

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chr11	86,334,369	86,344,081	FZD4	11q14	11q14 MicroDeletion	604579	Eye
chr12	65,029,066	65,359,020	GRIP1	12q14	12q14.3 Deletion	604597	Brain and dysmorphic
chr14	60,045,150	60,048,904	SIX6	14q22	14q22 MicroDeletion	606326	Eye
chr14	60,246,009	60,260,792	SIX4	14q22	14q22 MicroDeletion	606342	Eye
chr14	56,337,178	56,346,937	OTX2	14q22	14q22 MicroDeletion, Microphthalmia, Syndromic 5	600037	Eye
chr14	53,486,207	53,493,362	BMP4	14q22	14q22 MicroDeletion, Orofacial Cleft 11	112262	Facial
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
chr20	56,838,189	56,929,641	GNAS	20q13	Albright Hereditary Osteodystrophy	139320	Mild DD and DF
chrX	66,680,599	66,860,844	AR	Xq12	Androgen Insensitivity Syndrome	313700	Female Male
chr11	31,762,916	31,796,085	PAX6	11p13-p11	Aniridia	607108	Eye defect
chr1	145,694,956	145,712,108	GJA5	1q21	Atrial Fibrillation	121013	Heart defect
chr5	172,591,744	172,594,868	NKX2-5	5q35	Atrial Septal Defect with Atrioventricular Conduction Defects	600584	Heart defect
chr4	111,758,029	111,782,566	PITX2	4q25	Axenfeld-Rieger Syndrome	601542	Eye and teeth defects
chr11	1,962,981	1,985,640	H19	11p15	Beckwith-Wiedemann	103280	Cancer and overgrowth
chr11	2,096,925	2,136,469	IGF2	11p15	Beckwith-Wiedemann	147470	Cancer and overgrowth
chr11	2,127,584	2,148,999	INS	11p15	Beckwith-Wiedemann	176730, 19	Cancer and overgrowth
chr11	2,851,440	2,873,550	CDKN1C	11p15	Beckwith-Wiedemann	607542	Cancer and overgrowth
chr11	2,867,526	2,913,051	SLC22A18	11p15	Beckwith-Wiedemann	602631	Cancer and overgrowth
chr11	2,896,078	2,917,225	PHLDA2	11p15	Beckwith-Wiedemann	602631	Cancer and overgrowth
chrX	133,335,008	133,390,488	PHF6	Xq26	Borjeson-Forsman-Lehmann Syndrome	300414	Severe mental and physical
chr20	33,360,416	33,489,441	GDF5	20q11	Brachydactyl Type C	601146	Finger defects
chr13	90,848,888	92,317,488	GPC5	13q31	Brachydactyly	602446	Finger defects
chr13	92,677,096	93,853,948	GPC6	13q31	Brachydactyly	604404	Finger defects
chr8	72,272,222	72,437,021	EYA1	8q13	Branchio-Oto-Renal Syndrome	602064	Kidney failure
chr17	67,628,756	67,634,156	SOX9	17q24	Campomelic Dysplasia	608160	Bone defect
chr10	91,180,753	91,285,293	SLC16A12	10q23	Cataract, Juvenile, with Microcornea A and Glucosuria	611910	Eye defect
chr21	26,174,732	26,465,003	APP	21q21	Cerebral Amyloid Angiopathy	104760	Blood vessel failure
chr17	15,073,822	15,109,369	PMP22	17p11	Charcot-Marie Tooth Disease	601097	Motor and sensory failure – late onset
chr8	61,753,893	61,942,017	CHD7	8q12	Charge Syndrome	601653	Deaf/Blind ++
chrX	2,862,852	2,892,311	CDPX1(ARSE)	Xp22	Chondrodysplasia Punctata, X-linked Recessive	300180	Skeletal/Eye ++
chrX	85,002,841	85,189,222	CHM	Xp21	Choroideremia	300390	Eye defect
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
chr4	4,911,225	4,917,628	MSX1	4p16	Cleft Lip	142983	Facial defect
chr2	199,842,469	200,038,076	SATB2	2q32	Cleft Palate	608148	Facial defect
chr6	45,404,032	45,626,796	RUNX2	6p21	Cleidocranial Dysplasia	600211	Skeletal defects
chrX	20,077,950	20,194,671	RPS6KA3	Xp22	Coffin-Lowry Syndrome	300075	MCA
chr15	91,244,423	91,372,241	CHD2	15q26	Congenital Diaphragmatic Hernia	602119	Diaphragm/lung problems
chr15	94,670,161	94,683,047	NR2F2	15q26	Congenital Diaphragmatic Hernia	107773	Diaphragm/lung problems
chr8	106,400,323	106,885,943	ZFPM2(FOG2)	8q23	Congenital Heart	603693	Heart defect
chr5	36,912,742	37,101,678	NIPBL	5p13	Cornelia de Lange	608667	Severe mental and physical
chr11	15,949,900	16,454,494	SOX6	11p15.1	Craniosynostosis	607257	Skull defect
chr5	174,084,181	174,090,508	MSX2	5q35	Craniosynostosis	123101	Skull defect
chr5	1,306,287	1,348,162	TERT	5p15	Cri du Chat	187270	Severe Mental and Physical
chr13	31,211,679	31,275,009	LGR8(RXFP2)	13q13	Cryptorchidism, Unilateral or Bilateral	606655	Missing testes
chr7	156,479,506	156,496,108	HLXB9(MNX1)	7q36	Currarino Syndrome	142994	Skeletal defects
chrX	119,444,035	119,487,189	LAMP2	Xq24	Danon Disease	309606	Heart defect
chr16	65,620,551	65,692,459	CBFB	16q22	Delayed Cranial Ossification	121360	Skull defect
chr19	47,055,828	47,067,324	RPS19	19q13	Diamond Blackfan Anemia	603474	Blood and MCA
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

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chr21	37,517,596	37,561,703	DSCR3	21q22	Downs Syndrome Critical Region	605298	MCA
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
chr7	94,042,477	94,133,413	SGCE	7p21	Dystonia-11	604149	Skeletal defects
chr7	94,113,623	94,146,938	PEG10	7p21	Dystonia-11	604149	Skeletal defects
chr7	94,123,295	94,123,994	PEG10_ICRegion	7p21	Dystonia-11	604149	Skeletal defects
chrX	68,752,636	69,176,046	EDA	Xq13	Ectodermal Dysplasia	300451	Various skin
chr7	93,861,809	93,898,480	COL1A2	7q21	Ehlers -Danlos Syndrome	120160	Joint and skin defect
chrX	100,539,435	100,549,657	GLA	Xq22	Fabry Disease	300644	Kidney, heart, skin, eye
chr2	15,998,134	16,004,579	MYCN	2q24	Feingold	164840	Hands, feet, mental
chr1	221,168,406	221,245,958	DISP1	1q41	Fryns 1q41	229850	Severe physical, fatal
chr7	41,967,073	42,243,900	GLI3	7p14	Greig Cephalopolysindactyly Syndrome	165240	Physical and mild mental
chrX	62,771,573	62,922,138	ARHGFE9	Xq11	Heterotaxy	300429	Random placement of organs
chr10	42,892,523	42,945,803	RET	10q11	Hirschsprung Disease	164761	Bowel defect
chr13	77,367,617	77,447,665	EDNRB	13q22	Hirschsprung Disease	131244	Bowel defect
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
chr12	113,276,118	113,330,630	TBX5	12q14	Holt-Oram Syndrome	601620	Bone and heart
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
chrX	21,960,842	22,176,399	PHEX	Xp22	Hypophosphatemic Rickets	300550	Bone
chrX	153,423,653	153,446,455	IKBKG	Xq28	Incontinentia Pigmenti	300248	Hair, skin, teeth defect
chr11	17,371,008	17,455,025	ABCC8	11p15	Infantile Hyperinulinism, Enteropathy and Deafness (Ushers Disease)	600509	Deafness
chr11	17,472,018	17,522,539	USH1C	11p15	Infantile Hyperinulinism, Enteropathy and Deafness (Ushers Disease)	605242	Deafness
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
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
chr15	46,487,797	46,725,210	FBN1	15q21	Marfan Syndrome	134797	Tall, heart defects
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
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(LI)	17p13	Miller-Dieker Syndrome	601545	Severe Mental and Physical
chr1	68,274,232	68,299,047	DIRAS3	1p31	Monosomy 1p31	605193	Mild 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

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chrX	31,048,667	33,277,779	DMD	Xq28	Muscular Dystrophy	300777	Muscle wasting, early diagnosis key
chrX	149,487,727	149,592,272	MTM1	Xq28	Myotubular Myopathy	300415	Muscle, lung and heart defects
chr9	128,416,569	128,503,132	LMX1B	9q33	Nail-Patella Syndrome	602575	Nail and knee defect
chrX	17,303,464	17,664,034	NHS	Xp22	Nance-Horan Syndrome	300457	Eye defect
chr10	21,110,094	21,503,122	NEBL	10p12	Nebulette	605491	Cardiac, immune and DD
chrX	49,573,965	49,750,632	CLCN5	Xp11	Nephrolithiasis 2, X-linked, Dent Disease	300008	Kidney disease
chr2	110,237,195	110,319,883	NPHP1	2q13	Nephronophthisis 1	607100	Kidney disease
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 disorders
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
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
chr20	49,833,990	49,852,421	SALL4	20q13	Okihiro	607343	Eye and physical defects
chr14	36,196,533	36,216,762	PAX9	14q13	Oligodontia		Missing teeth
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-Kavaggia Syndrome	300188	MCA
chr3	190,831,910	191,097,756	TP73L	3q28	Optic Atrophy	603273	Eye defect
chrX	38,096,302	38,165,553	OTC	Xp11	Ornithine Transcarbamylase Deficiency	300461	Enzyme deficiency progressive
chrX	13,662,801	13,697,391	OFD1	Xp22	Orofaciodigital 1	300170	Face, mouth, fingers and kidney
chr17	45,616,456	45,633,992	COL1A1	17q21	Osteogenesis Imperfecta	120150	Bone defect
chr12	63,849,638	63,928,374	LEMD3	12q14	Osteopoikilosis	607844	Bone defect
chr18	51,045,967	51,454,183	TCF4	18q21	Pitt-Hopkins Syndrome	602272	Apnea, breathing, mental and physical problems
chr7	156,168,394	156,378,602	LMBR1	7q36	Polydactyl Preaxial 2	605522	Thumb defect
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, 607306	MCA Mental and Physical
chr2	31,603,160	31,659,544	SRD5A2	2p23	Pseudovaginal Perineoscrotal Hypospadias	607306	Sex between male and female
chrX	19,271,972	19,287,886	PDHA1	Xp22	Pyruvate Dehydrogenase Deficiency	300502	Enzyme deficiency progressive
chr17	33,120,548	33,179,182	TCF2(HNF1B)	17q12	Renal Cysts and Diabetes Syndrome	189907	Renal Cysts and Diabetes Syndrome
chr13	47,775,912	47,954,023	RB1	13q14	Retinoblastoma	180200	Eye disorder
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
chr17	39,186,625	39,191,682	SOST	17q21	Sclerostin	605740	Bone disease
chr6	100,943,471	101,018,272	SIM1	6q16	Severe Obesity	603128	Obesity
chr1	178,466,065	178,510,811	LHX4	1q25	Short Stature	602146	Short stature
chr7	50,615,258	50,838,651	GRB10	7p12	Silver-Russell Syndrome	161523	Treatable dwarfism
chrX	132,497,442	132,947,332	GPC3	Xq26	Simpson, Golabi, Behmel Syndrome Type 1	300037	overgrowth
chr11	70,823,105	70,837,125	DHCR7	11q13	Smith-Lemli-Opitz	602858	mile 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	46,653,018	46,684,528	COL2A1	12q13	Sticker 1	120140	Joint, hearing and tongue
chr1	103,115,583	103,346,640	COL11A1	1p21	Stickler Syndrome	120280	Joint, hearing and tongue
chr2	176,653,081	176,656,936	EVX2	2q31	Synpolydactyly	142991	Fused or extra fingers/toes
chr2	176,664,382	176,767,892	HOXDgenes	2q31	Synpolydactyly	142989	Fused or extra fingers/toes
chr2	176,665,778	176,668,912	HOXD13	2q31	Synpolydactyly	142989	Fused or extra fingers/toes
chr2	176,695,334	176,697,891	HOXD9	2q31	Synpolydactyly	142982	Fused or extra fingers/toes

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chr1	145,841,561	145,848,017	GJA8	1q21	Thrombocytopenia Absent Radius Syndrome	600897	Arm, blood and platelet deformities Treatable in utero.
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
chr12	113,592,442	113,606,352	TBX3	12q24	Ulnar-Mammary Syndrome	601621	Bone, genital and nipple defects.
chr1	208,027,885	208,046,102	IRF6	1q32	Van der Woude Syndrome	607199	Facial defects and mild learning disability
chr6	43,845,931	43,862,201	VEGFA	6p21	Vascular Endothelial Growth Factor Disorders	192240	Variety of physical disorders
chr2	130,995,137	131,002,053	CFC1	2q21	Visceral Heterotaxy	605194	Randomized organ and arterial placement
chr3	10,158,319	10,168,746	VHL	3p25	Von Hippel Lindau	608537	Cyst and tumour formation, cancer
chr2	222,772,851	222,871,944	PAX3	2q36	Waardenburg Syndrome I	606597	Hearing loss and pigmentation problems
chr3	69,871,323	70,100,177	MITF	3p14	Waardenburg Syndrome IIA	158645	Hearing loss and pigmentation problems
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	107,569,810	107,827,431	COL4A5	Xq22	X-linked Alport Syndrome	303630	Hearing, eye and kidney defects
chrX	106,758,415	106,780,912	PRPS1	Xq22	X-linked Charcot Marie Tooth Disease	311850	Progressive muscle and nerve wasting - late onset
chrX	53,417,795	53,466,343	SMC1L1(SMC 1A)	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
chrX	136,476,012	136,481,923	ZIC3	Xq26	X-linked Heterotaxy	300265	Left-right assymetry, severe organ defects
chrX	18,567,731	18,600,150	RS1	Xp22	X-linked Juvenile Retinoschisis	312700	Eye disorder
chrX	53,474,931	53,478,048	HADH2(HSD1 7B10)	Xp11	X-linked Mental Retardation	300256	Most common mental retardation phenotype with some dysmorphic features
chrX	38,305,683	38,433,116	TM4SF2(TSP AN7)	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

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
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
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

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chrX	118,592,527	118,602,407	UBE2A	Xq23	X-linked Mental Retardation	312180	Most common mental retardation
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 Christianson 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	53,576,476	53,730,398	HUWE1	Xp11	X-linked Mental Retardation, Turner Syndrome	300697	Heart defects, ovary, short stature. Usually normal intelligence.
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
chr9	126,283,337	126,309,520	NR5A1	9q3	XY Sex Reversal	184757	Genetically male, but female sex organs
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
chr18	42,798,570	42,820,446	TCEB3C	18q21		609522	Transcription factor - imprinting
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	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
chr11	2,671,987	2,683,633	KCNQ1_ICRegion	11p15		607542	Heart defects
chr14	20,897,000	20,932,000	MicroDeletion Region	14q11		605012	Heart failure
chr22	16,454,903	16,491,588	ATP6V1E1	22q11		108746	Linked to muscle abnormalities
chrX	505,079	540,146	SHOX	Xp22		400020	Short stature
chrY	2,714,821	2,716,600	SRY	Yp11		480000	Sexual defects

**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.