



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	8634369	86344001	FZD4	11q14	11q14 Microdeletion		●	●	●	#
chr11	114500000	130800000		11q23.3-q24.3	11q23.3 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	15q24.1 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		16p13.1	16p13.1 Microdeletion predisposition to autism		●	●	●	#
chr17	41217449	41289373	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 Microduplication		●	●	●	#
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	126206008	LMNB1	5q23	Adult-onset autosomal dominant leukodystrophy	169500	●	●	●	#
chrX	100491098	100532426	BTX	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	22819887	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	5819885	6150822	NLGN4X	Xp22	Autism	209850	●	●	●	#
chrY	13052929	13603474	AZFa	Yq11.2	AZFa region	415000	●	●	●	#
chrY	23663736	25513794	AZFb	Yq11.2	AZFb region	415000	●	●	●	#
chrY	23539798	23560998	BPY2	Yq11.2	AZFb region	415000	●	●	●	#
chrY	25173539	25194740	BPY2	Yq11.2	AZFb region	415000	●	●	●	#
chrY	25586438	25607639	BPY2	Yq11.2	AZFb region	415000	●	●	●	#
chrY	24600764	24603549	CDY1	Yq11.2	AZFb region	415000	●	●	●	#
chrY	26177652	26180436	CDY1	Yq11.2	AZFb region	415000	●	●	●	#
chrY	23684890	23754627	DAZ1	Yq11.2	AZFb region	415000	●	●	●	#
chrY	24765503	24770366	GOLGA2LY(AF332229)	Yq11.2	AZFb region	415000	●	●	●	#
chrY	26010846	26015709	GOLGA2LY(AF332229)	Yq11.2	AZFb region	415000	●	●	●	#
chr15	46285791	46382417	SLC12A1	15q21	Barter 1	601678	●	●	●	#
chr11	128213125	128242478	KCNJ1	11q24	Barter 2	241200	●	●	●	#
chr1	16221073	16256063	CLCNKB	1p36	Barter 3	607364	●	●	●	#
chr1	55237205	55247053	BSND	1p32	Barter 4A	602522	●	●	●	#
chr1	16217957	16233132	CLCNKA	1p36	Barter 4B	613090	●	●	●	#
chr3	121902590	123488032	CASR	3q13.33-q21.1	Barter with autosomal dominant hypocalcaemia	601199	●	●	●	#
chr11	2851440	2873550	CKN1C	11p15	Beckwith-Wiedemann	130650	○	●	●	#
chr11	1962981	1965640	H19	11p15	Beckwith-Wiedemann	130650	●	●	●	#
chr11	2069825	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	●	●	●	#
chr10	158000	330000	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	○	●	●	#

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 juvenile 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	47394994	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	PQB1	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	ZDHC15	Xq13	X-linked mental retardation		●	●	●	
chrX	128766594	128805805	ZDHC9	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	53793398	HUWE1	Xp11	X-linked mental retardation, turner	300706	○	●	●	#
chrX	67179440	67570372	OPHN1	Xq12	X-linked mental retardation, with cerebellar hypoplasia and distinctive facial appearance	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	FOXP1B	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	56876787	56880787	GNAS_ICRegion	20q13			●	●	●	
chr20	56859055	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

Summary for each array, plus coverage within each gene/targeted 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)

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