

Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chrX	30,232,507	30,237,413	NR0B1(DAX1)	Xp21	Adrenal Hypoplasia Congenita	300473	Sterility
chr20	10,566,334	10,602,590	JAG1	20p12	Alagille Syndrome	601920	Late onset organ failure
chr16	155,725	171,196	HBA1&HBA2	16p13	Alpha Thalassemia Mental Retardation	141850	Blood disorder
chrX	76,647,847	76,928,358	ATRX	Xq21	Alpha Thalassemia Mental Retardation Syndrome	300032	Blood disorder
chrY	13,052,929	13,603,474	AZFa	Yq11	AZFa Region	400005	Male sterility
chrY	23,539,798	23,560,998	BPY2	Yq11	AZFc Region	400013	Male sterility
chrY	23,663,736	25,513,794	AZFc	Yq11	AZFc Region	400003	Male sterility
chrY	23,684,890	23,754,627	DAZ1	Yq11	AZFc Region	400003	Male sterility
chrY	24,600,764	24,603,549	CDY1	Yq11	AZFc Region	400016	Male sterility
chrY	24,765,503	24,770,366	GOLGA2LY(A F332229)	Yq11	AZFc Region	400035	Male sterility
chrY	25,173,539	25,194,740	BPY2	Yq11	AZFc Region	400016	Male sterility
chrY	25,586,438	25,607,639	BPY2	Yq11	AZFc Region	400013	Male sterility
chrY	26,010,846	26,015,709	GOLGA2LY(A F332229)	Yq11	AZFc Region	400035	Male sterility
chrY	26,177,652	26,180,436	CDY1	Yq11	AZFc Region	400016	Male sterility
chr11	128,213,125	128,242,478	KCNJ1	11q24	Bartter 2	600359	Kidney and blood
chr1	16,221,073	16,256,063	CLCNKB	1p36	Bartter 3	602023	Kidney and blood
chr1	55,237,205	55,247,053	BSND	1p32	Bartter 4	606412	Kidney and blood
chr1	16,217,957	16,233,132	CLCNKA	1p36	Bartter 4	602024	Kidney and blood
chr15	46,285,791	46,382,417	SLC12A1	15q21	Bartter Type 1	600839	Kidney and blood
chr3	123,385,220	123,488,032	CASR	3q13	Bartter with Autosomal Dominant Hypocalcemia	601199	Kidney and blood
chr3	140,144,730	140,149,880	FOXL2	3q22	Blepharophimosis	605597	Ovary failure and cancer
chr8	72,272,222	72,437,021	EYA1	8q13	Branchio-Oto-Renal Syndrome	602064	Kidney failure
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
chrX	85,002,841	85,189,222	CHM	Xp21	Choroideremia	300390	Eye defect
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	37,524,264	37,557,658	CYBB	xp11	Chronic Granulomatous Disease	300481	Immune disorder
chr5	147,184,339	147,191,453	SPINK1	5q32	Chronic Pancreatitis	167790	Pancreas damage
chr6	32,044,642	32,162,690	CYP21A2	6p21	Congenital Adrenal Hyperplasia	201910	Infertility and sex disorders
chr10	88,506,376	88,674,925	BMPR1A	10q23	Cowden	601728	Polyps and cancer
chr17	3,486,511	3,513,146	CTNS	17p13	Cystinosis	606272	Kidney and eye defect
chr2	44,356,103	44,401,443	SLC3A1	2p21	Cystinuria with Mitochondrial Disease	104614	Kidney disease
chr2	44,399,406	44,442,448	PREPL	2p21	Cystinuria with Mitochondrial Disease	104614	Kidney disease
chrX	119,444,035	119,487,189	LAMP2	Xq24	Danon Disease	309606	Heart defect
chr13	19,659,605	19,665,114	GJB2(Connexin 26)	13q12	Deafness	121011	Deafness
chr5	149,717,428	149,760,063	TCOF1	5q33	Deafness	606847	Deafness
chrX	100,487,306	100,490,343	TIMM8A	Xq22	Deafness-Dystonia-Optic Neuropathy	300356	Deafness and eye
chrX	152,821,179	152,825,834	AVPR2	Xq28	Diabetes Insipidus, Nephrogenic, X-linked Syndrome	300538	Diabetes
chr6	144,293,129	144,427,428	ZAC(PLAGL1)	6q24	Diabetes Mellitus, Transient Neonatal, 1	603044	Diabetes
chr6	144,370,000	144,377,576	HYMAI	6q24	Diabetes Mellitus, Transient Neonatal, 1	603044	Diabetes
chr19	47,055,828	47,067,324	RPS19	19q13	Diamond Blackfan Anemia	603474	Blood and MCA
chr16	88,331,460	88,410,566	FANCA	16q24	Fanconi Anemia	607139	Blood, cancer
chr5	112,071,117	112,209,835	APC	5q22	Gardner Syndrome	601731	Polyps and cancer
chr16	55,456,620	55,507,263	SLC12A3	16q13	Gitelman Syndrome	610966	Kidney
chrX	30,581,461	30,658,645	GK	Xp21	Glycerol Kinase Deficiency	300474	Mild mental and metabolic
chrX	153,717,263	153,904,192	F8	Xq28	Hemophilia	306700	Blood disorder
chrX	138,440,561	138,473,283	F9	Xq27	Hemophilia B	300746	Blood disorder
chr18	46,810,581	46,865,409	MADH4(SMAD4)	18q21	Hereditary Hemorrhagic Telangiectasia Syndrome	600993	Blood vessel disorder
chr15	97,010,284	97,325,282	IGF1R	15q25	IGF-1 Resistance	147307	Insulin defect
chr15	41,612,952	41,664,382	KIAA0377(HISPPD2A)	15q15	Infertility and Deafness	601979	Infertility and deafness
chr15	41,710,064	41,747,192	CATSPER2	15q15	Infertility and Deafness	611102	Infertility and deafness
chr8	38,387,813	38,445,509	FGFR1	8p12	Kallman 1	136350	Sex hormone defect
chrX	8,456,915	8,660,227	KAL1	Xp22	Kallmann Syndrome	308700	Sex hormone defect
chrX	133,421,923	133,462,362	HPRT1	Xq26	Lesch-Nyhan Syndrome	308000	Progressive MD and gout
chr11	2,412,796	2,836,915	KCNQ1	11p15	Long QT Syndrome	607542	Heart rhythm defect
chrX	77,042,046	77,200,000	ATP7A	Xq21	Menkes Disease	300011	Copper deficiency leading to mental



Metabolic & Organ



Chromosome	start (bp)	stop (bp)	Gene	Band	Syndrome	OMIM	Keywords
chr16	971,809	976,980	SOX8	16p13	Mental Retardation associated with Alpha	605923	Mental retardation and alpha
chr22	49,410,315	49,413,453	ARSA	22q13	Metachromatic Leukodystrophy	607574	Progressive muscle failure and speech
chr15	25,673,628	26,018,061	OCA2	15q13	Oculocutaneous Albinism Type 2	611409	Albino
chrX	38,096,302	38,165,553	OTC	Xp11	Ornithine Transcarbamylase Deficiency	300461	Enzyme deficiency progressive
chrX	77,246,425	77,268,980	PGK1	Xp21	Phosphoglycerate Kinase Deficiency	311800	Anaemia and mental disorders
chr16	2,078,712	2,125,900	PKD1	16p13	Polycystic Kidney Disease	601313	Kidney problems
chr4	89,147,844	89,217,953	PKD2	4q22	Polycystic Kidney Disease	173910	Kidney problems
chr17	42,686,207	42,745,076	ITGB3	17q21	Thrombasthenia of Glanzmann and Naegeli	173470	Blood disorder
chrX	100,491,098	100,532,426	BTK	Xq22	X-linked agammaglobulinemia	300300	Immune disorder, treatable
chrX	107,569,810	107,827,431	COL4A5	Xq22	X-linked Alport Syndrome	303630	Hearing, eye and kidney defects
chrX	123,307,875	123,334,696	SH2D1A	Xq25	X-linked Lymphoproliferative Syndrome Type 1	300490	Immune disorder and cancer
chrX	122,821,729	122,875,503	BIRC4(XIAP)	Xq25	X-linked Lymphoproliferative Syndrome Type 2	300079	Immune disorder and cancer
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
chr18	42,798,570	42,820,446	TCEB3C	18q21		609522	Transcription factor - imprinting
chr14	20,897,000	20,932,000	MicroDeletion Region	14q11		605012	Heart failure
chr11	3,054,921	3,153,115	OSBPL5	11p15		606733	Binding protein - imprinting
chr14	20,889,476	20,922,265	SUPT16H	14q11		605012	Heart failure
chr2	202,949,916	203,140,719	BMPR2	2q32		600799	Causes high blood pressure in lungs

**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.