

Comparison of gene content of ISCA arrays

syndrome	chromosome	start (bp)	stop (bp)	Gene	Band	OMIM	ISCA 4x44k	ISCA 4x180k	ISCA 8x60k
1p36 deletion	chr1	2149994	2231416	SKI	1p36	164780	no	yes	yes
1p36 deletion	chr1	3548988	3649715	TP73	1p36	601990	no	yes	yes
Barter 4	chr1	16217957	16233132	CLCNKA	1p36	602024	no	yes	yes
Barter 3	chr1	16221073	16256063	CLCNKB	1p36	602023	no	yes	yes
Barter 4	chr1	55237205	55247053	BSND	1p32	606412	no	yes	yes
NFIA Haploinsufficiency	chr1	61320883	61694624	NFIA	1p31	600727	low coverage	yes	yes
monosomy 1p31 p22	chr1	68274232	68299047	DIRAS3	1p31	605193	no	yes	yes
Stickler syndrome	chr1	1.03E+08	1.03E+08	COL11A1	1p21	120280	yes	yes	yes
atrial fibrillation	chr1	1.46E+08	1.46E+08	GJA5	1q21	121013	yes	yes	yes
Thrombocytopenia absent radius syndrome	chr1	1.46E+08	1.46E+08	GJA8	1q21	600897	yes	yes	yes
Short stature	chr1	1.78E+08	1.79E+08	LHX4	1q25	602146	yes	yes	yes
Van der Woude syndrome	chr1	2.08E+08	2.08E+08	IRF6	1q32	607199	yes	yes	yes
Fryns 1q41 autism	chr1	2.21E+08	2.21E+08	DISP1	1q41	607502	no	yes	yes
	chr1	2.3E+08	2.3E+08	DISC1	1q42	605210	low coverage	yes	yes
MR	chr1	2.34E+08	2.34E+08	TBCE	1q42	604934	no	yes	yes
diGeorge 2	chr10	8136673	8157170	GATA3	10p14	131320	yes	yes	yes
Nebulette	chr10	21110094	21503122	NEBL	10p12	605491	yes	yes	yes
Hirschsprung Disease Plus	chr10	42892523	42945803	RET	10q11	164761	low coverage	yes	yes
autism	chr10	64241763	64246133	EGR2	10q21	129010	no	yes	yes
10q22-23 deletion	chr10	83625077	84735341	NRG3	10q22	605533	yes	yes	yes
10q22-23 deletion	chr10	87349292	88116230	GRID1	10q22	610659	low coverage	yes	yes
Cowden	chr10	88506376	88674925	BMPR1A	10q23	601728	no	yes	yes
MR	chr10	89613175	89718512	PTEN	10q23	612242	yes	yes	yes
CATARACT, JUVENILE, WITH MICROCORNEA AND GLUCOSURIA	chr10	91180753	91285293	SLC16A12	10q23	611910	no	yes	yes
Split/hand foot malformation -3	chr10	1.03E+08	1.03E+08	LBX1	10q24	604255	no	yes	yes
Split/hand foot malformation -3	chr10	1.03E+08	1.03E+08	BTRC	10q24	603482	no	yes	yes
Split/hand foot malformation -3	chr10	1.03E+08	1.03E+08	POLL	10q24	606343	no	yes	yes
Split/hand foot malformation -3	chr10	1.03E+08	1.03E+08	FBXW4	10q24	600095	yes	yes	yes
Schizencephaly	chr10	1.19E+08	1.19E+08	EMX2	10q26	600035	yes	yes	yes
Beckwith-Wiedemann	chr11	1962981	1985640	H19	11p15	103280	no	yes	yes
a non-coding region	chr11	1975651	1980951	H19_ICRegion	11p15		no	yes	yes
Beckwith-Wiedemann	chr11	2096925	2136469	IGF2	11p15	147470	yes	yes	yes
Beckwith-Wiedemann	chr11	2127584	2148999	INS	11p15	176730, 191290	no	yes	yes
long QT syndrome	chr11	2412796	2836915	KCNQ1	11p15	607542	yes	yes	yes
	chr11	2671987	2683633	KCNQ1_ICRegion	11p15	607542	low coverage	yes	yes
Beckwith-Wiedemann	chr11	2851440	2873550	CDKN1C	11p15	607542	low coverage	yes	yes
Beckwith-Wiedemann	chr11	2867526	2913051	SLC22A18	11p15	602631	no	yes	yes
Beckwith-Wiedemann	chr11	2896078	2917225	PHLDA2	11p15	602631	no	yes	yes
	chr11	3054921	3153115	OSBPL5	11p15	606733	no	yes	yes
Craniosynostosis	chr11	15949900	16454494	SOX6	11p15.1-p15.2	607257	low coverage	yes	yes
Infantile hyperinulinism, enteropathy and deafness (Ushers disease)	chr11	17371008	17455025	ABCC8	11p15	600509	no	yes	yes
Infantile hyperinulinism, enteropathy and deafness (Ushers disease)	chr11	17472018	17522539	USH1C	11p15	605242	no	yes	yes
Aniridia	chr11	31762916	31796085	PAX6	11p13-p14	607108	yes	yes	yes
WAGR Syndrome probes	chr11	32355900	32423662	WT1	11p13	607102	yes	yes	yes
Potocki-Shaffer syndrome	chr11	44073675	44223555	EXT2	11p11	608210	low coverage	yes	yes
Potocki-Shaffer syndrome	chr11	44242734	44288292	ALX4	11p11	605420, 601224	yes	yes	yes

Leukodystrophy	chr11	67130983	67136581	NDUFV1	11q13	161015	yes	yes	yes
Smith-Lemli-Opitz	chr11	70823105	70837125	DHCR7	11q13	602858	no	yes	yes
11q14 microdeletion	chr11	86334369	86344081	FZD4	11q14	604579	no	yes	yes
Bartter 2	chr11	1.28E+08	1.28E+08	KCNU1	11q24	600359	no	yes	yes
Timothy syndrome	chr12	1950490	2677376	CACNA1C	12p13	114205	low coverage	yes	yes
Sticker 1	chr12	46653018	46684528	COL2A1	12q13	120140	yes	yes	yes
Osteopokilosis	chr12	63849638	63928374	LEMD3	12q14	607844	no	yes	yes
12q14.3 deletion	chr12	65029066	65359020	GRIP1	12q14	604597	low coverage	yes	yes
Joubert 5	chr12	86966921	87060124	CEP290	12q21	610142	no	yes	yes
Noonan 1	chr12	1.11E+08	1.11E+08	PTPN11	12q24	176876	yes	yes	yes
Holt-Oram syndrome	chr12	1.13E+08	1.13E+08	TBX5	12q14	601620	no	yes	yes
Ulnar-Mammary	chr12	1.14E+08	1.14E+08	TBX3	12q24	601621	no	yes	yes
Deafness	chr13	19659605	19665114	GJB2(Connexin 26)	13q12	121011	no	yes	yes
CRYPTORCHIDISM, UNILATERAL OR BILATERAL	chr13	31211679	31275009	LGR8(RXFP2)	13q13	606655	no	yes	yes
Breast cancer	chr13	31787617	31871809	BRCA2	13q13	600185	no	yes	yes
Retinoblastoma	chr13	47775912	47954023	RB1	13q14	180200	yes	yes	yes
13q21.32 del	chr13	65774967	66702464	PCDH9	13q21	603581	yes	yes	yes
Protocadherin 9	chr13	77367617	77447665	EDNRB	13q22	131244	no	yes	yes
Hirschsprung Disease Plus	chr13	90848888	92317488	GPC5	13q31	602446	yes	yes	yes
brachydactyly	chr13	92677096	93853948	GPC6	13q31	604404	yes	yes	yes
Holopresencaphyl 5	chr13	99432320	99437019	ZIC2	13q32	603073	yes	yes	yes
14q11.2 deletion	chr14	20889476	20922265	SUPT16H	14q11	605012	no	yes	yes
14q11.2 deletion	chr14	20897000	20932000	MicrodeletionRegion	14q11	605012	no	yes	yes
14q11.2 deletion	chr14	20923198	20975244	CHD8	14q11	610528	no	yes	yes
14q12 deletion	chr14	28306038	28308622	FOXG1B	14q12		yes	yes	yes
Oligodontia	chr14	36196533	36216762	PAX9	14q13		yes	yes	yes
14q22 microdeletion, Orofacial cleft 11	chr14	53486207	53493362	BMP4	14q22	112262	no	yes	yes
14q22 microdeletion, Microphthalmia, syndromic 5	chr14	56337178	56346937	OTX2	14q22	600037	no	yes	yes
14q22 microdeletion	chr14	60045150	60048904	SIX6	14q22	606326	no	yes	yes
14q22 microdeletion	chr14	60182506	60185933	SIX1	14q22	601205	no	yes	yes
14q22 microdeletion	chr14	60246009	60260792	SIX4	14q22	606342	no	yes	yes
maternal UPD	chr14	1E+08	1E+08	DLK1	14q32	176290	no	yes	yes
	chr14	1E+08	1E+08	DLK1&MEG3_ICregion	14q32	176290	no	yes	yes
	chr14	1E+08	1E+08	MEG3	14q32	605636	no	yes	yes
Angelman / Prader Willi	chr15	20556790	20585849	NIPA2	15q11	608146	yes	yes	yes
Angelman / Prader Willi	chr15	20594720	20637877	NIPA1	15q11	608146	yes	yes	yes
Angelman / Prader Willi	chr15	21361547	21407800	MKRN3	15q11	603586	no	yes	yes
Angelman / Prader Willi	chr15	21429787	21452081	MAGEL2	15q11	605283	no	yes	yes
Angelman / Prader Willi	chr15	21471646	21493542	NDN	15q11	602117	no	yes	yes
Angelman / Prader Willi	chr15	22619887	22774822	SNRPN	15q11	182279	yes	yes	yes
Angelman / Prader Willi	chr15	22713304	22720548	PWS_ICregion	15q11	182279	no	yes	yes
Angelman / Prader Willi	chr15	22741227	22784821	SNURF	15q11	182279	low coverage	yes	yes
Angelman / Prader Willi	chr15	22747536	22751866	PWS_ICregion	15q11	182279	no	yes	yes
Angelman / Prader Willi	chr15	22751228	22774822	SNRPN	15q11	182279	no	yes	yes
Angelman / Prader Willi	chr15	22768233	22793232	SNORD107/64/108	15q11	182279	no	yes	yes
Angelman / Prader Willi	chr15	22835000	23010100	SnoRNA	15q11	601491	no	yes	yes
Angelman / Prader Willi	chr15	23064582	23084648	SNORD109B	15q11		no	yes	yes
MR	chr15	23123488	23245220	UBE3A	15q11	601623	yes	yes	yes
autism	chr15	23463512	23669962	ATP10A	15q12	605855	no	yes	yes
autism	chr15	24339787	24767950	GABRB3	15q12	137192	low coverage	yes	yes
Oculocutaneous albinism 2	chr15	25673628	26018061	OCA2	15q13	611409	low coverage	yes	yes
15q13.3 microdeletion syndr	chr15	30110018	30248527	CHRNA7	15q13	118511	no	yes	yes
15q15.3 infertility and deafness	chr15	41612952	41664382	KIAA0377(HISPPD2A)	15q15	601979	no	yes	yes
15q15.3 infertility and deafness	chr15	41710064	41747192	CATSPER2	15q15	611102	no	yes	yes
Bartter 1	chr15	46285791	46382417	SLC12A1	15q21	600839	no	yes	yes
MARFAN SYNDROME	chr15	46487797	46725210	FBN1	15q21	134797	yes	yes	yes
	chr15	49527231	49702259	DMXL2	15q21	612186	no	yes	yes
Congenital diaphragmatic hernia	chr15	91244423	91372241	CHD2	15q26	602119	yes	yes	yes
Congenital diaphragmatic hernia	chr15	94670161	94683047	NR2F2	15q26	107773	yes	yes	yes

IGF-1 resistance	chr15	97010284	97325282	IGF1R	15q25	147307	yes	yes	yes
Alpha thalassemia	chr16	155725	171196	HBA1&HBA2	16p13	141850	no	yes	yes
mental retardation									
Mental retardation associated with Alpha thalassaemia	chr16	971809	976980	SOX8	16p13	605923	yes	yes	yes
TUBEROUS SCLEROSIS, polycystic kidney disease	chr16	2038600	2078713	TSC2	16p13	191902	yes	yes	yes
Polycystic kidney disease	chr16	2078712	2125900	PKD1	16p13	601313	no	yes	yes
Rubinstein Taybi Syndrome	chr16	3642941	3648097	DNASE1	16p13	125505	no	yes	yes
RUBINSTEIN-TAYBI SYNDROME	chr16	3717720	3870723	CREBBP	16p13	600140	yes	yes	yes
16p13.2 del	chr16	6009133	7702500	A2BP1	16p13	605104	yes	yes	yes
Creatine deficiency syndrome / X-linked mental retardation (XLMR)	chr16	33686002	33694027	SLC6A8	16p11		no	yes	yes
Townes Brocks	chr16	49727387	49742684	SALL1	16q12	602218	no	yes	yes
Gitelman	chr16	55456620	55507263	SLC12A3	16q13	610966	yes	yes	yes
Bilateral frontoparietal polymicrogyria	chr16	56211151	56256445	GPR56	16q13	604110	no	yes	yes
DELAYED CRANIAL OSSIFICATION DUE TO CFBF HAPLOINSUFFICIENCY INCLUDED	chr16	65620551	65692459	CBFB	16q22	121360	no	yes	yes
FANCONI ANEMIA	chr16	88331460	88410566	FANCA	16q24	607139	no	yes	yes
Miller-Dieker	chr17	853509	1037366	ABR	17p13	600365	no	yes	yes
Miller-Dieker	chr17	1194593	1250267	YWHAE	17p13	605066	yes	yes	yes
Miller-Dieker	chr17	1368033	1412860	PITPNA	17p13	600174	no	yes	yes
Miller-Dieker	chr17	2443686	2535638	PAFAH1B1(LIS1)	17p13	601545	yes	yes	yes
Cystinosis	chr17	3486511	3513146	CTNS	17p13	606272	yes	yes	yes
Li-Fraumeni 1	chr17	7505822	7531642	TP53	17p13	191170	no	yes	yes
Charcot-Marie Tooth Disease	chr17	15073822	15109369	PMP22	17p11	601097	yes	yes	yes
Smith-Magenis	chr17	17525512	17655488	RAI1	17p11	607642	yes	yes	yes
NEUROTICISM	chr17	25549032	25586831	SLC6A4	17p11	182138	no	yes	yes
Neurofibromatosis 1	chr17	26446121	26728821	NF1	17q11	162200	low coverage	yes	yes
Neurofibromatosis 1	chr17	27288185	27352162	JJAZ1(SUZ12)	17q11	606245	no	yes	yes
RENAL CYSTS AND DIABETES SYNDROME	chr17	33120548	33179182	TCF2(HNF1B)	17q12	189907	yes	yes	yes
Sclerostin	chr17	39186625	39191682	SOST	17q21	605740	no	yes	yes
17q21.31	chr17	41217449	41268973	CRHR1	17q21	122561	no	yes	yes
17q21.31	chr17	41327544	41461546	MAPT	17q21	157140	yes	yes	yes
THROMBASTHENIA OF GLANZMANN AND NAEFELI	chr17	42686207	42745076	ITGB3	17q21	173470	no	yes	yes
Osteogenesis imperfecta	chr17	45616456	45633992	COL1A1	17q21	120150	yes	yes	yes
Campomelic dysplasia	chr17	67628756	67634156	SOX9	17q24	608160	yes	yes	yes
Holopresencephaly 4	chr18	3402072	3448406	TGIF1	18p31	602630	low coverage	yes	yes
	chr18	42798570	42820446	TCEB3C	18q21	609522	no	yes	yes
Dyggve-melchior-clausen disease	chr18	44824170	45250999	DYM	18q21	607461	yes	yes	yes
HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME	chr18	46810581	46865409	MADH4(SMAD4)	18q21	600993	no	yes	yes
Pitt Hopkins	chr18	51045967	51454183	TCF4	18q21	602272	low coverage	yes	yes
CHROMOSOME 18q DELETION SYNDROME	chr18	70416108	70906616	ZNF407	18q22		low coverage	yes	yes
	chr18	73090996	73111084	GALR1	18q23	600377	no	yes	yes
DIAMOND-BLACKFAN ANEMIA	chr19	47055828	47067324	RPS19	19q13	603474	no	yes	yes
	chr19	61967731	62053886	ZIM2	19q13	601483	no	yes	yes
	chr19	62005614	62053875	PEG3	19q13	601483	no	yes	yes
	chr19	62040958	62043979	PEG3_ICregion	19q13	601483	no	yes	yes
Zinc finger protein-264	chr19	62384680	62432350	ZNF264	19q13	604668	no	yes	yes
Feingold	chr2	15998134	16004579	MYCN	2q24	164840	yes	yes	yes
Pseudovaginal perineoscrotal hypospadias	chr2	31603160	31659544	SRD5A2	2p23	607306	no	yes	yes
Noonan 4	chr2	39066469	39201067	SOS1	2p22	182530	no	yes	yes
Cystinuria with mitochondrial disease	chr2	44356103	44401443	SLC3A1	2p21	104614	no	yes	yes

Cystinuria with mitochondrial disease	chr2	44399406	44442448	PREPL	2p21	104614	no	yes	yes
Holopresencephaly 2	chr2	45022365	45025945	SIX3	2p21	603714	yes	yes	yes
autism	chr2	50000922	51113178	NRXN1	2p16	600565	yes	yes	yes
2p15p16.1 microdeletion	chr2	61136239	61673319	MicrodeletionRegion	2p15	602559	low coverage	yes	yes
Nephronophthisis 1	chr2	1.1E+08	1.1E+08	NPHP1	2q13	607100	yes	yes	yes
Holoprosencephaly 9	chr2	1.21E+08	1.21E+08	GLI2	2q14	165230	low coverage	yes	yes
visceral heterotaxy	chr2	1.31E+08	1.31E+08	CFC1	2q21	605194	no	yes	yes
Mowat-Wilson syndrome	chr2	1.45E+08	1.45E+08	ZEB2	2q22	605802	low coverage	yes	yes
autism	chr2	1.62E+08	1.63E+08	SLC4A10	2q24	605556	low coverage	yes	yes
SCN1A-related seizures	chr2	1.67E+08	1.67E+08	SCN1A	2q24	182389	no	yes	yes
HYPOMYELINATION, GLOBAL CEREBRAL	chr2	1.72E+08	1.72E+08	SLC25A12	2q31	603667	no	yes	yes
Split/hand foot malformation -5	chr2	1.73E+08	1.73E+08	DLX1	2q31	600029	yes	yes	yes
Split/hand foot malformation -5	chr2	1.73E+08	1.73E+08	DLX2	2q31	126255	yes	yes	yes
Synpolydactyly	chr2	1.77E+08	1.77E+08	EVX2	2q31	142991	no	yes	yes
Synpolydactyly	chr2	1.77E+08	1.77E+08	HOXDgenes	2q31	142989	yes	yes	yes
Synpolydactyly	chr2	1.77E+08	1.77E+08	HOXD13	2q31	142989	yes	yes	yes
Synpolydactyly	chr2	1.77E+08	1.77E+08	HOXD9	2q31	142982	no	yes	yes
Cleft pallet	chr2	2E+08	2E+08	SATB2	2q32	608148	low coverage	yes	yes
2q32.2	chr2	2.03E+08	2.03E+08	BMPR2	2q32	600799	no	yes	yes
Waardenburg syndrome I	chr2	2.23E+08	2.23E+08	PAX3	2q36	606597	yes	yes	yes
2q37	chr2	2.4E+08	2.4E+08	HDAC4	2q37	605314	low coverage	yes	yes
Alaïlle Syndrome	chr20	10566334	10602590	JAG1	20p12	601920	yes	yes	yes
Brachydactyl type C	chr20	33360416	33489441	GDF5	20q11	601146	yes	yes	yes
	chr20	35573020	35595505	NNAT	20q11	603106	no	yes	yes
	chr20	41566487	41613948	L3MBTL	20q13	608802	no	yes	yes
Okhiro	chr20	49833990	49852421	SALL4	20q13	607343	no	yes	yes
	chr20	56676787	56680787	GNAS_ICregion	20q13	603666	no	yes	yes
Albright Hereditary osteodystrophy	chr20	56838189	56929641	GNAS	20q13	139320	no	yes	yes
	chr20	56859056	56861251	GNAS_ICregion	20q13	139320	no	yes	yes
	chr20	56896896	56899356	GNAS_ICregion	20q13	139320	no	yes	yes
Cerebral amyloid angiopathy	chr21	26174732	26465003	APP	21q21	104760	low coverage	yes	yes
DOWN SYNDROME CRITICAL REGION	chr21	34810654	34909252	DSCR1(RCAN1)	21q22	602917	no	yes	yes
DOWN SYNDROME CRITICAL REGION	chr21	35081968	36278917	RUNX1	21q22	151385	yes	yes	yes
DOWN SYNDROME CRITICAL REGION	chr21	37517596	37561703	DSCR3	21q22	605298	no	yes	yes
Downs Syndrome Critical Region" Probes	chr21	37661729	37809549	DYRK1A	21q22	600855	no	yes	yes
Holoprosencephaly 1	chr21	44256634	44350858	TMEM1	21q22	602103	yes	yes	yes
Holoprosencephaly 1 (HPE1)	chr21	46432788	46473119	LSS	21q22	600909	no	yes	yes
	chr22	16454903	16491588	ATP6V1E1	22q11	108746	no	yes	yes
diGeorge 1	chr22	17698224	17815220	HIRA	22q11	600637	yes	yes	yes
diGeorge 1	chr22	18124226	18151110	TBX1	22q11	602054	yes	yes	yes
diGeorge 1	chr22	19601714	19638035	CRKL	22q11	602007	no	yes	yes
22q11.2 distal deletion	chr22	21852552	21990224	BCR	22q11	151410	no	yes	yes
Meningioma / NF2	chr22	28329565	28424583	NF2	22q12	607379	no	yes	yes
METACHROMATIC LEUKODYSTROPHY	chr22	49410315	49413453	ARSA	22q13	607574	no	yes	yes
autism	chr22	49459936	49518507	SHANK3	22q13	606230	yes	yes	yes
autism spectrum disorder	chr3	2115550	3074645	CNTN4	3p25	607280	yes	yes	yes
Von Hippel Lindau	chr3	10158319	10168746	VHL	3p25	608537	no	yes	yes
Noonan syndrome 5 (NS5)	chr3	12600108	12680678	RAF1	3p25	164760	no	yes	yes
Loeys-Dietz syndrome	chr3	30622998	30710637	TGFBR2	3p24	190182	yes	yes	yes
MR	chr3	46594217	46598956	TGDF1	3p21	187395	yes	yes	yes
Waardenburg syndrome IIA	chr3	69871323	70100177	MITF	3p14	158645	yes	yes	yes
Bartter with autosomal dominant hypocalcemia	chr3	1.23E+08	1.23E+08	CASR	3q13	601199	no	yes	yes
Blepharophimosis	chr3	1.4E+08	1.4E+08	FOXL2	3q22	605597	yes	yes	yes
Dandy Walker	chr3	1.49E+08	1.49E+08	ZIC4	3q24	608948	yes	yes	yes
Dandy Walker	chr3	1.49E+08	1.49E+08	ZIC1	3q24	600470	yes	yes	yes
Microphthalmia	chr3	1.83E+08	1.83E+08	SOX2	3q26	184429	yes	yes	yes
Optic Atrophy	chr3	1.91E+08	1.91E+08	TP73L	3q28	603273	yes	yes	yes
3q29 microdeletion syndrome	chr3	1.98E+08	1.98E+08	PAK2	3q29	605022	no	yes	yes
Wolf-Hirschhorn	chr4	1784558	1827772	LETM1	4p16	604407	yes	yes	yes

Wolf-Hirschhorn	chr4	1842921	1953728	WHSC1	4p16	602952	yes	yes	yes
Wolf-Hirschhorn	chr4	1954251	1980547	WHSC2	4p16	606026	yes	yes	yes
cleft lip	chr4	4911225	4917628	MSX1	4p16	142983	yes	yes	yes
Polycystic kidney disease	chr4	89147844	89217953	PKD2	4q22	173910	no	yes	yes
Rieger	chr4	1.12E+08	1.12E+08	PITX2	4q25	601542	yes	yes	yes
Cri du chat	chr5	1306287	1348162	TERT	5p15	187270	no	yes	yes
Cornelia de Lange	chr5	36912742	37101678	NIPBL	5p13	608667	yes	yes	yes
Gardner	chr5	1.12E+08	1.12E+08	APC	5q22	601731	yes	yes	yes
Adult-onset autosomal dominant leukodystrophy	chr5	1.26E+08	1.26E+08	LMNB1	5q23	150340	no	yes	yes
Chronic pancreatitis	chr5	1.47E+08	1.47E+08	SPINK1	5q32	167790	no	yes	yes
deafness	chr5	1.5E+08	1.5E+08	TCOF1	5q33	606847	yes	yes	yes
Holoprosencephaly and preaxial polydactyly	chr5	1.71E+08	1.71E+08	FBXW11	5q35	605651	no	yes	yes
Atrial septal defect (ASD) with atrioventricular conduction defects	chr5	1.73E+08	1.73E+08	NKX2-5	5q35	600584	no	yes	yes
Craniosynostosis	chr5	1.74E+08	1.74E+08	MSX2	5q35	123101	yes	yes	yes
SOTOS	chr5	1.76E+08	1.77E+08	NSD1	5q35	606681	yes	yes	yes
CHROMOSOME 6pter-p24 DELETION SYNDROME	chr6	1555680	1559128	FKHL7(FOXC1)	6p24	609010	no	yes	yes
Congenital adrenal hyperplasia (CAH)	chr6	32044642	32162690	CYP21A2	6p21	201910	no	yes	yes
VEGFA disorders	chr6	43845931	43862201	VEGFA	6p21	192240	yes	yes	yes
Cleidocranial dysplasia	chr6	45404032	45626796	RUNX2	6p21	600211	yes	yes	yes
obesity, severe	chr6	1.01E+08	1.01E+08	SIM1	6q16	603128	yes	yes	yes
MENTAL RETARDATION, AUTOSOMAL RECESSIVE 6_MRT6	chr6	1.02E+08	1.03E+08	GRIK2	6q16	138244	low coverage	yes	yes
eye	chr6	1.36E+08	1.36E+08	AHI1	6q23	608894	no	yes	yes
diabetes mellitus, transient neonatal, 1	chr6	1.44E+08	1.44E+08	ZAC(PLAGL1)	6q24	603044	no	yes	yes
diabetes mellitus, transient neonatal, 1	chr6	1.44E+08	1.44E+08	HYMAI	6q24	603044	no	yes	yes
Hepatocellular carcinoma	chr6	1.6E+08	1.6E+08	IGF2R	6q25	147260	no	yes	yes
Saethre-Chotzen	chr7	19121616	19123550	TWIST1	7p21	601622	yes	yes	yes
GREIG CEPHALOPOLYSYNDACTYLY SYNDROME	chr7	41967073	42243900	GLI3	7p14	165240	yes	yes	yes
Silver-Russell	chr7	50615258	50838651	GRB10	7p12	161523	no	yes	yes
Williams-Beuren	chr7	73080407	73120966	ELN	7q11	130160	yes	yes	yes
Williams-Beuren	chr7	73136092	73174790	LIMK1	7q11	194050	yes	yes	yes
Williams-Beuren	chr7	73826245	73841595	NCF1	7q11	608512	yes	yes	yes
Ehlers - Danlos	chr7	93861809	93898480	COL1A2	7q21	120160	yes	yes	yes
Dystonia-11	chr7	94042477	94133413	SGCE	7p21	604149	no	yes	yes
Dystonia-11	chr7	94113623	94146938	PEG10	7p21	604149	no	yes	yes
Dystonia-11	chr7	94123295	94123994	PEG10_ICregion	7p21	604149	no	yes	yes
	chr7	94364911	94770958	PPP1R9A	7q21	602468	low coverage	yes	yes
Split/hand foot	chr7	96140173	96177139	SHFM1	7q21	183600	yes	yes	yes
Split/hand foot	chr7	96477643	96502078	DLX5	7q21	600028	no	yes	yes
#N/A	chr7	98084545	98097116	NPTX2	7q22	600750	no	yes	yes
Lissencephaly with cerebellar hypoplasia	chr7	1.03E+08	1.03E+08	RELN	7q22	600514	low coverage	yes	yes
Verbal dyspraxia	chr7	1.14E+08	1.14E+08	FOXP2	7q31	605317	yes	yes	yes
autism	chr7	1.16E+08	1.16E+08	MET	7q31	164860	no	yes	yes
autism	chr7	1.22E+08	1.22E+08	CADPS2	7q31	609978	yes	yes	yes
	chr7	1.3E+08	1.3E+08	CPA4	7q32	607635	no	yes	yes
	chr7	1.3E+08	1.3E+08	MEST	7q32	601029	no	yes	yes
	chr7	1.3E+08	1.3E+08	KLF14	7q32	609393	no	yes	yes
MR	chr7	1.42E+08	1.42E+08	PRSS1	7q34	601564	no	yes	yes
autism	chr7	1.45E+08	1.48E+08	CNTNAP2	7q35-36	276000	no	yes	yes
autism	chr7	1.55E+08	1.55E+08	EN2	7q36	131310	no	yes	yes
Holoprosencephaly 3	chr7	1.55E+08	1.55E+08	SHH	7q36	600725	yes	yes	yes
POLYDACTYLY, PREAXIAL II	chr7	1.56E+08	1.56E+08	LMBR1	7q36	605522	no	yes	yes
CURRARINO SYNDROME	chr7	1.56E+08	1.56E+08	HLXB9(MNX1)	7q36	142994	no	yes	yes
MR	chr8	1426975	1654048	DLGAP2	8p23	605438	low coverage	yes	yes
diGeorge	chr8	8679409	8788541	MFHAS1	8p23	605352	no	yes	yes
diGeorge	chr8	11571877	11654918	GATA4	8p23	600576	yes	yes	yes
Kabuki Syndrome	chr8	11737443	11763055	CTSB	8p23	116810	no	yes	yes
Kallman 1	chr8	38387813	38445509	FGFR1	8p12	136350	no	yes	yes
CHARGE syndrome	chr8	61753893	61942017	CHD7	8q12	601653	yes	yes	yes
Branchio-Oto-Renal Syndrome	chr8	72272222	72437021	EYA1	8q13	602064	yes	yes	yes
Manic depressive psychosis	chr8	82732751	82761115	IMPA1	8q21	602064	yes	yes	yes

congenital heart	chr8	1.06E+08	1.07E+08	ZFPM2(FOG2)	8q23	603693	low coverage	yes	yes
Trischororhinophlange al, Langer-Giedion Syndrome probes	chr8	1.16E+08	1.17E+08	TRPS1	8q23	604386	yes	yes	yes
Langer-Giedion Syndrome probes	chr8	1.19E+08	1.19E+08	EXT1	8q23	150230	yes	yes	yes
8q24.3	chr8	1.41E+08	1.41E+08	KCNK9	8q24	605874	no	yes	yes
CHROMOSOME 9p DELETION SYNDROME	chr9	831690	959090	DMRT1	9p24	602424	low coverage	yes	yes
CHROMOSOME 9p DELETION SYNDROME	chr9	1039858	1047552	DMRT2	9p24	604935	no	yes	yes
HOLOPROSENCEPHALY 7	chr9	97245085	97319068	PTCH1	9q22	601309	yes	yes	yes
9q22.32-q22.33 / Loeys-Dietz	chr9	1.01E+08	1.01E+08	TGFBR1	9q22	190181	yes	yes	yes
XY sex reversal	chr9	1.26E+08	1.26E+08	NR5A1	9q3	184757	yes	yes	yes
Nail-Patella Syndrome	chr9	1.28E+08	1.29E+08	LMX1B	9q33	602575	yes	yes	yes
Tuberous sclerosis	chr9	1.35E+08	1.35E+08	TSC1	9q34	605284	yes	yes	yes
9q34.3 syndrome	chr9	1.4E+08	1.4E+08	EHMT1	9q34	607001	yes	yes	yes
PAR1	chrX	1	2700000	PAR1	Xp22		yes	yes	yes
SHOX	chrX	505079	540146	SHOX	Xp22	400020	yes	yes	yes
Chondrodysplasia punctata, X-linked recessive	chrX	2862852	2892311	CDPX1(ARSE)	Xp22	300180	no	yes	yes
autism	chrX	5818085	6155888	NLGN4X	Xp22	300427	no	yes	yes
Steroid sulfatase	chrX	6461659	6463159	VCX3A	Xp22	300533	no	yes	yes
Steroid sulfatase	chrX	7075293	7282680	STS	Xp22	300747	yes	yes	yes
XLMR	chrX	8393346	8394346	VCX3A	Xp22		no	yes	yes
Kallmann	chrX	8456915	8660227	KAL1	Xp22	308700	yes	yes	yes
Opitz	chrX	10376166	10765667	MID1	Xp22	300552	yes	yes	yes
XLMR	chrX	11039373	11051122	HCCS	Xp22	300056	yes	yes	yes
Orofacioidigital 1	chrX	13662801	13697391	OFD1	Xp22	300170	yes	yes	yes
XLMR	chrX	14771450	14801105	FANCB	Xp22	300515	no	yes	yes
XLMR	chrX	15753850	15783021	AP1S2	Xp22	300629	no	yes	yes
Nance-Horan	chrX	17303464	17664034	NHS	Xp22	300457	yes	yes	yes
X linked spasms	chrX	18353678	18581666	CDKL5	Xp22	300203	yes	yes	yes
X linked juvenile retinoschisis	chrX	18567731	18600150	RS1	Xp22	312700	yes	yes	yes
Pyruvate dehydrogenase deficiency	chrX	19271972	19287886	PDHA1	Xp22	300502	no	yes	yes
Coffin-Lowry	chrX	20077950	20194671	RPS6KA3	Xp22	300075	no	yes	yes
XLMR	chrX	21868763	21922876	SMS	Xp22	300105	no	yes	yes
Hypophosphatemic rickets	chrX	21960842	22176399	PHEX	Xp22	300550	no	yes	yes
XLMR	chrX	24932213	24943775	ARX	Xp21	300382	yes	yes	yes
XLMR	chrX	28515480	29884757	IL1RAPL1	Xp21	300206	yes	yes	yes
Adrenal hypoplasia congenita	chrX	30232507	30237413	NR0B1(DAX1)	Xp21	300473	yes	yes	yes
Glycerol Kinase deficiency	chrX	30581461	30658645	GK	Xp21	300474	yes	yes	yes
Muscular dystrophy	chrX	31048667	33277779	DMD	Xq28	300777	yes	yes	yes
Chronic granulomatous disease	chrX	37524264	37557658	CYBB	xp11	300481	no	yes	yes
Ornithine transcarbamylase deficiency	chrX	38096302	38165553	OTC	Xp11	300461	yes	yes	yes
XLMR	chrX	38305683	38433116	TM4SF2(TSPAN7)	Xp11	300096	no	yes	yes
XLMR	chrX	39795561	39921526	BCOR	Xp11	300485	no	yes	yes
XLMR	chrX	40325160	40350832	ATP6AP2	Xp11	300566	no	yes	yes
XLMR	chrX	41264287	41667212	CASK	Xp11	300172	no	yes	yes
BRUNNER SYNDROME	chrX	43400353	43491012	MAOA	Xp11	309850	no	yes	yes
Norrie disease	chrX	43692968	43717694	NDP	Xp11	300658	no	yes	yes
Xp11.3 deletion	chrX	46243490	46289820	ZNF674	Xp11	300573	no	yes	yes
Xp11.3 deletion	chrX	46581319	46626733	RP2	Xp11	300757	yes	yes	yes
XLMR	chrX	47191350	47227289	ZNF41	Xp11	314995	no	yes	yes
XLMR	chrX	47316244	47364200	SYN1	Xp11	311040	no	yes	yes
XLMR	chrX	47379864	47394964	ELK1	Xp11	311040	no	yes	yes
XLMR	chrX	47581245	47666550	ZNF81	Xp11	314998	no	yes	yes
X-linked mental retardation (XLMR)	chrX	48201871	48213509	SLC38A5	Xp11	300649	no	yes	yes
XLMR	chrX	48219493	48229696	FTSJ1	Xp11	300499	no	yes	yes
XLMR	chrX	48252315	48264146	PORCN	Xp11	300651	no	yes	yes
XLMR	chrX	48640139	48645364	PQBP1	Xp11	300463	no	yes	yes
nephrolithiasis 2, X-linked, Dent disease	chrX	49573965	49750632	CLCN5	Xp11	300008	no	yes	yes
XLMR	chrX	50351387	50573784	SHROOM4(KIAA1202)	Xp11	300579	no	yes	yes
XLMR	chrX	53237378	53271329	JARID1C	Xp11	314690	no	yes	yes
X-linked Cornelia de Lange syndrome	chrX	53417795	53466343	SMC1L1(SMC1A)	Xp11	300040	yes	yes	yes
MENTAL RETARDATION, X-LINKED, SYNDROMIC 10: MRXS10	chrX	53474931	53478048	HADH2(HSD17B10)	Xp11	300256	no	yes	yes

Mental retardation, X-linked, syndromic, Turner	chrX	53576476	53730398	HUWE1	Xp11	300697	low coverage	yes	yes
XLMR	chrX	53979838	54088121	PHF8	Xp11	300560	no	yes	yes
AARSKOG-SCOTT SYNDROME	chrX	54488612	54539324	FGD1	Xp11	300564	no	yes	yes
XLMR	chrX	56275632	56328254	KLF8	Xp11	300286	no	yes	yes
Heterotaxy	chrX	62771573	62922138	ARHGGEF9	Xq11	300429	no	yes	yes
Androgen insensitivity syndrome	chrX	66680599	66680844	AR	Xq12	313700	yes	yes	yes
MENTAL RETARDATION, X-LINKED, WITH CEREBELLAR HYPOPLASIA AND DISTINCTIVE FACIAL APPEARANCE	chrX	67179440	67570372	OPHN1	Xq12	300127	yes	yes	yes
Ectodermal dysplasia	chrX	68752636	69176046	EDA	Xq13	300451	yes	yes	yes
XLMR	chrX	69581449	69642062	DLG3	Xq13	300189	no	yes	yes
Opitz - Kaveggia syndrome	chrX	70255131	70279029	MED12	Xq13	300188	no	yes	yes
autism	chrX	70281436	70307776	NLGN3	Xq13	300336	no	yes	yes
XLMR	chrX	70376199	70391660	ZNF261 (ZMYM3)	Xq13	30061	no	yes	yes
XIST deficiency	chrX	72863307	73063491	XIST	Xq13	314670	yes	yes	yes
Allan-Herndon-Dudley syndrome, X-linked, mental retardation (XLMR)	chrX	73557810	73670475	SLC16A2	Xq13	300095	no	yes	yes
XLMR	chrX	73870137	74061709	KIAA2022	Xq13	300524	low coverage	yes	yes
XLMR	chrX	74508786	74659600	ZDHHC15	Xq13	300576	no	yes	yes
ALPHA-THALASSEMIA/MENTAL RETARDATION SYNDROME	chrX	76647847	76928358	ATRX	Xq21	300032	yes	yes	yes
Menkes disease	chrX	77042046	77200000	ATP7A	Xq21	300011	yes	yes	yes
Phosphoglycerate kinase deficiency	chrX	77246425	77268980	PGK1	Xp21	311800	no	yes	yes
XLMR	chrX	79818351	79951889	BRWD3	Xp21	300553	no	yes	yes
Choroideremia	chrX	85002841	85189222	CHM	Xp21	300390	no	yes	yes
ROLANDIC EPILEPSY, MENTAL RETARDATION, AND SPEECH DYSPRAXIA, X-LINKED	chrX	99785819	99812952	SRPX2	Xq22	300642	no	yes	yes
Deafness-Dystonia-Optic Neuropathy	chrX	1E+08	1E+08	TIMM8A	Xq22	300356	no	yes	yes
X linked agammaglobulinemia	chrX	1E+08	1.01E+08	BTK	Xq22	300300	yes	yes	yes
Fabry disease	chrX	1.01E+08	1.01E+08	GLA	Xq22	300644	no	yes	yes
XLMR	chrX	1.01E+08	1.01E+08	NXF5	Xq22	300319	no	yes	yes
PELIZAEUS-MERZBACHER DISEASE	chrX	1.03E+08	1.03E+08	PLP1	Xq22	300401	yes	yes	yes
CHARCOT-MARIE-TOOTH DISEASE, X-LINKED RECESSIVE, 5, PRPP synthetase hyperactivity	chrX	1.07E+08	1.07E+08	PRPS1	Xq22	311850	no	yes	yes
Opitz / FP syndrome	chrX	1.07E+08	1.07E+08	MID2	Xq22	300204	no	yes	yes
Alport syndrome (X)	chrX	1.08E+08	1.08E+08	COL4A5	Xq22	303630	yes	yes	yes
XLMR	chrX	1.09E+08	1.09E+08	ACSL4	Xp22	300157	no	yes	yes
XLMR	chrX	1.1E+08	1.1E+08	PAK3	Xp22	300142	low coverage	yes	yes
MR	chrX	1.1E+08	1.11E+08	DCX	Xp22-p23	300121	yes	yes	yes
XLMR	chrX	1.15E+08	1.15E+08	AGTR2	Xq23	300034	no	yes	yes
XLMR	chrX	1.19E+08	1.19E+08	UBE2A	Xq23	312180	no	yes	yes
XLMR	chrX	1.19E+08	1.19E+08	UPF3B	Xq24	300298	low coverage	yes	yes
XLMR	chrX	1.19E+08	1.19E+08	NDUFA1	Xq24	300078	no	yes	yes
Danon disease	chrX	1.19E+08	1.19E+08	LAMP2	Xq24	309606	no	yes	yes
XLMR	chrX	1.2E+08	1.2E+08	CUL4B	Xq24	300304	no	yes	yes
XLMR	chrX	1.22E+08	1.22E+08	GRIA3	Xq24	305915	low coverage	yes	yes
LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 2	chrX	1.23E+08	1.23E+08	BIRC4(XIAP)	Xq25	300079	yes	yes	yes
LYMPHOPROLIFERATIVE SYNDROME, X-LINKED, 1	chrX	1.23E+08	1.23E+08	SH2D1A	Xq25	300490	yes	yes	yes
Lowe	chrX	1.29E+08	1.29E+08	OCRL	Xq25	300535	no	yes	yes
XLMR	chrX	1.29E+08	1.29E+08	ZDHHC9	Xq25	300646	no	yes	yes
Simpson - Golabi Behmel type 1	chrX	1.32E+08	1.33E+08	GPC3	Xq26	300037	low coverage	yes	yes

BORJESON-FORSSMAN-LEHMANN SYNDROME	chrX	1.33E+08	1.33E+08	PHF6	Xq26	300414	no	yes	yes
Lesch-Nyhan syndrome	chrX	1.33E+08	1.33E+08	HPRT1	Xq26	308000	yes	yes	yes
X-linked mental retardation (XLMR), syndromic, Chrystianson type	chrX	1.35E+08	1.35E+08	SLC9A6	Xq26	300231	no	yes	yes
XLMR	chrX	1.36E+08	1.36E+08	ARHGEF6	Xq26	300267	no	yes	yes
X linked heterotaxy	chrX	1.36E+08	1.36E+08	ZIC3	Xq26	300265	yes	yes	yes
HEMOPHILIA B	chrX	1.38E+08	1.38E+08	F9	Xq27	300746	no	yes	yes
Mental retardation X linked Xq27.1 dup and del	chrX	1.39E+08	1.39E+08	SOX3	Xq27	313430	yes	yes	yes
Fragile X (FMR1 and FMR2 deletion)	chrX	1.47E+08	1.47E+08	FMR1	Xq27	309550	yes	yes	yes
Fragile X (FMR1 and FMR2 deletion)	chrX	1.47E+08	1.48E+08	FMR2(AFF2)	Xq28	309548	low coverage	yes	yes
Hunter syndrome, Mucopolysaccharidosis type II	chrX	1.48E+08	1.48E+08	IDS	Xq28	309900	yes	yes	yes
MYOTUBULAR MYOPATHY	chrX	1.49E+08	1.5E+08	MTM1	Xq28	300415	no	yes	yes
Creatine deficiency syndrome, XLMR	chrX	1.53E+08	1.53E+08	SLC6A8	Xq28	300036	no	yes	yes
Adrenoleukodystrophy; (ALD)	chrX	1.53E+08	1.53E+08	ABCD1	Xq28	300475	no	yes	yes
HSAS, MASA, CRASH syndromes	chrX	1.53E+08	1.53E+08	L1CAM	Xq28	300371	no	yes	yes
Diabetes insipidus, nephrogenic, X-linked	chrX	1.53E+08	1.53E+08	AVPR2	Xq28	300538	no	yes	yes
Retts	chrX	1.53E+08	1.53E+08	MECP2	Xq28	300260	yes	yes	yes
HETEROTOPIA, PERIVENTRICULAR, X-LINKED DOMINANT	chrX	1.53E+08	1.53E+08	FLNA	Xq28	300017	no	yes	yes
XLMR	chrX	1.53E+08	1.53E+08	RPL10	Xq28	209850	no	yes	yes
XLMR	chrX	1.53E+08	1.53E+08	GDI1	Xq28	300104	no	yes	yes
INCONTINENTIA PIGMENTI	chrX	1.53E+08	1.53E+08	IKBKKG	Xq28	300248	yes	yes	yes
DYSKERATOSIS CONGENITA, X-LINKED	chrX	1.54E+08	1.54E+08	DKC1	Xq28	300125	no	yes	yes
Hemophilia	chrX	1.54E+08	1.54E+08	F8	Xq28	306700	yes	yes	yes
PAR2	chrX	1.55E+08	1.55E+08	PAR2	Xq28	300053	yes	yes	yes
SRY deletion	chrY	2714821	2716600	SRY	Yp11	480000	yes	yes	yes
AZFa	chrY	13052929	13603474	AZFa	Yq11	400005	yes	yes	yes
AZFc region	chrY	23539798	23560998	BPY2	Yq11	400013	no	yes	yes
AZFc region	chrY	23663736	25513794	AZFc	Yq11	400003	no	yes	yes
AZFc region	chrY	23684890	23754627	DAZ1	Yq11	400003	no	yes	yes
AZFc region	chrY	24600764	24603549	CDY1	Yq11	400016	no	yes	yes
AZFc region	chrY	24765503	24770366	GOLGA2LY(AF33222)	Yq11	400035	no	yes	yes
AZFc region	chrY	25173539	25194740	BPY2	Yq11	400016	no	no	yes
AZFc region	chrY	25586438	25607639	BPY2	Yq11	400013	no	no	yes
AZFc region	chrY	26010846	26015709	GOLGA2LY(AF33222)	Yq11	400035	no	no	yes
AZFc region	chrY	26177652	26180436	CDY1	Yq11	400016	no	no	yes

low coverage = probes are present in the region but not at a significant density  
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