

Signature PrenatalChip®TE Disorders Tested

Clinically recognized regions of the genome assayed by the Signature PrenatalChip®TE(v2)

The Signature PrenatalChip®TE (Targeted Enhanced) may identify copy number changes associated with genetic conditions, including adult-onset conditions and/or cancer predisposition that are potentially unrelated to the current clinical findings, but that may become apparent in childhood or 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				
1q21.1 Distal microdeletion *	612474	Multiple ACP6 candidate GJA5 candidate GJA8 candidate	1q21.1	12q14.1–q15 Microdeletion *		Multiple LEMD3 candidate GRIP1 candidate HMG2A candidate	12q14.1–q15
1q21.1 Microdeletion with susceptibility for thrombocytopenia-absent radius (TAR)	274000	Multiple	1q21.1	12q14.3-related primordial dwarfism/Russell-Silver-like		HMG2A	12q14.3
1q41–q42 Microdeletion/Fryns	612530 229850	Multiple	1q41–q42	12q24.21–q24.23 Microduplication *		Multiple	12q24.21–q24.23
1q44 Microdeletion		Multiple AKT3 candidate	1q44	14q11.2 Microdeletion		Multiple CHD8 candidate SUPT16H candidate	14q11.2
2p15–p16.1 Microdeletion	612513	Multiple	2p15–p16.1	14q12 Microduplication *	164874	Multiple FOXG1 candidate	14q12
2p16.3 Microdeletion/Pitt-Hopkins-like 2 *	600565	NRXN1	2p16.3	14q22–q23 Microdeletion		Multiple	14q22–q23
2p21 Microdeletion, autosomal recessive	606407	Multiple	2p21	14q32.2 Microdeletion causing upd(14)mat phenotype		Multiple DLK1 candidate RTL1 candidate	14q32.2
2q23.1 Microdeletion		MBD5 EPC2	2q23.1	14q32.2 Microdeletion causing upd(14)pat phenotype	608149	Multiple MEG3 candidate MEG8 candidate RTL1 candidate	14q32.2
2q32.2–q33 Microdeletion	119540	Multiple SATB2 candidate	2q33.1	15q11–q13 Microduplication *	608636	Multiple	15q11–q13
3q29 Microdeletion	609425	Multiple	3q29	15q13.3 Microdeletion	612001	Multiple CHRNA7 candidate	15q13.3
3q29 Microduplication *	611936	Multiple	3q29	15q24.1–q24.2 Microdeletion	613406	Multiple	15q24.1–q24.2
5p13.2 Microduplication *	613174	NIPBL	5p13.2	15q24.1–q24.2 Microduplication *	613406	Multiple	15q24.1–q24.2
5q35.2–q35.3 Microduplication *		NSD1	5q35.2–q35.3	16p11.2 Microdeletion *	611913	Multiple	16p11.2
6p25.3 Microdeletion	612582	Multiple	6p25.3	16p11.2–p12.2 Microdeletion *	613604	Multiple	16p11.2–p12.2
6q24.3 Microdeletion	612863	Multiple	6q24.3	16p12.1 Microdeletion		Multiple CDR2 candidate EEF2K candidate UQCRC2 candidate	16p12.1
6q25.2–q25.3 Microdeletion	612863	Multiple	6q25.2–q25.3	16p13.11 Microdeletion *		Multiple	16p13.1
7q11.23 Microduplication *	609757	Multiple	7q11.23	16p13.3 Microdeletion/Severe Rubinstein-Taybi	610543	Multiple CREBBP	16p13.3
8p23.1 Microdeletion/ Congenital diaphragmatic hernia 2 (CDH2)	222400	Multiple GATA4 candidate	8p23.1	16p13.3 Microduplication *	613458	CREBBP	16p13.3
8p23.1 Microduplication *		Multiple GATA4 candidate	8p23.1	16q11.2–q12.2 Microdeletion		Multiple SALL1 candidate ZNF423 candidate	16q11.2–q12.2
8q12 Microduplication *		Multiple CHD7 candidate	8q12.1–8q12.3				
9q22.32–q22.33 Microdeletion		Multiple TGFBRI candidate GABBR2 candidate	9q22.33				
9q34 Microdeletion/Kleefstra *	610253	EHMT1	9q34.3				
10q22.3–q23.31 Microdeletion		Multiple	10q22.3–q23.31				

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17p13.1 Microdeletion	613776	Multiple TP53	17p13.1	Basal cell nevus/Gorlin-Goltz	109400	PTCH1	9q22.32
17p13.3 Distal microdeletion		YWHAE CRK	17p13.3	Beckwith-Wiedemann, IGF2-related *	130650	IGF2	11p15.5
17p13.3 Distal microduplication, not including lissencephaly region (PAFAH1B1) *	613215	YWHAE CRK	17p13.3	Beckwith-Wiedemann, KCNQ1OT1-related	130650	KCNQ1OT1	11p15.5
17p13.3 Proximal microduplication, including lissencephaly region (PAFAH1B1) *	613215	PAFAH1B1 (LIS1) YWHAE CRK	17p13.3	Benign neonatal epilepsy	121200	KCNQ2	20q13.33
17q12 Microduplication *		Multiple HNF1B candidate LHX1 candidate	17q12	Beta thalassemia	141900	HBB	11p15.4
17q21.31 Microdeletion	610443	Multiple MAPT candidate	17q21.31	Bilateral frontoparietal polymicrogyria, autosomal recessive (BFPF)	606854	GPR56	16q13
17q21.31 Microduplication *	613533	Multiple	17q21.31	Blepharophimosis, ptosis, epicanthus inversus (BPE)	110100	FOXL2	3q22.3
17q23.1–q23.2 Microdeletion *	613355	Multiple TBX2 candidate TBX4 candidate	17q23.1–q23.2	Boston-type craniosynostosis/Craniosynostosis type 2 *	604757	MSX2	5q35.2
19q13.11 Microdeletion	613026	Multiple LSM14A candidate UBA2 candidate	19q13.11	Branchio-oto-renal (BOR)/Melnick-Fraser	113650	EYA1	8q13.3
22q11.2 Distal microdeletion *	611867	Multiple	22q11.2	Campomelic dysplasia (CMPD)	114290	SOX9	17q24.3
22q11.21 Microduplication *	608363	Multiple TBX1 candidate	22q11.21	Cat-eye *	115470	Multiple	22q11.1
22q13.3 Microdeletion *	606232	Multiple SHANK3 candidate	22q13.3	Cerebellar hypoplasia, VLDLR-related, autosomal recessive/Hutterite disequilibrium, autosomal recessive	224050	VLDLR	9p24.2
Xp11.22–p11.23 Microduplication *	300801	Multiple	Xp11.22–p11.23	Cerebral cavernous malformations, type 1 (CCM1)	116860	KRIT1	7q21.2
Xp11.22-linked intellectual disability/Mental retardation 17, X-linked/Mental retardation 31, X-linked *	300705	Multiple HSD17B10 candidate HUWE1 candidate	Xp11.22	Cerebral cavernous malformations, type 2 (CCM2)	603284	CCM2	7p13
Xp11.3 Microdeletion	300578	Multiple RP2 candidate ZNF674 candidate	Xp11.3	Cerebral cavernous malformations, type 3 (CCM3)	603285	PDCD10	3q26.1
Xp11.4–p21.2 Contiguous gene deletion		Multiple IL1RAPL1 OTC	Xp11.4–p21.2	CHARGE	214800	CHD7	8q12.2
Xq28 Microduplication *	300815	Multiple GDI1 candidate IKBKG candidate	Xq28	Chondrodysplasia 1, X-linked (CDPX1)	302950	ARSE	Xp22.33
Adrenal hypoplasia congenita, X-linked (AHC)	300200	NR0B1	Xp21.2	Choroideremia, X-linked	303100	CHM	Xq21.2
Agammaglobulinemia, X-linked/Bruton agammaglobulinemia, X-linked	300755	BTK	Xq22.1	Chronic granulomatous disease, X-linked	306400	CYBB	Xp11.4
Alagille	118450	JAG1	20p12.2	Cleidocranial dysplasia (CCD)	119600	RUNX2	6p12.3
Albright hereditary osteodystrophy-like/Brachydactyly with intellectual disability/2q37 Microdeletion	600430	HDAC4	2q37.3	Cohen, autosomal recessive *	216550	VPS13B	8q22.2
Alpha thalassemia with intellectual disability (AT-ID)	141750	Multiple HBA1 HBA2 SOX8 candidate	16p13.3	Congenital diaphragmatic hernia (CDH)	142340	Multiple CHD2 candidate NR2F2 candidate	15q26.1–q26.3
Alport, X-linked (ATS)	301050	COL4A5	Xq22.3	Congenital hemidysplasia with ichthyosiform erythroderma & limb defects (CHILD)	308050	NSDHL	Xq28
Alport plus diffuse leiomyomatosis, X-linked (ATS-DL)	301050	COL4A5 COL4A6	Xq22.3	Cornelia de Lange/Brachmann-de Lange	122470	NIPBL	5p13.2
Androgen insensitivity, X-linked	300068	AR	Xq12	Cowden/PTEN hamartoma tumor	158350	PTEN	10q23.31
Angelman	105830	UBE3A	15q11.2	Craniofrontonasal, X-linked	304110	EFNB1	Xq13.1
Aniridia II	106210	PAX6	11p13	Cri-du-chat	123450	Multiple TERT candidate CTNND2 candidate	5p15.2–p15.33
ANKRD11 haploinsufficiency/16q24.3 Microdeletion		ANKRD11	16q24.3	Curarino	176450	MXN1	7q36.3
Atrial septal defect (ASD) with atrioventricular conduction defects	108900	NKX2-5	5q35.2	Dandy-Walker malformation (DWM)	220200	Multiple ZIC1 candidate ZIC4 candidate	3q23–q25.1
Autism with intellectual disability, SHANK2-related	613436	SHANK2	11q13.3	Deafness 22, autosomal recessive	607039	OTOA	16p12.2
Bannayan-Riley-Ruvalcaba (BRRS)/PTEN hamartoma tumor	153480	PTEN	10q23.31	Diamond-Blackfan anemia 1	105650	RPS19	19q13.2
				DiGeorge/Velocardiofacial (VCF)/22q11.21 Microdeletion	188400/192430	TBX1	22q11.21
				DiGeorge 2	601362	Multiple	10p14
				Dopa-responsive dystonia (DRD)/Segawa	128230	GCH1	14q22.2–q22.3
				Dosage-sensitive sex reversal, X-linked *	300018	NR0B1	Xp21.2
				Early-onset ataxia with oculomotor apraxia & hypoalbuminemia, autosomal recessive	208920	APTX	9p13.3
				Emery-Dreifuss muscular dystrophy, X-linked (EDMD)	181350	EMD	Xq28
				EPHA7 haploinsufficiency/6q16.1 Microdeletion		EPHA7	6q16.1
				Epilepsy & intellectual disability restricted to females, X-linked/Juberg-Hellman, X-linked/Dravet-like, X-linked	300088	PCDH19	Xq22.1
				Facio-genital dysplasia, X-linked/Aarskog-Scott, X-linked	305400	FGD1	Xp11.22
				Familial adenomatous polyposis with intellectual disabilities (FAP-ID)/5q22 Microdeletion	175100	APC	5q22.2
				Feingold	164280	MYCN	2p24.3
				FMR1 microdeletion, X-linked	300624	FMR1	Xq27.3
				Focal dermal hypoplasia, X-linked/Goltz, X-linked	305600	PORCN	Xp11.23
				GLUT1 deficiency	606777	SLC2A1	1p34.2
				Glycerol kinase deficiency, X-linked (GKD)	300474	GK	Xp21.2

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Greig cephalopolysyndactyly	175700	GLI3	7p14.1	Leri-Weill dyschondrosteosis, X- & Y-linked (LWD)	127300	SHOX	Xp22.33 & Yp11.32
Growth hormone insensitivity, autosomal recessive/ Laron, autosomal recessive	262500	GHR	5p12	Lesch-Nyhan, X-linked (LNS)	300322	HPRT1	Xq26.2
Hemophilia A, X-linked	306700	F8	Xq28	Li-Fraumeni 1 (LFS)	151623	TP53	17p13.1
Hemophilia B, X-linked	306900	F9	Xq27.1	Lissencephaly, X-linked	300067	DCX	Xq22.3
Hereditary hemorrhagic telangiectasia, type 2	600376	ACVRL1	12q13.13	Lissencephaly 1	607432	PAFAH1B1 (LIS1)	17p13.3
Hereditary paraganglioma-pheochromocytoma, SDHB-related	115310	SDHB	1p36.13	Lowe, X-linked	309000	OCRL	Xq25
Hereditary paraganglioma-pheochromocytoma, SDHD-related	168000	SDHD	11q23.1	Lymphoproliferative, X-linked (XLP)	308240	SH2D1A	Xq25
Heterotaxy, X-linked	306955	ZIC3	Xq26.3	Marfan 1 (MFS1)	154700	FBN1	15q21.1
Hirschsprung, RET-related	142623	RET	10q11.21	McLeod neuroacanthocytosis, X-linked	314850	XK	Xp21.1
Holoprosencephaly 1	236100	Multiple TRAPP C10 candidate PWP2 candidate	21q22.3	Menkes, X-linked (MNK)	309400	ATP7A	Xq21.1
Holoprosencephaly 2	157170	SIX3	2p21	Mental retardation 1, autosomal dominant (MRD1)	156200	MBD5	2q23.1
Holoprosencephaly 3	142945	SHH	7q36.3	Mental retardation 5, autosomal dominant	612621	SYNGAP1	6p21.32
Holoprosencephaly 4	142946	TGIF1	18p11.31	Mental retardation 6, autosomal recessive	611092	GRIK2	6q16.3
Holoprosencephaly 5	609637	ZIC2	13q32.3	Mental retardation 7, autosomal recessive	611093	TUSC3	8p22
Holoprosencephaly 8	609408	Multiple	14q13.1-q13.2	Mental retardation 9, X-linked/Mental retardation 44, X-linked	309549	FTSJ1	Xp11.23
Holt-Oram *	142900	TBX5	12q24.21	Mental retardation 21, X-linked/Mental retardation 34, X-linked	300143	IL1RAPL1	Xp21.3
Hydrocephalus and nephrogenic diabetes insipidus, X-linked		L1CAM AVPR2	Xq28	Mental retardation 94, X-linked *	300699	GRIA3	Xq25
Hyper-IgE recurrent infection, autosomal recessive	243700	DOCK8	9p24.3	Mesomelic dysplasia Kantaputra type *	156232	HOXD gene cluster candidate	2q31.1
Hypohidrotic ectodermal dysplasia, X-linked (XHED)	305100	EDA	Xq13.1	Metachromatic leukodystrophy, autosomal recessive (MLD)/ Arylsulfatase A deficiency	250100	ARSA	22q13.3
Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	146255	GATA3	10p14	Microphthalmia 3	206900	SOX2	3q26.33
Hypospadias 2, X-linked	300758	MAMLD1	Xq28	Microphthalmia 7 with linear skin defects, X-linked	309801	HCCS	Xp22.2
Hypotonia-cystinuria, autosomal recessive	606407	SLC3A1 PREPL	2p21	Miller-Dieker	247200	PAFAH1B1 (LIS1)	17p13.3
Idiopathic short stature, X- & Y-linked (ISSX)	300582	SHOX	Xp22.33 & Yp11.32	Mohr-Tranebjaerg, X-linked/Deafness-dystonia-optic neuropathy, X-linked (DDON)	304700	TIMM8A	Xq22.1
Infantile hyperinsulinism with enteropathy & deafness, autosomal recessive	606528	USH1C ABCC8	11p15.1	Mowat-Wilson *	235730	ZEB2	2q22.3
Infantile spasms, CDKL5-related, X-linked/ Atypical Rett, CDKL5-related, X-linked	300672	CDKL5	Xp22.13	Myoclonus dystonia	159900	SGCE	7q21.3
Infantile spasms, MAGI2-related		MAGI2	7q21.11	Myotubular myopathy 1, X-linked	310400	MTM1	Xq28
Intellectual disability with cerebellar hypoplasia & distinctive facial appearance, X-linked/ Mental retardation 60, X-linked *	300486	OPHN1	Xq12	Nablus mask-like facial	608156	Multiple	8q22.1
Intellectual disability with isolated growth hormone deficiency, X-linked *	300123	SOX3	Xq27.1	Nail-patella (NPS)	161200	LMX1B	9q33.3
Intellectual disability with language impairment and autistic features	613670	FOXP1	3p14.1	Nephronophthisis 1, autosomal recessive	256100	NPHP1	2q13
Intellectual disability with microcephaly & disproportionate pontine and cerebellar hypoplasia, X-linked	300749	CASK	Xp11.4	Nephropathic cystinosis, autosomal recessive	219800	CTNS	17p13.3
Intellectual disability with stereotypical movements, epilepsy, and/or cerebral malformations/ 5q14.3 Microdeletion	613443	MEF2C	5q14.3	Neurofibromatosis 1 with intellectual disabilities (NF1-ID)*	162200	NF1	17q11.2
Jacobsen/11q terminal deletion	147791	Multiple	11q23-11q25	Neurofibromatosis 2 (NF2)	101000	NF2	22q12.2
Joubert 4, autosomal recessive	609583	NPHP1	2q13	Neurosensory deafness, autosomal recessive (DFNB1)	220290	GJB6	13q12.11
Juvenile polyposis (JPS), BMPR1A-related	174900	BMPR1A	10q23.2	NFIA haploinsufficiency/1p31.3 Microdeletion	613735	NFIA	1p31.3
Juvenile polyposis (JPS), SMAD4-related	174900	SMAD4	18q21.2	Norrie, X-linked *	310600	NDP	Xp11.3
Kallmann 1, X-linked	308700	KAL1	Xp22.31	Oculocutaneous albinism 2, autosomal recessive (OCA2) *	203200	OCA2	15q13.1
Langer-Giedion/Trichorhinophalangeal type II	150230	TRPS1 EXT1	8q23.3 8q24.11	Oculofaciocardiodental, X-linked/Microphthalmia 2	300166	BCOR	Xp11.4
Langer mesomelic dysplasia, X- & Y-linked (LMD)	249700	SHOX	Xp22.33 & Yp11.32	Okhiro/Duane radial ray (DRRS)	607323	SALL4	20q13.2
				Opitz GBBB, X-linked	300000	MID1	Xp22.2
				Ornithine transcarbamylase deficiency, X-linked (OTC)	311250	OTC	Xp11.4
				Osteopathia striata with cranial sclerosis, X-linked	300373	FAM123B	Xq11.1
				Oto-dental	166750	FGF3	11q13.3
				Pallister-Killian *	601803	Multiple	12p
				Pelizaeus-Merzbacher, X-linked *	312080	PLP1	Xq22.2
				Peutz-Jeghers (PJS)	175200	STK11	19p13.3

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Pitt-Hopkins	610954	TCF4	18q21.1	Toe syndactyly, telecanthus, anogenital & renal malformations, X-linked (STAR)	300707	FAM58A	Xq28
Pitt-Hopkins-like 1, autosomal recessive/Cortical dysplasia-focal epilepsy, autosomal recessive (CDFE)	610042	CNTNAP2	7q35-q36.1	Townes-Brocks 1	107480	SALL1	16q12.1
Polycystic kidney disease 1 (PKD1)	173900	PKD1	16p13.3	Trichorhinophalangeal 1	190350	TRPS1	8q23.3
Potocki-Lupski/17p11.2 Microduplication *	610883	Multiple RAI1 candidate	17p11.2	Tuberous sclerosis 1 (TSC1)	191100	TSC1	9q34.13
Potocki-Shaffer	601224	EXT2 ALX4	11p11.2	Tuberous sclerosis 2 (TSC2)	613254	TSC2	16p13.3
Prader-Willi (PWS)	176270	SNORD116 cluster candidate	15q11.2	Ulnar-mammary	181450	TBX3	12q24.21
Prader-Willi-like phenotype	176270	Multiple SIM1 candidate	6q16.1-q16.3	Usher IIC, autosomal recessive	605472	GPR98	5q14.3
Renal cysts and diabetes (RCAD)	137920	HNF1B	17q12	Van der Woude	119300	IRF6	1q32.2
Retinoblastoma with intellectual disability	180200	RB1	13q14.2	von Hippel-Lindau	193300	VHL	3p25.3
Rett, congenital variant	613454	FOXP1	14q12	Waardenburg I	193500	PAX3	2q36.1
Rieger 1 (RIEG1)/Axenfeld-Rieger	180500	PITX2	4q25	Waardenburg IIA	193510	MITF	3p14.1
Rubinstein-Taybi (RTS)	180849	CREBBP	16p13.3	Walker-Warburg, LARGE-related, autosomal recessive/ Muscular dystrophy-dystroglycanopathy A1, autosomal recessive	236670	LARGE	22q12.3
Saethre-Chotzen	101400	TWIST1	7p21.1	Williams-Beuren (WBS)	194050	ELN	7q11.23
Severe myoclonic epilepsy of infancy (SMEI)/Dravet	607208	SCN1A	2q24.3	Wilms Tumor 1 (WT1)	194070	WT1	11p13
Sex reversal, autosomal dominant 2 (SRA2)	154230	Multiple DMRT1 candidate	9p24.3	Wilms tumor-aniridia-genital anomalies-retardation (WAGR)	194072	PAX6 WT1	11p13
Short stature, pituitary and cerebellar defects, small sella turcica/Pituitary hormone deficiency, combined, 4	262700	LHX4	1q25.2	Wolf-Hirschhorn	194190	Multiple	4p16.3
Siderius type intellectual disability, X-linked	300263	PHF8	Xp11.22	XX male/SRY dosage abnormalities	278850	SRY	Yp11.31
Simpson-Golabi-Behmel, X-linked (SGBS)	312870	GPC3	Xq26.2	XY gonadal dysgenesis/ SRY dosage abnormalities	400044	SRY	Yp11.31
Smith-Magenis (SMS)	182290	RAI1	17p11.2	XY sex-reversal, +/- adrenal failure/ 46,XY sex reversal 3/ Adrenocortical insufficiency	612965	NR5A1	9q33.3
Sotos	117550	NSD1	5q35.3	All 43 unique pericentromeric regions		Multiple	43 sites
Speech & language disorder 1	602081	FOXP2	7q31.1	All 41 unique subtelomeric regions		Multiple	41 sites
Split-hand/foot malformation 1 (SHFM1)/Ectrodactyly	183600	SHFM1	7q21.3	Aneuploidy for 24 chromosomes		Multiple	24 chromosomes
Split-hand/foot malformation 3 (SHFM3)/Ectrodactyly *	246560	FBXW4	10q24.32				
Split-hand/foot malformation 5 (SHFM5)/Ectrodactyly	606708	EVX2 candidate HOXD gene cluster candidate	2q31.1				
Steroid sulfatase deficiency, X-linked/Ichthyosis, X-linked	308100	STS	Xp22.31				
Synpolydactyly/Syndactyly II	186000	HOXD gene cluster	2q31.1				

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