

SIGNATURECHIPOS™ / PRENATALCHIP® OS DISORDERS TESTED



Clinically recognized regions of the genome
assayed by SignatureChipOS™(V2) /
Signature PrenatalChip®OS(V2)

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The SignatureChipOS™ / PrenatalChip®OS is a “whole genome” array and therefore may identify genetic conditions not included in the representative list of Disorders Tested. This array may identify copy number changes associated with adult-onset conditions and/or cancer predisposition that are potentially unrelated to the patient’s current clinical findings, but that may become apparent later in life. In addition, if there is a family history of a known or suspected genetic condition unrelated to the reason for testing, please contact the laboratory to discuss prior to sample submission.

DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION	DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION
■ 1p36 Microdeletion *	607872	Multiple	1p36	■ 17q21.3 Microdeletion *	610443	Multiple MAPT candidate	17q21.3
■ 1q21.1 Microdeletion with susceptibility for thrombocytopenia- absent radius (TAR)	274000	Multiple	1q21.1	■ 22q11.2 Distal microdeletion *	611867	Multiple	22q11.2
■ 1q21.1 Microdeletion with susceptibility to mental retardation, autism, or congenital anomalies *	612474	Multiple ACP6 candidate GJA5 candidate GJA8 candidate	1q21.1	■ 22q11.21 Microduplication *	608363	Multiple TBX1 candidate	22q11.21
■ 1q41-q42 Microdeletion		Multiple DISP1 candidate	1q41	■ 22q13.3 Microdeletion *	606232	Multiple ARSA candidate SHANK3 candidate	22q13.3
■ 1q44 Microdeletion		Multiple AKT3 candidate	1q44	■ Xp11.22-linked mental retardation *		Multiple HSD17B10 candidate HUWE1 candidate	Xp11.22
■ 2p15-p16.1 Microdeletion		Multiple	2p15-p16.1	■ Xp11.3 Microdeletion	300578	Multiple RP2 candidate ZNF674 candidate	Xp11.3
■ 2p21 Microdeletion, homozygous	606407	Multiple	2p21	■ Xp11.4-p21.2 Contiguous gene deletion		Multiple IL1RAPL1 OTC	Xp11.4-p21.2
■ 2q32.2-q33 Microdeletion	119540	Multiple SATB2 candidate	2q33.1	■ Adrenal hypoplasia congenita (AHC)	300200	NR0B1	Xp21.2
■ 3q29 Microdeletion	609425	Multiple	3q29	■ Alagille	118450	JAG1	20p12.2
■ 6p25.3 Microdeletion		Multiple	6p25.3	■ Albright hereditary osteodystrophy- like/Brachydactyly-MR	600430	Multiple	2q37.3
■ 6q24.3 Microdeletion		Multiple	6q24.3	■ Alpha thalassemia mental retardation (ATR-16)	141750	HBA1 HBA2	16p13.3
■ 7q11.23 Microduplication *	609757	Multiple	7q11.23	■ Androgen insensitivity	300068	AR	Xq12
■ 8p23.1 Microdeletion *		Multiple GATA4 candidate	8p23.1	■ Angelman	105830	UBE3A	15q11.2
■ 9q22.32-q22.33 Microdeletion		Multiple TGFBRI1 candidate	9q22.33	■ Aniridia II	106210	PAX6	11p13
■ 9q34 Microdeletion *	610253	Multiple EHMT1 candidate	9q34.3	■ Atrial septal defect (ASD) with atrioventricular conduction defects	108900	NKX2-5	5q35.2
■ 10q22.3-q23.31 Microdeletion		Multiple	10q22.3- q23.31	■ Bannayan-Riley-Ruvalcaba (BRRS)	153480	PTEN	10q23.31
■ 12q14.1-q15 Microdeletion *		Multiple LEMD3 candidate GRIP1 candidate	12q14.3	□ Bartter, antenatal 1	601678	SLC12A1	15q21.1
■ 12q24.21-q24.23 Microduplication *		Multiple	12q24.21- q24.23	□ Bartter, antenatal 2	241200	KCNJ1	11q24.3
■ 14q11.2 Microdeletion		Multiple CHD8 candidate SUPT164 candidate	14q11.2	□ Bartter 3 (classic)	607364	CLCNKB	1p36.13
■ 14q22-q23 Microdeletion	600037	Multiple	14q22-q23	□ Bartter 4 (infantile with sensorineural deafness)	602522	BSND CLCNKA & CLCNKB	1p32.3 1p36.13
■ 15q11-q13 Microduplication *	608636	Multiple	15q11-q13	■ Basal cell nevus/Gorlin-Goltz	109400	PTCH1	9q22.32
■ 15q13.3 Microdeletion	612001	Multiple CHRNA7 candidate	15q13.3	■ Beckwith-Wiedemann, IGF2-related *	130650	IGF2	11p15.5
■ 15q24.1-q24.3 Microdeletion		Multiple	15q24.1-q24.3	□ Beckwith-Wiedemann, KCNQ1OT1- related	130650	KCNQ1OT1	11p15.5
■ 16p11.2 Microdeletion *	611913	Multiple	16p11.2	□ Benign neonatal epilepsy	121200	KCNQ2	20q13.33
■ 16p11.2-p12.2 Microdeletion *		Multiple	16p11.2-p12.2	□ Bilateral frontoparietal polymicrogyria (BFPP)	606854	GPR56	16q13
■ 16p13.1 Microdeletion predisposing to autism and/or mental retardation *		Multiple	16p13.1	■ Blepharophimosis, ptosis, epicanthus inversus (BPE)	110100	FOXL2	3q22.3
■ 16p13.3 Microdeletion/Severe Rubinstein-Taybi	610543	CREBBP DNASE1	16p13.3	■ Boston-type craniosynostosis *	604757	MSX2	5q35.2
■ 16q11.2-q12.2 Microdeletion		Multiple SALL1 candidate ZNF423 candidate	16q11.2-q12.2	■ Branchio-oto-renal (BOR)/ Melnick-Fraser	113650	EYA1	8q13.3
				□ Buschke-Ollendorff	166700	LEMD3	12q14.3

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DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION	DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION
■ Campomelic dysplasia (CMPD)	114290	SOX9	17q24.3	□ Holoprosencephaly 7	610828	PTCH1	9q22.32
■ Cat-eye *	115470	Multiple	22q11.1	■ Holoprosencephaly 8	609408	Multiple	14q13.1-q13.2
■ Cerebellar hypoplasia, VLDLR-related/ Hutterite dysequilibrium	224050	VLDLR	9p24.2	□ Holoprosencephaly and preaxial polydactyly *	605651	FBXW11	5q35.1
■ Cerebral cavernous malformations, type 1 (CCM1)	116860	KRIT1	7q21.2	□ Holt-Oram *	142900	TBX5	12q24.21
■ Cerebral cavernous malformations, type 2 (CCM2)	603284	CCM2	7p13	■ Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	146255	GATA3	10p14
□ Cerebral cavernous malformations, type 3 (CCM3)	603285	PDCD10	3q26.1	■ Hypotonia-cystinuria	606407	SLC3A1 PREPL	2p21
■ CHARGE	214800	CHD7	8q12.2	■ Infantile hyperinsulinism with enteropathy & deafness	606528	USH1C ABCC8	11p15.1
■ Choroideremia	303100	CHM	Xq21.2	■ Infantile spasms, MAGI2-related	606382	MAGI2	7q21.11
■ Cleidocranial dysplasia (CCD)	119600	RUNX2	6p12.3	■ Jacobsen/11q terminal deletion disorder	147791	Multiple	11q23-11qter
□ Congenital deafness with inner ear agenesis, microtia, & microdontia	610706	FGF3	11q13.3	□ Joubert 3	608629	AHI1	6q23.3
■ Congenital diaphragmatic hernia (CDH)	142340	CHD2 NR2F2	15q26.1 15q26.2	■ Joubert 4	609583	NPHP1	2q13
■ Congenital diaphragmatic hernia 2 (CDH2) *	222400	GATA4 candidate	8p23.1	□ Joubert 5	610188	CEP290	12q21.32
□ Cornelia de Lange	122470	NIPBL	5p13.2	■ Juvenile polyposis (JPS), BMPR1A-related	174900	BMPR1A	10q23.2
■ Cowden	158350	PTEN	10q23.31	■ Juvenile polyposis (JPS), SMAD4-related	174900	SMAD4	18q21.2
■ Craniofrontonasal	304110	EFNB1	Xq13.1	■ Kallmann 1	308700	KAL1	Xp22.31
■ Cri-du-Chat	123450	Multiple	5p15.2	■ Langer-Giedion	150230	TRPS1 EXT1	8q23.3 8q24.11
■ Currarino	176450	MNX1	7q36.3	■ Langer mesomelic dysplasia (LMD)	249700	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
■ Dandy-Walker malformation (DWM)	220200	ZIC1 ZIC4	3q24	□ Leber congenital amaurosis X (LCAx)	611755	CEP290	12q21.32
■ DiGeorge/Velocardiofacial (VCF)	188400	HIRA TBX1	22q11.21	■ Leri-Weill dyschondrosteosis (LWD)	127300	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
■ DiGeorge 2	601362	Multiple	10p14	■ Li-Fraumeni 1 (LFS)	151623	TP53	17p13.1
■ Dosage-sensitive sex reversal *	300018	NR0B1	Xp21.2	■ Lissencephaly 1	607432	PFAFH1B1 (LIS1)	17p13.3
■ Down syndrome critical region (DSCR) *	602917	Multiple	21q22.13	□ Lissencephaly with cerebellar hypoplasia	257320	RELN	7q22.1
■ Familial adenomatous polyposis (FAP)/ Gardner/MR	175100	APC	5q22.2	□ Loeyes-Dietz (LDS), TGFBR1-related	609192	TGFBR1	9q22.33
□ Familial hypocalciuric hypercalcemia 1 (HHC1)	145980	CASR	3q21.1	□ Loeyes-Dietz (LDS), TGFBR2-related	610168	TGFBR2	3p24.1
■ Feingold	164280	MYCN	2p24.3	■ Lowe	309000	OCRL	Xq25
□ FG 5 *	300581	MID2	Xq22.3	□ Macrocephaly-autism	605309	PTEN	10q23.31
■ FMR1 Microdeletion	300624	FMR1	Xq27.3	■ Marfan 1 (MFS1)	154700	FBN1	15q21.1
■ Focal dermal hypoplasia/Goltz	305600	PORCN	Xp11.23	□ Marfan 2 (MFS2)	610380	TGFBR2	3p24.1
■ Fryns	229850	DISP1 candidate	1q41	■ McLeod	314850	XK	Xp21.1
□ Generalized epilepsy with febrile seizures plus 2 (GEFS+2)	604233	SCN1A	2q24.3	□ Meckel 4	611134	CEP290	12q21.32
□ Gitelman	263800	SLC12A3	16q13	■ Menkes (MNK)	309400	ATP7A	Xq21.1
■ Glycerol kinase deficiency (GKD)	300474	GK	Xp21.2	■ Microphthalmia 3	206900	SOX2	3q26.33
■ Greig cephalopolysyndactyly	175700	GLI3	7p14.1	■ Microphthalmia 7 with linear skin defects	309801	Multiple	Xp22.2
■ Hemophilia A	306700	F8	Xq28	■ Miller-Dieker	247200	PFAFH1B1 (LIS1)	17p13.3
■ Hemophilia B	306900	F9	Xq27.1	■ Mohr-Tranebjaerg	304700	TIMM8A	Xq22.1
■ Hereditary hemorrhagic telangiectasia, type 2	600376	ACVRL1	12q13.13	■ Mowat-Wilson *	235730	ZEB2	2q22.3
□ Holoprosencephaly 1	236100	TRAPPC10	21q22.3	■ Myoclonus dystonia	159900	SGCE	7q21.3
■ Holoprosencephaly 2	157170	SIX3	2p21	■ Nablus mask-like facial	608156	Multiple	8q21.3-q22.1
■ Holoprosencephaly 3	142945	SHH	7q36.3	■ Nail-patella (NPS)	161200	LMX1B	9q33.3
■ Holoprosencephaly 4	142946	TGIF1	18p11.31	□ Neonatal severe primary hypoparathyroidism (NSHPT)	239200	CASR	3q21.1
■ Holoprosencephaly 5	609637	ZIC2	13q32.3	■ Nephronophthisis 1	256100	NPHP1	2q13
□ Holoprosencephaly 6	605934	Multiple	2q37.1-q37.3				

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DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION	DISORDER	OMIM#	GENE(S)/LOCUS	LOCATION
■ Nephropathic cystinosis	219800	CTNS	17p13.3	■ SRY dosage abnormalities	278850/ 306100	SRY	Yp11.31
■ Neurofibromatosis 1 (NF1)/MR	162200	NF1	17q11.2	■ Steroid sulfatase deficiency	308100	STS	Xp22.31
■ Neurofibromatosis 2 (NF2)	101000	NF2	22q12.2	□ Stickler I	108300	COL2A1	12q13.11
■ Neurosensory deafness, autosomal recessive (DFNB1)	220290	GJB6	13q12.11	■ Synpolydactyly/Syndactyly II	186000	HOXD gene cluster	2q31.1
■ NFIA haploinsufficiency	600727	NFIA	1p31.3	■ Townes-Brocks 1	107480	SALL1	16q12.1
□ Noonan 1	163950	PTPN11	12q24.13	■ Trichorhinophalangeal 1	190350	TRPS1	8q23.3
□ Noonan 4	610733	SOS1	2p22.1	■ Tuberous sclerosis 1 (TSC1)	191100	TSC1	9q34.13
■ Norrie	310600	NDP	Xp11.3	■ Tuberous sclerosis 2 (TSC2)	191100	TSC2	16p13.3
■ Oculocutaneous albinism 2 (OCA2) *	203200	OCA2	15q13.1	■ Ulnar-mammary	181450	TBX3	12q24.21
■ Okihiro	607323	SALL4	20q13.2	■ Van der Woude	119300	IRF6	1q32.2
■ Opitz	300000	MID1	Xp22.2	□ Vascular endothelial growth factor (VEGFA)-related disorders	192240	VEGFA	6p21.1
■ Ornithine transcarbamylase deficiency (OTC)	311250	OTC	Xp11.4	■ von Hippel-Lindau	193300	VHL	3p25.3
□ Oro-facio-digital 1 (OFD1)	311200	OFD1	Xp22.2	■ Waardenburg I	193500	PAX3	2q36.1
■ Oto-dental	166750	FGF3	11q13.3	■ Waardenburg IIA	193510	MITF	3p14.1
□ Oto-facio-cervical (OFC)	166780	EYA1	8q13.3	■ WAGR	194072	PAX6 WT1	11p13
■ Pallister-Killian *	601803	Multiple	12p	□ Walker-Warburg, LARGE-related	236670	LARGE	22q12.3
□ Parietal foramina 1	168500	MSX2	5q35.2	■ Williams-Beuren	194050	ELN	7q11.23
■ Pelizaeus-Merzbacher *	312080	PLP1	Xq22.2	■ Wilms Tumor 1	194070	WT1	11p13
■ Peutz-Jeghers (PJS)	175200	STK11	19p13.3	■ Wolf-Hirschhorn	194190	Multiple	4p16.3
■ Pitt-Hopkins	610954	TCF4	18q21.1	■ X-linked agammaglobulinemia	300755	BTK	Xq22.1
■ Polycystic kidney disease 1 (PKD1)	601313	PKD1	16p13.3	■ X-linked Alport (ATS)	301050	COL4A5	Xq22.3
■ Potocki-Lupski/17p11.2 Microduplication *	610883	Multiple	17p11.2	■ X-linked Alport plus diffuse leiomyomatosis (ATS-DL)	301050	COL4A5 COL4A6	Xq22.3
■ Potocki-Shaffer	601224	EXT2 ALX4	11p11.2	■ X-linked chronic granulomatous disease	306400	CYBB	Xp11.4
■ Prader-Willi (PWS)	176270	SNRPN	15q11.2	■ X-linked heterotaxy	306955	ZIC3	Xq26.3
■ Prader-Willi-like phenotype	176270	SIM1	6q16.3	■ X-linked hydrocephalus and nephrogenic diabetes insipidus		L1CAM AVPR2	Xq28
□ Proteus/Proteus-like	176920	PTEN	10q23.31	■ X-linked idiopathic short stature (ISSX)	300582	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
■ PTEN hamartoma tumor	158350	PTEN	10q23.31	□ X-linked infantile spasms, ARX-related	308350	ARX	Xp21.3
■ Renal cysts and diabetes (RCAD) *	137920	HNF1B	17q12	■ X-linked infantile spasms, CDKL5-related	300672	CDKL5	Xp22.13
■ Retinoblastoma/MR	180200	RB1	13q14.2	□ X-linked juvenile retinoschisis	312700	RS1	Xp22.13
■ Rieger 1 (RIEG1)	180500	PITX2	4q25	□ X-linked lissencephaly	300067	DCX	Xq22.3
■ Rubinstein-Taybi (RTS)	180849	CREBBP	16p13.3	□ X-linked lissencephaly with ambiguous genitalia	300215	ARX	Xp21.3
■ Saethre-Chotzen	101400	TWIST1	7p21.1	■ X-linked lymphoproliferative (XLP)	308240	SH2D1A	Xq25
□ Schizencephaly	269160	EMX2	10q26.11	■ X-linked mental retardation 21	300143	IL1RAPL1	Xp21.3
□ Senior-Loken 6	610189	CEP290	12q21.32	□ X-linked mental retardation 30	300558	PAK3	Xq22.3
■ Severe myoclonic epilepsy of infancy (SMEI)	607208	SCN1A	2q24.3	□ X-linked mental retardation 54	300419	ARX	Xp21.3
■ Sex reversal, autosomal dominant 2 (SRA2)	154230	Multiple DMRT1 candidate	9p24.3	■ X-linked mental retardation with isolated growth hormone deficiency *	300123	SOX3	Xq27.1
□ Short stature, pituitary and cerebellar defects, small sella turcica	262700	LHX4	1q25.2	■ X-linked mental retardation with microcephaly & disproportionate pontine and cerebellar hypoplasia	300749	CASK	Xp11.4
■ Simpson-Golabi-Behmel (SGBS)	312870	GPC3	Xq26.2	■ XX male	278850	SRY	Yp11.31
□ Smith-Lemli-Opitz (SLOS)	270400	DHCR7	11q13.4	■ XY gonadal dysgenesis	400044	SRY	Yp11.31
■ Smith-Magenis (SMS)	182290	RAI1	17p11.2	□ XY sex-reversal, +/- adrenal failure	184757	NR5A1	9q33.3
■ Sotos	117550	NSD1	5q35.3	N/A All 41 unique subtelomeric regions		Multiple	41 sites
■ Speech & language disorder 1	602081	FOXP2	7q31.1	N/A All 43 unique pericentromeric regions/marker chromosomes		Multiple	43 sites
■ Split-hand/foot malformation 1 (SHFM1)	183600	SHFM1	7q21.3	N/A Aneuploidy for 24 chromosomes		Multiple	24 chromosomes
■ Split-hand/foot malformation 3 (SHFM3) *	600095	FBXW4	10q24.32				
□ Split-hand/foot malformation 4 (SHFM4)	605289	TP63	3q28				
■ Split-hand/foot malformation 5 (SHFM5)	606708	DLX1 DLX2	2q31.1				

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