

Prenatal Targeted Oligo Array CGH Disease Regions Screened

Associated Condition	Gene	Chr/Band	OMIM #	Associated Condition	Gene	Chr/Band	OMIM #
1p31 Deletion	DIRAS3	1p31	605193	Bartter 2	KCNJ1	11q24	241200
1p36 Microdeletion	SKI	1p36	607872	Bartter 3	CLCNKB	1p36	607364
1p36 Microdeletion	TP73	1p36	607872	Bartter 4A	BSND	1p32	602522
2p15-16.1 Microdeletion	Multiple	2p15-16.1	612513	Bartter 4B	CLCNKA	1p36	613090
2q37.3 Deletion	HDAC4	2q37	600430	Bartter with autosomal dominant hypocalcemia	CASR	3q13.33-q21.1	601199
3q29 Microdeletion	PAK2	3q29	609425	Beckwith-Wiedemann	H19 IC	11p15	130650
6p24 Microdeletion	FKHL7	6p24	612852	Beckwith-Wiedemann	KCNQ1 IC	11p15	130650
8p23.1 Deletion	CTSB	8p23	116810	Beckwith-Wiedemann	CDKN1C	11p15	130650
9p Deletion	DMRT1	9p24	158170	Beckwith-Wiedemann	H19	11p15	130650
9p Deletion	DMRT2	9p24	158170	Beckwith-Wiedemann	IGF2	11p15	130650
9q34.3 Microdeletion	EHMT1	9q34.3	610253	Beckwith-Wiedemann	INS	11p15	130650
10q22-23 Deletion	GRID1	10q22	610659	Beckwith-Wiedemann	PHLDA2	11p15	130650
12q14.1-q15 Deletion	GRIP1	12q14	604597	Beckwith-Wiedemann	SLC22A18	11p15	130650
13q Deletion	GPC5	13q31	602446	Beckwith-Wiedemann	GPR56	16q13	606854
13q Deletion	GPC6	13q31	604404	Bilateral frontoparietal polymicrogyria	FOXL2	3q22	110100
13q Deletion	PCDH9	13q21	603581	Blepharophimosis	SOX9	17q24	114290
14q11-q22 Deletion	CHD8	14q11	613457	Branchiootorenal	EYA1	8q13	113650
14q11-q22 Deletion	SUPT16H	14q11	613457	Campomelic dysplasia	SOX9	17q24	114290
14q22 Microdeletion, Branchiootic syndrome 3	SIX1	14q22	608389	Cat-Eye Syndrome (Type I)	ATP6V1E1	22q11	115470
14q22 Microdeletion, Branchiootic syndrome 3	SIX4	14q22	608389	CHARGE	CHD7	8q12	214800
14q22 Microdeletion, Microphthalmia, isolated, with cataract 2	SIX6	14q22	212550	Choroeremia	CHM	Xp21	303100
14q22 Microdeletion, Microphthalmia, syndromic 5	OTX2	14q22	610125	Chronic granulomatous disease	CYBB	Xp11	306400
14q22 Microdeletion, Orofacial Cleft 11	BMP4	14q22	600625	Cleidocranial dysplasia	RUNX2	6p21	119600
15q13.3 Microdeletion	CHRNA7	15q13	612001	Congenital diaphragmatic hernia	NR2F2	15q26	142340
17q21.31 Microdeletion	CRHR1	17q21.31	610443	Cornelia de Lange 1	NIPBL	5p13	122470
17q21.31 Microduplication	MAPT	17q21.31	613533	Cornelia de Lange 2	SMC1L1	Xp11	300590
18q Microdeletion	TCEB3B	18q21	609522	Cowden	BMPR1A	10q23	158350
18q Deletion	ZNF407	18q22	301808	Cri-du-Chat	TERT	5p15	123450
18q Deletion	GALR1	18q23	600377	Dandy-Walker	ZIC1	3q24	220200
22q11.2 Deletion	BCR	22q11	611867	Dandy-Walker	ZIC4	3q24	220200
1q44 Microdeletion	AKT3			DiGeorge 1	CRKL	22q11	188400
Adrenal hypoplasia congenital	NR0B1	Xp21	300200	DiGeorge 1	HIRA	22q11	188400
Agammaglobulinemia, X-linked	BTK	Xq22.1	300755	DiGeorge 1	TBX1	22q11	188400
Alagille	JAG1	20p12	118450	DiGeorge 2	GATA3	10p14	601362
Alpha-thalassemia mental retardation	HBA1 & HBA2	16p13	141750	DiGeorge 2	NEBL	10p12	601362
Alpha-thalassemia mental retardation	SOX8	16p13.3	141750	Down syndrome critical region	DSCR1	21q22	190685
Alpha-thalassemia mental retardation, X-linked	ATRX	Xq21	300032	Down syndrome critical region	DSCR3	21q22	190685
Alport, X-linked	COL4A5	Xq22.3	301050	Down syndrome critical region	DYRK1A	21q22	190685
Androgen insensitivity	AR	Xq12	300068	Down syndrome critical region	RUNX1	21q22	190685
Angelman / Prader-Willi	MAGEL2	15q11	105830	Duchenne muscular dystrophy	DMD	Xp21.1-p21.2	310200
Angelman / Prader-Willi	MKRN3	15q11	105830	Feingold	MYCN	2q24	164280
Angelman / Prader-Willi	NDN	15q11	105830	Focal dermal hypoplasia	PORCN	Xp11	300651
Angelman / Prader-Willi	NIPA1	15q11	105830	FMR1 microdeletion syndrome	FMR1	Xq27	300624
Angelman / Prader-Willi	NIPA2	15q11	105830	FMR1 microdeletion syndrome	FMR2	Xq28	300624
Angelman / Prader-Willi	PWS IC	15q11	105830	Fryns 1q41	DISP1	1q41	229850
Angelman / Prader-Willi	SNORD10	15q11	105830	Gitelman	SLC12A3	16q13	263800
Angelman / Prader-Willi	SNORD10 7/64/108			Glycerol kinase deficiency	GK	Xp21	307030
Angelman / Prader-Willi	SNORD10 9B	15q11	105830	Gonadal Dysgenesis	SRY	Yp11.31	480000
Angelman / Prader-Willi	SnoRNA	15q11	105830	Greig cephalopolysyndactyly	GLI3	7p14	175700
Angelman / Prader-Willi	SNRPN	15q11	105830	Hemophilia A	F8	Xq28	306700
Angelman / Prader-Willi	SNRPN	15q11	105830	Hemophilia B	F9	Xq27	306900
Angelman / Prader-Willi	SNURF	15q11	105830	Hereditary hemorrhagic telangiectasia	MADH4	18q21	175050
Angelman / Prader-Willi	UBE3A	15q11	105830	Holoprosencephaly 1	LSS	21q22	600909
Aniridia	PAX6	11p13-p14	106210	Holoprosencephaly 1	TMEM1	21q22	602103
Atrial septal defect	NKX2-5	5q35	108900	Holoprosencephaly 2	SIX3	2p21	157170
Bartter 1	SLC12A1	15q21	601678	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 polydactyly	FBXW11	5q35	264480

Associated Condition	Gene	Chr/Band	OMIM #	Associated Condition	Gene	Chr/Band	OMIM #
Holt-Oram	TBX5	12q14	142900	Split-hand / foot malformation 3	BTRC	10q24.32	600095
Ichthyosis, X-linked	STS	Xp22.31	308100	Split-hand / foot malformation 3	FBXW4	10q24.32	600095
Ichthyosis, X-linked	VCX3A	Xp22	308100	Split-hand / foot malformation 3	LBX1	10q24.32	600095
Joubert 3	AHI1	6q23	608894	Split-hand / foot malformation 3	POLL	10q24.32	600095
Joubert 5	CEP290	12q21	610188	Split-hand / foot malformation 5	DLX1	2q31	606708
Kallmann 1	KAL1	Xp22.31	308700	Split-hand / foot malformation 5	DLX2	2q31	606708
Langer-Giedion	EXT1	8q24.11	150230	Stickler 1	COL2A1	12q13.11	108300
Leri-Weill Dyschondrosteosis	PAR1	Xp22	127300	Synpolydactyly	EVX2	2q31	186000
Li-Fraumeni 1	TP53	17p13.1	151623	Synpolydactyly	HOXD13	2q31.1	186000
Lissencephaly with cerebellar hypoplasia	RELN	7q22.1	257320	Synpolydactyly	HOXD9	2q31	186000
Lissencephaly, X-linked	DCX	Xq22.3-q23	300067	Townes-Brocks	SALL1	16q12.1	107480
Loeys-Dietz, type 1A	TGFBR1	9q22	609192	Trichorhinophalangeal	TRPS1	8q23.3	150230
Loeys-Dietz, type 2B	TGFBR2	3p24	610380	Tuberous sclerosis 1	TSC1	9q34.13	191100
Lowe	OCRL	Xq25	309000	Tuberous sclerosis 2	TSC2	16p13.3	613254
Macrocephaly/Autism	PTEN	10q23	605309	Ulnar-mammary	TBX3	12q24.21	181450
Marfan 1	FBN1	15q21.1	154700	van der Woude	IRF6	1q32.2	119300
Menkes disease	ATP7A	Xq21.1	309400	Vascular endothelial growth factor disorders	VEGFA	6p21	192240
Microphthalmia	SOX2	3q26.3-q27	206900	von Hippel-Lindau	VHL	3p25.3	193300
Miller-Dieker	ABR	17p13.3	247200	Waardenburg I	PAX3	2q36.1	193500
Miller-Dieker	PFAH1B1	17p13.3	247200	Waardenburg IIA	MITF	3p14.1	193510
Miller-Dieker	PITPNA	17p13.3	247200	WAGR	WT1	11p13	194072
Miller-Dieker	YWHAE	17p13.3	247200	Williams-Beuren	ELN	7q11.23	194050
Mowat-Wilson	ZEB2	2q22	235730	Williams-Beuren	GTF2I	7q11.23	194050
Nail-patella	LMX1B	9q33.3	161200	Williams-Beuren	GTF2IRD1	7q11.23	194050
Nephronophthisis 1	NPHP1	2q13	256100	Williams-Beuren	LIMK1	7q11.23	194050
Neurofibromatosis Type 1	JJAZ1	17q11.2	162200	Williams-Beuren	NCF1	7q11.23	194050
Neurofibromatosis Type 1	NF1	17q11.2	162200	Wolf-Hirschhorn	LETM1	4p16.3	194190
Noonan 1	PTPN11	12q24	163950	Wolf-Hirschhorn	WHSC1	4p16.3	194190
Noonan 4	SOS1	2p22.1	610733	Wolf-Hirschhorn	WHSC2	4p16.3	194190
Noonan 5	RAF1	3p25	611553	X-linked heterotaxy	ZIC3	Xq26.3	306955
Norrie disease	NDP	Xp11	310600	X-linked juvenile retinoschisis	RS1	Xp22	312700
Opitz G	MID1	Xq22	300000	X-linked lymphoproliferative type 1	SH2D1A	Xq25	308240
Ornithine transcarbamylase deficiency	OTC	Xp11.4	311250	X-linked lymphoproliferative type 2	BIRC4	Xq25	308240
Orofacioidigital 1	OFD1	Xp22.2	311200	X-linked mental retardation	ARX	Xp21	300419
Pelizaeus-Merzbacher disease	PLP1	Xq22.2	312080	X-linked mental retardation	IL1RAPL1	Xp21	300143
Pitt-Hopkins	TCF4	18q21.2	610954	X-linked mental retardation	PAK3	Xp22	300558
Polycystic kidney disease 1	PKD1	16p13	601313	X-linked mental retardation	SOX3	Xq27	300123
Polycystic kidney disease 2	PKD2	4q22.1	613095	X-linked spasms	CDKL5	Xp22.13	300672
Potocki-Shaffer	ALX4	11p11.2	601224	Xp11.3 Deletion	RP2	Xp11	300578
Potocki-Shaffer	EXT2	11p11.2	601224	Xp11.3 Deletion	ZNF674	Xp11	300578
Renal cysts and diabetes	TCF2	17q12	137920	XY sex reversal	NR5A1	9q33.3	184757
Retinoblastoma	RB1	13q14.2	180200	Unique subtelomeric regions	Multiple	43 sites	
Rieger	PITX2	4q25	180500	Unique pericentromeric regions	Multiple	43 sites	
Rubinstein-Taybi	CREBBP	16p13	610543	Aneuploidy	Multiple	Any individual chromosome	
Rubinstein-Taybi	DNASE1	16p13	610543				
Short stature	LHX4	1q25.2	262700				
SHOX	SHOX	Xp22.33	312865				
Simpson-Golabi-Behmel, type 1	GPC3	Xq26	312870				
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				

NOTE: The Prenatal Targeted oligonucleotide array offered by the Genetics Center is a diagnostic screening tool designed to evaluate targeted disease regions of the genome for gains or losses of genomic material. As of March 2011, the above listed genetic conditions have been associated with the indicated regions being evaluated by the Prenatal Targeted array. Copy number losses of more than 2 Mb throughout the genome will also be reported. 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 Prenatal Targeted array may detect copy number variations of uncertain clinical significance, or changes associated with cancer predisposing disorders that may manifest later in life and be unrelated to prenatal ultrasound findings. Also, imbalances in highly repetitive regions (pericentromeric and telomeric regions) may not be detected. For certain conditions, additional testing may be warranted. Please contact the Genetics Center to discuss patient samples if there are any questions.