Syndrome/Locus	OMIM	Position	Candidate Gene(s)
16p11.2-p12.2 microdeletion syndrome	613604	16p11.2-p12.2	Multiple
16p12.1 microdeletion syndrome	136570	16p12.1	Multiple
16p13.1 microdeletion predisposing to autism and/or mental retardation	--	16p13.1	Multiple
16p13.3 microdeletion	180849	16p13.3	CREBBP, DNASE1
16q11.2-q12.2 microdeletion syndrome	602218	16q11.2-q12.2	SALL1, ZNF423
16q24.3 microdeletion syndrome	148050	16q24.3	ANKRD11
17p13.1 deletion syndrome	613776	17p13.1	Multiple
17p13.3 centromeric duplication syndrome	613215	17p13.3	Multiple
17p13.3 microdeletion syndrome	247200	17p13.3	PAFAH1B1, YWHAE
17q11.2 deletion syndrome	673615	17q11.2	NF1
17q12 microdeletion syndrome	610443	17q21.3	MAPT
17q21.3 microdeletion/duplication	610443	17q21.3	MAPT
17q22 deletion syndrome	--	17q22	NOG
17q24.2-q24.3 deletion syndrome	--	17q24.2-q24.3	Multiple
19p13.13 deletion syndrome	613638	19p13.13	Multiple
19q13.11 deletion syndrome	613026	19q13.11	Multiple
20p12.3 microdeletion syndromic	--	20p12.3	Multiple
22q11.2 distal microdeletion	611867	22q11.2	Multiple
22q11.21 microduplication	608363	22q11.21	TBX1
22q13.3 microdeletion	606232	22q13.3	ARSA, SHANK3
46,XX sex reversal 2	278850	17q24.3	SOX9
46,XY sex reversal 3	612965	9q33.3	NR5A1
A Alagille syndrome 1	118450	20p12.2	JAG1
Alpha thalassemia/mental retardation syndrome	141750	16p13.3	HBA1, HBA2
Alveolar capillary dysplasia with misalignment of pulmonary veins	265380	16q24.1	FOXF1
Androgen insensitivity syndrome	300068	Xq12	AR
Angelman syndrome	105830	15q11.2	Multiple
Aniridia	106210	11p13	PAX6
Aural atresia, congenital	607842	18q22.3	TSHZ1
Autism spectrum disorder, intellectual disability	613670	3p13	FOXP1
Autism, intellectual disability	600565	2p16.3	NRNX1
Axenfeld-Rieger syndrome type 1	180500	4q25	PITX2
B Bannayan-Riley-Ruvalcaba syndrome	153480	10q23.31	PTEN
Basal cell nevus syndrome	109400	9q22.32	PTCH1
Becker muscular dystrophy	300376	Xp21	DMD
Beckwith-Wiedemann syndrome	130650	11p15.5	IGF2
Brachydactyly-mental retardation syndrome	600430	2q37.3	Multiple
Branchiootorenal syndrome 1	113650	8q13.3	EYA1
C Campomelic dysplasia	114290	17q24.3	SOX9
Cat-eye syndrome	115470	22q11.21	Multiple
Cerebral cavernous malformations 2	603284	7p13	CCM2
Cerebral cavernous malformations, type 1	116860	7q21.2	KRIT1
Charcot-Marie-Tooth disease type 1A	118220	17p12	PMP22
CHARGE syndrome	214800	8q12.2	CHD7
Choroideremia	303100	Xq21.2	CHM
Chromosome 6q25-q25 deletion syndrome	612863	6q25.3	Multiple

Syndrome/Locus	OMIM	Position	Candidate Gene(s)
Cleidocranial dysplasia	119600	6p21.1	RUNX2
Coffin-Lowry syndrome	303600	Xp22.12	RPS6KA3
Congenital adrenal hypoplasia	300200	Xp21.2	NR0B1
Congenital heart defects, nonsyndromic 2	614980	6q25.1	TAB2
Cornelia de Lange syndrome 1	122470	5p13.2	NIPBL
Cowden syndrome 1	158350	10q23.31	PTEN
Craniofrontonasal syndrome	304110	Xq13.1	EFNB1
Craniosynostosis type 2	604757	5q35.1	MSX2
CRASH syndrome	303350	Xq28	L1CAM
Cri-du-chat syndrome	123450	5p15.2	Multiple
Currarino syndrome	176450	7q36.3	MNX1
D Deafness, autosomal recessive 1A (DFNB1A)	220290	13q12.11	GJB6
DiGeorge syndrome	188400	22q11.21	HIRA, TBX1
DiGeorge syndrome 2	601362	10p14	Multiple
Dosage sensitive sex reversal 2	300018	Xp21.1	NR0B1
Double-outlet right ventricle	217095	19p13.11	GDF1
Dravet syndrome	607208	2q24.3	SCN1A
Duane-radial ray syndrome	607323	20q13.2	SALL4
Duchenne muscular dystrophy	300376	Xp21	DMD
E Epileptic encephalopathy, childhood-onset	615369	15q26.1-q26.2	CHD2
Epileptic encephalopathy, early infantile 2	300672	Xp21.3	CDKL5
Epileptic encephalopathy, early infantile 4	612164	9q34.11	STXBP1
Epileptic encephalopathy, early infantile 6	607208	2q24.3	SCN1A
F Familial adenomatous polyposis 1	175100	5q22.2	APC
Feingold syndrome 1	164280	2p24.3	MYCN
FGFR2 related disorders	176943	10q26.3	FGFR2
FMR1-associated disorders	300624	Xq27.3	FMR1
Focal dermal hypoplasia	305600	Xp11.23	PORCN
Fragile X syndrome	300624	Xq27.3	FMR1
G Generalized epilepsy with febrile seizures plus, type 2	604403	2q24.3	SCN1A
Glass syndrome	612313	2q33.1	SATB2
Glycerol kinase deficiency	300474	Xp21.1	GK
Goltz syndrome	305600	Xp11.23	PORCN
Gorlin syndrome	109400	9q22.32	PTCH1
Greig cephalopolysyndactyly syndrome	175700	7p14.1	GLI3
H Hemophilia A	306700	Xq28	F8
Hemophilia B	306900	Xq27.1	F9
Hereditary hemorrhagic telangiectasia type 1	187300	9q34.11	ENG
Hereditary hemorrhagic telangiectasia type 2	600376	12q13.13	ACVRL1
Hereditary neuropathy with pressure palsies	162500	17p11.2	PMP22
Holoprosencephaly and preaxial polydactyly	264480	5q35.1	FBXW11
Holoprosencephaly type 2	157170	2p21	SIX3
Holoprosencephaly type 3	142945	7q36	SHH
Holoprosencephaly type 4	142946	18p11.3	TGIF1
Holoprosencephaly type 5	609637	13q32.3	ZIC2
Holoprosencephaly type 7	610828	9q22.32	PTCH1

Syndrome/Locus	OMIM	Position	Candidate Gene(s)
Holoprosencephaly type 10	612530	1q41	DISP1
Holt-Oram-related disorders	142900	12q24.21	TBX5
Hypoparathyroidism, sensorineural deafness, and renal disease (HDR syndrome)	146255	10p14	GATA3
Hypotonia-cystinuria syndrome	606407	2p21	SLC3A1, PREPL
I Infantile spasms, MAGI2-related	606382	7q21.11	MAGI2
J Jacobsen syndrome	147791	11q23-11q25	Multiple
Juvenile polyposis of infancy	612242	10q23	PTEN, BMPR1A
Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	175050	18q21.2	SMAD4
K Kallmann syndrome 1	308700	Xp22.31	KAL1
KBG syndrome	148050	16q24.3	ANKRD11
Kleefstra syndrome	610253	9q34	EHMT1
L Langer mesomelic dysplasia	249700	Xp22.33	SHOX
Langer-Giedion syndrome	150230	8q24.11	TRPS1, EXT1
Leri-Weill dyschondrosteosis	127300	Xp22.33	SHOX
Lissencephaly 1	607432	17p13.3	PAFAH1B1 (LIS1)
Loeys-Dietz syndrome 1	609192	9q22.33	TGFBR1
Loeys-Dietz syndrome type 2	610168	3p24.1p23	TGFBR2
Loeys-Dietz syndrome type 4	614816	1q41	TGFB2
Lowe oculocerebrorenal syndrome	309000	Xq25	OCRL
M Marfan syndrome	154700	15q21.1	FBN1
MASA syndrome	303350	Xq28	L1CAM
MECP2 deletion syndrome	180200	Xq28	MECP2
MECP2 duplication	300815	Xq28	MECP2
Melnick-Fraser syndrome	113650	8q13.3	EYA1
Menkes syndrome	309400	Xq21.1	ATP7A
Mental retardation, autosomal dominant 1	156200	2q23.1	MBD5
Mental retardation, autosomal dominant 6	613970	12p13.1	GRIN2B
Mental retardation, autosomal dominant 12	614562	6q25.3	ARID1B
Mental retardation, autosomal dominant 20	613443	5q14.3	MEF2C
Mental retardation, autosomal dominant 22	612337	1q43-q44	ZBTB18
Mental retardation , autosomal dominant 26	615834	7q11.22	KIAA0442
Mesomelia-synostoses syndrome	600383	8q13.2q13.3	SULF1, SLCO5A1
Mesomelic dysplasia, Savarirayan-type	605274	2q11.2	AFF3
Microphthalmia type 7 with linear skin defects	309801	Xp22.3	HCCS
Miller-Dieker lissencephaly syndrome	247200	17p13.3	PAFAH1B1, YWHAE
Mowat-Wilson syndrome	235730	2q22.3	ZEB2
Mucopolysaccharidosis type II	309900	Xq28	IDS
Myoclonus dystonia	159900	7q21.3	SGCE
N Nail-patella syndrome	161200	9q33.3	LMX1B
Nance-Horan syndrome	302350	Xp22.13	NHS
Neurofibromatosis type 1	162200	17q11.2	NF1
Neurofibromatosis type 2	101000	22q12.2	NF2
NFIA haploinsufficiency	613735	1p31.3	NFIA
Noonan syndrome type 4	610733	2p22.1	SOS1
Norrie disease	310600	Xp11.3	NDP
O Opitz GBBB syndrome	300000	Xp22	MID1

Syndrome/Locus	OMIM	Position	Candidate Gene(s)
Optic atrophy 1	125250	3q29	OPA1
Oralfacialdigital syndrome type 1	311200	Xp22.2	OFD1
Ornithine transcarbamylase deficiency	311250	Xp11.4	OTC
Osteopathia striata with cranial sclerosis	300373	Xq11.2	AMER1
Otodental dysplasia and coloboma due to 11q13.3 microdeletion	166750	11q13.3	FGF3, FADD
Otofacialcervical syndrome	166780	8q13.3	EYA1
P Pallister-Killian syndrome	601803	12p	Multiple
Papillorenal syndrome	120330	10q24.31	PAX2
Parietal foramina 1	168500	5q35.1	MSX2
Pelizaeus-Merzbacher syndrome	312080	Xq22.2	PLP1
Phelan-McDermid syndrome	606232	22q13.3	SHANK3
Pitt-Hopkins syndrome	610954	18q21.1	TCF4
Pitt-Hopkins-like syndrome 2	614325	2p16.3	NRXN1
Polycystic kidney disease 1	173900	16p13.3	PKD1
Potocki-Lupski syndrome	610883	17p11.2	Multiple
Potocki-Shaffer syndrome	601224	11p11.2	EXT2, ALX4
Prader-Willi syndrome	176270	15q11.2	Multiple
Pseudotrisomy 13 syndrome	264480	5q35.1	FBXW11
PTEN-related disorders	158350	10q23.31	PTEN
R Renal cysts and diabetes syndrome	137920	17q12	HNF1B
Retinoblastoma	180200	13q14.2	RB1
Rett syndrome	180200	Xq28	MECP2
Rett syndrome, congenital variant	613454	14q12	FOXG1
Rubenstein-Taybi syndrome 1	180849	16p13.3	CREBBP
S Saethre-Chotzen syndrome	101400	7p21.1	TWIST1
Severe obesity, early-onset	601665	6q16.3	SIM1
Simpson-Golabi-Behmel syndrome type 1	312870	Xq26.2	GPC3
Smith-Lemli-Opitz syndrome	270400	11q13.4	DHCR7
Smith-Magenis syndrome	182290	17p11.2	RAI1
Sotos syndrome 1	117550	5q35.3	NSD1
Spastic paraplegia 4	182601	2p22.3	SPAST
Speech and language disorder type 1	602081	7q31.1	FOXP2
Split-hand/foot malformation 1	183600	7q21.3	Multiple
Split-hand/foot malformation 4	605289	3q28	TP63
Steroid sulfatase deficiency	308100	Xp22.31	STS
Synpolydactyly 1	186000	2q31.1	HOXD13
T Townes-Brocks syndrome 1	107480	16q12.1	SALL1
Trichorhinophalangeal syndrome type 1	190350	8q23.3	TRPS1
Trichorhinophalangeal syndrome type 2	150230	8q24.11	TRPS1, EXT1
Trigonocephaly	190440	8p11.23p11.22	FGFR1
Tuberous sclerosis type 1	191100	9q34.13	TSC1
Tuberous sclerosis type 2	191100	16p13.3	TSC2
U Ulnar-mammary syndrome	181450	12q24.21	TBX3
V Van der Woude syndrome	119300	1q32.2	IRF6
W Waardenburg syndrome type I	193500	2q36.1	PAX3
Waardenburg syndrome type 2A	193510	3p14.1-p13	MITF
Waardenburg syndrome type 2E	611584	22q13.1	SOX10
WAGR syndrome	194072	11p13	PAX6, WT1
Williams-Beuren syndrome	194050	7q11.23	ELN
Wilms Tumor 1	194070	11p13	WT1
Wolf-Hirschhorn syndrome	194190	4p16.3	Multiple
X X-linked agammaglobulinemia	300755	Xq22.1	BTK
X-linked alpha-thalassemia/mental retardation syndrome	301040	Xq21.1	ATRX
X-linked Alport syndrome	301050	Xq22.3	COL4A5
X-linked chronic granulomatous disease	306400	Xp11.4	CYBB
X-linked cleft palate with or without ankyloglossia	303400	Xq21.1	TBX22
X-linked heterotaxy	306955	Xq26.3	ZIC3
X-linked ichthyosis	308100	Xp22.31	STS
X-linked infantile spasm syndrome	300672	Xp21.3	CDKL5
X-linked juvenile retinoschisis	312700	Xp22.13	RS1
X-linked lissencephaly	300067	Xq23	DCX
X-linked lymphoproliferative syndrome 1	308240	Xq25	SH2D1A
X-linked mental retardation	300495	Xp22.32-p22.32	NLGN4X
X-linked mental retardation with microcephaly and pontine and cerebellar hypoplasia	300749	Xp11.4	CASK
X-linked mental retardation, Siderius type	300263	Xp11.22	PHF8
X-linked mental retardation, types 17 and31	300705	Xp11.22	HSD17B10, HUWE1
X-linked mental retardation, type 21	300143	Xp21.3	IL1RAPL1
X-linked mental retardation, type 30	300558	Xq22.3	PAK3
X-linked mental retardation, type 94	300699	Xq25	GRIA3
X-linked mental retardation, with panhypopituitarism	300123	Xq27.1	SOX3
X-linked mental retardation, FRAXE type	309548	Xq28	AFF2
X-linked short stature	300582	Xp22.33 / Yp11.32	SHOX
Xp11.22 microduplication	300705	Xp11.22	HSD17B10, HUWE1
Xp11.3 microdeletion	300578	Xp11.3	RP2, ZNF674
Xp21 microdeletion syndrome	300679	Xp21	NR0B1, IL1RAPL1, GK
Xp21 microdeletion syndrome	310200	Xp21	DMD
Xq28 microduplication	300815	Xq28	MECP2

Test Information

In >200 well-characterized regions of known clinical significance the SNP markers are enriched to provide an average resolution of approximately 20 Kb for the detection of copy number changes. The average resolution outside of these regions is approximately 1 Mb. In order to reduce the likelihood of a VOUS result, the following copy number variants (CNVs) are typically not reported unless considered to be clinically relevant to the specific case: CNVs that do not contain any OMIM Morbid genes, heterozygous CNVs that contain only autosomal recessive OMIM Morbid genes, or CNVs considered to be *risk factors* for neurodevelopmental and/or neurobehavioral disorders such as CNVs limited to the BP1-BP2 region of 15q11.2 and microduplications of 16p11.2. In addition, partial or whole-gene duplications of commonly deleted genes such as STS gene at Xp22.31 are not reported. Due to its targeted nature, this test is not recommended for delineation of regions of homozygosity.

Test Limitations

As with all microarray testing, the CombiSNP Targeted Array for Prenatal Analysis does not detect: very small intragenic deletions or duplications, balanced chromosomal aberrations such as Robertsonian or reciprocal translocations, balanced inversions, balanced insertions, or imbalances in genomic regions that are not represented on the microarray. This test does not routinely evaluate for consanguinity. Although this test can detect uniparental isodisomy, it cannot detect uniparental heterodisomy. Please remember, normal microarray results do not rule out the possibility of a genetic disorder or syndrome that is due to a genetic alteration not detected or evaluated by this test.

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CombiSNP Prenatal Targeted Array Disorder List

The CombiSNP Targeted Array for Prenatal Analysis delivers high-density coverage of over 200 regions of known clinical significance while minimizing the likelihood of identifying Variants of Unclear Clinical Significance (VOUS). The following is a list of disorders and clinically relevant regions targeted by this array. All the chromosome band and genomic positions are based on UCSC Human Genome Build 19 (NCBI build 37, Feb 2009). Please refer to the "Test Information" section at the end of this list for additional details about the benefits and limitations of this test.

Syndrome/Locus	OMIM	Position	Candidate Gene(s)
1p36 deletion syndrome	607872	1p36	Multiple
1q21.1 deletion syndrome, 1.35 Mb	612474	1q21.1-q21.2	Multiple
1q21.1 deletion with susceptibility to TAR	274000	1q21.1-q21.2	Multiple
1q21.1 duplication syndrome	612475	1q21.1-q21.2	Multiple
1q41-q42 deletion syndrome	612530	1q41-q42.12	Multiple
1q43-q44 deletion syndrome	612337	1q43-q44	AKT3, ZBTB18
2p16.1-p15 deletion syndrome	612513	2p16.1-p15	Multiple
2p21 microdeletion, homozygous	606407	2p21	Multiple
2q32-q33 deletion syndrome	612313	2q32-q33	SATB2
2q33.1 deletion syndrome	612313	2q33.1	SATB2
2q37 deletion syndrome	600430	2q37.3	HDAC4
3q13.31 deletion syndrome	615433	3q13.31	ZBTB20
3q26.33-3q27.2 deletion syndrome	(none)	3q26.33-3q27.2	Multiple
3q27.3 deletion syndrome	(none)	3q27.3	Multiple
3q29 deletion syndrome	609425	3q29	Multiple
4q21 deletion syndrome	613509	4q21	Multiple
5q14.3 deletion syndrome	613443	5q14.3	MEF2C
6pter-p24 deletion syndrome	612582	6pter-p24	Multiple
7q11.23 duplication syndrome	609757	7q11.23	Multiple
8p23.1 deletion/duplication syndrome	600576	8p23.1	GATA4
9q22.3 deletion syndrome	601309	9q22.3	PTCH1, FANCC
9q34.3 deletion syndrome	610253	9q34	EHMT1
10q23 deletion syndrome	612242	10q23	PTEN, BMPR1A
10q26 deletion syndrome	609625	10q26	DOCK1, C10ORF90
11q terminal deletion syndrome	147791	11q23-11q25	Multiple
12q14.3 microdeletion syndrome	--	12q14.13	LEMD3, HMGA2, and GRIP1
14q11.2 microdeletion	--	14q11.2	CHD8, SUPT164
14q22-q23 microdeletion	--	14q22.1-q23.1	Multiple
14q32.2 microdeletion	--	14q32.2q32.31	MEG3, DLK1
15q13.3 microdeletion syndrome	612001	15q13.3	CHRNA7
15q24 microdeletion/microduplication	613406	15q24	Multiple
15q26.3 overgrowth syndrome	147370	15q26.3	IGF1R
16p11.2 microdeletion syndrome	611913	16p11.2	Multiple