

180k ISCA Oligo Array CGH Disease Regions Targeted

Associated Condition	Gene	Chr/Band	OMIM #	Associated Condition	Gene	Chr/Band	OMIM #
1p31 Deletion	DIRAS3	1p31	605193	Chondrodysplasia punctata, X-linked recessive	CDPX1	Xp22	302950
1p36 Microdeletion	SKI	1p36	607872	Choroideremia	CHM	Xp21	303100
1p36 Microdeletion	TP73	1p36	607872	Chronic granulomatous disease	CYBB	Xp11	306400
2p15-16.1 Microdeletion	Multiple	2p15-16.1	612513	Chronic pancreatitis	SPINK1	5q32	167800
2q37.3 Deletion	HDAC4	2q37	600430	Cleft lip	MSX1	4p16	608874
3q29 Microdeletion	PAK2	3q29	609425	Cleft palate	SATB2	2q32	119540
6p24 Microdeletion	FKHL7	6p24	612852	Cleidocranial dysplasia	RUNX2	6p21	119600
8p23.1 Deletion	CTSB	8p23	116810	Coffin-Lowry	RPS6KA3	Xp22	303600
9p Deletion	DMRT1	9p24	158170	Congenital adrenal hyperplasia (CAH)	CYP21A2	6p21.32	201910
9p Deletion	DMRT2	9p24	158170	Congenital diaphragmatic hernia	NR2F2	15q26	142340
9q34.3 Microdeletion	EHMT1	9q34.3	610253	Cornelia de Lange 1	NIPBL	5p13	122470
10q22-23 Deletion	GRID1	10q22	610659	Cornelia de Lange 2	SMC1L1	Xp11	300590
12q14.1-q15 Deletion	GRIP1	12q14	604597	Cowden	MLPR1A	10q23	158350
13q Deletion	GPC5	13q31	602446	Craniosynostosis	MSX2	5q35	604757
13q Deletion	GPC6	13q31	604404	Craniosynostosis	SOX6	11p15.1-p15.2	218350
13q Deletion	PCDH9	13q21	603581	Creatine deficiency / X-linked mental retardation	SLC6A8	16p11	300352
14q11-q22 Deletion	CHD8	14q11	613457	Creatine deficiency / X-linked mental retardation	SLC6A8	16p11	300352
14q11-q22 Deletion	SUPT16H	14q11	613457	Cri-du-Chat	TERT	5p15	123450
14q22 Microdeletion, Branchiootic syndrome 3	SIX1	14q22	608389	Cryptorchidism, idiopathic	LGR8	13q13	219050
14q22 Microdeletion, Branchiootic syndrome 3	SIX4	14q22	608389	Currarino	HLXB9	7q36	176450
14q22 Microdeletion, Microphthalmia, isolated, with cataract 2	SIX6	14q22	212550	Cystinosis	CTNS	17p13	219800
14q22 Microdeletion, Microphthalmia, syndromic 5	OTX2	14q22	610125	Cystinuria with mitochondrial disease	PREPL	2p21	606407
14q22 Microdeletion, Orofacial Cleft 11	BMP4	14q22	600625	Cystinuria with mitochondrial disease	SLC3A1	2p21	606407
15q13.3 Microdeletion	CHRNA7	15q13	612001	Dandy-Walker	ZIC1	3q24	222020
17q21.31 Microdeletion	CRHR1	17q21.31	610443	Dandy-Walker	ZIC4	3q24	222020
17q21.31 Microduplication	MAPT	17q21.31	613533	Danon disease	LAMP2	Xq24	300257
18q Microdeletion	TCEB3B	18q21	609522	Deafness, autosomal recessive 1A	GJB2	13q12	220290
18q Deletion	ZNF407	18q22	301808	Deafness, sensorineural, and male infertility	CATSPER2	15q15	611102
18q Deletion	GALR1	18q23	600377	Deafness-dystonia-optic neuropathy	TIMM8A	Xq22	304700
22q11.2 Deletion	BCR	22q11	611867	Delayed cranial ossification	CBFB	16q22	121360
Aarskog-Scott	FGD1	Xp11	305400	Diabetes insipidus, nephrogenic, X-linked	AVPR2	Xq28	304800
Adrenal hypoplasia congenital	NR0B1	Xp21	300200	Diabetes mellitus, transient neonatal, 1	ZAC	6q24.2	601410
Adult-onset autosomal dominant leukodystrophy	LMNB1	5q23	169500	Diabetes mellitus, transient neonatal, 1	HYMAI	6q24	601410
Agammaglobulinemia, X-linked	BTK	Xq22.1	300755	Diamond-Blackfan anemia	RPS19	19q13	105650
Alagille	JAG1	20p12	118450	Diaphragmatic hernia 3	ZFPM2	8q23	610187
Alexander disease	NDUFV1	11q13	203450	DiGeorge 1	CRKL	22q11	188400
Alpha-thalassemia mental retardation	HBA1 & HBA2	16p13	141750	DiGeorge 1	GATA4	8p23	188400
Alpha-thalassemia mental retardation	SOX8	16p13.3	141750	DiGeorge 1	HIRA	22q11	188400
Alpha-thalassemia mental retardation, X-linked	ATRX	Xq21	300032	DiGeorge 1	MFHAS1	8p23	188400
Alport, X-linked	COL4A5	Xq22.3	301050	DiGeorge 1	TBX1	22q11	188400
Androgen insensitivity	AR	Xq12	300068	DiGeorge 2	GATA3	10p14	601362
Angelman / Prader-Willi	MAGEL2	15q11	105830	DiGeorge 2	NEBL	10p12	601362
Angelman / Prader-Willi	MKRN3	15q11	105830	Down syndrome critical region	DSCR1	21q22	190685
Angelman / Prader-Willi	NDN	15q11	105830	Down syndrome critical region	DSCR3	21q22	190685
Angelman / Prader-Willi	NIPA1	15q11	105830	Down syndrome critical region	DYRK1A	21q22	190685
Angelman / Prader-Willi	NIPA2	15q11	105830	Down syndrome critical region	RUNX1	21q22	190685
Angelman / Prader-Willi	PWS1C	15q11	105830	Duane-radial ray	SALL4	20q13	607323
Angelman / Prader-Willi	SNORD107/64/108	15q11	105830	Duchenne muscular dystrophy	DMD	Xp21.1-p21.2	310200
Angelman / Prader-Willi	SNORD109B	15q11	105830	Dyggve-Melchior-Clausen disease	DYM	18q21	223800
Angelman / Prader-Willi	SnoRNA	15q11	105830	Dystonia-11	PEG10	7p21	159900
Angelman / Prader-Willi	SNRPN	15q11	105830	Dystonia-11	PEG10 IC	7p21	159900
Angelman / Prader-Willi	SNRPN	15q11	105830	Ectodermal dysplasia	SCGE	7p21	159900
Angelman / Prader-Willi	SNRPN	15q11	105830	Ehlers-Danlos	EDA	Xq13	305100
Angelman / Prader-Willi	SNURF	15q11	105830	Exudative vitreoretinopathy	COL1A2	7q21	225320
Angelman / Prader-Willi	UBE3A	15q11	105830	Fabry disease	FZD4	11q14	133780
Aniridia	PAX6	11p13-p14	106210	Fanconi anemia	GLA	Xq22	301500
Anxiety-related personality traits	SLC6A4	17p11	607834	Fanconi anemia	FANCA	16q24	227650
Atrial fibrillation	GJA5	1q21	121013	Fanconi anemia	FANCB	Xp22	300515
Atrial septal defect	NKX2-5	5q35	108900	Feingold	MYCN	2q24	164280
Autism	A2BP1	16p13	209850	FG syndrome 4	CASK	Xp11	300172
Autism	ATP10A	15q12	209850	Focal dermal hypoplasia	PORCN	Xp11	300651
Autism	CADPS2	7q31	209850	Forebrain defects	TDGF1	3p21	187395
Autism	CNTN4	3p25	209850	Fragile X	FMR1	Xq27	300624
Autism	CNTNAP2	7q35-36	209850	Fragile X	FMR2	Xq28	300624
Autism	DLGAP2	8p23	209850	Fryns 1q41	DISP1	1q41	229850
Autism	EGR2	10q21	209850	Gardner	APC	5q22	175100
Autism	EN2	7q36	209850	Gitelman	SLC12A3	16q13	263800
Autism	GABRB3	15q12	209850	Glycerol kinase deficiency	GK	Xp21	307030
Autism	MET	7q31	209850	Golabi-Ito-Hall	PQBP1	Xp11	300463
Autism	NLGN3	Xq13	209850	Gonadal Dysgenesis	SRY	Yp11.31	480000
Autism	NLGN4X	Xp22	209850	Greig cephalopolysyndactyly	GLI3	7p14	175700
Autism	SHANK3	22q13	209850	Hemophilia A	F8	Xq28	306700
Autism	SLC4A10	2q24	209850	Hemophilia B	F9	Xq27	306900
Autism	DISC1	1q42	209850	Hepatocellular carcinoma	IGF2R	6q25	114550
Autism	NRXN1	2p16	209850	Hereditary hemorrhagic telangiectasia	MADH4	18q21	175050
Autism	NPTX2	7q22	600750	Hereditary pancreatitis	PRSS1	7q34	276000
Barter 1	SLC12A1	15q21	601678	Heterotopia, periventricular, X-linked dominant	FLNA	Xq28	300049
Barter 2	KCNJ1	11q24	241200	Hirschsprung disease	EDNRB	13q22	600155
Barter 3	CLCNKB	1p36	607364	Hirschsprung disease plus	RET	10q11	142623
Barter 4A	BSND	1p32	602522	Holoprosencephaly 1	LSS	21q22	600909
Barter 4B	CLCNKA	1p36	613090	Holoprosencephaly 1	TMEM1	21q22	602103
Barter with autosomal dominant hypocalcemia	CASR	3q13.33-q21.1	601199	Holoprosencephaly 2	SIX3	2p21	157170
Beckwith-Wiedemann	H19 IC	11p15	130650	Holoprosencephaly 3	SHH	7q36	142945
Beckwith-Wiedemann	KCNQ1 IC	11p15	130650	Holoprosencephaly 4	TGIF1	18p31	142946
Beckwith-Wiedemann	CDKN1C	11p15	130650	Holoprosencephaly 5	ZIC2	13q32	609637
Beckwith-Wiedemann	H19	11p15	130650	Holoprosencephaly 7	PTCH1	9q22	610828
Beckwith-Wiedemann	IGF2	11p15	130650	Holoprosencephaly 9	GLI2	2q14	610829
Beckwith-Wiedemann	INS	11p15	130650	Holoprosencephaly and preaxial polydactyly	FBXW11	5q35	264480
Beckwith-Wiedemann	PHLDA2	11p15	130650	Holt-Oram	TBX5	12q14	142900
Beckwith-Wiedemann	SLC22A18	11p15	130650	HSAS, MASA, CRASH	L1CAM	Xq28	303350
Bilateral frontoparietal polymicrogyria	GPR56	16q13	606854	Hunter, mucopolysaccharidosis type 2	IDS	Xq28	309900
Birk-Barel	KCNK9	8q24	605874	Hyperekplexia and Epilepsy	ARHGEF9	Xq11	300607
Blepharophimosis	FOXL2	3q22	110100	Hypomyelination, global, cerebral	SLC25A12	2q31	612949
Borjeson-Forsman-Lehmann	PHF6	Xq26	301900	Hypophosphatemic rickets	PHEX	Xp22	307800
Brachydactyly type C	GDF5	20q11	113100	Ichthyosis, X-linked	STS	Xp22.31	308100
Branchiootorenal	EYA1	8q13	113650	Ichthyosis, X-linked	VX3A	Xp22	308100
Breast cancer	BRCA2	13q13	114480	IGF-1 resistance	IGF1R	15q25	147370
Brunner	MAOA	Xp11	300615	Incontinentia pigmenti	IKBK	Xq28	308300
Campomelic dysplasia	SOX9	17q24	114290	Infertility and deafness	KIAA0377 (HISPPD2A)	15q15	611102
Carnitine biosynthesis pathway	PAR2	Xq28	300777	Joubert 3	AHI1	6q23	608894
Cataract, juvenile, with microcornea A and glucosuria	SLC16A12	10q23	612018	Joubert 5	CEP290	12q21	610188
Cat-Eye Syndrome (Type I)	ATP6V1E1	22q11	115470	Kallmann 1	KAL1	Xp22.31	308700
Cerebral amyloid angiopathy	APP	21q21	605714	Kallmann 2	FGFR1	8p12	147950
Charcot-Marie-Tooth disease, X-linked	PRPS1	Xq22	311070	Kenny-Caffey	TBCE	1q42	244460
Charcot-Marie-Tooth disease, type 1A	PMP22	17p11	118220	Langer-Giedion	EXT1	8q24.11	150230
CHARGE	CHD7	8q12	214800	Leri-Weill Dyschondrosteosis	PAR1	Xp22	127300

Associated Condition	Gene	Chr/Band	OMIM #	Associated Condition	Gene	Chr/Band	OMIM #
Lesch-Nyhan	HPRT1	Xq26.2	300322	Stickler 1	COL2A1	12q13.11	108300
Li-Fraumeni 1	TP53	17p13.1	151623	Stickler 2	COL11A1	1p21	604841
Lissencephaly with cerebellar hypoplasia	RELN	7q22.1	257320	Synpolydactyly	EVX2	2q31	186000
Lissencephaly, X-linked	DCX	Xq22.3-q23	300067	Synpolydactyly	HOXD13	2q31.1	186000
Loeys-Dietz, type 1A	TGFBR1	9q22	609192	Synpolydactyly	HOXD9	2q31	186000
Loeys-Dietz, type 2B	TGFBR2	3p24	610380	Thrombasthenia of Glanzmann and Naegeli	ITGB3	17q21	273800
Long QT	KCNQ1	11p15	192500	Thrombocytopenia absent radius	GJA8	1q21.1	274000
Lowe	OCRL	Xq25	309000	Timothy	CACNA1C	12p13	601005
Macrocephaly/Autism	PTEN	10q23	605309	Townes-Brocks	SALL1	16q12.1	107480
Major affective disorder	IMPA1	8q21	125480	Treacher Collins mandibulofacial dysostosis	TCOF1	5q33	154500
Marfan 1	FBN1	15q21.1	154700	Trichorhinophalangeal	TRPS1	8q23.3	150230
Meningioma	NF2	22q12	607174	Tuberous sclerosis 1	TSC1	9q34.13	191100
Menkes disease	ATP7A	Xq21.1	309400	Tuberous sclerosis 2	TSC2	16p13.3	613254
Mental Retardation	DMXL2	15q21	612186	Ulnar-mammary	TBX3	12q24.21	181450
Mental Retardation, Autosomal Dominant 5	SYNGAP1	6p21.3	612621	Usher	ABCC8	11p15	276904
Mental Retardation, Autosomal Recessive 6	GRIK2	6q16	611092	Usher	USH1C	11p15	276904
Metachromatic leukodystrophy	ARSA	22q13	250100	van der Woude	IRF6	1q32.2	119300
Microphthalmia, syndromic 2	BCOR	Xp11	300166	Vascular endothelial growth factor disorders	VEGFA	6p21	192240
Microphthalmia, syndromic 7	HCCS	Xp22	300056	Verbal dyspraxia	FOXP2	7q31.1	602081
Microphthalmia	SOX2	3q26.3-q27	206900	Visceral heterotaxy	CFC1	2q21	605376
Migraine, familial hemiplegic, 3	SCN1A	2q24.3	609634	Visceral heterotaxy	CFC1	2q21	605376
Miller-Dieker	ABR	17p13.3	247200	von Hippel-Lindau	VHL	3p25.3	193300
Miller-Dieker	PAFAH1B1	17p13.3	247200	Waardenburg I	PAX3	2q36.1	193500
Miller-Dieker	PITPNA	17p13.3	247200	Waardenburg IIA	MITF	3p14.1	193510
Miller-Dieker	YWHAE	17p13.3	247200	WAGR	WT1	11p13	194072
Mitochondrial Complex I Deficiency	NDUFA1	Xq24	252010	Williams-Beuren	ELN	7q11.23	194050
Mowat-Wilson	ZEB2	2q22	235730	Williams-Beuren	GTF2I	7q11.23	194050
Myotubular myopathy	MTM1	Xq28	310400	Williams-Beuren	GTF2IRD1	7q11.23	194050
Nail-patella	LMX1B	9q33.3	612100	Williams-Beuren	LMMK1	7q11.23	194050
Nance-Horan	NHS	Xp22	302350	Williams-Beuren	NCF1	7q11.23	194050
Nephroblastoma 1	CLCN5	Xp11	310468	Wolf-Hirschhorn	LETM1	4p16.3	194190
Nephronophthisis 1	NPHP1	2q13	256100	Wolf-Hirschhorn	WHSC1	4p16.3	194190
Neural development	NNAT	20q11	603106	Wolf-Hirschhorn	WHSC2	4p16.3	194190
Neurite formation	PPP1R9A	7q21	602468	X-inactivation, familial skewed	XIST	Xq13	314670
Neurofibromatosis Type 1	JAZ1	17q11.2	162200	X-linked dyskeratosis congenita	DKC1	Xq28	305000
Neurofibromatosis Type 1	NF1	17q11.2	162200	X-linked heterotaxy	ZIC3	Xq26.3	306955
NFIA haploinsufficiency	NFIA	1p31.3	600727	X-linked juvenile retinoschisis	RS1	Xp22	312700
Noonan 1	PTPN11	12q24	163950	X-linked lymphoproliferative type 1	SH2D1A	Xq25	308240
Noonan 4	SOS1	2p22.1	610733	X-linked lymphoproliferative type 2	BIRC4	Xq25	308240
Noonan 5	RAF1	3p25	611553	X-linked mental retardation	ACSL4	Xp22	300387
Norrie disease	NDP	Xp11	310600	X-linked mental retardation	AGTR2	Xq23	300034
Obesity, severe	SIM1	6q16	603128	X-linked mental retardation	AP1S2	Xp22	300630
Oculocutaneous albinism type 2	OCA2	15q13	203200	X-linked mental retardation	ARHGFE6	Xq26	300436
Oligodontia	PAX9	14q13	604625	X-linked mental retardation	ARX	Xp21	300419
Opitz G	MID1	Xq22	300000	X-linked mental retardation	ATP6AP2	Xp11	300423
Opitz G	MID2	Xq22	300204	X-linked mental retardation	BRWD3	Xp21	300659
Opitz-Kaveggia	MED12	Xq13.1	305450	X-linked mental retardation	CUL4B	Xp24	300354
Optic atrophy	TP73L	3q28	165500	X-linked mental retardation	DLG3	Xq13	300189
Ornithine transcarbamylase deficiency	OTC	Xp11.4	311250	X-linked mental retardation	FTSJ1	Xp11	309549
Orofaciodigital 1	OFD1	Xp22.2	311200	X-linked mental retardation	GD1I	Xq28	309541
Osteogenesis imperfecta	COL1A1	17q21	120150	X-linked mental retardation	GRIA3	Xq24	300699
Osteopetrosis	LEMD3	12q14.3	166705	X-linked mental retardation	HADH2	Xp11	300220
Pelizaeus-Merzbacher disease	PLP1	Xq22.2	312080	X-linked mental retardation	IL1RAPL1	Xp21	300143
Phosphoglycerate kinase deficiency	PGK1	Xp21	300653	X-linked mental retardation	JARID1C	Xp11	300534
Pitt-Hopkins	TCF4	18q21.2	610954	X-linked mental retardation	KIAA2022	Xq13	300524
Polycystic kidney disease 1	PKD1	16p13	601313	X-linked mental retardation	OPHN1	Xq12	300486
Polycystic kidney disease 2	PKD2	4q22.1	613095	X-linked mental retardation	PAK3	Xp22	300558
Polydactyly preaxial 2	LMBR1	7q36	174500	X-linked mental retardation	PHF8	Xp11	300263
Potocki-Shaffer	ALX4	11p11.2	601224	X-linked mental retardation	SLC38A5	Xp11	309549
Potocki-Shaffer	EXT2	11p11.2	601224	X-linked mental retardation	SMS	Xp22	309583
Pseudohypoparathyroidism Ia	GNAS	20q13	103580	X-linked mental retardation	SOX3	Xq27	300123
Pseudohypoparathyroidism Ia	GNAS IC	20q13	103580	X-linked mental retardation	SYN1	Xp11	300491
Pseudovaginal perineoscrotal hypospadias	SRD5A2	2p23	264600	X-linked mental retardation	TM4SF2	Xp11	300210
Pulmonary venoocclusive disease	BMPR2	2q32	265450	X-linked mental retardation	UBE2A	Xq23	312180
Pyruvate dehydrogenase deficiency	PDHA1	Xp22	312170	X-linked mental retardation	UPF3B	Xq24	300676
Renal cysts and diabetes	TCF2	17q12	137920	X-linked mental retardation	VCX3A	Xp22	300533
Retinoblastoma	RB1	13q14.2	180200	X-linked mental retardation	ZDHHC15	Xq13	300577
Rett syndrome, congenital variant	FOXG1B	14q12	613454	X-linked mental retardation	ZDHC9	Xq25	300799
Rett	MECP2	Xq28	312750	X-linked mental retardation	ZNF261	Xq13	300061
Rieger	PITX2	4q25	180500	X-linked mental retardation	ZNF41	Xp11	314995
Rolandic epilepsy, mental retardation, and speech dyspraxia	SRPX2	Xq22	300643	X-linked mental retardation	ZNF81	Xp11	300498
Rubinstein-Taybi	CREBBP	16p13	610543	X-linked mental retardation, Allan-Herndon-Dudley	SLC16A2	Xq13	300523
Rubinstein-Taybi	DNASE1	16p13	610543	X-linked mental retardation, Christianson type	SLC9A6	Xq26	300243
Russell-Silver	CPA4	7q32	607635	X-linked mental retardation, Stocco dos Santos	SHROOM4	Xp11	300434
Russell-Silver	GRB10	7p12	180860	X-linked mental retardation, Turner	HUWE1	Xp11	300706
Schizophrenia 11	NRG3	10q22	608078	X-linked spasms	CDKL5	Xp22.13	300672
Sclerosteosis	SOST	17q21.31	269500	Xp11.3 Deletion	RP2	Xp11	300578
Scoliosis	CHD2	15q26	602119	Xp11.3 Deletion	ZNF674	Xp11	300578
Short stature	LHX4	1q25.2	262700	Xq28 Microdeletion	ABCD1	Xq28	300475
SHOX	SHOX	Xp22.33	312865	Xq28 Microduplication	RPL10	Xq28	300815
Simpson-Golabi-Behmel, type 1	GPC3	Xq26	312870	XY sex reversal	NR5A1	9q33.3	184757
Small size	PEG3	19q13	601483	Areas of clinical interest	ELK1	Xp11	311040
Small size	PEG3 IC	19q13			KLF8	Xp11	300286
Small size	ZIM2	19q13	601483		NXF5	Xp11	300319
Smith-Lemli-Opitz	DHCR7	11q13.4	270400		KLF14	7q32	609393
Smith-Magenis	RAI1	17p11.2	182290		MEST	7q32	601029
Sotos	NSD1	5q35.3	117550		OSBPL5	11p15	606733
Split-hand / foot malformation 1 with deafness	DLX5	7q21.3	220600		DLK1&MEG3 IC	14q32	608149
Split-hand / foot malformation 1 with deafness	SHFM1	7q21.3	220600		DLK1	14q32	176290
Split-hand / foot malformation 3	BTRC	10q24.32	600095		MEG3	14q32	605636
Split-hand / foot malformation 3	FBXW4	10q24.32	600095		ZNF264	19q13	604668
Split-hand / foot malformation 3	LBX1	10q24.32	600095		L3MBTL	20q13	608802
Split-hand / foot malformation 3	POLL	10q24.32	600095	Unique subtelomeric regions	Multiple	43 sites	
Split-hand / foot malformation 5	DLX1	2q31	606708	Unique pericentromeric regions	Multiple	43 sites	
Split-hand / foot malformation 5	DLX2	2q31	606708	Aneuploidy	Multiple	Any individual chromosome	

NOTE: The 180k ISCA oligonucleotide array offered by the Genetics Center is a diagnostic screening tool designed to evaluate the entire genome for gains or losses of genomic material. As of October 2010, 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.