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Prenatal Targeted Oligo + SNP 60k Array CGH
Disease Regions Screened

Associated Condition	Gene	Chr/Band	OMIM #
1p32-31 Deletion	DIRAS3	1p32-p31	613735
1p36 Deletion	Multiple	1p36	607872
1q21.1 Deletion (1.35 Mb)	Multiple	1q21.1	612474
1q21.1 Deletion (200 Kb)	Multiple	1q21.1	274000
1q41-42 Deletion	Multiple	1q41-q42	612530
1q43-44 Deletion	AKT3	1q43-q44	612337
2p16.1-p15 Deletion	Multiple	2p16.1-15	612513
2p16.3 Deletion	NRXN1	2p16.3	600565
2p21 Deletion	Multiple	2p21	606407
2q23.1 Deletion	EPC2, MBD5	2q23.1	611000, 611472
2q32.2–q33 Deletion	SATB2	2q32.2q33	119540
2q37.3 Deletion	HDAC4	2q37.3	600430
3q29 Deletion	PAK2	3q29	609425
3q29 Duplication	Multiple	3q29	611936
5p13.2 Duplication	NIPBL	5p13.2	613174
5q35.2–q35.3 Duplication	NSD1	5q35.2–q35.3	606681
6pter-p24 Deletion	FOXC1	6p24	612582
6q24.3 Deletion	Multiple	6q24.3	612863
6q25.2–q25.3 Deletion	Multiple	6q25.2-q25.3	612863
7q11.23 Duplication	Multiple	7q11.23	609757
8p23.1 Deletion	CTSB	8p23.1	116810
8p23.1 Deletion / Congenital diaphragmatic hernia 2 (CDH2)	Multiple	8p23.1	222400
8p23.1 Duplication	Multiple	8p23.1	
8q12 Duplication	CHD7	8q12.1-q12.2	608892
9p Deletion	Multiple	9p	158170
9q22.32–q22.33 Deletion	GABBR2, TGFBR1	9q22.32–q22.33	607340, 190181
9q34.3 Deletion / Kleefstra	EHMT1	9q34.3	610253
10q22-23 Deletion	Multiple	10q22-q23	
12q14.1-q15 Deletion	GRIP1, HMGA2, LEMD3	12q14.1-q15	604597, 600698, 607844
12q14.3-Related primordial dwarfism / Russell-Silver-like	HMGA2	12q14.3	600698
12q24.21–q24.23 Duplication	Multiple	12q24.21–q24.23	
13q14 Deletion	Multiple	13q14	613884
14q11-q22 Deletion	Multiple	14q11-q22	613457
14q12 Duplication	FOXG1	14q12	164874
14q22–q23 Deletion	Multiple	14q22-q23	
14q32.2 Deletion causing UPD(14) maternal phenotype	DLK1, RTL1	14q32.2	176290, 611896
14q32.2 Deletion causing UPD(14) paternal phenotype	MEG3, MEG8, RTL1	14q32.2	605636, 613648, 611896
15q11–q13 Duplication	Multiple	15q11-q13	608636
15q13.3 Deletion	Multiple	15q13.3	612001
15q24 Deletion	Multiple	15q24	613406
15q24.1–q24.2 Duplication	Multiple	15q24.1–q24.2	
16p11.2 Deletion	Multiple	16p11.2	611913
16p12.1 Deletion	Multiple	16p12.1	136570
16p12.2-p11.2 Deletion	Multiple	16p12.2-p11.2	613604
16p13.11 Deletion	Multiple	16p13.11	
16p13.3 Deletion	Multiple	16p13.3	610543
16p13.3 Duplication	CREBBP	16p13.3	613458

Associated Condition	Gene	Chr/Band	OMIM #
16q11.2–q12.2 Deletion	Multiple	16q11.2–q12.2	
17p13.1 Deletion	Multiple	17p13.1	613776
17p13.3 Distal deletion	Multiple	17p13.3	
17p13.3 Distal duplication, not including PAFAH1B1	Multiple	17p13.3	613215
17p13.3 Proximal duplication, including PAFAH1B1	Multiple	17p13.3	613215
17q11.2 Deletion	NF1	17q11.2	613675
17q12 Deletion	Multiple	17q12	614527
17q12 Duplication	Multiple	17q12	614526
17q21.31 Deletion	Multiple	17q21.31	610443
17q21.31 Duplication	Multiple	17q21.31	613533
17q23.1–q23.2 Deletion	Multiple	17q23.1-q23.2	613355
18q Deletion	Multiple	18q	601808
18q21.1 Deletion	TCEB3B	18q21.1	609522
19q13.11 Deletion	Multiple	19q13.11	613026
22q11.2 Distal deletion	Multiple	22q11.2	611867
22q11.2 Duplication	Multiple	22q11.2	608363
22q13.3 Deletion	Multiple	22q13.3	606232
Xp11.23–p11.22 Duplication	Multiple	Xp11.23-p11.22	300801
Xp11.3 Deletion	Multiple	Xp11.3	300578
Xp21.2-p11.4 Deletion	Multiple	Xp21.2-p11.4	
Xq28 Duplication	Multiple	Xq28	300815
Aarskog-Scott syndrome	FGD1	Xp11.22	305400
Adrenal hypoplasia congenital	NR0B1	Xp21.2	300200
Agammaglobulinemia, X-linked	BTK	Xq22.1	300755
Alagille	JAG1	20p12.2	118450
Albright hereditary osteodystrophy-like syndrome/2q37 deletion syndrome	HDAC4	2q37.3	600430
Alpha-thalassemia mental retardation	HBA1, HBA2, SOX8	16p13.3	141750
Alpha-thalassemia mental retardation, X-linked	ATRX	Xq21.1	300032
Alport plus diffuse leiomyomatosis, X-linked	COL4A6	Xq22.3	308940
Alport, X-linked	COL4A5	Xq22.3	301050
Androgen insensitivity	AR	Xq12	300068
Angelman / Prader-Willi	MAGEL2, MKRN3, NDN, NIPA1, NIPA2, PWS IC, SNORD 107/64/108, SNORD 109B, SNORD 116 cluster, SnoRNA, SNRPN, SNURF, UBE3A	15q11.2	105830, 176270, 105830
Aniridia	PAX6	11p13	106210
ANKRD11 haploinsufficiency	ANKRD11	16q24.3	611192
Atrial septal defect 7	NKX2-5	5q35.1	108900

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Associated Condition	Gene	Chr/Band	OMIM #
Autism, SHANK2-related	SHANK2	11q13.3-q13.4	613436
Autistic Features, X-linked, susceptibility	NLGN4	Xp22.32-p22.31	
AZF Regions	Multiple	Yq11.21	415000
Bannayan-Riley-Ruvalcaba	PTEN	10q23.31	153480
Bartter 1	SLC12A1	15q21.1	601678
Bartter 2	KCNJ1	11q24.3	241200
Bartter 3	CLCNKB	1p36.13	607364
Bartter 4A	BSND	1p32.3	602522
Bartter 4B	CLCNKA	1p36.13	613090
Bartter with autosomal dominant hypocalcemia	CASR	3q21.1	601199
Basal cell nevus	PTCH1	9q22.32	109400
Beckwith-Wiedemann	CDKN1C, H19, H19 IC, IGF2, INS, KCNQ1 IC, PHLDA2, SLC22A18	11p15.4, 11p15.5	130650
Benign familial neonatal seizures	KCNQ2	20q13.33	121200
Beta thalassemia	HBB	11p15.4	613985
Bilateral frontoparietal polymicrogyria	GPR56	16q21	606854
Blepharophimosis	FOXL2	3q22.3	110100
Branchiootic syndrome 3	SIX1	14q23.1	608389
Branchiootorenal	EYA1	8q13.3	113650
Campomelic dysplasia	SOX9	17q24.3	114290
Cat-eye syndrome	ATP6V1E1	22q11	115470
Cerebellar ataxia, mental retardation, and dysequilibrium	VLDLR	9p24.2	224050
Cerebral cavernous malformations 1	KRIT1	7q21.2	116860
Cerebral cavernous malformations 2	CCM2	7p13	603284
Cerebral cavernous malformations 3	PDCD10	3q26.1	603285
Charcot Marie Tooth Type 1A	PMP22	17p12	118220
CHARGE	CHD7	8q12.1-q12.2	214800
Chondrodysplasia punctata 1, X-linked	ARSE	Xp22.33	302950
Choroideremia	CHM	Xq21.2	303100
Chronic granulomatous disease	CYBB	Xp11.4	306400
Cleidocranial dysplasia	RUNX2	6p21.1	119600
Cohen syndrome	VPS13B	8q22.2	216550
Congenital diaphragmatic hernia	Multiple	15q26.1	142340
Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD)	NSDHL	Xq28	308050
Cornelia de Lange 1	NIPBL	5p13.2	122470
Cornelia de Lange 2	SMC1A	Xp11.22	300590
Cortical dysplasia-focal epilepsy	CNTNAP2	7q35–q36	610042
Cowden disease	PTEN	10q23.31	158350
Craniofrontonasal	EFNB1	Xq13.1	304110
Craniosynostosis type 2	MSX2	5q35.2	604757
Cri-du-Chat	TERT	5p15.2	123450
Currarino	MNX1	7q36.3	176450
Dandy-Walker	Multiple	3q22-q24	220200
Deafness, autosomal recessive 22	OTOA	16p12.2	607039
DiGeorge 1	CRKL, HIRA, TBX1	22q11.21	188400
DiGeorge 2	Multiple	10p14-p13	601362
Down syndrome critical region	Multiple	21q22.3	190685
Dravet	SCN1A	2q24.3	607208

Associated Condition	Gene	Chr/Band	OMIM #
Duane radial ray	SALL4	20q13.2	607323
Duchenne muscular dystrophy	DMD	Xp21.2-p21.1	310200
Early-onset ataxia with oculomotor apraxia and hypoalbuminemia	APTX	9p21.1	208920
Emery-Dreifuss muscular dystrophy 1, X-linked (EDMD1)	EMD	Xq28	300384
EPHA7 haploinsufficiency	EPHA7	6q16.1	602190
Epileptic encephalopathy, early infantile	PCDH19	Xq22.1	300088
Familial adenomatous polyposis 1	APC	5q22.2	175100
Feingold	MYCN	2p24.3	164280
Focal dermal hypoplasia	PORCN	Xp11.23	305600
Fragile X	AFF2, FMR1	Xq28, Xq27.3	300624
Fryns	DISP1	1q41	229850
Gitelman	SLC12A3	16q13	263800
GLUT1 deficiency	SLC2A1	1p34.2	606777
Glycerol kinase deficiency	GK	Xp21.2	307030
Gonadal Dysgenesis	SRY	Yp11.31	480000
Greig cephalopolysyndactyly	GLI3	7p14.1	175700
Hemophilia A	F8	Xq28	306700
Hemophilia B	F9	Xq27.1	306900
Hereditary hemorrhagic telangiectasia	SMAD4	18q21.2	175050
Hereditary hemorrhagic telangiectasia 2	ACVRL1	12q13.13	600376
Heterotaxy, visceral 1	ZIC3	Xq26.3	306955
Hirschsprung disease, susceptibility to, 1	RET	10q11.21	142623
Holoprosencephaly 1	LSS, TRAPPC10	21q22.3	236100
Holoprosencephaly 2	SIX3	2p21	157170
Holoprosencephaly 3	SHH	7q36.3	142945
Holoprosencephaly 4	TGIF1	18p11.31	142946
Holoprosencephaly 5	ZIC2	13q32.3	609637
Holoprosencephaly 6	Multiple	2q37.1-q37.3	605934
Holoprosencephaly 7	PTCH1	9q22.32	610828
Holoprosencephaly 8	Multiple	14q13.1–q13.2	609408
Holoprosencephaly 9	GLI2	2q14.2	610829
Holoprosencephaly and preaxial polydactly	FBXW11	5q35.1	264480
Holt-Oram	TBX5	12q24.21	142900
Hydrocephalus and nephrogenic diabetes insipidus	AVPR2, L1CAM	Xq28	
Hyper-IgE recurrent infection	DOCK8	9p24.3	243700
Hypohidrotic ectodermal dysplasia (XHED)	EDA	Xq13.1	305100
Hypoparathyroidism, sensorineural deafness, renal disease	GATA3	10p14	146255
Hypospadius 2	MAMLD1	Xq28	300758
Hypotonia-cystinuria	PREPL, SLC3A1	2p21	606407
Ichthyosis, X-linked	STS, VCX3A	Xp22.31	308100
Idiopathic short stature (ISSX)	SHOX	Xp22.33, Yp11.32	300582
Incontinentia Pigmenti	IKBKG	Xq28	308300
Infantile hyperinsulinism with enteropathy and deafness	ABCC8, KCNJ11, USH1C	11p15-p14	606528
Infantile spasms, CDKL5-related / Atypical Rett	CDKL5	Xp22.13	300672
Infantile spasms, MAGI2-related	MAGI2	7q21.11	606382

(more associated syndromes on the next page)

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Prenatal Targeted Oligo + SNP 60k Array CGH
Disease Regions Screened (continued)

Associated Condition	Gene	Chr/Band	OMIM #
Intellectual disability with cerebellar hypoplasia and distinctive facial appearance	OPHN1	Xq12	300486
Intellectual disability with language impairment and autistic features	FOXP1	3p13	613670
Intellectual disability with microcephaly with pontine and cerebellar hypoplasia	CASK	Xp11.4	300749
Intellectual disability with panhypopituitarism	SOX3	Xq27.1	300123
Intellectual disability with stereotypical movements, epilepsy, and/or cerebral malformations	MEF2C	5q14.3	613443
Jacobsen	Multiple	11q23	147791
Joubert 3	AHI1	6q23.3	608629
Joubert 4	NPHP1	2q13	609583
Joubert 5	CEP290	12q21.32	610188
Juvenile polyposis, BMPR1A-related	BMPR1A	10q23.2	174900
Juvenile polyposis, SMAD4-related	SMAD4	18q21.2	174900
Kallmann 1	KAL1	Xp22.31	308700
Langer Mesomelic Dysplasia	SHOX	Xp22.33, Yp11.32	249700
Langer-Giedion	Multiple	8q24.11-q24.13	150230
Laron syndrome	GHR	5p13-p12	262500
Leri-Weill Dyschondrosteosis	SHOX	Xp22.33, Yp11.32	127300
Lesch-Nyhan	HPRT1	Xq26.2-q26.3	300322
Leukodystrophy	Multiple	11q14.2-q14.3	
Li-Fraumeni 1	TP53	17p13.1	151623
Lissencephaly 1	PAFAH1B1	17p13.3	607432
Lissencephaly 2	RELN	7q22.1	257320
Lissencephaly, X-linked	DCX	Xq23	300067
Loeys-Dietz, type 1A	TGFBR1	9q22.33	609192
Loeys-Dietz, type 2B	TGFBR2	3p24.1	610380
Long QT	KCNQ1	11p15.5-p15.4	192500
Lowe	OCRL	Xq25-q26	309000
Lymphoproliferative, X-linked	SH2D1A	Xq25	308240
Macrocephaly/Autism	PTEN	10q23.31	605309
Marfan 1	FBN1	15q21.1	154700
McLeod	XK	Xp21.1	300842
Menkes	ATP7A	Xq21.1	309400
Mental retardation 1	MBD5	2q23.1	156200
Mental retardation 21	IL1RAPL1	Xp21.3-p21.2	300143
Mental retardation 5	SYNGAP1	6p21.32	612621
Mental retardation 6	GRIK2	6q16.3	611092
Mental retardation 7	TUSC3	8p22	611093
Mental retardation 9	FTSJ1	Xp11.23	309549
Mental retardation 94	GRIA3	Xq25	300699
Mesomelic dysplasia, Kantaputra type	HOXD gene cluster	2q24-q32	156232
Metachromatic leukodystrophy	ARSA	22q13.33	250100
Microphthalmia 2	BCOR	Xp11.4	300166
Microphthalmia 3	SOX2	3q26.33	206900
Microphthalmia 5	OTX2	14q22.3	610125
Microphthalmia 7	HCCS	Xp22.2	309801

Associated Condition	Gene	Chr/Band	OMIM #
Microphthalmia, isolated, with cataract 2	SIX6	14q23.1	212550
Miller-Dieker	ABR, PAFAH1B1, PITPNA, YWHAE	17p13.3	247200
Mohr-Tranebjaerg	TIMM8A	Xq22.1	304700
Mowat-Wilson	ZEB2	2q22.3	235730
Muscular Dystrophy-Dystroglycanopathy A6	LARGE	22q12.3	613154
Myoclonus dystonia	SGCE	7q21.3	159900
Myotubular myopathy 1	MTM1	Xq28	310400
Nablus mask-like facial	Multiple	8q22.1	608156
Nail-patella (NPS)	LMX1B	9q33.3	161200
Nephronophthisis 1	NPHP1	2q13	256100
Nephropathic cystinosis	CTNS	17p13.2	219800
Neurofibromatosis 1	JJAZ1, NF1	17q11.2	162200
Neurofibromatosis 2	NF2	22q12.2	101000
Neurosensory deafness (DFNB1)	GJB6	13q12.11	220290
NFIA haploinsufficiency	NFIA	1p31.3	600727
Noonan 1	PTPN11	12q24.13	163950
Noonan 4	SOS1	2p22.1	610733
Noonan 5	RAF1	3p25.2	611553
Norrie disease	NDP	Xp11.3	310600
Oculocutaneous albinism 2 (OCA2)	OCA2	15q12-q13	203200
Opitz G	MID1	Xp22.2	300000
Ornithine transcarbamylase deficiency	OTC	Xp11.4	311250
Orofacial Cleft 11	BMP4	14q22.2	600625
Orofaciodigital 1	OFD1	Xp22.2	311200
Osteogenesis Imperfecta	COL1A1	17q21.33	166200
Osteopathia striata with cranial sclerosis	FAM123B	Xq11.2	300373
Otodental dysplasia	FGF3	11q13	166750
Pallister-Killian	Multiple	12p	601803
Paragangliomas 1	SDHD	11q23.1	168000
Paragangliomas 4	SDHB	1p36.13	115310
Pelizaeus-Merzbacher disease	PLP1	Xq22.2	312080
Peutz-Jeghers	STK11	19p13.3	175200
Pitt-Hopkins	TCF4	18q21.2	610954
Polycystic kidney disease 1	PKD1	16p13.3	601313
Polycystic kidney disease 2	PKD2	4q22.1	613095
Potocki-Lupski	Multiple	17p11.2	610883
Potocki-Shaffer	Multiple	11p11.2	601224
Prader-Willi-like phenotype	SIM1	6q16.3	176270
Renal cysts and diabetes	HNF1B	17q12	137920
Retinoblastoma	RB1	13q14.2	180200
Rett Syndrome	MECP2	Xq28	312750
Rett syndrome, congenital variant	FOXG1	14q12	613454
Rieger	PITX2	4q25	180500
Rubinstein-Taybi	CREBBP, DNASE1	16p13	180849
Russell-Silver	CPA4, GRB1	7q32.2, 7p11.2	180860
Saethre-Chotzen	TWIST1	7p21.1	101400
Short stature	LHX4	1q25.2	262700
SHOX	SHOX	Xp22.33	312865
Siderius type intellectual disability	PHF8	Xp11.22	300263
Simpson-Golabi-Behmel 1	GPC3	Xq26.2	312870
Smith-Lemli-Opitz	DHCR7	11q13.4	270400
Smith-Magenis	RAI1	17p11.2	182290

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Associated Condition	Gene	Chr/Band	OMIM #
Sotos	NSD1	5q35.2-q35.3	117550
Speech-language disorder 1	FOXP2	7q31.1	602081
Split-hand / foot malformation 1	SHFM1	7q21.2-q21.3	183600
Split-hand / foot malformation 1 with deafness	DLX5	7q21.3	220600
Split-hand / foot malformation 3	BTRC, FBXW4, LBX1, POLL	10q24	246560
Split-hand / foot malformation 4	TP63	3q28	605289
Split-hand / foot malformation 5	DLX1, DLX2	2q31	606708
Stickler 1	COL2A1	12q13.11	108300
Stickler 2	COL11A1	1p21.1	604841
Synpolydactyly	EVX2, HOXD13, HOXD9	2q31.1	186000
Timothy Syndrome	CACNA1C	12p13.33	601005
Toe syndactyly, telecanthus, anogenital and renal malformations (STAR)	FAM58A	Xq28	300707
Townes-Brocks	SALL1	16q12.1	107480
Treacher-Collins	TCOF1	5q32	154500
Tuberous sclerosis 1	TSC1	9q34.13	191100
Tuberous sclerosis 2	TSC2	16p13.3	613254
Ulnar-mammary	TBX3	12q24.21	181450
Usher IIC	GPR98	5q14.3	605472
Van der Woude	IRF6	1q32.2	119300
Vascular endothelial growth factor disorders	VEGFA	6p21.1	192240
Velocardiofacial	TBX1	22q11.21	192430
Von Hippel-Lindau	VHL	3p25.3	193300
Waardenburg I	PAX3	2q36.1	193500
Waardenburg IIA	MITF	3p14.1-p13	193510
WAGR	PAX6, WT1	11p13	194072
Williams-Beuren	ELN, GTF2I, GTF2IRD1, LIMK1, NCF1	7q11.23	194050
Wilms Tumor 1	WT1	11p13	194070
Wolf-Hirschhorn	LETM1, WHSC1, WHSC2	4p16.3	194190
X-linked juvenille retinoschisis	RS1	Xp22.13	312700
X-linked lymphoproliferative type 2	XIAP	Xq25	300635
X-linked mental retardation 30	PAK3	Xq23	300558
X-linked mental retardation, ARX-related	ARX	Xp21.3	300419
Xp11.22 X-linked mental retardation 17	Multiple	Xp11.22	300705
XX Sex Reversal 2	SOX9	17q24.3	278850
XY Sex Reversal 1	SRY	Yp11.31	400044
XY Sex Reversal 2	NR0B1	Xp21.2	300018
XY Sex Reversal 3	NR5A1	9q33.3	612965
XY Sex Reversal 4	DMRT1	9p24.3	
Unique pericentromeric regions	Multiple	43 sites	
Unique subtelomeric regions	Multiple	41 sites	
Aneuploidy	Multiple	Any individual chromosome	
Uniparental Disomy (UPD)	Multiple	Chromosomes 7,11,14,15, 16 & 20	

NOTE: The Prenatal Targeted oligonucleotide array offered by the Genetics Center is a diagnostic screening tool designed to evaluate targeted childhood-onset disease regions of the genome for gains or losses of genomic material. This specific array will also detect whole chromosome loss of herterozygosity (LOH) for chromosomes 7, 11, 14, 15, 16, and 20. As of March 2013, 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 thorough out 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.

Prenatal Targeted array is derived from the International Standards for Cytogenomic Arrays (ISCA) Consortium (now ICCG) design and database as of March 2013.