Mental retardation
chr15	23,064,582	23,084,648	SNORD109B	15q11	Angelman / Prader Willi		Mental retardation
chr15	21,471,646	21,493,542	NDN	15q11	Angelman / Prader Willi	602117	Mental retardation
chr10	64,241,763	64,246,133	EGR2	10q21	Autism	129010	Autism
chr15	23,463,512	23,669,962	ATP10A	15q12	Autism	605855	Autism
chr15	24,339,787	24,767,950	GABRB3	15q12	Autism	137192	Autism
chr1	229,829,184	230,243,641	DISC1	1q42	Autism	605210	Autism
chr22	49,459,936	49,518,507	SHANK3	22q13	Autism	606230	Autism
chr2	50,000,992	51,113,178	NRXN1	2p16	Autism	600565	Autism
chr2	162,189,192	162,548,489	SLC4A10	2q24	Autism	605556	Autism
chr7	116,099,695	116,225,676	MET	7q31	Autism	164860	Autism
chr7	121,746,687	122,313,750	CADPS2	7q31	Autism	609978	Autism
chr7	145,444,386	147,749,019	CNTNAP2	7q35-36	Autism	276000	Autism
chr7	154,943,160	154,950,579	EN2	7q36	Autism	131310	Autism
chrX	5,818,085	6,155,888	NLGN4X	Xp22	Autism	300427	Autism
chrX	70,281,436	70,307,776	NLGN3	Xq13	Autism	300336	Autism
chr3	2,115,550	3,074,645	CNTN4	3p25	Autism Spectrum Disorder	607280	Autism
chr16	56,211,151	56,256,445	GPR56	16q13	Bilateral Frontoparietal Polymicrogyria	604110	Severe mental
chrX	133,335,008	133,390,488	PHF6	Xq26	Borjeson-Forsman-Lehmann Syndrome	300414	Severe mental and physical
chrX	43,400,353	43,491,012	MAOA	Xp11	Brunner Syndrome	309850	Mild mental – aggressive
chr8	61,753,893	61,942,017	CHD7	8q12	Charge Syndrome	601653	Deaf/Blind ++
chr18	70,416,108	70,906,616	ZNF407	18q22	Chromosome 18q Deletion Syndrome		MCA
chr6	1,555,680	1,559,128	FKHL7(FOXC1)	6p24	Chromosome 6p24 Deletion Syndrome	609010	MCA
chr9	831,690	959,090	DMRT1	9p24	Chromosome 9p Deletion Syndrome	158170	MCA
chr9	1,039,858	1,047,552	DMRT2	9p24	Chromosome 9p Deletion Syndrome	604935	MCA
chrX	20,077,950	20,194,671	RPS6KA3	Xp22	Coffin-Lowry Syndrome	300075	MCA
chrX	152,606,586	152,615,234	SLC6A8	Xq28	Creatine Deficiency Syndrome	300036	Mental retardation
chr16	33,686,002	33,694,027	SLC6A8	16p11	Creatine Deficiency Syndrome / X-linked Mental Retardation		Mental retardation
chr16	34,877,510	35,027,509		16p11	Creatine Deficiency Syndrome / X-linked Mental Retardation		Mental retardation
chr5	1,306,287	1,348,162	TERT	5p15	Cri du Chat	187270	Severe Mental and Physical
chr3	148,586,527	148,607,097	ZIC4	3q24	Dandy Walker	608948	Severe mental
chr3	148,609,871	148,617,196	ZIC1	3q24	Dandy Walker	600470	Severe mental
chr22	17,698,224	17,815,220	HIRA	22q11	diGeorge 1	600637	MCA
chr22	18,124,226	18,151,110	TBX1	22q11	diGeorge 1	602054	MCA
chr22	19,601,714	19,638,035	CRKL	22q11	diGeorge 1	602007	MCA
chr10	8,136,673	8,157,170	GATA3	10p14	diGeorge 2	131320	MCA
chr8	8,679,409	8,788,541	MFHAS1	8p23	diGeorge Syndrome	605352	MCA
chr8	11,571,877	11,654,918	GATA4	8p23	diGeorge Syndrome	600576	MCA
chr21	34,810,654	34,909,252	DSCR1(RCAN1)	21q22	Downs Syndrome Critical Region	602917	MCA
chr21	35,081,968	36,278,917	RUNX1	21q22	Downs Syndrome Critical Region	151385	MCA
chr21	37,517,596	37,561,703	DSCR3	21q22	Downs Syndrome Critical Region	605298	MCA

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chr21	37,661,729	37,809,549	DYRK1A	21q22	Downs Syndrome Critical Region	600855	MCA
chr18	44,824,170	45,250,999	DYM	18q21	Dyggve-Melchior-Clausen Disease	607461	Severe Mental and Physical
chr2	15,998,134	16,004,579	MYCN	2q24	Feingold	164840	Hands, feet, mental
chrX	146,801,201	146,840,303	FMR1	Xq27	Fragile X Syndrome	309550	Severe Mental retardation
chrX	147,389,831	147,889,899	FMR2(AFF2)	Xq28	Fragile X Syndrome	309548	Severe Mental retardation
chrX	30,581,461	30,658,645	GK	Xp21	Glycerol Kinase Deficiency	300474	Mild mental and metabolic
chr7	41,967,073	42,243,900	GLI3	7p14	Greig Cephaloplysindactyly Syndrome	165240	Physical and mild mental
chrX	153,230,091	153,256,123	FLNA	Xq28	Heterotopia, Periventricular, X-linked Dominant Syndrome	300017	Epilepsy
chr2	45,022,365	45,025,945	SIX3	2p21	Holoprosencephaly 2	603714	Brain defect
chr18	3,402,072	3,448,406	TGIF1	18p31	Holoprosencephaly Type 4	602630	Brain defect
chr13	99,432,320	99,437,019	ZIC2	13q32	Holoprosencephaly 5	603073	Brain defect
chr21	44,256,634	44,350,858	TMEM1	21q22	Holoprosencephaly 1	602103	Brain defect
chr21	46,432,788	46,473,119	LSS	21q22	Holoprosencephaly 1	600909	Brain defect
chr7	155,288,319	155,297,728	SHH	7q36	Holoprosencephaly 3	600725	Brain defect
chr9	97,245,085	97,319,068	PTCH1	9q22	Holoprosencephaly 7	601309	Brain defect
chr2	121,210,293	121,466,699	GLI2	2q14	Holoprosencephaly 9	165230	Brain defect
chr5	171,221,161	171,366,482	FBXW11	5q35	Holoprosencephaly and Preaxial Polydactyly	605651	Brain defect
chrX	152,780,581	152,804,778	L1CAM	Xq28	HSAS, MASA, CRASH Syndromes	300371	Physical and mental
chrX	148,368,203	148,423,359	IDS	Xq28	Hunter Syndrome, Mucopolysaccharidosis Type 2	309900	Progressive MCA
chr2	172,349,126	172,458,979	SLC25A12	2q31	Hypomyelination, Global, Cerebral	603667	Mental retardation
chr12	86,966,921	87,060,124	CEP290	12q21	Joubert 5	610142	Brain defect
chr6	135,646,817	135,860,576	AHI1	6q23	Joubert Syndrome	608894	Brain defect
chr8	11,737,443	11,763,055	CTSB	8p23	Kabuki Syndrome	116810	Severe Mental and Physical
chr8	118,880,787	119,193,239	EXT1	8q23	Langer-Giedion Syndrome	150230	Mild DD and PD
chrX	133,421,923	133,462,362	HPRT1	Xq26	Lesch-Nyhan Syndrome	308000	Progressive MD and gout
chr11	67,130,983	67,136,581	NDUFV1	11q13	Leukodystrophy	161015	Progressive MD
chr7	102,899,473	103,417,198	RELN	7q22	Lissencephaly with Cerebellar Hypoplasia	600514	Severe Mental and Physical
chr3	30,622,998	30,710,637	TGFBR2	3p24	Loeys-Dietz Syndrome	190182	Severe Mental and Physical – can be reduced following surgery
chr9	100,907,233	100,956,294	TGFBR1	9q22	Loeys-Dietz Syndrome	190181	Severe Mental and Physical – can be reduced following surgery
chrX	128,501,933	128,554,211	OCRL	Xq25	Lowe	300535	Eye and Brain defects
chr8	82,732,751	82,761,115	IMPA1	8q21	Manic Depressive Psychosis	602064	Manic Depressive Psychosis
chr14	100,252,981	100,281,225	DLK1	14q32	Maternal UPD	176290	Growth retardation and minor dysmorphic
chr22	28,329,565	28,424,583	NF2	22q12	Meningioma / NF2	607379	Mental retardation and microcephaly
chrX	77,042,046	77,200,000	ATP7A	Xq21	Menkes Disease	300011	Copper deficiency leading to mental and physical
chr10	89,613,175	89,718,512	PTEN	10q23	Mental Retardation	612242	Mental retardation and physical
chr15	23,123,488	23,245,220	UBE3A	15q11	Mental Retardation	601623	Mental retardation and physical
chr1	233,597,351	233,678,903	TBCE	1q42	Mental Retardation	604934	Mental retardation and physical
chr3	46,594,217	46,598,956	TDGF1	3p21	Mental Retardation	187395	Mental retardation and minor dysmorphic
chr7	142,136,904	142,140,495	PRSS2	7q34	Mental Retardation	601564	Mental retardation and physical
chr8	1,426,975	1,654,048	DLGAP2	8p23	Mental Retardation	605438	Mental retardation and physical
chr16	971,809	976,980	SOX8	16p13	Mental Retardation associated with Alpha Thalaessamia	605923	Mental retardation and alpha thalassaemia
chr6	101,953,675	102,623,474	GRIK2	6q16	Mental Retardation, Autosomal Recessive 6	138244	Mental retardation and physical
chr22	49,410,315	49,413,453	ARSA	22q13	Metachromatic Leukodystrophy	607574	Progressive muscle failure and speech
chr3	182,903,176	182,926,972	SOX2	3q26	Microphthalmia	184429	Brain defect leading to severe developmental delay
chr17	853,509	1,037,366	ABR	17p13	Miller-Dieker Syndrome	600365	Severe Mental and Physical
chr17	1,194,593	1,250,267	YWHAE	17p13	Miller-Dieker Syndrome	605066	Severe Mental and Physical
chr17	1,368,033	1,412,860	PITPNA	17p13	Miller-Dieker Syndrome	600174	Severe Mental and Physical
chr17	2,443,686	2,535,638	PAFAH1B1(LIS1)	17p13	Miller-Dieker Syndrome	601545	Severe Mental and Physical
chr1	68,274,232	68,299,047	DIRAS3	1p31	Monosomy 1p31	605193	Mile DD and PD
chr2	144,862,055	144,994,386	ZEB2	2q22	Mowat-Wilson Syndrome	605802	Mental retardation and physical, required early intervention with speech therapy and physical therapy
chr10	21,110,094	21,503,122	NEBL	10p12	Nebulette	605491	Cardiac, immune and DD
chr17	26,446,121	26,728,821	NF1	17q11	Neurofibromatosis Type 1	162200	Skeletal defects and learning disorders
chr17	27,288,185	27,352,162	JJAZ1(SUZ12)	17q11	Neurofibromatosis Type 1	606245	Skeletal defects and learning
chr17	25,549,032	25,586,831	SLC6A4	17p11	Neuroticism	182138	Behavioural disorder
chr1	61,320,883	61,694,624	NFIA	1p31	NFIA Haploinsufficiency	600727	Mental and physical
chr12	111,340,919	111,432,099	PTPN11	12q24	Noonan 1	176876	Heart, short stature, dysmorphic.

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chr3	12,600,108	12,680,678	RAF1	3p25	Noonan Syndrome 5	164760	Heart, short stature, dysmorphic.
chr2	39,066,469	39,201,067	SOS1	2p22	Noonan Syndrome Type 4	182530	Heart, short stature, dysmorphic.
chrX	43,692,968	43,717,694	NDP	Xp11	Norrie Disease	300658	Eye, ear and some DD
chrX	106,956,116	107,057,077	MID2	Xq22	Opitz / FP Syndrome	300204	MCA
chrX	10,376,166	10,765,667	MID1	Xp22	Opitz Syndrome	300552	MCA
chrX	70,255,131	70,279,029	MED12	Xq13	Opitz-Kaveggia Syndrome	300188	MCA
chrX	38,096,302	38,165,553	OTC	Xp11	Ornithine Transcarbamylase Deficiency	300461	Enzyme deficiency progressive
chrX	102,918,410	102,934,201	PLP1	Xq22	Pelizaeus-Merzbacher Disease	300401	Central nervous system defect
chrX	77,246,425	77,268,980	PGK1	Xp21	Phosphoglycerate Kinase Deficiency	311800	Anaemia and mental disorders
chr18	51,045,967	51,454,183	TCF4	18q21	Pitt-Hopkins Syndrome	602272	Apnea, breathing, mental and physical problems
chr11	44,073,675	44,223,555	EXT2	11p11	Potocki-Shaffer Syndrome	608210	MCA Mental and Physical
chr11	44,242,734	44,288,292	ALX4	11p11	Potocki-Shaffer Syndrome	605420, 608210	MCA Mental and Physical
chr13	65,774,967	66,702,464	PCDH9	13q21	Protocadherin 9 - EFMR Disorder	603581	Mental retardation and Epilepsy
chrX	19,271,972	19,287,886	PDHA1	Xp22	Pyruvate Dehydrogenase Deficiency	300502	Enzyme deficiency progressive
chrX	152,940,458	153,016,323	MECP2	Xq28	Retts Syndrome	300260	Various mental and physical defects.
chrX	99,785,819	99,812,952	SRPX2	Xq22	Rolandic Epilepsy, X-linked Mental Retardation, and Speech Dyspraxia,	300642	Mental and physical
chr16	3,642,941	3,648,097	DNASE1	16p13	Rubinstein Taybi Syndrome	125505	Mental, physical and cancer
chr16	3,717,720	3,870,723	CREBBP	16p13	Rubinstein Taybi Syndrome	600140	Mental, physical and cancer
chr7	19,121,616	19,123,550	TWIST1	7p21	Saethre-Chotzen	601622	Mostly physical, mild mental
chr10	119,291,946	119,299,043	EMX2	10q26	Schizencephaly	600035	Mental and physical
chr2	166,553,916	166,638,395	SCN1A	2q24	SCN1A Related Seizures	182389	Seizures
chr11	70,823,105	70,837,125	DHCR7	11q13	Smith-Lemli-Opitz	602858	mild to severe mental and physical
chr17	17,525,512	17,655,488	RAI1	17p11	Smith-Magenis Syndrome	607642	Physical, behavioural and mental
chr5	176,492,686	176,659,820	NSD1	5q35	Sotos Syndrome	606681	Overgrowth, behavioural and some cancer
chr7	96,140,173	96,177,139	SHFM1	7q21	Split Hand and Foot	183600	Abnormal hand/feet some mental
chr7	96,477,643	96,502,078	DLX5	7q21	Split Hand and Foot	600028	Abnormal hand/feet some mental
chr10	103,103,815	103,307,060	BTRC	10q24	Split Hand and Foot Malformation-3	603482	Abnormal hand/feet some mental
chr10	103,328,629	103,338,004	POLL	10q24	Split Hand and Foot Malformation-3	606343	Abnormal hand/feet some mental
chr10	103,360,412	103,444,733	FBXW4	10q24	Split Hand and Foot Malformation-3	600095	Abnormal hand/feet some mental
chr10	102,976,723	102,978,707	LBX1	10q24	Split Hand and Foot Malformation-3	604255	Abnormal hand/feet some mental
chr2	172,658,454	172,662,647	DLX1	2q31	Split Hand and Foot Malformation-5	600029	Abnormal hand/feet some mental
chr2	172,672,412	172,675,724	DLX2	2q31	Split Hand and Foot Malformation-5	126255	Abnormal hand/feet some mental
chrX	6,461,659	6,463,159	VCX3A	Xp22	Steroid Sulfatase	300533	Physical, behavioural and mental
chrX	7,075,293	7,282,680	STS	Xp22	Steroid Sulfatase	300747	Physical, behavioural and mental
chr12	1,950,490	2,677,376	CACNA1C	12p13	Timothy Syndrome	114205	Heart rhythm defects and Autism like features
chr16	49,727,387	49,742,684	SALL1	16q12	Townes Brocks Syndrome	602218	Various physical defects and some learning disorders
chr8	116,489,900	116,750,429	TRPS1	8q23	Trischororhinophlangeal, Langer-Giedion Syndrome	604386	Bone abnormalities, cancer and some learning disorders
chr9	134,756,557	134,809,841	TSC1	9q34	Tuberous Sclerosis	605284	Abnormal benign tumour growth and learning disorders
chr16	2,038,600	2,078,713	TSC2	16p13	Tuberous Sclerosis, Polycystic Kidney Disease	191902	Abnormal benign tumour growth, kidney and learning disorders
chr7	113,513,618	114,118,328	FOXP2	7q31	Verbal Dyspraxia	605317	Severe speech and language disorder
chr11	32,355,900	32,423,662	WT1	11p13	WAGR Syndrome	607102	Various mental, visual and physical defects.
chr7	73,080,407	73,120,966	ELN	7q11	Williams-Beuren Syndrome	130160	Mental and physical
chr7	73,136,092	73,174,790	LIMK1	7q11	Williams-Beuren Syndrome	194050	Mental and physical
chr7	73,826,245	73,841,595	NCF1	7q11	Williams-Beuren Syndrome	608512	Mental and physical
chr4	1,784,558	1,827,772	LETM1	4p16	Wolf-Hirschhorn Syndrome	604407	Severe to profound mental retardation, microcephaly, seizures, poor muscle and cleft lip and/or cleft palate
chr4	1,842,921	1,953,728	WHSC1	4p16	Wolf-Hirschhorn Syndrome	602952	Severe to profound mental retardation, microcephaly, seizures, poor muscle and cleft lip and/or cleft palate
chr4	1,954,251	1,980,547	WHSC2	4p16	Wolf-Hirschhorn Syndrome	606026	Severe to profound mental retardation, microcephaly, seizures, poor muscle and cleft lip and/or cleft palate
chrX	72,863,307	73,063,491	XIST	Xq13	X Inactivation Specific Transcript	314670	Severe physical and mental
chrX	53,417,795	53,466,343	SMC1L1(SMC1A)	Xp11	X-linked Cornelia de Lange Syndrome	300040	Broad spectrum physical and mental
chrX	153,644,344	153,659,154	DKC1	Xq28	X-linked Dyskeratosis Congenita	300125	Various physical defects and some learning disorders

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chrX	53,474,931	53,478,048	HADH2(HSD17B10)	Xp11	X-linked Mental Retardation	300256	Most common mental retardation phenotype with some dysmorphic features
chrX	38,305,683	38,433,116	TM4SF2(TSPAN7)	Xp11	X-linked Mental Retardation	300096	Most common mental retardation phenotype with some dysmorphic features
chrX	39,795,561	39,921,526	BCOR	Xp11	X-linked Mental Retardation	300485	Most common mental retardation phenotype with some dysmorphic features
chrX	40,325,160	40,350,832	ATP6AP2	Xp11	X-linked Mental Retardation	300566	Most common mental retardation phenotype with some dysmorphic features
chrX	41,264,287	41,667,212	CASK	Xp11	X-linked Mental Retardation	300172	Most common mental retardation phenotype with some dysmorphic features
chrX	47,191,350	47,227,289	ZNF41	Xp11	X-linked Mental Retardation	314995	Most common mental retardation phenotype with some dysmorphic features
chrX	47,316,244	47,364,200	SYN1	Xp11	X-linked Mental Retardation	311040	Most common mental retardation phenotype with some dysmorphic features
chrX	47,379,864	47,394,964	ELK1	Xp11	X-linked Mental Retardation	311040	Most common mental retardation phenotype with some dysmorphic features
chrX	47,581,245	47,666,550	ZNF81	Xp11	X-linked Mental Retardation	314998	Most common mental retardation phenotype with some dysmorphic features
chrX	48,201,871	48,213,509	SLC38A5	Xp11	X-linked Mental Retardation	300649	Most common mental retardation phenotype with some dysmorphic features
chrX	48,219,493	48,229,696	FTSJ1	Xp11	X-linked Mental Retardation	300499	Most common mental retardation phenotype with some dysmorphic features
chrX	48,252,315	48,264,146	PORCN	Xp11	X-linked Mental Retardation	300651	Most common mental retardation phenotype with some dysmorphic features
chrX	48,640,139	48,645,364	PQBP1	Xp11	X-linked Mental Retardation	300463	Most common mental retardation phenotype with some dysmorphic features
chrX	50,351,387	50,573,784	SHROOM4(KIAA1202)	Xp11	X-linked Mental Retardation	300579	Most common mental retardation phenotype with some dysmorphic features
chrX	53,237,378	53,271,329	JARID1C	Xp11	X-linked Mental Retardation	314690	Most common mental retardation phenotype with some dysmorphic features
chrX	53,979,838	54,088,121	PHF8	Xp11	X-linked Mental Retardation	300560	Most common mental retardation phenotype with some dysmorphic features
chrX	56,275,632	56,328,254	KLF8	Xp11	X-linked Mental Retardation	300286	Most common mental retardation phenotype with some dysmorphic features
chrX	24,932,213	24,943,775	ARX	Xp21	X-linked Mental Retardation	300382	Most common mental retardation phenotype with some dysmorphic features
chrX	28,515,480	29,884,757	IL1RAPL1	Xp21	X-linked Mental Retardation	300206	Most common mental retardation phenotype with some dysmorphic features
chrX	79,818,351	79,951,889	BRWD3	Xp21	X-linked Mental Retardation	300553	Most common mental retardation phenotype with some dysmorphic features
chrX	11,039,373	11,051,122	HCCS	Xp22	X-linked Mental Retardation	300056	Most common mental retardation phenotype with some dysmorphic features
chrX	14,771,450	14,801,105	FANCB	Xp22	X-linked Mental Retardation	300515	Most common mental retardation phenotype with some dysmorphic features
chrX	15,753,850	15,783,021	AP1S2	Xp22	X-linked Mental Retardation	300629	Most common mental retardation phenotype with some dysmorphic features
chrX	8,393,346	8,394,346	VCX3A	Xp22	X-linked Mental Retardation		Most common mental retardation phenotype with some dysmorphic features
chrX	21,868,763	21,922,876	SMS	Xp22	X-linked Mental Retardation	300105	Most common mental retardation phenotype with some dysmorphic features
chrX	108,771,220	108,863,275	ACSL4	Xp22	X-linked Mental Retardation	300157	Most common mental retardation phenotype with some dysmorphic features

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chrX	110,074,169	110,350,829	PAK3	Xp22	X-linked Mental Retardation	300142	Most common mental retardation phenotype with some dysmorphic features
chrX	110,423,663	110,541,962	DCX	Xp22-p2	X-linked Mental Retardation	300121	Most common mental retardation phenotype with some dysmorphic features
chrX	69,581,449	69,642,062	DLG3	Xq13	X-linked Mental Retardation	300189	Most common mental retardation phenotype with some dysmorphic features
chrX	70,376,199	70,391,660	ZNF261 (ZMYM3)	Xq13	X-linked Mental Retardation	30061	Most common mental retardation phenotype with some dysmorphic features
chrX	73,870,137	74,061,709	KIAA2022	Xq13	X-linked Mental Retardation	300524	Most common mental retardation phenotype with some dysmorphic features
chrX	74,508,786	74,659,600	ZDHHC15	Xq13	X-linked Mental Retardation	300576	Most common mental retardation phenotype with some dysmorphic features
chrX	100,973,741	100,999,205	NXF5	Xq22	X-linked Mental Retardation	300319	Most common mental retardation phenotype with some dysmorphic features
chrX	115,216,031	115,219,848	AGTR2	Xq23	X-linked Mental Retardation	300034	Most common mental retardation phenotype with some dysmorphic features
chrX	118,592,527	118,602,407	UBE2A	Xq23	X-linked Mental Retardation	312180	Most common mental retardation phenotype with some dysmorphic features
chrX	118,852,017	118,870,996	UPF3B	Xq24	X-linked Mental Retardation	300298	Most common mental retardation phenotype with some dysmorphic features
chrX	118,889,762	118,894,646	NDUFA1	Xq24	X-linked Mental Retardation	300078	Most common mental retardation phenotype with some dysmorphic features
chrX	119,542,474	119,593,712	CUL4B	Xq24	X-linked Mental Retardation	300304	Most common mental retardation phenotype with some dysmorphic features
chrX	122,145,839	122,450,474	GRIA3	Xq24	X-linked Mental Retardation	305915	Most common mental retardation phenotype with some dysmorphic features
chrX	128,766,594	128,805,805	ZDHHC9	Xq25	X-linked Mental Retardation	300646	Most common mental retardation phenotype with some dysmorphic features
chrX	135,575,377	135,691,169	ARHGEF6	Xq26	X-linked Mental Retardation	300267	Most common mental retardation phenotype with some dysmorphic features
chrX	153,279,912	153,283,874	RPL10	Xq28	X-linked Mental Retardation	209850	Most common mental retardation phenotype with some dysmorphic features
chrX	153,318,715	153,325,009	GDI1	Xq28	X-linked Mental Retardation	300104	Most common mental retardation phenotype with some dysmorphic features
chrX	139,411,973	139,415,436	SOX3	Xq27	X-linked Mental Retardation	313430	Most common mental retardation phenotype with some dysmorphic features
chrX	134,895,252	134,957,094	SLC9A6	Xq26	X-linked Mental Retardation Syndrome Chrystianson Type	300231	Mental retardation, small head and speech defect
chrX	73,557,810	73,670,475	SLC16A2	Xq13	X-linked Mental Retardation, Allan-Herndon-Dudley Syndrome	300095	Severe mental and weak muscle tone
chrX	67,179,440	67,570,372	OPHN1	Xq12	X-linked Mental Retardation, with Cerebellar Hypoplasia and Distinctive Facial Appearance	300127	Various physical defects and learning disorders
chrX	18,353,678	18,581,666	CDKL5	Xp22	X-linked Spasms	300203	Severe seizures and developmental delay.
chrX	46,243,490	46,289,820	ZNF674	Xp11	Xp11.3 Deletion	300573	Mental retardation
chrX	46,581,319	46,626,733	RP2	Xp11	Xp11.3 Deletion	300757	Mental retardation
chr19	62,384,680	62,432,350	ZNF264	19q13	Zinc Finger Protein-264	604668	Zinc finger protein - imprinting
chr11	1,975,651	1,980,951	H19_ICRegion	11p15			Imprinting centre
chr14	20,923,198	20,975,244	CHD8	14q11		610528	Growth retardation
chr14	28,306,038	28,308,622	FOXG1B	14q12			Microcephaly and mental retardation
chr14	100,261,144	100,364,369	DLK1&MEG3_ICRegion	14q32		176290	UPD, mental and physical disorders
chr15	49,527,231	49,702,259	DMXL2	15q21		612186	Involved in cortex
chr16	6,009,133	7,702,500	A2BP1	16p13		605104	Mental retardation and epilepsy
chr17	41,217,449	41,268,973	CRHR1	17q21		122561	Mental and eating disorders
chr17	41,327,544	41,461,546	MAPT	17q21		157140	Developmental delay, Alzheimers
chr18	42,798,570	42,820,446	TCEB3C	18q21		609522	Transcription factor - imprinting

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chr18	73,090,996	73,111,084	GALR1	18q23		600377	Behavioural disorders, cancer, Alzheimers link
chr20	56,676,787	56,680,787	GNAS_ICRegion	20q13		603666	Broad spectrum physical and mental and cancer
chr20	56,859,056	56,861,251	GNAS_ICRegion	20q13		139320	Broad spectrum physical and mental and cancer
chr20	56,896,896	56,899,356	GNAS_ICRegion	20q13		139320	Broad spectrum physical and mental and cancer
chr2	239,634,801	239,987,580	HDAC4	2q37		605314	Causes mental retardation and dysmorphism
chr7	94,364,911	94,770,958	PPP1R9A	7q21		602468	Gene involved in brain function
chr7	98,084,545	98,097,116	NPTX2	7q22		600750	Brain and immune function
chr7	129,710,229	129,761,249	CPA4	7q32		607635	Growth retardation and dysmorphic
chr7	129,903,281	129,943,368	MEST	7q32		601029	Growth retardation and minor dysmorphic
chr8	140,683,985	140,794,480	KCNK9	8q24		605874	Known as Birk-Barel Syndrome. Mental retardation and dysmorphism
chrX	1	2,700,000	PAR1	Xp22			Mental and physical
chrX	154,595,003	154,913,754	PAR2	Xq28		300053	Bipolar disorder

**Important:** The testing and interpretation of Sengenics Sdn Bhd DNA analyses are based on the current understanding of the molecular genetics and basis for these conditions. DNA-based testing is highly accurate but due to the specific base pair locations selected for probes, a negative result for any of the syndromes tested cannot eliminate the possibility that the individual carries a mutation at base pair locations not covered by the test. By submitting DNA samples and orders to Sengenics Sdn Bhd the client has agreed to use the DNA-test results and interpretations at its own responsibility and to indemnify, hold harmless and defend Sengenics Sdn Bhd from any claims which may arise due to any non-detection of syndromes.