GENETICS CENTER

180k ISCA Oligo-SNP Array CGH
Disease Regions Targeted

Associated Condition	Gene	Chr/Band	OMIM #
1p31 Deletion	DIRAS3	1p31	605193
1p36 Microdeletion	SKI	1p36	607872
1p36 Microdeletion	TP73	1p36	607872
2p15-16.1 Microdeletion	Multiple	2p15-16.1	612513
2q37.3 Deletion	HDAC4	2q37	600430
3q29 Microdeletion	PAK2	3q29	609425
6p24 Microdeletion	FKHL7	6p24	612852
8p23.1 Deletion	CTSB	8p23	116810
9p Deletion	DMRT1	9p24	158170
9p Deletion	DMRT2	9p24	158170
9q34.3 Microdeletion	EHMT1	9q34.3	610253
10q22-23 Deletion	GRID1	10q22	610659
12q14.1-q15 Deletion	GRIP1	12q14	604597
13q Deletion	GPC5	13q31	602446
13q Deletion	GPC6	13q31	604404
13q Deletion	PCDH9	13q21	603581
14q11-q22 Deletion	CHD8	14q11	613457
14q11-q22 Deletion	SUPT16H	14q11	613457
14q22 Microdeletion, Branchiootic syndrome 3	SIX1	14q22	608389
14q22 Microdeletion, Branchiootic syndrome 3	SIX4	14q22	608389
14q22 Microdeletion, Microphthalmia, isolated, with cataract 2	SIX6	14q22	212550
14q22 Microdeletion, Microphthalmia, syndromic 5	OTX2	14q22	610125
14q22 Microdeletion, Orofacial Cleft 11	BMP4	14q22	600625
15q13.3 Microdeletion	CHRNA7	15q13	612001
17q21.31 Microdeletion	CRHR1	17q21.31	610443
17q21.31 Microduplication	MAPT	17q21.31	613533
18q Microdeletion	TCEB3B	18q21	609522
18q Deletion	ZNF407	18q22	301808
18q Deletion	GALR1	18q23	600377
22q11.2 Deletion	BCR	22q11	611867
Aarskog-Scott	FGD1	Xp11	305400
Adrenal hypoplasia congenital	NR0B1	Xp21	300200
Adult-onset autosomal dominant leukodystrophy	LMNB1	5q23	169500
Agammaglobulinemia, X-linked	BTK	Xq22.1	300755
Alagille	JAG1	20p12	118450
Alexander disease	NDUFV1	11q13	203450
Alpha-thalassemia mental retardation	HBA1 & HBA2	16p13	141750
Alpha-thalassemia mental retardation	SOX8	16p13.3	141750
Alpha-thalassemia mental retardation, X-linked	ATRX	Xq21	300032
Alport, X-linked	COL4A5	Xq22.3	301050
Androgen insensitivity	AR	Xq12	300068
Angelman / Prader-Willi	MAGEL2	15q11	105830
Angelman / Prader-Willi	MKRN3	15q11	105830
Angelman / Prader-Willi	NDN	15q11	105830
Angelman / Prader-Willi	NIPA1	15q11	105830
Angelman / Prader-Willi	NIPA2	15q11	105830
Angelman / Prader-Willi	PWS IC	15q11	105830
Angelman / Prader-Willi	SNORD107/64/108	15q11	105830
Angelman / Prader-Willi	SNORD109B	15q11	105830
Angelman / Prader-Willi	SnoRNA	15q11	105830
Angelman / Prader-Willi	SNRPN	15q11	105830
Angelman / Prader-Willi	SNRPN	15q11	105830
Angelman / Prader-Willi	SNURF	15q11	105830
Angelman / Prader-Willi	UBE3A	15q11	105830
Aniridia	PAX6	11p13-p14	106210
Anxiety-related personality traits	SLC6A4	17p11	607834
Atrial fibrillation	GJA5	1q21	121013
Atrial septal defect	NKX2-5	5q35	108900
Autism	A2BP1	16p13	209850
Autism	ATP10A	15q12	209850
Autism	CADPS2	7q31	209850
Autism	CNTN4	3p25	209850
Autism	CNTNAP2	7q35-36	209850
Autism	DLGAP2	8p23	209850
Autism	EGR2	10q21	209850
Autism	EN2	7q36	209850
Autism	GABRB3	15q12	209850
Autism	MET	7q31	209850
Autism	NLGN3	Xq13	209850
Autism	NLGN4X	Xp22	209850
Autism	SHANK3	22q13	209850
Autism	SLC4A10	2q24	209850
Autism	DISC1	1q42	209850
Autism	NRXN1	2p16	209850
Autism	NPTX2	7q22	600750
Bartter 1	SLC12A1	15q21	601678
Bartter 2	KCNJ1	11q24	241200
Bartter 3	CLCNKB	1p36	607364
Bartter 4A	BSND	1p32	602522
Bartter 4B	CLCNKA	1p36	613090
Bartter with autosomal dominant hypocalcemia	CASR	3q13.33-q21.1	601199
Beckwith-Wiedemann	H19 IC	11p15	130650
Beckwith-Wiedemann	KCNQ1 IC	11p15	130650
Beckwith-Wiedemann	CDKN1C	11p15	130650
Beckwith-Wiedemann	H19	11p15	130650
Beckwith-Wiedemann	IGF2	11p15	130650
Beckwith-Wiedemann	INS	11p15	130650
Beckwith-Wiedemann	PHLDA2	11p15	130650
Beckwith-Wiedemann	SLC22A18	11p15	130650
Bilateral frontoparietal polymicrogyria	GPR56	16q13	606854
Birk-Barel	KCNK9	8q24	605874
Blepharophimosis	FOXL2	3q22	110100
Borjeson-Forssman-Lehmann	PHF6	Xq26	301900
Brachydactyly type C	GDF5	20q11	113100
Branchiootorenal	EYA1	8q13	113650
Breast cancer	BRCA2	13q13	114480
Brunner	MAOA	Xp11	300615
Campomelic dysplasia	SOX9	17q24	114290
Carnitine biosynthesis pathway	PAR2	Xq28	300777
Cataract, juvenile, with microcornea A and glucosuria	SLC16A12	10q23	612018
Cat-Eye Syndrome (Type I)	ATP6V1E1	22q11	115470
Cerebral amyloid angiopathy	APP	21q21	605714
Charcot-Marie-Tooth disease, X-linked	PRPS1	Xq22	311070
Charcot-Marie-Tooth disease, type 1A	PMP22	17p11	118220
CHARGE	CHD7	8q12	214800

Associated Condition	Gene	Chr/Band	OMIM #
Chondrodysplasia punctata, X-linked recessive	CDPX1	Xp22	302950
Choroideremia	CHM	Xp21	303100
Chronic granulomatous disease	CYBB	Xp11	306400
Chronic pancreatitis	SPINK1	5q32	167800
Cleft lip	MSX1	4p16	608874
Cleft palate	SATB2	2q32	119540
Cleidocranial dysplasia	RUNX2	6p21	119600
Coffin-Lowry	RPS6KA3	Xp22	303600
Congenital adrenal hyperplasia (CAH)	CYP21A2	6p21.32	201910
Congenital diaphragmatic hernia	NR2F2	15q26	142340
Cornelia de Lange 1	NIPBL	5p13	122470
Cornelia de Lange 2	SMC1L1	Xp11	300590
Cowden	BMPR1A	10q23	158350
Craniosynostosis	MSX2	5q35	604757
Craniosynostosis	SOX6	11p15.1-p15.2	218350
Creatine deficiency / X-linked mental retardation	SLC6A8	16p11	300352
Creatine deficiency / X-linked mental retardation	SLC6A8	16p11	300352
Cri-du-Chat	TERT	5p15	123450
Cryptorchidism, idiopathic	LGR8	13q13	219050
Currarino	HLXB9	7q36	176450
Cystinosis	CTNS	17p13	219800
Cystinuria with mitochondrial disease	PREPL	2p21	606407
Cystinuria with mitochondrial disease	SLC3A1	2p21	606407
Dandy-Walker	ZIC1	3q24	220200
Dandy-Walker	ZIC4	3q24	220200
Danon disease	LAMP2	Xq24	300257
Deafness, autosomal recessive 1A	GJB2	13q12	220290
Deafness, sensorineural, and male infertility	CATSPER2	15q15	611102
Deafness-dystonia-optic neuronopathy	TIMM8A	Xq22	304700
Delayed cranial ossification	CBFB	16q22	121360
Diabetes insipidus, nephrogenic, X-linked	AVPR2	Xq28	304800
Diabetes mellitus, transient neonatal, 1	ZAC	6q24.2	601410
Diabetes mellitus, transient neonatal, 1	HYMAI	6q24	601410
Diamond-Blackfan anemia	RPS19	19q13	105650
Diaphragmatic hernia 3	ZFPM2	8q23	610187
DiGeorge 1	CRKL	22q11	188400
DiGeorge 1	GATA4	8p23	188400
DiGeorge 1	HIRA	22q11	188400
DiGeorge 1	MFHAS1	8p23	188400
DiGeorge 1	TBX1	22q11	188400
DiGeorge 2	GATA3	10p14	601362
DiGeorge 2	NEBL	10p12	601362
Down syndrome critical region	DSCR1	21q22	190685
Down syndrome critical region	DSCR3	21q22	190685
Down syndrome critical region	DYRK1A	21q22	190685
Down syndrome critical region	RUNX1	21q22	190685
Duane-radial ray	SALL4	20q13	607323
Duchenne muscular dystrophy	DMD	Xp21.1-p21.2	310200
Dyggve-Melchior-Clausen disease	DYM	18q21	223800
Dystonia-11	PEG10	7p21	159900
Dystonia-11	PEG10 IC	7p21	159900
Dystonia-11	SGCE	7p21	159900
Ectodermal dysplasia	EDA	Xq13	305100
Ehlers-Danlos	COL1A2	7q21	225320
Exudative vitreoretinopathy	FZD4	11q14	133780
Fabry disease	GLA	Xq22	301500
Fanconi anemia	FANCA	16q24	227650
Fanconi anemia	FANCB	Xp22	300515
Feingold	MYCN	2q24	164280
FG syndrome 4	CASK	Xp11	300172
Focal dermal hypoplasia	PORCN	Xp11	300651
Forebrain defects	TDGF1	3p21	187395
Fragile X	FMR1	Xq27	300624
Fragile X	FMR2	Xq28	300624
Fryns 1q41	DISP1	1q41	229850
Gardner	APC	5q22	175100
Gitelman	SLC12A3	16q13	263800
Glycerol kinase deficiency	GK	Xp21	307030
Golabi-Ito-Hall	PQBP1	Xp11	300463
Gonadal Dysgenesis	SRY	Yp11.31	480000
Greig cephalopolysyndactyly	GLI3	7p14	175700
Hemophilia A	F8	Xq28	306700
Hemophilia B	F9	Xq27	306900
Hepatocellular carcinoma	IGF2R	6q25	114550
Hereditary hemorrhagic telangiectasia	MADH4	18q21	175050
Hereditary pancreatitis	PRSS1	7q34	276000
Heterotopia, periventricular, X-linked dominant	FLNA	Xq28	300049
Hirschsprung disease	EDNRB	13q22	600155
Hirschsprung disease plus	RET	10q11	142623
Holoprosencephaly 1	LSS	21q22	600909
Holoprosencephaly 1	TMEM1	21q22	602103
Holoprosencephaly 2	SIX3	2p21	157170
Holoprosencephaly 3	SHH	7q36	142945
Holoprosencephaly 4	TGIF1	18p31	142946
Holoprosencephaly 5	ZIC2	13q32	609637
Holoprosencephaly 7	PTCH1	9q22	610828
Holoprosencephaly 9	GLI2	2q14	610829
Holoprosencephaly and preaxial polydactly	FBXW11	5q35	264480
Holt-Oram	TBX5	12q14	142900
HSAS, MASA, CRASH	L1CAM	Xq28	303350
Hunter, mucopolysaccharidosis type 2	IDS	Xq28	309900
Hyperekplexia and Epilepsy	ARHGEF9	Xq11	300607
Hypomyelination, global, cerebral	SLC25A12	2q31	612949
Hypophosphatemic rickets	PHEX	Xp22	307800
Ichthyosis, X-linked	STS	Xp22.31	308100
Ichthyosis, X-linked	VCX3A	Xp22	308100
IGF-1 resistance	IGF1R	15q25	147370
Incontinentia pigmenti	IKBKG	Xq28	308300
Infertility and deafness	KIAA0377 (HISPPD2A)	15q15	611102
Joubert 3	AHI1	6q23	608894
Joubert 5	CEP290	12q21	610188
Kallmann 1	KAL1	Xp22.31	308700
Kallmann 2	FGFR1	8p12	147950
Kenny-Caffey	TBCE	1q42	244460
Langer-Giedion	EXT1	8q24.11	150230
Leri-Weill Dyschondrosteosis	PAR1	Xp22	127300

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(more associated syndromes on the reverse side)

Associated Condition	Gene	Chr/Band	OMIM #
Lesch-Nyhan	HPRT1	Xq26.2	300322
Li-Fraumeni 1	TP53	17p13.1	151623
Lissencephaly with cerebellar hypoplasia	RELN	7q22.1	257320
Lissencephaly, X-linked	DCX	Xq22.3-q23	300067
Loeys-Dietz, type 1A	TGFBR1	9q22	609192
Loeys-Dietz, type 2B	TGFBR2	3p24	610380
Long QT	KCNQ1	11p15	192500
Lowe	OCRL	Xq25	309000
Macrocephaly/Autism	PTEN	10q23	605309
Major affective disorder	IMPA1	8q21	125480
Marfan 1	FBN1	15q21.1	154700
Meningioma	NF2	22q12	607174
Menkes disease	ATP7A	Xq21.1	309400
Mental Retardation	DMXL2	15q21	612186
Mental Retardation, Autosomal Dominant 5	SYNGAP1	6p21.3	612621
Mental Retardation, Autosomal Recessive 6	GRIK2	6q16	611092
Metachromatic leukodystrophy	ARSA	22q13	250100
Microphthalmia, syndromic 2	BCOR	Xp11	300166
Microphthalmia, syndromic 7	HCCS	Xp22	300056
Microphthalmia	SOX2	3q26.3-q27	206900
Migraine, familial hemiplegic, 3	SCN1A	2q24.3	609634
Miller-Dieker	ABR	17p13.3	247200
Miller-Dieker	PAFAH1B1	17p13.3	247200
Miller-Dieker	PITPNA	17p13.3	247200
Miller-Dieker	YWHAE	17p13.3	247200
Mitochondiral Complex I Deficiency	NDUFA1	Xq24	252010
Mowat-Wilson	ZEB2	2q22	235730
Myotubular myopathy	MTM1	Xq28	310400
Nail-patella	LMX1B	9q33.3	161200
Nance-Horan	NHS	Xp22	302350
Nephrolithiasis 1	CLCN5	Xp11	310468
Nephronophthisis 1	NPHP1	2q13	256100
Neural development	NNAT	20q11	603106
Neurite formation	PPP1R9A	7q21	602468
Neurofibromatosis Type 1	JJAZ1	17q11.2	162200
Neurofibromatosis Type 1	NF1	17q11.2	162200
NFIA haploinsufficiency	NFIA	1p31.3	600727
Noonan 1	PTPN11	12q24	163950
Noonan 4	SOS1	2p22.1	610733
Noonan 5	RAF1	3p25	611553
Norrie disease	NDP	Xp11	310600
Obesity, severe	SIM1	6q16	603128
Oculocutaneous albinism type 2	OCA2	15q13	203200
Oligodontia	PAX9	14q13	604625
Opitz G	MID1	Xq22	300000
Opitz G	MID2	Xq22	300204
Opitz-Kaveggia	MED12	Xq13.1	305450
Optic atrophy	TP73L	3q28	165500
Ornithine transcarbamylase deficiency	OTC	Xp11.4	311250
Orofaciodigital 1	OFD1	Xp22.2	311200
Osteogenesis imperfecta	COL1A1	17q21	120150
Osteopoikilosis	LEMD3	12q14.3	166705
Pelizaeus-Merzbacher disease	PLP1	Xq22.2	312080
Phosphoglycerate kinase deficiency	PGK1	Xp21	300653
Pitt-Hopkins	TCF4	18q21.2	610954
Polycystic kidney disease 1	PKD1	16p13	601313
Polycystic kidney disease 2	PKD2	4q22.1	613095
Polydactyly preaxial 2	LMBR1	7q36	174500
Potocki-Shaffer	ALX4	11p11.2	601224
Potocki-Shaffer	EXT2	11p11.2	601224
Pseudohypoparathyroidism Ia	GNAS	20q13	103580
Pseudohypoparathyroidism Ia	GNAS IC	20q13	103580
Pseudovaginal perineoscrotal hypospadias	SRD5A2	2p23	264600
Pulmonary venoocclusive disease	BMPR2	2q32	265450
Pyruvate dehydrogenase deficiency	PDHA1	Xp22	312170
Renal cysts and diabetes	TCF2	17q12	137920
Retinoblastoma	RB1	13q14.2	180200
Rett syndrome, congenital variant	FOXG1B	14q12	613454
Rett	MECP2	Xq28	312750
Rieger	PITX2	4q25	180500
Rolandic epilepsy, mental retardation, and speech dyspraxia	SRPX2	Xq22	300643
Rubinstein-Taybi	CREBBP	16p13	610543
Rubinstein-Taybi	DNASE1	16p13	610543
Russell-Silver	CPA4	7q32	607635
Russell-Silver	GRB10	7p12	180860
Schizophrenia 11	NRG3	10q22	608078
Sclerosteosis	SOST	17q21.31	269500
Scoliosis	CHD2	15q26	602119
Short stature	LHX4	1q25.2	262700
SHOX	SHOX	Xp22.33	312865
Simpson-Golabi-Behmel, type 1	GPC3	Xq26	312870
Small size	PEG3	19q13	601483
Small size	PEG3 IC	19q13	
Small size	ZIM2	19q13	601483
Smith-Lemli-Opitz	DHCR7	11q13.4	270400
Smith-Magenis	RAI1	17p11.2	182290
Sotos	NSD1	5q35.3	117550
Split-hand / foot malformation 1 with deafness	DLX5	7q21.3	220600
Split-hand / foot malformation 1 with deafness	SHFM1	7q21.3	220600
Split-hand / foot malformation 3	BTRC	10q24.32	600095
Split-hand / foot malformation 3	FBXW4	10q24.32	600095
Split-hand / foot malformation 3	LBX1	10q24.32	600095
Split-hand / foot malformation 3	POLL	10q24.32	600095
Split-hand / foot malformation 5	DLX1	2q31	606708
Split-hand / foot malformation 5	DLX2	2q31	606708

Associated Condition	Gene	Chr/Band	OMIM #
Stickler 1	COL2A1	12q13.11	108300
Stickler 2	COL11A1	1p21	604841
Synpolydactyly	EVX2	2q31	186000
Synpolydactyly	HOXD13	2q31.1	186000
Synpolydactyly	HOXD9	2q31	186000
Thrombasthenia of Glanzmann and Naegeli	ITGB3	17q21	273800
Thrombocytopenia absent radius	GJA8	1q21.1	274000
Timothy	CACNA1C	12p13	601005
Townes-Brocks	SALL1	16q12.1	107480
Treacher Collins mandibulofacial dysostosis	TCOF1	5q33	154500
Trichorhinophalangeal	TRPS1	8q23.3	150230
Tuberous sclerosis 1	TSC1	9q34.13	191100
Tuberous sclerosis 2	TSC2	16p13.3	613254
Ulnar-mammary	TBX3	12q24.21	181450
Usher	ABCC8	11p15	276904
Usher	USH1C	11p15	276904
van der Woude	IRF6	1q32.2	119300
Vascular endothelial growth factor disorders	VEGFA	6p21	192240
Verbal dyspraxia	FOXP2	7q31.1	602081
Visceral heterotaxy	CFC1	2q21	605376
Visceral heterotaxy	CFC1	2q21	605376
von Hippel-Lindau	VHL	3p25.3	193300
Waardenburg I	PAX3	2q36.1	193500
Waardenburg IIA	MITF	3p14.1	193510
WAGR	WT1	11p13	194072
Williams-Beuren	ELN	7q11.23	194050
Williams-Beuren	GTF2I	7q11.23	194050
Williams-Beuren	GTF2IRD1	7q11.23	194050
Williams-Beuren	LIMK1	7q11.23	194050
Williams-Beuren	NCF1	7q11.23	194050
Wolf-Hirschhorn	LETM1	4p16.3	194190
Wolf-Hirschhorn	WHSC1	4p16.3	194190
Wolf-Hirschhorn	WHSC2	4p16.3	194190
X-inactivation, familial skewed	XIST	Xq13	314670
X-linked dyskeratosis congenita	DKC1	Xq28	305000
X-linked heterotaxy	ZIC3	Xq26.3	306955
X-linked juvenille retinoschisis	RS1	Xp22	312700
X-linked lymphoproliferative type 1	SH2D1A	Xq25	308240
X-linked lymphoproliferative type 2	BIRC4	Xq25	308240
X-linked mental retardation	ACSL4	Xp22	300387
X-linked mental retardation	AGTR2	Xq23	300034
X-linked mental retardation	AP1S2	Xp22	300630
X-linked mental retardation	ARHGEF6	Xq26	300436
X-linked mental retardation	ARX	Xp21	300419
X-linked mental retardation	ATP6AP2	Xp11	300423
X-linked mental retardation	BRWD3	Xp21	300659
X-linked mental retardation	CUL4B	Xq24	300354
X-linked mental retardation	DLG3	Xq13	300189
X-linked mental retardation	FTSJ1	Xp11	309549
X-linked mental retardation	GDI1	Xq28	309541
X-linked mental retardation	GRIA3	Xq24	300699
X-linked mental retardation	HADH2	Xp11	300220
X-linked mental retardation	IL1RAPL1	Xp21	300143
X-linked mental retardation	JARID1C	Xp11	300534
X-linked mental retardation	KIAA2022	Xq13	300524
X-linked mental retardation	OPHN1	Xq12	300486
X-linked mental retardation	PAK3	Xp22	300558
X-linked mental retardation	PHF8	Xp11	300263
X-linked mental retardation	SLC38A5	Xp11	309549
X-linked mental retardation	SMS	Xp22	309583
X-linked mental retardation	SOX3	Xq27	300123
X-linked mental retardation	SYN1	Xp11	300491
X-linked mental retardation	TM4SF2	Xp11	300210
X-linked mental retardation	UBE2A	Xq23	312180
X-linked mental retardation	UPF3B	Xq24	300676
X-linked mental retardation	VCX3A	Xp22	300533
X-linked mental retardation	ZDHHC15	Xq13	300577
X-linked mental retardation	ZDHHC9	Xq25	300799
X-linked mental retardation	ZNF261	Xq13	300061
X-linked mental retardation	ZNF41	Xp11	314995
X-linked mental retardation	ZNF81	Xp11	300498
X-linked mental retardation, Allan-Herndon-Dudley	SLC16A2	Xq13	300523
X-linked mental retardation, Christianson type	SLC9A6	Xq26	300243
X-linked mental retardation, Stocco dos Santos	SHROOM4	Xp11	300434
X-linked mental retardation, Turner	HUWE1	Xp11	300706
X-linked spasms	CDKL5	Xp22.13	300672
Xp11.3 Deletion	RP2	Xp11	300578
Xp11.3 Deletion	ZNF674	Xp11	300578
Xq28 Microdeletion	ABCD1	Xq28	300475
Xq28 Microduplication	RPL10	Xq28	300815
XY sex reversal	NR5A1	9q33.3	184757
Areas of clinical interest	ELK1	Xp11	311040
	KLF8	Xp11	300286
	NXF5	Xp11	300319
	KLF14	7q32	609393
	MEST	7q32	601029
	OSBPL5	11p15	606733
	DLK1&MEG3 IC	14q32	608149
	DLK1	14q32	176290
	MEG3	14q32	605636
	ZNF264	19q13	604668
	L3MBTL	20q13	608802
Unique subtelomeric regions	Multiple	43 sites	
Unique pericentromeric regions	Multiple	43 sites	
Aneuploidy	Multiple	Any individual chromosome	

NOTE: The 180k ISCA v.2 oligonucleotide-SNP array offered by the Genetics Center is a diagnostic screening tool designed to evaluate the entire genome for gains or losses of genomic material and loss of heterozygosity that could indicate uniparental disomy. As of September 2013, the above listed conditions have been associated with the indicated regions being targeted by the 180k ISCA array. This listing is not all-inclusive and is subject to change as more is discovered about the associations between syndromes, genes, and the targeted regions. The 180k ISCA array may detect copy number variations of uncertain clinical significance, or changes associated with adult onset or cancer predisposing disorders that may manifest later in life and be unrelated to the patient's current clinical picture. Also, small imbalances (less than 200Kb or 10 probes in length) and imbalances in highly repetitive regions (pericentromeric and telomeric regions) may not be detected. The ability of this assay to detect mosaicism is limited, as the assay was not designed for this purpose. For certain conditions, additional testing may be warranted. Please contact the Genetics Center to discuss patient samples if there are any questions.