

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords	Learning & Behavioural	Physical	Metabolic & Organ	Hereditary Cancers	Syndromes Selected
chr10	83,625,077	84,735,341	NRG3	10q22	10q22-23 Deletion	605533	Brain	X				
chr10	87,349,292	88,116,230	GRID1	10q22	10q22-23 Deletion	610659	Brain	X				
chr11	86,334,369	86,344,081	FZD4	11q14	11q14 MicroDeletion	604579	Eye		X			
chr12	65,029,066	65,359,020	GRIP1	12q14	12q14.3 Deletion	604597	Brain and dysmorphic	X	X			
chr14	60,045,150	60,048,904	SIX6	14q22	14q22 MicroDeletion	606326	Eye		X			
chr14	60,246,009	60,260,792	SIX4	14q22	14q22 MicroDeletion	606342	Eye		X			
chr14	60,182,506	60,185,933	SIX1	14q22	14q22 MicroDeletion	601205	Kidney/hearing					
chr14	56,337,178	56,346,937	OTX2	14q22	14q22 MicroDeletion, Microphthalmia, Syndromic 5	600037	Eye		X			
chr14	53,486,207	53,493,362	BMP4	14q22	14q22 MicroDeletion, Orofacial Cleft 11	112262	Facial		X			
chr15	30,110,018	30,248,527	CHRNA7	15q13	15q13.3 MicroDeletion Syndrome	118511	Schizophrenia/Epilepsy	X				
chr22	21,852,552	21,990,224	BCR	22q11	22q11.2 Distal Deletion	151410	Leukaemia				X	
chr2	61,136,239	61,673,319	MicroDeletion Region	2p15	2p15p16.1 MicroDeletion	602559	Multiple	X	X			
chr3	197,951,312	198,043,751	PAK2	3q29	3q29 MicroDeletion Syndrome	605022	Multiple	X	X			
chr9	139,725,238	139,850,397	EHMT1	9q34	9q34.3 Syndrome	607001	Multiple	X	X			
chrX	54,488,612	54,539,324	FGD1	Xp11	Aarskog-Scott Syndrome	300564	Multiple	X	X			
chrX	30,232,507	30,237,413	NROB1(DAX1)	Xp21	Adrenal Hypoplasia Congenita	300473	Sterility			X		
chrX	152,643,530	152,663,374	ABCD1	Xq28	Adrenoleukodystrophy	300475	Late onset brain disease	X				
chr5	126,140,732	126,200,608	LMNB1	5q23	Adult-onset Autosomal Dominant Leukodystrophy	150340	Late onset brain disease	X				
chr20	10,566,334	10,602,590	JAG1	20p12	Alagille Syndrome	601920	Late onset organ failure			X		
chr20	56,838,189	56,929,641	GNAS	20q13	Albright Hereditary Osteodystrophy	139320	Mild DD and DF	X	X			
chr16	155,725	171,196	HBA1&HBA2	16p13	Alpha Thalassemia Mental Retardation	141850	Blood disorder			X		
chrX	76,647,847	76,928,358	ATRX	Xq21	Alpha Thalassemia Mental Retardation Syndrome	300032	Blood disorder			X		
chrX	66,680,599	66,860,844	AR	Xq12	Androgen Insensitivity Syndrome	313700	Female Male		X			
chr15	20,556,790	20,585,849	NIPA2	15q11	Angelman / Prader Willi	608146	Mental retardation	X				
chr15	20,594,720	20,637,877	NIPA1	15q11	Angelman / Prader Willi	608146	Mental retardation	X				
chr15	21,361,547	21,407,800	MKRN3	15q11	Angelman / Prader Willi	603586	Mental retardation	X				
chr15	21,429,787	21,452,081	MAGEL2	15q11	Angelman / Prader Willi	605283	Mental retardation	X				
chr15	22,619,887	22,774,822	SNRPN	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,713,304	22,720,548	PWS_ICRegion	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,741,227	22,784,821	SNURF	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,747,536	22,751,866	PWS_ICRegion	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,751,228	22,774,822	SNRPN	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,768,233	22,793,232	SNORD107/64/108	15q11	Angelman / Prader Willi	182279	Mental retardation	X				
chr15	22,835,000	23,010,100	SnoRNA	15q11	Angelman / Prader Willi	601491	Mental retardation	X				
chr15	23,064,582	23,084,648	SNORD109B	15q11	Angelman / Prader Willi		Mental retardation	X				
chr15	21,471,646	21,493,542	NDN	15q11	Angelman / Prader Willi	602117	Mental retardation	X				
chr11	31,762,916	31,796,085	PAX6	11p13-p11	Aniridia	607108	Eye defect		X			
chr1	145,694,956	145,712,108	GJA5	1q21	Atrial Fibrillation	121013	Heart defect		X			
chr5	172,591,744	172,594,868	NKX2-5	5q35	Atrial Septal Defect with Atrioventricular Conduction Defects	600584	Heart defect		X			
chr1	229,829,184	230,243,641	DISC1	1q42	Autism	605210	Autism	X				
chr10	64,241,763	64,246,133	EGR2	10q21	Autism	129010	Autism	X				
chr15	23,463,512	23,669,962	ATP10A	15q12	Autism	605855	Autism	X				
chr15	24,339,787	24,767,950	GABRB3	15q12	Autism	137192	Autism	X				
chr2	50,000,992	51,113,178	NRXN1	2p16	Autism	600565	Autism	X				
chr2	162,189,192	162,548,489	SLC4A10	2q24	Autism	605556	Autism	X				
chr22	49,459,936	49,518,507	SHANK3	22q13	Autism	606230	Autism	X				
chr7	116,099,695	116,225,676	MET	7q31	Autism	164860	Autism	X				
chr7	121,746,687	122,313,750	CADPS2	7q31	Autism	609978	Autism	X				
chr7	145,444,386	147,749,019	CNTNAP2	7q35-36	Autism	276000	Autism	X				
chr7	154,943,160	154,950,579	EN2	7q36	Autism	131310	Autism	X				
chrX	5,818,085	6,155,888	NLGN4X	Xp22	Autism	300427	Autism	X				
chrX	70,281,436	70,307,776	NLGN3	Xq13	Autism	300336	Autism	X				
chr3	2,115,550	3,074,645	CNTN4	3p25	Autism Spectrum Disorder	607280	Autism	X				
chr4	111,758,029	111,782,566	PITX2	4q25	Axenfeld-Rieger Syndrome	601542	Eye and teeth defects		X			
chrY	13,052,929	13,603,474	AZFa	Yq11	AZFa Region	400005	Male sterility			X		
chrY	23,539,798	23,560,998	BPY2	Yq11	AZFc Region	400013	Male sterility			X		
chrY	23,663,736	25,513,794	AZFc	Yq11	AZFc Region	400003	Male sterility			X		
chrY	23,684,890	23,754,627	DAZ1	Yq11	AZFc Region	400003	Male sterility			X		
chrY	24,600,764	24,603,549	CDY1	Yq11	AZFc Region	400016	Male sterility			X		
chrY	24,765,503	24,770,366	GOLGA2LY(A F332229)	Yq11	AZFc Region	400035	Male sterility			X		
chrY	25,173,539	25,194,740	BPY2	Yq11	AZFc Region	400016	Male sterility			X		
chrY	25,586,438	25,607,639	BPY2	Yq11	AZFc Region	400013	Male sterility			X		
chrY	26,010,846	26,015,709	GOLGA2LY(A F332229)	Yq11	AZFc Region	400035	Male sterility			X		
chrY	26,177,652	26,180,436	CDY1	Yq11	AZFc Region	400016	Male sterility			X		
chr11	128,213,125	128,242,478	KCNJ1	11q24	Barter 2	600359	Kidney and blood			X		
chr1	16,221,073	16,256,063	CLCNKB	1p36	Barter 3	602023	Kidney and blood			X		
chr1	55,237,205	55,247,053	BSND	1p32	Barter 4	606412	Kidney and blood			X		
chr1	16,217,957	16,233,132	CLCNKA	1p36	Barter 4	602024	Kidney and blood			X		
chr15	46,285,791	46,382,417	SLC12A1	15q21	Barter Type 1	600839	Kidney and blood			X		
chr3	123,385,220	123,488,032	CASR	3q13	Barter with Autosomal Dominant Hypocalcemia	601199	Kidney and blood			X		
chr11	1,962,981	1,985,640	H19	11p15	Beckwith-Wiedemann	103280	Cancer and overgrowth		X		X	
chr11	2,096,925	2,136,469	IGF2	11p15	Beckwith-Wiedemann	147470	Cancer and overgrowth		X		X	
chr11	2,127,584	2,148,999	INS	11p15	Beckwith-Wiedemann	176730, 19	Cancer and overgrowth		X		X	
chr11	2,851,440	2,873,550	CDKN1C	11p15	Beckwith-Wiedemann	607542	Cancer and overgrowth		X		X	
chr11	2,867,526	2,913,051	SLC22A18	11p15	Beckwith-Wiedemann	602631	Cancer and overgrowth		X		X	
chr11	2,896,078	2,917,225	PHLDA2	11p15	Beckwith-Wiedemann	602631	Cancer and overgrowth		X		X	
chr16	56,211,151	56,256,445	GPR56	16q13	Bilateral Frontoparietal Polymicrogyria	604110	Severe mental	X				
chr3	140,144,730	140,149,880	FOXL2	3q22	Blepharophimosis	605597	Ovary failure and cancer			X	X	
chrX	133,335,008	133,390,488	PHF6	Xq26	Borjeson-Forsman-Lehmann Syndrome	300414	Severe mental and physical	X	X			
chr20	33,360,416	33,489,441	GDF5	20q11	Brachydactyl Type C	601146	Finger defects		X			
chr13	90,848,888	92,317,488	GPC5	13q31	Brachydactyly	602446	Finger defects		X			
chr13	92,677,096	93,853,948	GPC6	13q31	Brachydactyly	604404	Finger defects		X			
chr8	72,272,222	72,437,021	EYA1	8q13	Branchio-Oto-Renal Syndrome	602064	Kidney failure			X		
chr13	31,787,617	31,871,809	BRCA2	13q13	Breast Cancer	600185	Cancer				X	
chrX	43,400,353	43,491,012	MAOA	Xp11	Brunner Syndrome	309850	Mild mental - aggressive	X				
chr17	67,628,756	67,634,156	SOX9	17q24	Campomelic Dysplasia	608160	Bone defect		X			
chr10	91,180,753	91,285,293	SLC16A12	10q23	Cataract, Juvenile, with Microcornea A and Glucosuria	611910	Eye defect		X			
chr21	26,174,732	26,465,003	APP	21q21	Cerebral Amyloid Angiopathy	104760	Blood vessel failure		X	X		
chr17	15,073,822	15,109,369	PMP22	17p11	Charcot-Marie Tooth Disease	601097	Motor and sensory failure - late onset		X	X		
chr8	61,753,893	61,942,017	CHD7	8q12	Charge Syndrome	601653	Deaf/Blind ++	X	X			
chrX	2,862,852	2,892,311	CDPX1(ARSE)	Xp22	Chondrodysplasia Punctata, X-linked Recessive	300180	Skeletal/Eye ++		X			
chrX	85,002,841	85,189,222	CHM	Xp21	Choroideremia	300390	Eye defect		X	X		

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chr18	70,416,108	70,906,616	ZNF407	18q22	Chromosome 18q Deletion Syndrome		MCA	X	X			
chr6	1,555,680	1,559,128	FKHL7(FOXC1)	6p24	Chromosome 6p24 Deletion Syndrome	609010	MCA	X	X			
chr9	831,690	959,090	DMRT1	9p24	Chromosome 9p Deletion Syndrome	158170	MCA	X	X	X		
chr9	1,039,858	1,047,552	DMRT2	9p24	Chromosome 9p Deletion Syndrome	604935	MCA	X	X	X		
chrX	37,524,264	37,557,658	CYBB	xp11	Chronic Granulomatous Disease	300481	Immune disorder			X		
chr5	147,184,339	147,191,453	SPINK1	5q32	Chronic Pancreatitis	167790	Pancreas damage			X		
chr4	4,911,225	4,917,628	MSX1	4p16	Cleft Lip	142993			X			
chr2	199,842,469	200,038,076	SATB2	2q32	Cleft Palate	608148	Facial defect		X			
chr6	45,404,032	45,626,796	RUNX2	6p21	Cleidocranial Dysplasia	600211	Skeletal defects		X			
chrX	20,077,950	20,194,671	RPS6KA3	Xp22	Coffin-Lowry Syndrome	300075	MCA	X	X			
chr15	91,244,423	91,372,241	CHD2	15q26	Congenital Diaphragmatic Hernia	602119	Diaphragm/lung problems		X			
chr15	94,670,161	94,683,047	NR2F2	15q26	Congenital Diaphragmatic Hernia	107773	Diaphragm/lung problems		X			
chr8	106,400,323	106,885,943	ZFPM2(FOG2)	8q23	Congenital Heart	603693	Heart defect		X			
chr6	32,044,642	32,162,690	CYP21A2	6p21	Congenital Adrenal Hyperplasia	201910	Infertility and sex disorders			X		
chr5	36,912,742	37,101,678	NIPBL	5p13	Cornelia de Lange	608667	Severe mental and physical		X			
chr10	88,506,376	88,674,925	BMPR1A	10q23	Cowden	601728	Polyps and cancer			X	X	
chr11	15,949,900	16,454,494	SOX6	11p15.1	Craniosynostosis	607257	Skull defect		X			
chr5	174,084,181	174,090,508	MSX2	5q35	Craniosynostosis	123101	Skull defect		X			
chrX	152,606,586	152,615,234	SLC6A8	Xq28	Creatine Deficiency Syndrome	300036	Mental retardation	X				
chr16	33,686,002	33,694,027	SLC6A8	16p11	Creatine Deficiency Syndrome / X-linked Mental Retardation		Mental retardation	X				
chr16	34,877,510	35,027,509		16p11	Creatine Deficiency Syndrome / X-linked Mental Retardation		Mental retardation	X				
chr5	1,306,287	1,348,162	TERT	5p15	Cri du Chat	187270	Severe Mental and Physical	X	X			
chr13	31,211,679	31,275,009	LGR8(RXFP2)	13q13	Cryptorchidism, Unilateral or Bilateral	606655	Missing testes		X			
chr7	156,479,506	156,496,108	HLXB9(MNX1)	7q36	Currarino Syndrome	142994	Skeletal defects		X			
chr17	3,486,511	3,513,146	CTNS	17p13	Cystinosis	606272	Kidney and eye defect			X		
chr2	44,356,103	44,401,443	SLC3A1	2p21	Cystinuria with Mitochondrial Disease	104614	Kidney disease			X		
chr2	44,399,406	44,442,448	PREPL	2p21	Cystinuria with Mitochondrial Disease	104614	Kidney disease			X		
chr3	148,586,527	148,607,097	ZIC4	3q24	Dandy Walker	608948	Severe mental	X				
chr3	148,609,871	148,617,196	ZIC1	3q24	Dandy Walker	600470	Severe mental	X				
chrX	119,444,035	119,487,189	LAMP2	Xq24	Danon Disease	309606	Heart defect		X	X		
chr13	19,659,605	19,665,114	GJB2(Connexin 26)	13q12	Deafness	121011	Deafness			X		
chr5	149,717,428	149,760,063	TCOF1	5q33	Deafness	606847	Deafness			X		
chrX	100,487,306	100,490,343	TIMM8A	Xq22	Deafness-Dystonia-Optic Neuropathy	300356	Deafness and eye			X		
chr16	65,620,551	65,692,459	CBFB	16q22	Delayed Cranial Ossification	121360	Skull defect		X			
chrX	152,821,179	152,825,834	AVPR2	Xq28	Diabetes Insipidus, Nephrogenic, X-linked Syndrome	300538	Diabetes			X		
chr6	144,293,129	144,427,428	ZAC(PLAGL1)	6q24	Diabetes Mellitus, Transient Neonatal, 1	603044	Diabetes			X		
chr6	144,370,000	144,377,576	HYMA1	6q24	Diabetes Mellitus, Transient Neonatal, 1	603044	Diabetes			X		
chr19	47,055,828	47,067,324	RPS19	19q13	Diamond Blackfan Anemia	603474	Blood and MCA		X	X		
chr22	17,698,224	17,815,220	HIRA	22q11	diGeorge 1	600637	MCA	X	X			
chr22	18,124,226	18,151,110	TBX1	22q11	diGeorge 1	602054	MCA	X	X			
chr22	19,601,714	19,638,035	CRKL	22q11	diGeorge 1	602007	MCA	X	X			
chr10	8,136,673	8,157,170	GATA3	10p14	diGeorge 2	131320	MCA	X	X			
chr8	8,679,409	8,788,541	MFHAS1	8p23	diGeorge Syndrome	605352	MCA	X	X			
chr8	11,571,877	11,654,918	GATA4	8p23	diGeorge Syndrome	600576	MCA	X	X			
chr21	34,810,654	34,909,252	DISC1(RCAN1)	21q22	Downs Syndrome Critical Region 1	602917	MCA	X	X			
chr21	35,081,968	36,278,917	RUNX1	21q22	Downs Syndrome Critical Region	151385	MCA	X	X			
chr21	37,517,596	37,561,703	DISC3	21q22	Downs Syndrome Critical Region	605298	MCA	X	X			
chr21	37,661,729	37,809,549	DYRK1A	21q22	Downs Syndrome Critical Region	600855	MCA	X	X			
chr18	44,824,170	45,250,999	DYM	18q21	Dyggve-Melchior-Clausen Disease	607461	Severe Mental and Physical	X	X			
chr7	94,042,477	94,133,413	SGCE	7p21	Dystonia-11	604149	Skeletal defects		X			
chr7	94,113,623	94,146,938	PEG10	7p21	Dystonia-11	604149	Skeletal defects		X			
chr7	94,123,295	94,123,994	PEG10_ICR region	7p21	Dystonia-11	604149	Skeletal defects		X			
chrX	68,752,636	69,176,046	EDA	Xq13	Ectodermal Dysplasia	300451	Various skin		X			
chr7	93,861,809	93,898,480	COL1A2	7q21	Ehlers -Danlos Syndrome	120160	Joint and skin defect		X			
chrX	100,539,435	100,549,657	GLA	Xq22	Fabry Disease	300644	Kidney, heart, skin, eye		X			
chr16	88,331,460	88,410,566	FANCA	16q24	Fanconi Anemia	607139	Blood, cancer			X	X	
chr2	15,998,134	16,004,579	MYCN	2q24	Feingold	164840	Hands, feet, mental	X	X			
chrX	146,801,201	146,840,303	FMR1	Xq27	Fragile X Syndrome	309550	Severe Mental retardation	X				
chrX	147,389,831	147,889,899	FMR2(AFF2)	Xq28	Fragile X Syndrome	309548	Severe Mental retardation	X				
chr1	221,168,406	221,245,958	DISP1	1q41	Fryns 1q41	229850	Severe physical, fatal		X			
chr5	112,071,117	112,209,835	APC	5q22	Gardner Syndrome	601731	Polyps and cancer			X	X	
chr16	55,456,620	55,507,263	SLC12A3	16q13	Gitelman Syndrome	610966	Kidney			X		
chrX	30,581,461	30,658,645	GK	Xp21	Glycerol Kinase Deficiency	300474	Mild mental and metabolic	X		X		
chr7	41,967,073	42,243,900	GLI3	7p14	Greig Cephalopolysyndactyly Syndrome	165240	Physical and mild mental	X	X			
chrX	153,717,263	153,904,192	F8	Xq28	Hemophilia	306700	Blood disorder			X		
chrX	138,440,561	138,473,283	F9	Xq27	Hemophilia B	300746	Blood disorder			X		
chr6	160,341,410	160,352,419	IGF2R	6q25	Hepatocellular Carcinoma	147260	Cancer				X	
chr18	46,810,581	46,865,409	MADH4(SMA D4)	18q21	Hereditary Hemorrhagic Telangiectasia Syndrome	600993	Blood vessel disorder			X		
chrX	62,771,573	62,922,138	ARHGFE9	Xq11	Heterotaxy	300429	Random placement of organs		X			
chrX	153,230,091	153,256,123	FLNA	Xq28	Heterotopia, Periventricular, X-linked Dominant Syndrome	300017	Epilepsy	X				
chr10	42,892,523	42,945,803	RET	10q11	Hirschsprung Disease	164761	Bowel defect		X			
chr13	77,367,617	77,447,665	EDNRB	13q22	Hirschsprung Disease	131244	Bowel defect		X			
chr2	45,022,365	45,025,945	SIX3	2p21	Holoprosencephaly 2	603714	Brain defect	X	X			
chr18	3,402,072	3,444,406	TGIF1	18p31	Holoprosencephaly Type 4	602630	Brain defect	X	X			
chr13	99,432,320	99,437,019	ZIC2	13q32	Holoprosencephaly 5	603073	Brain defect	X	X			
chr21	44,256,634	44,350,858	TMEM1	21q22	Holoprosencephaly 1	602103	Brain defect	X	X			
chr21	46,432,788	46,473,119	LSS	21q22	Holoprosencephaly 1	600909	Brain defect	X	X			
chr7	155,288,319	155,297,728	SHH	7q36	Holoprosencephaly 3	600725	Brain defect	X	X			
chr9	97,245,085	97,319,068	PTCH1	9q22	Holoprosencephaly 7	601309	Brain defect	X	X			
chr2	121,210,293	121,466,699	GLI2	2q14	Holoprosencephaly 9	165230	Brain defect	X	X			
chr5	171,221,161	171,366,482	FBXW11	5q35	Holoprosencephaly and Preaxial Polydactyly	605651	Brain defect	X	X			
chr12	113,276,118	113,330,630	TBX5	12q14	Holt-Oram Syndrome	601620	Bone and heart		X			
chrX	152,780,581	152,804,778	L1CAM	Xq28	HSAS, MASA, CRASH Syndromes	300371	Physical and mental	X	X			
chrX	148,368,203	148,423,359	IDS	Xq28	Hunter Syndrome, Mucopolysaccharidosis Type 2	309900	Progressive MCA	X	X			
chr2	172,349,126	172,458,979	SLC25A12	2q31	Hypomyelination, Global, Cerebral	603667	Mental retardation	X				
chrX	21,960,842	22,176,399	PHEX	Xp22	Hypophosphatemic Rickets	300550	Bone		X			
chr15	97,010,284	97,325,282	IGF1R	15q25	IGF-1 Resistance	147307	Insulin defect			X		
chrX	153,423,653	153,446,455	IKBKG	Xq28	Incontinentia Pigmenti	300248	Hair, skin, teeth defect		X			
chr11	17,371,008	17,455,025	ABCC8	11p15	Infantile Hyperinulinism, Enteropathy and Deafness (Ushers Disease)	600509	Deafness		X			
chr11	17,472,018	17,522,539	USH1C	11p15	Infantile Hyperinulinism, Enteropathy and Deafness (Ushers Disease)	605242	Deafness		X			
chr15	41,612,952	41,664,382	KIAA0377(HISPPD2A)	15q15	Infertility and Deafness	601979	Infertility and deafness			X		

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords	Learning & Behavioural	Physical	Metabolic & Organ	Hereditary Cancers	Syndromes Selected
chr15	41,710,064	41,747,192	CATSPER2	1q15	Infertility and Deafness	611102	Infertility and deafness					
chr12	86,966,921	87,060,124	CEP290	12q21	Joubert 5	610142	Brain defect	X		X		
chr6	135,646,817	135,860,576	AHI1	6q23	Joubert Syndrome	608894	Brain defect	X				
chr8	11,737,443	11,763,055	CTSB	8p23	Kabuki Syndrome	116810	Severe Mental and Physical	X	X			
chr8	38,387,813	38,445,509	FGFR1	8p12	Kallman 1	136350	Sex hormone defect			X		
chrX	8,456,915	8,660,227	KAL1	Xp22	Kallmann Syndrome	308700	Sex hormone defect			X		
chr8	118,880,787	119,193,239	EXT1	8q23	Langer-Giedion Syndrome	150230	Mild DD and PD	X	X			
chrX	133,421,923	133,462,362	HPRT1	Xq26	Lesch-Nyhan Syndrome	308000	Progressive MD and gout	X		X		
chr11	67,130,983	67,136,581	NDUFV1	11q13	Leukodystrophy	161015	Progressive MD	X				
chr17	7,505,822	7,531,642	TP53	17p13	Li-Fraumeni Type 1	191170	Cancer				X	
chr7	102,899,473	103,417,198	RELN	7q22	Lissencephaly with Cerebellar Hypoplasia	600514	Severe Mental and Physical	X	X			
chr3	30,622,998	30,710,637	TGFBR2	3p24	Loeys-Dietz Syndrome	190182	Severe Mental and Physical – can be reduced following surgery	X	X			
chr9	100,907,233	100,956,294	TGFBR1	9q22	Loeys-Dietz Syndrome	190181	Severe Mental and Physical – can be reduced following surgery	X	X			
chr11	2,412,796	2,836,915	KCNQ1	11p15	Long QT Syndrome	607542	Heart rhythm defect			X		
chrX	128,501,933	128,554,211	OCRL	Xq25	Lowe	300535	Eye and Brain defects	X	X			
chr8	82,732,751	82,761,115	IMPA1	8q21	Manic Depressive Psychosis	602064	Manic Depressive Psychosis	X				
chr15	46,487,797	46,725,210	FBN1	15q21	Marfan Syndrome	134797	Tall, heart defects		X			
chr14	100,252,981	100,281,225	DLK1	14q32	Maternal UPD	176290	Growth retardation and minor dysmorphic	X	X			
chr22	28,329,565	28,424,583	NF2	22q12	Meningioma / NF2	607379	Mental retardation and microcephaly	X	X			
chrX	77,042,046	77,200,000	ATP7A	Xq21	Menkes Disease	300011	Copper deficiency leading to mental and physical	X	X	X		
chr3	46,594,217	46,598,956	TDGF1	3p21	Mental Retardation	187395	Mental retardation and minor dysmorphic	X	X			
chr1	233,597,351	233,678,903	TBCE	1q42	Mental Retardation	604934	Mental retardation and physical	X	X			
chr10	89,613,175	89,718,512	PTEN	10q23	Mental Retardation	612242	Mental retardation and physical	X	X			
chr15	23,123,488	23,245,220	UBE3A	15q11	Mental Retardation	601623	Mental retardation and physical	X	X			
chr7	142,136,904	142,140,495	PRSS2	7q34	Mental Retardation	601564	Mental retardation and physical	X	X			
chr8	1,426,975	1,654,048	DLGAP2	8p23	Mental Retardation	605438	Mental retardation and physical	X	X			
chr16	971,809	976,980	SOX8	16p13	Mental Retardation associated with Alpha Thalaesamnia	605923	Mental retardation and alpha thalassaemia	X	X	X		
chr6	101,953,675	102,623,474	GRIK2	6q16	Mental Retardation, Autosomal Recessive 6	138244	Mental retardation and physical	X	X			
chr22	49,410,315	49,413,453	ARSA	22q13	Metachromatic Leukodystrophy	607574	Progressive muscle failure and speech	X	X	X		
chr3	182,903,176	182,926,972	SOX2	3q26	Microphthalmia	184429	Brain defect leading to severe developmental delay	X				
chr17	853,509	1,037,366	ABR	17p13	Miller-Dieker Syndrome	600365	Severe Mental and Physical	X	X			
chr17	1,194,593	1,250,267	YWHAE	17p13	Miller-Dieker Syndrome	605066	Severe Mental and Physical	X	X			
chr17	1,368,033	1,412,860	PITPNA	17p13	Miller-Dieker Syndrome	600174	Severe Mental and Physical	X	X			
chr17	2,443,686	2,535,638	PAFAH1B1(L1 S1)	17p13	Miller-Dieker Syndrome	601545	Severe Mental and Physical	X	X			
chr1	68,274,232	68,299,047	DIRAS3	1p31	Monosomy 1p31	605193	Mile DD and PD	X	X			
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	X	X			
chrX	31,048,667	33,277,779	DMD	Xq28	Muscular Dystrophy	300777	Muscle wasting, early diagnosis key		X			
chrX	149,487,727	149,592,272	MTM1	Xq28	Myotubular Myopathy	300415	Muscle, lung and heart defects		X			
chr9	128,416,569	128,503,132	LXM1B	9q33	Nail-Patella Syndrome	602575	Nail and knee defect		X			
chrX	17,303,464	17,664,034	NHS	Xp22	Nance-Horan Syndrome	300457	Eye defect		X			
chr10	21,110,094	21,503,122	NEBL	10p12	Nebulette	605491	Cardiac, immune and DD	X	X			
chrX	49,573,965	49,750,632	CLCN5	Xp11	Nephrolithiasis 2, X-linked, Dent Disease	300008	Kidney disease		X			
chr2	110,237,195	110,319,883	NPHP1	2q13	Nephronophthisis 1	607100	Kidney disease		X			
chr17	26,446,121	26,728,821	NF1	17q11	Neurofibromatosis Type 1	162200	Skeletal defects and learning disorders	X	X			
chr17	27,288,185	27,352,162	JJAZ1(SUZ12)	17q11	Neurofibromatosis Type 1	606245	Skeletal defects and learning disorders	X	X			
chr17	25,549,032	25,596,831	SLC6A4	17p11	Neuroticism	182138	Behavioural disorder	X				
chr1	61,320,883	61,694,624	NFIA	1p31	NFIA Haploinsufficiency	600727	Mental and physical	X	X			
chr12	111,340,919	111,432,099	PTPN11	12q24	Noonan 1	176876	Heart, short stature, dysmorphic.	X	X			
chr3	12,600,108	12,680,678	RAF1	3p25	Noonan Syndrome 5	164760	Heart, short stature, dysmorphic.	X	X			
chr2	39,066,469	39,201,067	SOS1	2p22	Noonan Syndrome Type 4	182530	Heart, short stature, dysmorphic.	X	X			
chrX	43,692,968	43,717,694	NDP	Xp11	Norrie Disease	300658	Eye, ear and some DD	X	X			
chr15	25,673,628	26,018,061	OCA2	15q13	Oculocutaneous Albinism Type 2	611409	Albino			X		
chr20	49,833,990	49,852,421	SALL4	20q13	Okhiro	607343	Eye and physical defects		X			
chr14	36,196,533	36,216,762	PAX9	14q13	Oligodontia		Missing teeth		X			
chrX	106,956,116	107,057,077	MID2	Xq22	Opitz / FP Syndrome	300204	MCA	X	X			
chrX	10,376,166	10,765,667	MID1	Xp22	Opitz Syndrome	300552	MCA	X	X			
chrX	70,255,131	70,279,029	MED12	Xq13	Opitz-Kavagga Syndrome	300188	MCA	X	X			
chr3	190,831,910	191,097,756	TP73L	3q28	Optic Atrophy	603273	Eye defect		X			
chrX	38,096,302	38,165,553	OTC	Xp11	Ornithine Transcarbamylase Deficiency	300461	Enzyme deficiency progressive	X	X	X		
chrX	13,662,801	13,697,391	OFD1	Xp22	Orofaciodigital 1	300170	Face, mouth, fingers and kidney		X			
chr17	45,616,456	45,633,992	COL1A1	17q21	Osteogenesis Imperfecta	120150	Bone defect		X			
chr12	63,849,638	63,928,374	LEMD3	12q14	Osteopoikilosis	607844	Bone defect		X			
chrX	102,918,410	102,934,201	PLP1	Xq22	Pelizaeus-Merzbacher Disease	300401	Central nervous system defect	X				
chrX	77,246,425	77,268,980	PGK1	Xp21	Phosphoglycerate Kinase Deficiency	311800	Anaemia and mental disorders	X		X		
chr18	51,045,967	51,454,183	TCF4	18q21	Pitt-Hopkins Syndrome	602272	Apnea, breathing, mental and physical problems	X	X			
chr16	2,078,712	2,125,900	PKD1	16p13	Polycystic Kidney Disease	601313	Kidney problems			X		
chr4	89,147,844	89,217,953	PKD2	4q22	Polycystic Kidney Disease	173910	Kidney problems			X		
chr7	156,168,394	156,378,602	LMBR1	7q36	Polydactyl Preaxial 2	605522	Thumb defect		X			
chr11	44,073,675	44,223,555	EXT2	11p11	Potocki-Shaffer Syndrome	608210	MCA Mental and Physical	X	X			
chr11	44,242,734	44,288,292	ALX4	11p11	Potocki-Shaffer Syndrome	605420, 60	MCA Mental and Physical	X	X			
chr13	65,774,967	66,702,464	PCDH9	13q21	Protocadherin 9 - EFMR Disorder	603581	Mental retardation and Epilepsy	X				
chr2	31,603,160	31,659,544	SRD5A2	2p23	Pseudovaginal Perineoscrotal Hypospadias	607306	Sex between male and female		X			
chrX	19,271,972	19,287,886	PDHA1	Xp22	Pyruvate Dehydrogenase Deficiency	300502	Enzyme deficiency progressive	X	X			
chr17	33,120,548	33,179,182	TCF2(HNF1B)	17q12	Renal Cysts and Diabetes Syndrome	189907	Renal Cysts and Diabetes Syndrome		X			
chr13	47,775,912	47,954,023	RB1	13q14	Retinoblastoma	180200	Eye disorder		X			
chrX	152,940,458	153,016,323	MECP2	Xq28	Retts Syndrome	300260	Various mental and physical defects.	X	X			
chrX	99,785,819	99,812,952	SRPX2	Xq22	Rolandic Epilepsy, X-linked Mental Retardation, and Speech Dyspraxia.	300642	Mental and physical	X	X			
chr16	3,642,941	3,648,097	DNASE1	16p13	Rubinstein Taybi Syndrome	125505	Mental, physical and cancer	X	X		X	
chr16	3,717,720	3,870,723	CREBBP	16p13	Rubinstein Taybi Syndrome	600140	Mental, physical and cancer	X	X		X	
chr7	19,121,616	19,123,550	TWIST1	7p21	Saethre-Chotzen	601622	Mostly physical, mild mental	X	X			
chr10	119,291,946	119,299,043	EMX2	10q26	Schizencephaly	600335	Mental and physical	X	X			
chr17	39,186,625	39,191,682	SOST	17q21	Sclerostin	605740	Bone disease		X			
chr2	166,553,916	166,638,395	SCN1A	2q24	SCN1A Related Seizures	182389	Seizures	X				
chr6	100,943,471	101,018,272	SIM1	6q16	Severe Obesity	603128	Obesity		X			
chr1	178,466,065	178,510,811	LHX4	1q25	Short Stature	602146	Short stature		X			

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords	Learning & Behavioural	Physical	Metabolic & Organ	Hereditary Cancers	Syndromes Selected
chr7	50,615,258	50,838,651	GRB10	7p12	Silver-Russell Syndrome	161523	Treatable dwarfism		X			
chrX	132,497,442	132,947,332	GPC3	Xq26	Simpson, Golabi, Behmel Syndrome Type 1	300037	overgrowth		X			
chr11	70,823,105	70,837,125	DHCR7	11q13	Smith-Lemli-Opitz	602858	mild to severe mental and physical	X	X			
chr17	17,525,512	17,655,488	RAH1	17p11	Smith-Magenis Syndrome	607642	Physical, behavioural and mental	X	X			
chr5	176,492,686	176,659,820	NSD1	5q35	Sotos Syndrome	606681	Overgrowth, behavioural and some cancer	X	X		X	
chr7	96,140,173	96,177,139	SHFM1	7q21	Split Hand and Foot	183600	Abnormal hand/feet some mental	X	X			
chr7	96,477,643	96,502,078	DLX5	7q21	Split Hand and Foot	600028	Abnormal hand/feet some mental	X	X			
chr10	103,103,815	103,307,060	BTRC	10q24	Split Hand and Foot Malformation-3	603482	Abnormal hand/feet some mental	X	X			
chr10	103,328,629	103,338,004	POLL	10q24	Split Hand and Foot Malformation-3	606343	Abnormal hand/feet some mental	X	X			
chr10	103,360,412	103,444,733	FBXW4	10q24	Split Hand and Foot Malformation-3	600095	Abnormal hand/feet some mental	X	X			
chr10	102,976,723	102,978,707	LBX1	10q24	Split Hand and Foot Malformation-3	604255	Abnormal hand/feet some mental	X	X			
chr2	172,658,454	172,662,647	DLX1	2q31	Split Hand and Foot Malformation-5	600029	Abnormal hand/feet some mental	X	X			
chr2	172,672,412	172,675,724	DLX2	2q31	Split Hand and Foot Malformation-5	126255	Abnormal hand/feet some mental	X	X			
chrX	6,461,659	6,463,159	VCX3A	Xp22	Steroid Sulfatase	300533	Physical, behavioural and mental	X	X			
chrX	7,075,293	7,282,680	STS	Xp22	Steroid Sulfatase	300747	Physical, behavioural and mental	X	X			
chr12	46,653,018	46,684,528	COL2A1	12q13	Stickler 1	120140	Joint, hearing and tongue		X			
chr1	103,115,583	103,346,640	COL11A1	1p21	Stickler Syndrome	120280	Joint, hearing and tongue		X			
chr2	176,653,081	176,656,936	EVX2	2q31	Synpolydactyly	142991	Fused or extra fingers/toes		X			
chr2	176,664,382	176,767,892	HOXDgenes	2q31	Synpolydactyly	142989	Fused or extra fingers/toes		X			
chr2	176,665,778	176,668,912	HOXD13	2q31	Synpolydactyly	142989	Fused or extra fingers/toes		X			
chr2	176,695,334	176,697,891	HOXD9	2q31	Synpolydactyly	142982	Fused or extra fingers/toes		X			
chr17	42,686,207	42,745,076	ITGB3	17q21	Thrombasthenia of Glanzmann and Naegeli	173470	Blood disorder			X		
chr1	145,841,561	145,848,017	GJA8	1q21	Thrombocytopenia Absent Radius Syndrome	600897	Arm, blood and platelet deformities Treatable in utero.		X			
chr12	1,950,490	2,677,376	CACNA1C	12p13	Timothy Syndrome	114205	Heart rhythm defects and Autism like features	X	X			
chr16	49,727,387	49,742,684	SALL1	16q12	Townes Brocks Syndrome	602218	Various physical defects and some learning disorders	X	X			
chr8	116,489,900	116,750,429	TRPS1	8q23	Trischororhinophlangeal, Langer-Giedion Syndrome	604386	Bone abnormalities, cancer and some learning disorders	X	X		X	
chr9	134,756,557	134,809,841	TSC1	9q34	Tuberous Sclerosis	605284	Abnormal benign tumour growth and learning disorders	X	X			
chr16	2,038,600	2,078,713	TSC2	16p13	Tuberous Sclerosis, Polycystic Kidney Disease	191902	Abnormal benign tumour growth, kidney and learning disorders	X	X			
chr12	113,592,442	113,606,352	TBX3	12q24	Ulnar-Mammary Syndrome	601621	Bone, genital and nipple defects.		X			
chr1	208,027,885	208,046,102	IRF6	1q32	Van der Woude Syndrome	607199	Facial defects and mild learning disability		X			
chr6	43,845,931	43,862,201	VEGFA	6p21	Vascular Endothelial Growth Factor Disorders	192240	Variety of physical disorders		X			
chr7	113,513,618	114,118,328	FOXP2	7q31	Verbal Dyspraxia	605317	Severe speech and language disorder	X				
chr2	130,995,137	131,002,053	CFC1	2q21	Visceral Heterotaxy	605194	Randomized organ and arterial placement		X			
chr3	10,158,319	10,168,746	VHL	3p25	Von Hippel Lindau	608537	Cyst and tumour formation, cancer		X		X	
chr2	222,772,851	222,871,944	PAX3	2q36	Waardenburg Syndrome I	606597	Hearing loss and pigmentation problems		X			
chr3	69,871,323	70,100,177	MITF	3p14	Waardenburg Syndrome IIA	158645	Hearing loss and pigmentation problems		X			
chr11	32,355,900	32,423,662	WT1	11p13	WAGR Syndrome	607102	Various mental, visual and physical defects.	X	X			
chr7	73,080,407	73,120,966	ELN	7q11	Williams-Beuren Syndrome	130160	Mental and physical	X	X			
chr7	73,136,092	73,174,790	LIMK1	7q11	Williams-Beuren Syndrome	194050	Mental and physical	X	X			
chr7	73,826,245	73,841,595	NCF1	7q11	Williams-Beuren Syndrome	608512	Mental and physical	X	X			
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	X	X			
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	X	X			
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	X	X			
chrX	72,863,307	73,063,491	XIST	Xq13	X Inactivation Specific Transcript	314670	Severe physical and mental	X	X			
chrX	100,491,098	100,532,426	BTK	Xq22	X-linked agammaglobulinemia	300300	Immune disorder, treatable			X		
chrX	107,569,810	107,827,431	COL4A5	Xq22	X-linked Alport Syndrome	303630	Hearing, eye and kidney defects		X	X		
chrX	106,758,415	106,780,912	PRPF1	Xq22	X-linked Charcot Marie Tooth Disease	311850	Progressive muscle and nerve wasting - late onset		X			
chrX	53,417,795	53,466,343	SMC1L1(SMC1A)	Xp11	X-linked Cornelia de Lange Syndrome	300040	Broad spectrum physical and mental	X	X			
chrX	153,644,344	153,659,154	DKC1	Xq28	X-linked Dyskeratosis Congenita	300125	Various physical defects and some learning disorders	X	X			
chrX	136,476,012	136,481,923	ZIC3	Xq26	X-linked Heterotaxy	300265	Left-right assymetry, severe organ defects		X			
chrX	18,567,731	18,600,150	RS1	Xp22	X-linked Juvenile Retinoschisis	312700	Eye disorder		X			
chrX	123,307,875	123,334,696	SH2D1A	Xq25	X-linked Lymphoproliferative Syndrome Type 1	300490	Immune disorder and cancer			X	X	
chrX	122,821,729	122,875,033	BIRC4(XIAP)	Xq25	X-linked Lymphoproliferative Syndrome Type 2	300079	Immune disorder and cancer			X	X	
chrX	53,474,931	53,478,048	HADH2(HSD17B10)	Xp11	X-linked Mental Retardation	300256	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	38,305,683	38,433,116	TM4SF2(TSPAN7)	Xp11	X-linked Mental Retardation	300096	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	39,795,561	39,921,526	BCOR	Xp11	X-linked Mental Retardation	300485	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	40,325,160	40,350,832	ATP6AP2	Xp11	X-linked Mental Retardation	300566	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	41,264,287	41,667,212	CASK	Xp11	X-linked Mental Retardation	300172	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	47,191,350	47,227,289	ZNF41	Xp11	X-linked Mental Retardation	314995	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	47,316,244	47,364,200	SYN1	Xp11	X-linked Mental Retardation	311040	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	47,379,864	47,394,964	ELK1	Xp11	X-linked Mental Retardation	311040	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	47,581,245	47,666,550	ZNF81	Xp11	X-linked Mental Retardation	314998	Most common mental retardation phenotype with some dysmorphic features	X	X			

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chrX	48,201,871	48,213,509	SLC38A5	Xp11	X-linked Mental Retardation	300649	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	48,219,493	48,229,696	FTSJ1	Xp11	X-linked Mental Retardation	300499	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	48,252,315	48,264,146	PORCN	Xp11	X-linked Mental Retardation	300651	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	48,640,139	48,645,364	PQBP1	Xp11	X-linked Mental Retardation	300463	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	50,351,387	50,573,784	SHROOM4(KI AA1202)	Xp11	X-linked Mental Retardation	300579	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	53,237,378	53,271,329	JARID1C	Xp11	X-linked Mental Retardation	314690	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	53,979,838	54,088,121	PHF8	Xp11	X-linked Mental Retardation	300560	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	56,275,632	56,328,254	KLF8	Xp11	X-linked Mental Retardation	300286	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	24,932,213	24,943,775	ARX	Xp21	X-linked Mental Retardation	300382	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	28,515,480	29,884,757	IL1RAPL1	Xp21	X-linked Mental Retardation	300206	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	79,818,351	79,951,889	BRWD3	Xp21	X-linked Mental Retardation	300553	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	11,039,373	11,051,122	HCCS	Xp22	X-linked Mental Retardation	300056	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	14,771,450	14,801,105	FANCB	Xp22	X-linked Mental Retardation	300515	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	15,753,850	15,783,021	AP1S2	Xp22	X-linked Mental Retardation	300629	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	8,393,346	8,394,346	VCX3A	Xp22	X-linked Mental Retardation		Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	21,868,763	21,922,876	SMS	Xp22	X-linked Mental Retardation	300105	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	108,771,220	108,863,275	ACSL4	Xp22	X-linked Mental Retardation	300157	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	110,074,169	110,350,829	PAK3	Xp22	X-linked Mental Retardation	300142	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	110,423,663	110,541,962	DCX	Xp22-p2	X-linked Mental Retardation	300121	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	69,581,449	69,642,062	DLG3	Xq13	X-linked Mental Retardation	300189	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	70,376,199	70,391,660	ZNF261(ZMY M3)	Xq13	X-linked Mental Retardation	30061	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	73,870,137	74,061,709	KIAA2022	Xq13	X-linked Mental Retardation	300524	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	74,508,786	74,659,600	ZDHHC15	Xq13	X-linked Mental Retardation	300576	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	100,973,741	100,999,205	NXF5	Xq22	X-linked Mental Retardation	300319	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	115,216,031	115,219,848	AGTR2	Xq23	X-linked Mental Retardation	300034	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	118,592,527	118,602,407	UBE2A	Xq23	X-linked Mental Retardation	312180	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	118,852,017	118,870,996	UPF3B	Xq24	X-linked Mental Retardation	300298	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	118,889,762	118,894,646	NDUFA1	Xq24	X-linked Mental Retardation	300078	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	119,542,474	119,593,712	CUL4B	Xq24	X-linked Mental Retardation	300304	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	122,145,839	122,450,474	GRIA3	Xq24	X-linked Mental Retardation	305915	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	128,766,594	128,805,805	ZDHHC9	Xq25	X-linked Mental Retardation	300646	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	135,575,377	135,691,169	ARHGEF6	Xq26	X-linked Mental Retardation	300267	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	153,279,912	153,283,874	RPL10	Xq28	X-linked Mental Retardation	209850	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	153,318,715	153,325,009	GDI1	Xq28	X-linked Mental Retardation	300104	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	139,411,973	139,415,436	SOX3	Xq27	X-linked Mental Retardation	313430	Most common mental retardation phenotype with some dysmorphic features	X	X			
chrX	134,895,252	134,957,094	SLC9A6	Xq26	X-linked Mental Retardation Syndrome Christianson Type	300231	Mental retardation, small head and speech defect	X	X			
chrX	73,557,810	73,670,475	SLC16A2	Xq13	X-linked Mental Retardation, Allan-Herndon-Dudley Syndrome	300095	Severe mental and weak muscle tone	X	X			
chrX	53,576,476	53,730,398	HUWE1	Xp11	X-linked Mental Retardation, Turner Syndrome	300697	Heart defects, ovary, short stature. Usually normal intelligence.		X			
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	X	X			

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords	Learning & Behavioural	Physical	Metabolic & Organ	Hereditary Cancers	Syndromes Selected
chrX	18,353,678	18,581,666	CDKL5	Xp22	X-linked Spasms	300203	Severe seizures and developmental delay.	X				
chrX	46,243,490	46,289,820	ZNF674	Xp11	Xp11.3 Deletion	300573	Mental retardation	X				
chrX	46,581,319	46,626,733	RP2	Xp11	Xp11.3 Deletion	300757	Mental retardation	X				
chr9	126,283,337	126,309,520	NR5A1	9q3	XY Sex Reversal	184757	Genetically male, but female sex organs		X			
chr19	62,384,680	62,432,350	ZNF264	19q13	Zinc Finger Protein-264	604668	Zinc finger protein - imprinting	X	X	X	X	
chr18	73,090,996	73,111,084	GALR1	18q23		600377	Behavioural disorders, cancer, Alzheimers link	X			X	
chr11	3,054,921	3,153,115	OSBP15	11p15		606733	Binding protein - imprinting			X		
chrX	154,595,003	154,913,754	PAR2	Xq28		300053	Bipolar disorder	X				
chr7	98,084,545	98,097,116	NPTX2	7q22		600750	Brain and immune function	X				
chr20	56,676,787	56,680,787	GNAS_ICRegion	20q13		603666	Broad spectrum physical and mental and cancer	X	X		X	
chr20	56,859,056	56,861,251	GNAS_ICRegion	20q13		139320	Broad spectrum physical and mental and cancer	X	X		X	
chr20	56,896,896	56,899,356	GNAS_ICRegion	20q13		139320	Broad spectrum physical and mental and cancer	X	X		X	
chr7	130,058,017	130,079,399	KLF14	7q32		609393	Can cause Basal cell carcinoma				X	
chr1	2,149,994	2,231,416	SKI	1p36		164780	Cancer				X	
chr1	3,548,988	3,649,715	TP73	1p36		601990	Cancer				X	
chr19	61,967,731	62,053,886	ZIM2	19q13		601483	Cancer				X	
chr19	62,005,614	62,053,875	PEG3	19q13		601483	Cancer				X	
chr19	62,040,958	62,043,979	PEG3_ICRegion	19q13		601483	Cancer				X	
chr20	35,573,020	35,595,505	NNAT	20q11		603106	Cancer				X	
chr2	202,949,916	203,140,719	BMPR2	2q32		600799	Causes high blood pressure in lungs			X		
chr2	239,634,801	239,987,580	HDAC4	2q37		605314	Causes mental retardation and dysmorphism	X	X			
chr17	41,327,544	41,461,546	MAPT	17q21		157140	Developmental delay, Alzheimers	X				
chr7	94,364,911	94,770,958	PPP1R9A	7q21		602468	Gene involved in brain function	X				
chr14	20,923,198	20,975,244	CHD8	14q11		610528	Growth retardation	X	X			
chr7	129,710,229	129,761,249	CPA4	7q32		607635	Growth retardation and dysmorphic	X	X			
chr7	129,903,281	129,943,368	MEST	7q32		601029	Growth retardation and minor dysmorphic	X	X			
chr11	2,671,987	2,683,633	KCNQ1_ICRegion	11p15		607542	Heart defects		X			
chr14	20,889,476	20,922,265	SUPT16H	14q11		605012	Heart failure			X		
chr14	20,897,000	20,932,000	MicroDeletion Region	14q11		605012	Heart failure		X	X		
chr11	1,975,651	1,980,951	H19_ICRegion	11p15			Imprinting centre	X	X	X	X	
chr20	41,566,487	41,613,948	L3MBTL	20q13		608802	Involved in cancers				X	
chr15	49,527,231	49,702,259	DMXL2	15q21		612186	Involved in cortex	X				
chr8	140,683,985	140,794,480	KCNK9	8q24		605874	Known as Birk-Barel Syndrome. Mental retardation and dysmorphism	X	X			
chr14	100,352,214	100,407,118	MEG3	14q32		605636	Linked to cancer				X	
chr22	16,454,903	16,491,588	ATP6V1E1	22q11		108746	Linked to muscle abnormalities		X			
chr17	41,217,449	41,268,973	CRHR1	17q21		122561	Mental and eating disorders	X				
chrX	1	2,700,000	PAR1	Xp22			Mental and physical	X	X			
chr16	6,009,133	7,702,500	A2BP1	16p13		605104	Mental retardation and epilepsy	X				
chr14	28,306,038	28,308,622	FOXG1B	14q12			Microcephaly and mental retardation	X	X			
chrY	2,714,821	2,716,600	SRY	Yp11		480000	Sexual defects		X			
chrX	505,079	540,146	SHOX	Xp22		400020	Short stature		X			
chr18	42,798,570	42,820,446	TCEB3C	18q21		609522	Transcription factor - imprinting	X	X	X	X	
chr14	100,261,144	100,364,369	DLK1&MEG3_ICRegion	14q32		176290	UPD, mental and physical disorders	X	X			

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.