CytoSure™

Comparison of gene content of ISCA and Syndrome Plus arrays

Summary for each array, plus coverage within each gene/targetted region:

Application	No. of arrays/slide	No. of probes	Genome/ backbone probe density	No. of targeted regions	Probe resolution	Scanner compatibility	Product name
Whole genome screening	8	60,000	Probe every 60kb	498	Probe every 31kb within genes, 4 probes per gene (on average)	High res.	CytoSure ISCA v2 (8x60k)
Whole genome screening	4	180,000	Probe every 25kb	501	Probe every 14kb within genes, 6 probes per gene (on average)	High res.	CytoSure ISCA v2 (4x180k)
Whole genome screening	2	105,000	Probe every 40kb	610	Probe every 3kb within genes, 33 probes per gene (on average)	High res. & 5 µm	CytoSure Syndrome Plus v2 (2x105k)
X chromosome analysis	4	44,000	Probe every 4kb	1118	Probe every 380bp within genes, 81 probes per gene (on average)	High res. & 5 µm	CytoSure Chromosome X (4x44k)

Chromosome	Start (bp)	Stop (bp)	Gene	Band	Syndrome	OMIM	ISCA 4x44k	ISCA & ISCA UPD 4x180k	ISCA 8x60k	Syndrome Plus 2x105k	
chr10	87349292	88116230	GRID1	10q22	10q22-23 Deletion		○	●	●	●	#
chr10	83625077	84735341	NRG3	10q22	10q22-23 Deletion		●	●	●	●	
chr11	86334369	86344081	FZD4	11q14	11q14 Microdeletion			●	●	●	#
chr11	114500000	130800000		11q23.3-q24.3	11q23 Jacobsen deletion	147791				●	#
chr12	65029066	65359020	GRIP1	12q14	12q14.3 Deletion		○	●	●		#
chr13	47300000	59600000		13q14.2-q21.1	13q14 Deletion					●	#
chr14	60182506	60185933	SIX1	14q22	14q22 Microdeletion		●	●	●	●	#
chr14	60246009	60260792	SIX4	14q22	14q22 Microdeletion		●	●	●	●	#
chr14	60045150	60048904	SIX6	14q22	14q22 Microdeletion		●	●	●	●	#
chr14	56337178	56346937	OTX2	14q22	14q22 Microdeletion, microphthalmia, syndromic 5		●	●	●	●	#
chr14	53486207	53493362	BMP4	14q22	14q22 Microdeletion, Orofacial Cleft 11		●	●	●	●	#
chr15	30110018	30248527	CHRNA7	15q13	15q13.3 Microdeletion	612001	●	●	●	●	#
chr15	41612952	41664382	KIAA0377 (HISPPD2A)	15q15.3	15q15.3 Infertility and deafness					●	#
chr15	72700000	75700000		15q24.1	15q24 Microdeletion					●	#
chr16	28100000	34600000		16p11.2	16p11.2 Deletion	611913				●	#
chr16	21200000	34600000		16p11.2-p12.2	16p11.2-p12.2 Microdeletion					●	#
chr16	14800000	16800000		16p13.11	16p13.1 Microdeletion predisposition to autism					●	#
chr17	41217449	41268973	CRHR1	17q21.31	17q21.31 Microdeletion	610443		●	●	●	#
chr17	41327544	41461546	MAPT	17q21.31	17q21.31 Microduplication	610443	●	●	●	●	#
chr18	70416108	70906616	ZNF407	18q22	18q Deletion	301808	○	●	●		#
chr1	2149994	2231416	SKI	1p36	1p36 Microdeletion	607872		●	●	●	#
chr1	3548988	3646715	TP73	1p36	1p36 Microdeletion	607872		●	●	●	#
chr22	21852552	21990224	BCR	22q11	22q11.2 Distal deletion	611867		●	●		#
chr22	17900000	25900000		22q11.2	22q11.2 Microduplication	608363				●	#
chr22	49400000	51300000	SHANK3	22q13.33	22q13 Microdeletion (Phelan McDermid)	606232				●	#
chr2	61136239	61673319	MicroDeletionRegion	2p15	2p15-p16.1 Microdeletion		○	●	●		#
chr2	239634801	239987580	HDAC4	2q37	2q37.3 Monosomy		○	●	●	●	
chr3	197951312	198043751	PAK2	3q29	3q29 Microdeletion	609425		●	●	●	#
chr6	1555680	1559128	FKHL7(FOXC1)	6p24	6p24 Deletion	612852		●	●		#
chr6	155500001	161000000		6p25.3	6p25.3 Microdeletion	220210				●	#
chr8	11571877	11654918		8p23.1	8p23.1 Microdeuplication					●	#
chr9	831690	959090	DMental RetardationT1	9p24	9p Deletion	158170	○	●	●		#
chr9	1039858	1047552	DMental RetardationT2	9p24	9p Deletion	158170		●	●		#
chr9	140000000	140000000	EHMT1	9q34.3	9q34.3 Microdeletion	610253	●	●	●	●	#
chrX	54488612	54539324	FGD1	Xp11	Aarskog-Scott	305400		●	●		#
chr15	98500000	102531392		15q26.3	Abnormal growth					●	
chrX	30232507	30237413	NR0B1(DAX1)	Xp21	Adrenal hypoplasia congenita	300200	●	●	●	●	#
chrX	152643530	152663374	ABCD1	Xq28	Adrenoleukodystrophy	300475		●	●		#
chr5	126140732	126200608	LMNB1	5q23	Adult-onset autosomal dominant leukodystrophy	169500		●	●	●	#
chrX	100491098	100532426	BTK	Xq22.1	Agammaglobulinemia, X-linked	300755	●	●	●	●	#
chr20	10566334	10602590	JAG1	20p12	Alagille	118450	●	●	●	●	#
chr20	56838189	56929641	GNAS	20q13	Albright hereditary osteodystrophy	103580		●	●	●	#
chr16	155725	171196	HBA1&HBA2	16p13	Alpha thalassemia mental retardation	141750		●	●	●	#
chrX	76647847	76928358	ATRX	Xq21	Alpha thalassemia mental retardation	301040	●	●	●		#
chrX	107569810	107827431	COL4A5	Xq22.3	Alport, X-linked	301050	●	●	●	●	#
chrX	66680599	66860844	AR	Xq12	Androgen insensitivity	300068	●	●	●	●	#
chr15	21429787	21452081	MAGEL2	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	21361547	21407800	MKRN3	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	20594720	20637877	NIPA1	15q11	Angelman / Prader Willi	105830/176270	●	●	●	●	#
chr15	20556790	20585849	NIPA2	15q11	Angelman / Prader Willi	105830/176270	●	●	●	●	#
chr15	22713304	22720548	PWS_ICRegion	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	22747536	22751866	PWS_ICRegion	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	22768233	22793232	SNORD107/64/108	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	23064582	23084648	SNORD109B	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	22835000	23010100	SnoRNA	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	22619887	22774822	SNRPN	15q11	Angelman / Prader Willi	105830/176270	●	●	●	●	#
chr15	22751228	22774822	SNRPN	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr15	22741227	22784821	SNURF	15q11	Angelman / Prader Willi	105830/176270	○	●	●	●	#
chr15	21471646	21493542	NDN	15q11	Angelman / Prader Willi	105830/176270		●	●	●	#
chr11	31762916	31796085	PAX6	11p13-p14	Aniridia	106210	●	●	●	●	#
chr1	145694956	145712108	GJA5	1q21	Atrial fibrillation		●	●	●		
chr5	172591744	172594868	NKX2-5	5q35	Atrial septal defect with atrioventricular conduction defects	108900		●	●	●	#
chr1	229829184	230243641	DISC1	1q42	Autism	209850	○	●	●	●	
chr2	50000992	51113178	NRXN1	2p16	Autism	209850	●	●	●	●	
chr2	162189192	162548489	SLC4A10	2q24	Autism	209850	○	●	●	●	
chr3	2115550	3074645	CNTN4	3p25	Autism	209850	●	●	●		
chr7	121746687	122313750	CADPS2	7q31	Autism	209850	●	●	●	●	
chr7	145444386	147749019	CNTNAP2	7q35-36	Autism	209850		●	●	●	
chr7	154943160	154950579	EN2	7q36	Autism	209850		●	●	●	
chr7	116099695	116225676	MET	7q31	Autism	209850		●	●	●	
chr10	64241763	64246133	EGR2	10q21	Autism	209850		●	●	●	
chr15	23463512	23669962	ATP10A	15q12	Autism	209850		●	●	●	
chr15	24339787	24767950	GABRB3	15q12	Autism	209850	○	●	●	●	
chr22	49459936	49518507	SHANK3	22q13	Autism	209850	●	●	●	●	
chrX	70281436	70307776	NLGN3	Xq13	Autism	209850		●	●	●	
chrX	5818085	6155888	NLGN4X	Xp22	Autism	209850		●	●	●	
chrY	13052929	13603474	AZFa	Yq11.2	AZFa region	415000	●	●	●	●	#
chrY	23663736	25513794	AZFc	Yq11.2	AZFa region	415000		●	●	●	#
chrY	23539798	23560998	BPY2	Yq11.2	AZFa region	415000		●	●	●	#
chrY	25173539	25194740	BPY2	Yq11.2	AZFa region	415000			●	●	#
chrY	25586438	25607639	BPY2	Yq11.2	AZFa region	415000			●	●	#
chrY	24600764	24603549	CDY1	Yq11.2	AZFa region	415000		●	●	●	#
chrY	26177652	26180436	CDY1	Yq11.2	AZFa region	415000			●	●	#
chrY	23684890	23754627	DAZ1	Yq11.2	AZFa region	415000		●	●	●	#
chrY	24765503	24770366	GOLGA2LY(AF332229)	Yq11.2	AZFa region	415000		●	●	●	#
chrY	26010846	26015709	GOLGA2LY(AF332229)	Yq11.2	AZFa region	415000			●	●	#
chr15	46285791	46382417	SLC12A1	15q21	Bartter 1	601678		●	●	●	#
chr11	128213125	128242478	KCNJ1	11q24	Bartter 2	241200		●	●	●	
chr1	16221073	16256063	CLCNKB	1p36	Bartter 3	607364		●	●	●	
chr1	55237205	55247053	BSND	1p32	Bartter 4A	602522		●	●	●	
chr1	16217957	16233132	CLCNKA	1p36	Bartter 4B	613090		●	●		
chr3	121902530	123488032	CASR	3q13.33-q21.1	Bartter with autosomal dominant hypocalcemia	601199		●	●	●	#
chr11	2851440	2873550	CDKN1C	11p15	Beckwith-Wiedemann	130650	○	●	●	●	#
chr11	1962981	1985640	H19	11p15	Beckwith-Wiedemann	130650		●	●	●	#
chr11	2096925	2136469	IGF2	11p15	Beckwith-Wiedemann	130650	●	●	●	●	#
chr11	2127584	2148999	INS	11p15	Beckwith-Wiedemann	130650		●	●	●	#
chr11	2896078	2917225	PHLDA2	11p15	Beckwith-Wiedemann	130650		●	●	●	#
chr11	2867526	2913051	SLC22A18	11p15	Beckwith-Wiedemann	130650		●	●	●	#
chr16	56211151	56256445	GPR56	16q13	Bilateral frontoparietal polymicrogyria	606854		●	●	●	#
chr3	140144730	140149880	FOXL2	3q22	Blepharophimosis	110100	●	●	●	●	#
chrX	133335008	133390488	PHF6	Xq26	Borjeson-forssman-lehmann	301900		●	●		#
chr13	90848888	92317488	GPC5	13q31	Brachydactyly		●	●	●		
chr13	92677096	93853948	GPC6	13q31	Brachydactyly		●	●	●		
chr20	33360416	33489441	GDF5	20q11	Brachydactyly type C	113100	●	●	●		#
chr8	72272222	72437021	EYA1	8q13	Branchio-oto-renal	113650	●	●	●	●	#
chr13	31787617	31871809	BRCA2	13q13	Breast cancer	114480		●	●		#
chrX	43400353	43491012	MAOA	Xp11	Brunner	300615		●	●		#
chr19	15270445	15311792	NOTCH3	19p13.12	CADASIL	125310				●	#
chr17	67628756	67634156	SOX9	17q24	Campomelic dysplasia	114290	●	●	●	●	#
chr17	158000	3300000	ASPA	17p13.3	Canavan	271900				●	#
chr10	91180753	91285293	SLC16A12	10q23	Cataract, juvenile, with microcornea A and glucosuria	612018		●	●		#
chr22	14700000	17900000		22q11.1	Cat-eye	115470				●	#

chr21	26174732	26465003	APP	21q21	Cerebral amyloid angiopathy	605714	○	●	●		#
chr3	160700001	167600000	PDCD10	3q26.1	Cerebral carvernous malformations (CCM)	603285				●	#
chr7	43300001	45400000	PDCD10	7p13	Cerebral carvernous malformations (CCM)	603285				●	#
chr7	91100001	92800000	PDCD10	7q21.2	Cerebral carvernous malformations (CCM)	603285				●	#
chr17	15073822	15109369	PMP22	17p11	Charcot marie tooth disease	118220	●	●	●	●	#
chrX	106758415	106780912	PRPS1	Xq22	Charcot marie tooth disease, X-linked	311070		●	●		#
chr8	61753893	61942017	CHD7	8q12	CHARGE	214800	●	●	●	●	#
chrX	2862852	2892311	CDPX1(ARSE)	Xp22	Chondrodysplasia punctata, X-linked recessive	302950		●	●		#
chrX	85002841	85189222	CHM	Xp21	Choroideremia	303100		●	●		#
chrX	37524264	37557658	CYBB	Xp11	Chronic granulomatous disease	306400		●	●		#
chr5	147184339	147191453	SPINK1	5q32	Chronic pancreatitis	167800		●	●		#
chr4	4911225	4917628	MSX1	4p16	Cleft lip	608874	●	●	●		#
chr2	199842469	200038076	SATB2	2q32	Cleft pallet	119540	○	●	●	●	#
chr6	45404032	45626796	RUNX2	6p21	Cleidocranial dysplasia	119600	●	●	●	●	#
chrX	20077950	20194671	RPS6KA3	Xp22	Coffin-lowry	303600		●	●		#
chr6	32044642	32162690	CYP21A2	6p21.32	Congenital adrenal hyperplasia (CAH)	201910		●	●	●	
chr15	91244423	91372241	CHD2	15q26	Congenital diaphragmatic hernia	142340	●	●	●	●	
chr15	94670161	94683047	NR2F2	15q26	Congenital diaphragmatic hernia	142340	●	●	●	●	
chr8	106400323	106885943	ZFPM2(FOG2)	8q23	Congenital heart	610187	○	●	●		#
chr5	36912742	37101678	NIPBL	5p13	Cornelia de Lange	122470	●	●	●	●	#
chrX	53417795	53466343	SMC1L1(SMC1A)	Xp11	Cornelia de Lange, X-linked	300590	●	●	●		#
chr10	88506376	88674925	BMPR1A	10q23	Cowden	158350		●	●	●	#
chr5	174084181	174090508	MSX2	5q35	Craniosynostosis	604757	●	●	●	●	#
chr11	15949900	16454494	SOX6	11p15.1-p15.2	Craniosynostosis	128350	○	●	●	●	
chr16	34877510	35027509	Centromere	16p11	Creatine deficiency / X-linked mental retardation	300352	●	●	●		#
chr16	33686002	33694027	SLC6A8	16p11	Creatine deficiency / X-linked mental retardation	300352		●	●		#
chrX	152606586	152615234	SLC6A8	Xq28	Creatine deficiency / X-linked mental retardation	300352		●	●		#
chr5	1306287	1348162	TERT	5p15	Cri-du-Chat	123450		●	●	●	#
chr13	31211679	31275009	LGR8(RXFP2)	13q13	Cryptorchidism, unilateral or bilateral	219050		●	●		#
chr7	156479506	156496108	HLXB9(MNX1)	7q36	Currarino	176450		●	●	●	#
chr7	117120017	117308716	CFTR	7q31.2	Cystic fibrosis	219700				●	#
chr17	3486511	3513146	CTNS	17p13	Cystinosis	219800	●	●	●	●	#
chr19	33321421	33360683	SLC7A9	19q13.11	Cystinuria	220100				●	#
chr2	44399406	44442448	PREPL	2p21	Cystinuria with mitochondrial disease	606407		●	●	●	#
chr2	44356103	44401443	SLC3A1	2p21	Cystinuria with mitochondrial disease	606407		●	●	●	#
chr3	148609871	148617196	ZIC1	3q24	Dandy-Walker	220200	●	●	●	●	
chr3	148586527	148607097	ZIC4	3q24	Dandy-Walker	220200	●	●	●	●	
chrX	119444035	119487189	LAMP2	Xq24	Danon disease	300257		●	●		#
chrX	1.53E+08	1.53E+08	DCX	Xq22.3-q23	DCX-associated lissencephaly/subcortical band heterotopia	300067	●	●	●	●	#
chr5	149717428	149760063	TCOF1	5q33	Deafness	154500	●	●	●		#
chr13	19659605	19665114	GJB2(Connexin 26)	13q12	Deafness	220290		●	●		#
chrX	100487306	100490343	TIMM8A	Xq22	Deafness-dystonis-optic neuronopathy	304700		●	●	●	#
chr16	65620551	65692459	CBFB	16q22	Delayed cranial ossification			●	●		
chrX	152821179	152825834	AVPR2	Xq28	Diabetes insipidus, nephrogenic, X-linked	304800		●	●		#
chr6	144370000	144377576	HYMAI	6q24	Diabetes mellitus, transient neonatal, 1	601410		●	●	●	#
chr6	144293129	144427428	ZAC(PLAGL1)	6q24.2	Diabetes mellitus, transient neonatal, 1	601410		●	●	●	#
chr19	47055828	47067324	RPS19	19q13	Diamond blackfan anemia	105650		●	●		#
chr8	11571877	11654918	GATA4	8p23	diGeorge 1	188400	●	●	●	●	#
chr8	8679409	8788541	MFHAS1	8p23	diGeorge 1	188400		●	●	●	#
chr22	19601714	19638035	CRKL	22q11	diGeorge 1	188400		●	●	●	#
chr22	17698224	17815220	HIRA	22q11	diGeorge 1	188400	●	●	●	●	#
chr22	18124226	18151110	TBX1	22q11	diGeorge 1	188400	●	●	●	●	#
chr10	8136673	8157170	GATA3	10p14	diGeorge 2	601362	●	●	●	●	#
chrX	31048667	33277779	DMD	Xp21.1-p21.2	DMD	310200	●	●	●	●	#
chr14	54100000	58100000		14q22.2-q22.3	Dopa resistant dystonia / parkinsons	168600				●	#
chr21	34810654	34909252	DSCR1(RCAN1)	21q22	Downs syndrome critical region	190685		●	●	●	#
chr21	37517596	37561703	DSCR3	21q22	Downs syndrome critical region	190685		●	●	●	#
chr21	37661729	37809549	DYRK1A	21q22	Downs syndrome critical region	190685		●	●	●	#
chr21	35081968	36278917	RUNX1	21q22	Downs syndrome critical region	190685	●	●	●	●	#
chr18	44824170	45250999	DYM	18q21	Dyggve-delchior-claussen disease	223800	●	●	●		#
chr7	94113623	94146938	PEG10	7p21	Dystonia-11	159900		●	●		#
chr7	94123295	94123994	PEG10_ICRegion	7p21	Dystonia-11	159900		●	●		#
chr7	94042477	94133413	SGCE	7p21	Dystonia-11	159900		●	●		#
chrX	68752636	69176046	EDA	Xq13	Ectodermal dysplasia	305100	●	●	●		#
chr7	93861809	93898480	COL1A2	7q21	Ehlers-danlos	225320	●	●	●		#
chrX	147100000	155270560	EMD	Xq28	Emery-dreifuss muscular dystrophy	310300				●	#
chrX	100539435	100549657	GLA	Xq22	Fabry disease	301500		●	●		#
chr3	10068113	10141343	FANCD2	3p25.3	Fanconi anemia	227650				●	#
chr16	88331460	88410566	FANCA	16q24	Fanconi anemia	227650		●	●		#
chr2	15998134	16004579	MYCN	2q24	Feingold	164280	●	●	●	●	#
chrX	146801201	146840303	FMR1	Xq27	Fragile X	300624	●	●	●	●	#
chrX	147389831	147889899	FMR2(AFF2)	Xq28	Fragile X	300624	○	●	●	●	#
chr1	221168406	221245958	DISP1	1q41	Fryns 1q41	229850		●	●	●	
chr5	112071117	112209835	APC	5q22	Gardner	175100	●	●	●	●	#
chr16	55456620	55507263	SLC12A3	16q13	Gitelman	263800	●	●	●		#
chr1	40100001	44100000	GLUT1	1p34.2	Glucose transport defect	606777				●	#
chrX	30581461	30658645	GK	Xp21	Glycerol kinase deficiency	307030	●	●	●	●	#
chr7	41967073	42243900	GLI3	7p14	Greig cephaloplysindactyly	175700	●	●	●	●	#
chrX	153717263	153904192	F8	Xq28	Hemophilia A	306700	●	●	●		#
chrX	138440561	138473283	F9	Xq27	Hemophilia B	306900		●	●		#
chr6	160341410	160352419	IGF2R	6q25	Hepatocellular carcinoma	114550		●	●		#
chr18	46810581	46865409	MADH4(SMAD4)	18q21	Hereditary hemorrhagic telangiectasia	175050		●	●		#
chr9	130300001	133500000	ENG	9q34.11	Hereditary hemorrhagic telangiectasia	187300				●	#
chr2	47630263	47710360	MMR	2p16.3-21	Hereditary non-polyposis colon cancer	120435				●	#
chrX	62771573	62922138	ARHGEF9	Xq11	Heterotaxy			●	●		
chrX	153230091	153256123	FLNA	Xq28	Heterotopia, periventricular, X-linked dominant	300049		●	●	●	#
chr2	144100001	148700000	RET	2q22.3	Hirschsprung disease plus	142623				●	#
chr10	42892523	42945803	RET	10q11	Hirschsprung disease plus	142623	○	●	●		#
chr13	77367617	77447665	EDNRB	13q22	Hirschsprung disease plus	600155		●	●		#
chr17	10700000	22200000		17p11.2-p12	HNPP	162500				●	#
chr2	45022365	45025945	SIX3	2p21	Holopresencaphaly 2	157170	●	●	●	●	#
chr18	3402072	3448406	TGIF1	18p31	Holopresencaphaly 4	142946	○	●	●	●	#
chr13	99432320	99437019	ZIC2	13q32	Holopresencaphaly 5	609637	●	●	●	●	#
chr21	46432788	46473119	LSS	21q22	Holoprosencephaly 1	236100		●	●		
chr21	44256634	44350858	TMEM1	21q22	Holoprosencephaly 1	236100	●	●	●	●	
chr7	155288319	155297728	SHH	7q36	Holoprosencephaly 3	142945	●	●	●	●	#
chr9	97245085	97319068	PTCH1	9q22	Holoprosencephaly 7	610828	●	●	●	●	#
chr2	121210293	121466699	GLI2	2q14	Holoprosencephaly 9	610829	○	●	●		#
chr5	171221161	171366482	FBXW11	5q35	Holoprosencephaly and preaxial polydactly	264480		●	●	●	
chr12	113276118	113330630	TBX5	12q14	Holt-Oram	142900		●	●	●	#
chrX	152780581	152804778	L1CAM	Xq28	HSAS, MASA, CRASH	307000, 303350		●	●		#
chrX	148368203	148423359	IDS	Xq28	Hunter, Mucopolysaccharidosis type 2	309900	●	●	●		
chr2	172349126	172458979	SLC25A12	2q31	Hypomyelination, global, cerebral	612949		●	●		#
chrX	21960842	22176399	PHEX	Xp22	Hypophosphatemic rickets	307800		●	●		#
chr15	97010284	97325282	IGF1R	15q25	IGF-1 resistance	147370	●	●	●		#
chrX	153423653	153446455	IKBKG	Xq28	Incontinentia pigmenti	308300	●	●	●		#
chr11	17371008	17455025	ABCC8	11p15	Infantile hyperinulinism, enteropathy and deafness (Ushers Disease)	606528 / 276904		●	●	●	#
chr11	17472018	17522539	USH1C	11p15	Infantile hyperinulinism, enteropathy and deafness (Ushers Disease)	606528 / 276904		●	●	●	#
chr15	41710064	41747192	CATSPER2	15q15	Infertility and deafness	611102		●	●	●	#
chr15	41612952	41664382	KIAA0377(HISPPD2A)	15q15	Infertility and deafness	611102		●	●		#
chr6	135646817	135860576	AHI1	6q23	Joubert 3	608692		●	●		#
chr12	86966921	87060124	CEP290	12q21	Joubert 5	610188		●	●	●	#
chr8	11737443	11763055	CTSB	8p23	Kabuki	147920		●	●		
chrX	8456915	8660227	KAL1	Xp22.31	Kallmann 1	308700	●	●	●	●	#
chr8	38387813	38445509	FGFR1	8p12	Kallmann 2	147950		●	●		#
chr8	118880787	119193239	EXT1	8q24.11	Langer-Giedion	150230	●	●	●	●	#
chrX	133421923	133462362	HPRT1	Xq26.2	Lesch-Nyhan	300322	●	●	●	●	#
chr11	67130983	67136581	NDUFV1	11q13	Leukodystrophy		●	●	●		
chr17	7505822	7531642	TP53	17p13.1	Li-Fraumeni 1	151623		●	●	●	#
chr7	102899473	103417198	RELN	7q22.1	Lissencephaly with cerebellar hypoplasia	257320	○	●	●	●	#
chr1	156084461	156109878	LMNA	1q22	LMNA	150330				●	#
chr3	30622998	30710637	TGFBR2	3p24	Loeys-Dietz	610380	●	●	●		#
chr9	100907233	100956294	TGFBR1	9q22	Loeys-Dietz	609192	●	●	●	●	#
chr11	2412796	2836915	KCNQ1	11p15	Long QT	192500	●	●	●		#
chrX	128501933	128554211	OCRL	Xq25	Lowe	309000		●	●	●	#
chr8	82732751	82761115	IMPA1	8q21	Manic depressive psychosis	125480	●	●	●		#
chr15	46487797	46725210	FBN1	15q21.1	Marfan 1	154700	●	●	●	●	#
chr3	30647994	30735631	TGFBR2	3p24.1	Marfan 2	154700				●	#
chr22	28329565	28424583	NF2	22q12	Meningioma / NF2	607174/101000		●	●	●	#
chrX	77042046	77200000	ATP7A	Xq21.1	Menkes	309400	●	●	●	●	#
chr1	233597351	233678903	TBCE	1q42	Mental retardation			●	●	●	
chr3	46594217	46598956	TDGF1	3p21	Mental retardation		●	●	●	●	
chr7	142136904	142140495	PRSS1	7q34	Mental retardation			●	●	●	
chr8	1426975	1654048	DLGAP2	8p23	Mental retardation		○	●	●	●	
chr10	89613175	89718512	PTEN	10q23	Mental retardation		●	●	●	●	

chr15	23123488	23245220	UBE3A	15q11	Mental retardation			●	●	●	●	
chr16	971809	976980		16p13	Mental retardation			●	●	●	●	
chr6	101953675	102623474	GRIK2	6q16	Mental retardation, autosomal recessive 6	611092		○	●	●	●	#
chr22	49410315	49413453	ARSA	22q13	Metachromatic leukodystrophy	250100			●	●		#
chrX	9500000	17100000	HCCS	Xp22.2	Microphthalmia with linear skin defects	309801					●	#
chr3	182903176	182926972	SOX2	3q26.3-q27	Microphthalmia	206900		●	●	●	●	#
chr17	853509	1037366	ABR	17p13.3	Miller-Dieker	247200			●	●	●	#
chr17	2443686	2535638	PAFAH1B1(LIS1)	17p13.3	Miller-Dieker	247200		●	●	●	●	#
chr17	1368033	1412860	PITPNA	17p13.3	Miller-Dieker	247200			●	●	●	#
chr17	1194593	1250267	YWHAE	17p13.3	Miller-Dieker	247200		●	●	●	●	#
chr1	68274232	68299047	DIRAS3	1p31	Monosomy 1p31				●	●		
chr2	144862055	144994386	ZEB2	2q22	Mowat-Wilson	235730		○	●	●		#
chr4	1795039	1810599	FGFR3	4p16.3	Muenke / achondroplasia	602849, 100800					●	#
chr1	46654354	46664121	POMGNT1	1p34.1	Muscle-eye-brain disease	253280					●	#
chrX	149487727	149592272	MTM1	Xq28	Myotubular myopathy	310400			●	●		#
chr9	128416569	128503132	LMX1B	9q33.3	Nail-patella	161200		●	●	●	●	#
chrX	17303464	17664034	NHS	Xp22	Nance-horan	302350		●	●	●		#
chr10	21110094	21503122	NEBL	10p12	Nebulette	605491		●	●	●		
chrX	49573965	49750632	CLCN5	Xp11	Nephrolithiasis 2, X-linked, dent disease	310468			●	●	●	#
chr2	110237195	110319883	NPHP1	2q13	Nephronophthisis 1	256100		●	●	●	●	#
chr17	27288185	27352162	JJAZ1(SUZ12)	17q11.2	Neurofibromatosis Type 1	162200			●	●	●	#
chr17	26446121	26728821	NF1	17q11.2	Neurofibromatosis Type 1	162200		○	●	●	●	#
chr17	25549032	25586831	SLC6A4	17p11	Neuroticism	607834			●	●		
chr1	61320883	61694624	NFIA	1p31.3	NFIA haploinsufficiency	600727		○	●	●	●	
chr12	111340919	111432099	PTPN11	12q24	Noonan 1	163950		●	●	●	●	#
chr2	39066469	39201067	SOS1	2p22.1	Noonan 4	610733			●	●	●	#
chr3	12600108	12680678	RAF1	3p25	Noonan 5	611553			●	●		#
chrX	43692968	43717694	NDP	Xp11	Norrie	310600			●	●	●	#
chr6	100943471	101018272	SIM1	6q16	Obesity, severe			●	●	●	●	
chr15	25673628	26018061	OCA2	15q13	Oculocutaneous albinism Type 2	203200		○	●	●		#
chr20	49833990	49852421	SALL4	20q13	Okihiro	607323			●	●	●	#
chr14	36196533	36216762	PAX9	14q13	Oligodontia	604625		●	●	●	●	#
chrX	10376166	10765667	MID1	Xq22	Opitz	300000		●	●	●	●	#
chrX	106956116	107057077	MID2	Xq22	Opitz / FP	300204			●	●		#
chrX	70255131	70279029	MED12	Xq13.1	Opitz-Kaveggia	305450			●	●	●	#
chr3	190831910	191097756	TP73L	3q28	Optic atrophy	165500		●	●	●	●	#
chrX	38096302	38165553	OTC	Xp11.4	Ornithine transcarbamylase deficiency	311250		●	●	●	●	#
chrX	13662801	13697391	OFD1	Xp22.2	Orofaciodigital 1	311200		●	●	●	●	#
chr17	45616456	45633992	COL1A1	17q21	Osteogenesis imperfecta			●	●	●		
chr12	63849638	63928374	LEMD3	12q14.3	Osteopoikilosis	166700			●	●	●	#
chr12	21300001	26500000		12p12.1	Pallistar-killian	601803					●	#
chrX	102918410	102934201	PLP1	Xq22.2	Pelizaeus-merzbacher disease	312080		●	●	●	●	#
chr13	31774112	31906409	B3GALTL	13p13-q12.3	Peters plus	261540					●	#
chr19	1000000	6900000	STK11	19p13.3	Peutz-Jeghers	175200					●	#
chrX	77246425	77268980	PGK1	Xp21	Phosphoglycerate kKinase deficiency	300653			●	●		#
chr18	51045967	51454183	TCF4	18q21.2	Pitt-Hopkins	610954		○	●	●	●	#
chr4	89147844	89217953	PKD2	4q22.1	Polycystic kidney disease	613095			●	●	●	#
chr16	2078712	2125900	PKD1	16p13	Polycystic kidney disease	601313			●	●		#
chr7	156168394	156378602	LMBR1	7q36	Polydactyl preaxial 2	174500			●	●		#
chr17	25800000	31800000		17q11.2	Potocki-Lupski	610883					●	#
chr11	44242734	44288292	ALX4	11p11.2	Potocki-Shaffer	601224		●	●	●	●	#
chr11	44073675	44223555	EXT2	11p11.2	Potocki-Shaffer	601224		○	●	●	●	#
chrX	1	2700000	PAR1	Xp22	Pseudoautosomal			●	●	●		
chrX	154595003	154913754	PAR2	Xq28	Pseudoautosomal			●	●	●		
chr2	31603160	31659544	SRD5A2	2p23	Pseudovaginal perineoscrotal dypospadias	264600			●	●		#
chrX	19271972	19287886	PDHA1	Xp22	Pyruvate dehydrogenase deficiency	312170			●	●		#
chr17	33120548	33179182	TCF2(HNF1B)	17q12	Reanal cysts and diabetes	137920		●	●	●	●	#
chr13	47775912	47954023	RB1	13q14.2	Retinoblastoma	180200		●	●	●	●	#
chrX	152940458	153016323	MECP2	Xq28	Retts	312750		●	●	●	●	#
chr4	111758029	111782566	PITX2	4q25	Rieger	180500		●	●	●	●	#
chrX	99785819	99812952	SRPX2	Xq22	Rolandic epilepsy, X-linked mental retardation, and speech dyspraxia	300643			●	●	●	#
chr16	3717720	3870723	CREBBP	16p13	Rubinstein taybi	610543		●	●	●	●	#
chr16	3642941	3648097	DNASE1	16p13	Rubinstein taybi	610543			●	●	●	#
chr7	19121616	19123550	TWIST1	7p21.1	Saethre-Chotzen	101400		●	●	●	●	#
chr10	119291946	119299043	EMX2	10q26.11	Schizencephaly	269160		●	●	●	●	#
chr17	39186625	39191682	SOST	17q21.31	Sclerostin	269500			●	●	●	#
chr2	166553916	166638395	SCN1A	2q24.3	SCN1A related seizures	609634			●	●	●	#
chr1	178466065	178510811	LHX4	1q25.2	Short stature	262700		●	●	●	●	#
chrX	505079	540146	SHOX	Xp22.33	SHOX	312865		●	●	●	●	#
chr7	50615258	50838651	GRB10	7p12	Silver-Russell	180860			●	●		#
chrX	132497442	132947332	GPC3	Xq26	Simpson, golabi, behmel, type 1	312870		○	●	●		#
chr11	70823105	70837125	DHCR7	11q13.4	Smith-lemli-opitz	270400			●	●	●	#
chr17	17525512	17655488	RAI1	17p11.2	Smith-magenis	182290		●	●	●	●	#
chr5	176492686	176659820	NSD1	5q35.3	Sotos	117550		●	●	●	●	#
chr7	96477643	96502078	DLX5	7q21.3	Split hand and foot	220600			●	●	●	
chr7	96140173	96177139	SHFM1	7q21.3	Split hand and foot	220600		●	●	●	●	
chr10	103103815	103307060	BTRC	10q24.32	Split hand and foot malformation 3	600095			●	●	●	#
chr10	103360412	103444733	FBXW4	10q24.32	Split hand and foot malformation 3	600095		●	●	●	●	#
chr10	102976723	102978707	LBX1	10q24.32	Split hand and foot malformation 3	600095			●	●	●	#
chr10	103328629	103338004	POLL	10q24.32	Split hand and foot malformation 3	600095			●	●	●	#
chr2	172658454	172662647	DLX1	2q31	Split hand and foot malformation 5	606708		●	●	●	●	#
chr2	172672412	172675724	DLX2	2q31	Split hand and foot malformation 5	606708		●	●	●	●	#
chrX	7075293	7282680	STS	Xp22.31	Steroid sulfatase	308100		●	●	●	●	#
chrX	6461659	6463159	VCX3A	Xp22.31	Steroid sulfatase	308100			●	●	●	#
chr12	46653018	46684528	COL2A1	12q13.11	Stickler 1	108300		●	●	●	●	#
chr1	103115583	103346640	COL11A1	1p21	Stickler 2	604841		●	●	●		#
chr2	176653081	176656936	EVX2	2q31	Synpolydactyly	186000			●	●	●	#
chr2	176665778	176668912	HOXD13	2q31.1	Synpolydactyly	186000		●	●	●	●	#
chr2	176695334	176697891	HOXD9	2q31	Synpolydactyly	186000			●	●	●	#
chr2	176664382	176767892	HOXDgenes	2q31	Synpolydactyly	186000		●	●	●	●	#
chr17	42686207	42745076	ITGB3	17q21	Thrombasthenia of glanzmann and naegeli	273800			●	●		#
chr1	145841561	145848017	GJA8	1q21.1	Thrombocytopenia absent radius	274000		●	●	●	●	#
chr12	1950490	2677376	CACNA1C	12p13	Timothy	601005		○	●	●		#
chr16	49727387	49742684	SALL1	16q12.1	Townes brocks	107480			●	●	●	#
chr8	116489900	116750429	TRPS1	8q23.3	Trischororhinophlangeal, langer-giedion	150230		●	●	●	●	#
chr9	134756557	134809841	TSC1	9q34.13	Tuberous sclerosis 1	191100		●	●	●	●	#
chr16	2038600	2078713	TSC2	16p13.3	Tuberous sclerosis 2	613254		●	●	●	●	#
chr12	113592442	113606352	TBX3	12q24.21	Ulnar-mammary	181450			●	●	●	#
chr1	208027885	208046102	IRF6	1q32.2	Van der Woude	119300		●	●	●	●	#
chr6	43845931	43862201	VEGFA	6p21	Vascular endothelial growth factor disorders			●	●	●	●	
chr7	113513618	114118328	FOXP2	7q31.1	Verbal dyspraxia	602081		●	●	●	●	#
chr2	130995137	131002053	CFC1	2q21	Visceral heterotaxy	605376			●	●		#
chr3	10158319	10168746	VHL	3p25.3	Von hippel lindau	193300			●	●	●	#
chr22	38368319	38380539	SOX10	22q13.1	Waardenburg 4	609136					●	#
chr2	222772851	222871944	PAX3	2q36.1	Waardenburg I	193500		●	●	●	●	#
chr3	69871323	70100177	MITF	3p14.1	Waardenburg IIA	193510		●	●	●	●	#
chr11	32355900	32423662	WT1	11p13	WAGR	194072		●	●	●	●	#
chr9	136501485	136524467	POMT1/POMT2	9q34.13	Walker/Warburg	236670					●	#
chr7	73080407	73120966	ELN	7q11.23	Williams-Beuren	194050		●	●	●	●	#
chr7	73136092	73174790	LIMK1	7q11.23	Williams-Beuren	194050		●	●	●	●	#
chr7	73826245	73841595	NCF1	7q11.23	Williams-Beuren	194050		●	●	●	●	#
chr7	73080407	73841595		7q11.23	Williams-Beuren microduplication	609757					●	#
chr13	52506806	52585630	ATP7B	13q14.3	Wilson disease	277900					●	#
chr4	1784558	1827772	LETM1	4p16.3	Wolf-hirschhorn	194190		●	●	●	●	#
chr4	1842921	1953728	WHSC1	4p16.3	Wolf-hirschhorn	194190		●	●	●	●	#
chr4	1954251	1980547	WHSC2	4p16.3	Wolf-hirschhorn	194190		●	●	●	●	#
chrX	72863307	73063491	XIST	Xq13	X inactivation specific transcript			●	●	●		
chrX	153644344	153659154	DKC1	Xq28	X-linked dyskeratosis congenita	305000			●	●		#
chrX	136476012	136481923	ZIC3	Xq26.3	X-linked heterotaxy	306955		●	●	●	●	#
chrX	18567731	18600150	RS1	Xp22	X-linked juvenille retinoschisis	312700		●	●	●	●	#
chrX	123307875	123334696	SH2D1A	Xq25	X-linked lymphoproliferative type 1	308240		●	●	●	●	#
chrX	122821729	122875503	BIRC4(XIAP)	Xq25	X-linked lymphoproliferative type 2	308240		●	●	●	●	#
chrX	108771220	108863275	ACSL4	Xp22	X-linked mental retardation				●	●	●	
chrX	115216031	115219848	AGTR2	Xq23	X-linked mental retardation				●	●	●	
chrX	15753850	15783021	AP1S2	Xp22	X-linked mental retardation				●	●	●	
chrX	135575377	135691169	ARHGEF6	Xq26	X-linked mental retardation				●	●	●	
chrX	24932213	24943775	ARX	Xp21	X-linked mental retardation			●	●	●	●	
chrX	40325160	40350832	ATP6AP2	Xp11	X-linked mental retardation				●	●	●	
chrX	39795561	39921526	BCOR	Xp11	X-linked mental retardation				●	●	●	
chrX	79818351	79951889	BRWD3	Xp21	X-linked mental retardation				●	●	●	
chrX	41264287	41667212	CASK	Xp11	X-linked mental retardation				●	●	●	
chrX	119542474	119593712	CUL4B	Xq24	X-linked mental retardation				●	●	●	
chrX	69581449	69642062	DLG3	Xq13	X-linked mental retardation				●	●	●	
chrX	47379864	47394964	ELK1	Xp11	X-linked mental retardation				●	●	●	
chrX	14771450	14801105	FANCB	Xp22	X-linked mental retardation				●	●	●	
chrX	48219493	48229696	FTSJ1	Xp11	X-linked mental retardation				●	●	●	

chrX	153318715	153325009	GDI1	Xq28	X-linked mental retardation				●	●	●	
chrX	122145839	122450474	GRIA3	Xq24	X-linked mental retardation			○	●	●	●	
chrX	53474931	53478048	HADH2(HSD17B10)	Xp11	X-linked mental retardation				●	●	●	
chrX	11039373	11051122	HCCS	Xp22	X-linked mental retardation			●	●	●	●	
chrX	28515480	29884757	IL1RAPL1	Xp21	X-linked mental retardation			●	●	●	●	
chrX	53237378	53271329	JARID1C	Xp11	X-linked mental retardation				●	●	●	
chrX	73870137	74061709	KIAA2022	Xq13	X-linked mental retardation			○	●	●	●	
chrX	56275632	56328254	KLF8	Xp11	X-linked mental retardation				●	●	●	
chrX	118889762	118894646	NDUFA1	Xq24	X-linked mental retardation				●	●	●	
chrX	100973741	100999205	NXF5	Xq22	X-linked mental retardation				●	●	●	
chrX	110074169	110350829	PAK3	Xp22	X-linked mental retardation			○	●	●	●	
chrX	53979838	54088121	PHF8	Xp11	X-linked mental retardation				●	●	●	
chrX	48252315	48264146	PORCN	Xp11	X-linked mental retardation				●	●	●	
chrX	48640139	48645364	PQBP1	Xp11	X-linked mental retardation				●	●	●	
chrX	153279912	153283874	RPL10	Xq28	X-linked mental retardation				●	●	●	
chrX	50351387	50573784	SHROOM4(KIAA1202)	Xp11	X-linked mental retardation				●	●	●	
chrX	48201871	48213509	SLC38A5	Xp11	X-linked mental retardation				●	●	●	
chrX	21868763	21922876	SMS	Xp22	X-linked mental retardation				●	●	●	
chrX	139411973	139415436	SOX3	Xq27	X-linked mental retardation			●	●	●	●	
chrX	47316244	47364200	SYN1	Xp11	X-linked mental retardation				●	●	●	
chrX	38305683	38433116	TM4SF2(TSPAN7)	Xp11	X-linked mental retardation				●	●	●	
chrX	118592527	118602407	UBE2A	Xq23	X-linked mental retardation				●	●	●	
chrX	118852017	118870996	UPF3B	Xq24	X-linked mental retardation			○	●	●	●	
chrX	8393346	8394346	VCX3A	Xp22	X-linked mental retardation				●	●	●	
chrX	74508786	74659600	ZDHHC15	Xq13	X-linked mental retardation				●	●	●	
chrX	128766594	128805805	ZDHHC9	Xq25	X-linked mental retardation				●	●	●	
chrX	70376199	70391660	ZNF261(ZMYM3)	Xq13	X-linked mental retardation				●	●	●	
chrX	47191350	47227289	ZNF41	Xp11	X-linked mental retardation				●	●	●	
chrX	47581245	47666550	ZNF81	Xp11	X-linked mental retardation				●	●	●	
chrX	73557810	73670475	SLC16A2	Xq13	X-linked mental retardation, allan-herndon-dudley		300523		●	●	●	#
chrX	134895252	134957094	SLC9A6	Xq26	X-linked mental retardation, christianson type		300243		●	●	●	#
chrX	53576476	53730398	HUWE1	Xp11	X-linked mental retardation, turner		300706	○	●	●	●	#
chrX	67179440	67570372	OPHN1	Xq12	X-linked mental retardation, with cerebellar hypoplasia distinctive facial appearance	and	300486	●	●	●	●	#
chrX	18353678	18581666	CDKL5	Xp22.13	X-linked spasms		300672	●	●	●	●	#
chrX	46581319	46626733	RP2	Xp11	Xp11.3 deletion		300578	●	●	●		#
chrX	46243490	46289820	ZNF674	Xp11	Xp11.3 deletion		300578		●	●	●	#
chr9	126283337	126309520	NR5A1	9q33.3	XY Sex Reversal		184757	●	●	●	●	
chr19	62384680	62432350	ZNF264	19q13	Zinc finger protein-264				●	●		
chr2	202949916	203140719	BMPR2	2q32					●	●		
chr7	129710229	129761249	CPA4	7q32					●	●		
chr7	130058017	130079399	KLF14	7q32					●	●		
chr7	129903281	129943368	MEST	7q32					●	●		
chr7	98084545	98097116	NPTX2	7q22					●	●		
chr7	94364911	94770958	PPP1R9A	7q21				○	●	●		
chr8	140683985	140794480	KCNK9	8q24					●	●		
chr11	1975651	1980951	H19_ICRegion	11p15					●	●		
chr11	2671987	2683633	KCNQ1_ICRegion	11p15				○	●	●		
chr11	3054921	3153115	OSBPL5	11p15					●	●		
chr13	65774967	66702464	PCDH9	13q21				●	●	●		
chr14	20923198	20975244	CHD8	14q11					●	●		
chr14	100252981	100281225	DLK1	14q32					●	●		
chr14	100261144	100364369	DLK1&MEG3_ICRegion	14q32					●	●		
chr14	28306038	28308622	FOXG1B	14q12				●	●	●		
chr14	100352214	100407118	MEG3	14q32					●	●		
chr14	20897000	20932000	MicroDeletionRegion	14q11					●	●		
chr14	20889476	20922265	SUPT16H	14q11					●	●		
chr15	49527231	49702259	DMXL2	15q21					●	●		
chr16	6009133	7702500	A2BP1	16p13				●	●	●		
chr18	73090996	73111084	GALR1	18q23					●	●		
chr18	42798570	42820446	TCEB3C	18q21					●	●		
chr19	62005614	62053875	PEG3	19q13					●	●		
chr19	62040958	62043979	PEG3_ICRegion	19q13					●	●		
chr19	61967731	62053886	ZIM2	19q13					●	●		
chr20	56676787	56680787	GNAS_ICRegion	20q13					●	●		
chr20	56859056	56861251	GNAS_ICRegion	20q13					●	●		
chr20	56896896	56899356	GNAS_ICRegion	20q13					●	●		
chr20	41566487	41613948	L3MBTL	20q13					●	●		
chr20	35573020	35595505	NNAT	20q11					●	●		
chr22	16454903	16491588	ATP6V1E1	22q11					●	●		
chrY	2714821	2716600	SRY	Yp11.31				●	●	●	●	#

#	Microdeletions or microduplications have been associated with this condition
○	Low coverage of syndrome
●	Coverage of syndrome
	No Coverage of Syndrome

Each design includes coverage for all aneuploidies and all sub-telemeres

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