

DISORDERS TESTED	OMIM#	LOCUS	BAND(S)
<input checked="" type="checkbox"/> 1p36 Microdeletion *	607872	(M)	1p36
<input checked="" type="checkbox"/> 1q21.1 Microdeletion with susceptibility for thrombocytopenia-absent radius (TAR)	274000	(M)	1q21.1
<input checked="" type="checkbox"/> 1q41-q42 Microdeletion		(M) DISP1 candidate	1q41
<input checked="" type="checkbox"/> 2p15-2p16.1 Microdeletion		(M)	2p15-p16.1
<input checked="" type="checkbox"/> 2q32.2-q33 Microdeletion	119540	(M) SATB2 candidate	2q33.1
<input checked="" type="checkbox"/> 3q29 Microdeletion	609425	(M)	3q29
<input checked="" type="checkbox"/> 6p25.3 Microdeletion		(M)	6p25.3
<input checked="" type="checkbox"/> 8p23.1 Microdeletion *		(M) GATA4 candidate	8p23.1
<input checked="" type="checkbox"/> 9q22.32-q22.33 Microdeletion		(M) TGFBFR1 candidate	9q22.33
<input checked="" type="checkbox"/> 9q34 Microdeletion*	610253	(M) EHMT1 candidate	9q34.3
<input checked="" type="checkbox"/> 10q22.3-q23.31 Microdeletion		(M)	10q22.3-q23.31
<input checked="" type="checkbox"/> 12q14.1-q15 Microdeletion *		(M) LEMD3 candidate GRIP1 candidate	12q14.3
<input checked="" type="checkbox"/> 12q24.21-12q24.23 Microduplication *		(M)	12q24.21-q24.23
<input checked="" type="checkbox"/> 14q22-14q23 Microdeletion	600037	(M)	14q21-14q22
<input checked="" type="checkbox"/> 15q24.1-q24.3 Microdeletion		(M)	15q24.1-q24.3
<input checked="" type="checkbox"/> 16p11.2-p12.2 Microdeletion *		(M)	16p11.2-p12.2
<input checked="" type="checkbox"/> 16p13.1 Microdeletion predisposing to autism and/or mental retardation *		(M)	16p13.1
<input checked="" type="checkbox"/> 16p13.3 Microdeletion/Severe Rubinstein-Taybi	610543	CREBBP DNASE1	16p13.3
<input checked="" type="checkbox"/> 17q21.3 Microdeletion *	610443	(M) MAPT candidate	17q21.3
<input checked="" type="checkbox"/> 22q13.3 Microdeletion	606232	(M) ARSA candidate SHANK3 candidate	22q13.3
<input checked="" type="checkbox"/> Xp11.3 Microdeletion	300578	(M) RP2 candidate ZNF674 candidate	Xp11.3
<input checked="" type="checkbox"/> Adrenal hypoplasia congenita (AHC) *	300200	NROB1	Xp21.2
<input checked="" type="checkbox"/> Adult-onset autosomal dominant leukodystrophy (ADLD)* (FISH)	169500	LMNB1	5q23.2
<input checked="" type="checkbox"/> Alagille	118450	JAG1	20p12.2
<input checked="" type="checkbox"/> Albright hereditary osteodystrophy-like/Brachydactyly-MR	600430	(M)	2q37.3
<input checked="" type="checkbox"/> Alpha thalassemia mental retardation (ATR-16) (OS)	141750	HBA1, HBA2	16pter-16p13.3
<input checked="" type="checkbox"/> Alzheimer disease, early onset with cerebral amyloid angiopathy* (FISH)	104300	APP	21q21.3
<input checked="" type="checkbox"/> Angelman *	105830	UBE3A	15q11.2
<input checked="" type="checkbox"/> Aniridia II	106210	PAX6	11p13
<input checked="" type="checkbox"/> Atrial septal defect (ASD) with atrioventricular conduction defects	108900	NKX2-5	5q35.2
<input checked="" type="checkbox"/> Bannayan-Riley-Ruvalcaba (BRRS)	153480	PTEN	10q23.31
<input type="checkbox"/> Bartter, antenatal 1	146200	SLC12A1	15q21.1
<input type="checkbox"/> Bartter, antenatal 2	241200	KCNJ1	11q24.3

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<input type="checkbox"/> Bartter 3 (classic)	607364	CLCNKB	1p36.13
<input type="checkbox"/> Bartter 4 (infantile with sensorineural deafness)	602522	BSND CLCNKA & CLCNKB	1p32.3 1p36.13
<input checked="" type="checkbox"/> Basal cell nevus/Gorlin-Goltz	109400	PTCH1	9q22.32
<input checked="" type="checkbox"/> Beckwith-Wiedemann *	130650	IGF2	11p15.5
<input type="checkbox"/> Bilateral frontoparietal polymicrogyria (BFPP)	606854	GPR56	16q13
<input checked="" type="checkbox"/> Blepharophimosis, ptosis, epicanthus inversus (BPE)	110100	FOXL2	3q22.3
<input checked="" type="checkbox"/> Branchio-oto-renal (BOR)/Melnick-Fraser	113650	EYA1	8q13.3
<input type="checkbox"/> Buschke-Ollendorff	166700	LEMD3	12q14.3
<input checked="" type="checkbox"/> Campomelic dysplasia (CMPD)	114290	SOX9	17q24.3
<input checked="" type="checkbox"/> Cat-eye *	115470	(M)	22q11.1
<input checked="" type="checkbox"/> CHARGE	214800	CHD7	8q12.2
<input checked="" type="checkbox"/> Cleidocranial dysplasia (CCD)	119600	RUNX2	6p12.3
<input checked="" type="checkbox"/> Congenital adrenal hyperplasia (CAH) (OS)	201910	CYP21A2	6p21.32
<input checked="" type="checkbox"/> Congenital diaphragmatic hernia (CDH)	142340	CHD2 NR2F2	15q26.1 15q26.2
<input checked="" type="checkbox"/> Congenital diaphragmatic hernia 2 (CDH2) *	222400	GATA4	8p23.1
<input type="checkbox"/> Cornelia de Lange	122470	NIPBL	5p13.2
<input checked="" type="checkbox"/> Cowden	158350	PTEN	10q23.31
<input checked="" type="checkbox"/> Cri-du-Chat	123450	(M)	5p15.2
<input checked="" type="checkbox"/> Currarino	176450	HLXB9	7q36.3
<input checked="" type="checkbox"/> Dandy-Walker malformation (DWM)	220200	ZIC1, ZIC4	3q24
<input checked="" type="checkbox"/> DiGeorge/Velocardiofacial (VCF) *	188400	HIRA, TBX1	22q11.21
<input checked="" type="checkbox"/> DiGeorge 2	601362	(M)	10p14
<input checked="" type="checkbox"/> Dosage-sensitive sex reversal *	300018	NROB1	Xp21.2
<input checked="" type="checkbox"/> Down syndrome critical region (DSCR) *	602917	(M)	21q22.13
<input checked="" type="checkbox"/> Familial adenomatous polyposis (FAP)/Gardner/MR	175100	APC	5q22.2
<input type="checkbox"/> Familial hypocalciuric hypercalcemia 1 (HHC1)	145980	CASR	3q21.1
<input checked="" type="checkbox"/> Feingold	164280	MYCN	2p24.3
<input type="checkbox"/> FG 5 *	300581	MID2	Xq22.3
<input checked="" type="checkbox"/> FMR1 Microdeletion	300624	FMR1	Xq27.3
<input checked="" type="checkbox"/> Frys	229850	DISP1 candidate	1q41
<input type="checkbox"/> Generalized epilepsy with febrile seizures plus 2 (GEFS+2)	604233	SCN1A	2q24.3
<input type="checkbox"/> Gitelman	263800	SLC12A3	16q13
<input checked="" type="checkbox"/> Glycerol kinase deficiency (GKD)	300474	GK	Xp21.2
<input checked="" type="checkbox"/> Greig cephalopolysyndactyly	175700	GLI3	7p14.1
<input type="checkbox"/> Holoprosencephaly 1	236100	TMEM1	21q22.3
<input checked="" type="checkbox"/> Holoprosencephaly 2	157170	SIX3	2p21
<input checked="" type="checkbox"/> Holoprosencephaly 3	142945	SHH	7q36.3
<input checked="" type="checkbox"/> Holoprosencephaly 4	142946	TGIF	18p11.31
<input checked="" type="checkbox"/> Holoprosencephaly 5	609637	ZIC2	13q32.3
<input type="checkbox"/> Holoprosencephaly 6	605934	(M)	2q37.1-q37.3
<input type="checkbox"/> Holoprosencephaly 7	610828	PTCH1	9q22.32
<input checked="" type="checkbox"/> Holoprosencephaly 8	609408	(M)	14q13.1-q13.2
<input type="checkbox"/> Holoprosencephaly and preaxial polydactyly *	605651	FBXW11	5q35.1

Symbol key



Microdeletions or microduplications have been associated with this condition



Microdeletions or microduplications are rare or not yet associated with this condition

(FISH)

\* microduplication syndrome available only through SignatureFISH™ testing

(M) multiple genes

(OS) based on clinical indication, SignatureChipOS™ oligonucleotide microarray testing may yield better results

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<input type="checkbox"/> Holt-Oram *	142900	TBX5	12q24.21
<input checked="" type="checkbox"/> Hypoparathyroidism, sensorineural deafness, renal disease (HDR)	146255	GATA3	10p14
<input checked="" type="checkbox"/> Jacobsen/11q terminal deletion disorder	147791	(M)	11q23-11qter
<input type="checkbox"/> Joubert 3	608629	AHI1	6q23.3
<input checked="" type="checkbox"/> Joubert 4	609583	NPHP1	2q13
<input type="checkbox"/> Joubert 5	610188	CEP290	12q21.32
<input checked="" type="checkbox"/> Juvenile polyposis syndrome (JPS), BMPR1A-related	174900	BMPR1A	10q23.2
<input checked="" type="checkbox"/> Juvenile polyposis syndrome (JPS), MADH4-related	174900	MADH4 (SMAD4)	18q21.2
<input checked="" type="checkbox"/> Kallmann 1	308700	KAL1	Xp22.31
<input checked="" type="checkbox"/> Langer-Giedion	150230	TRPS1 EXT1	8q23.3 8q24.11
<input checked="" type="checkbox"/> Langer mesomelic dysplasia (LMD) (OS)	249700	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
<input type="checkbox"/> Leber congenital amaurosis X (LCAX)	610142	CEP290	12q21.32
<input checked="" type="checkbox"/> Leri-Weill dyschondrosteosis (LWD) (OS)	127300	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
<input type="checkbox"/> Li-Fraumeni 1 (LFS)	151623	TP53	17p13.1
<input checked="" type="checkbox"/> Lissencephaly 1	607432	PAFAH1B1 (LIS1)	17p13.3
<input type="checkbox"/> Lissencephaly with cerebellar hypoplasia	257320	RELN	7q22.1
<input type="checkbox"/> Loeyes-Dietz (LDS), TGFBR1-related	609192	TGFBR1	9q22.33
<input type="checkbox"/> Loeyes-Dietz (LDS), TGFBR2-related	609192	TGFBR2	3p24.1
<input checked="" type="checkbox"/> Lowe	309000	OCRL	Xq25
<input type="checkbox"/> Macrocephaly-autism	605309	PTEN	10q23.31
<input checked="" type="checkbox"/> Marfan 1 (MFS1)	154700	FBN1	15q21.1
<input type="checkbox"/> Marfan 2 (MFS2)	154705	TGFBR2	3p24.1
<input type="checkbox"/> Meckel 4	611134	CEP290	12q21.32
<input checked="" type="checkbox"/> Microphthalmia 3	206900	SOX2	3q26.33
<input checked="" type="checkbox"/> Microphthalmia 7 with linear skin defects	309801	(M)	Xp22.2
<input checked="" type="checkbox"/> Miller-Dieker	247200	PAFAH1B1 (LIS1)	17p13.3
<input checked="" type="checkbox"/> Mohr-Tranebjaerg	304700	TIMM8A	Xq22.1
<input checked="" type="checkbox"/> Mowat-Wilson *	235730	ZFX1B	2q22.3
<input checked="" type="checkbox"/> Nablus mask-like facial	608156	(M)	8q21.3-q22.1
<input checked="" type="checkbox"/> Nail-patella (NPS)	161200	LMX1B	9q33.3
<input checked="" type="checkbox"/> Nephronophthisis 1	256100	NPHP1	2q13
<input type="checkbox"/> Neonatal severe primary hypoparathyroidism (NSHPT)	239200	CASR	3q21.1
<input checked="" type="checkbox"/> Neurofibromatosis 1 (NF1)/MR	162200	NF1	17q11.2
<input checked="" type="checkbox"/> Neurofibromatosis 2 (NF2)	101000	NF2	22q12.2
<input checked="" type="checkbox"/> NFIA Haploinsufficiency	600727	NFIA	1p31.3
<input type="checkbox"/> Noonan 1	163950	PTPN11	12q24.13
<input type="checkbox"/> Noonan 4	610733	SOS1	2p22.1
<input checked="" type="checkbox"/> Oculocutaneous albinism 2 (OCA2) * (OS)	203200	OCA2	15q13.1
<input checked="" type="checkbox"/> Okihiro	607323	SALL4	20q13.2
<input checked="" type="checkbox"/> Ornithine transcarbamylase deficiency (OTC)	311250	OTC	Xp11.4
<input type="checkbox"/> Oro-facio-digital 1 (OFD1)	311200	OFD1	Xp22.2
<input type="checkbox"/> Oto-facio-cervical (OFC)	166780	EYA1	8q13.3
<input checked="" type="checkbox"/> Pallister-Killian *	601803	(M)	12p
<input checked="" type="checkbox"/> Pelizaeus-Merzbacher *	312080	PLP1	Xq22.2
<input checked="" type="checkbox"/> Polycystic kidney disease 1 (PKD1) (OS)	601313	PKD1	16p13.3
<input checked="" type="checkbox"/> Potocki-Shaffer	601224	EXT2, ALX4	11p11.2
<input checked="" type="checkbox"/> Prader-Willi (PWS)*	176270	SNRPN	15q11.2
<input checked="" type="checkbox"/> Prader-Willi-like phenotype	176270	SIM1	6q16.3
<input type="checkbox"/> Proteus/Proteus-like	176920	PTEN	10q23.31
<input checked="" type="checkbox"/> PTEN hamartoma tumor	158350	PTEN	10q23.31

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<input checked="" type="checkbox"/> Renal cysts and diabetes (RCAD) *	137920	TCF2	17q12
<input checked="" type="checkbox"/> Retinoblastoma/MR	180200	RB1	13q14.2
<input checked="" type="checkbox"/> Rieger 1 (RIEG1)	180500	PITX2	4q25
<input checked="" type="checkbox"/> Rubinstein-Taybi (RTS)	180849	CREBBP	16p13.3
<input checked="" type="checkbox"/> Saethre-Chotzen	101400	TWIST1	7p21.1
<input type="checkbox"/> Schizencephaly	269160	EMX2	10q26.11
<input type="checkbox"/> Senior-Loken 6	610189	CEP290	12q21.32
<input checked="" type="checkbox"/> Severe myoclonic epilepsy of infancy (SMEI)	607208	SCN1A	2q24.3
<input type="checkbox"/> Short stature, pituitary and cerebellar defects, small sella turcica	606606	LHX4	1q25.2
<input type="checkbox"/> Smith-Lemli-Opitz (SLOS)	270400	DHCR7	11q13.4
<input checked="" type="checkbox"/> Smith-Magenis (SMS) *	182290	RAI1	17p11.2
<input checked="" type="checkbox"/> Sotos	117550	NSD1	5q35.3
<input checked="" type="checkbox"/> Split-hand/foot malformation 1 (SHFM1)	183600	SHFM1	7q21.3
<input checked="" type="checkbox"/> Split-hand/foot malformation 3 (SHFM3) *	600095	FBXW4	10q24.32
<input type="checkbox"/> Split-hand/foot malformation 4 (SHFM4)	605289	TP73L	3q28
<input checked="" type="checkbox"/> Split-hand/foot malformation 5 (SHFM5)	606708	DLX1, DLX2	2q31.1
<input checked="" type="checkbox"/> SRY dosage abnormalities	278850/ 306100	SRY	Yp11.31
<input checked="" type="checkbox"/> Steroid sulfatase deficiency	308100	STS	Xp22.31
<input type="checkbox"/> Stickler I	108300	COL2A1	12q13.11
<input checked="" type="checkbox"/> Synpolydactyly/Syndactyly II	186000	HOXD gene cluster	2q31.1
<input checked="" type="checkbox"/> Townes-Brocks 1	107480	SALL1	16q12.1
<input checked="" type="checkbox"/> Trichorhinophalangeal 1	190350	TRPS1	8q23.3
<input checked="" type="checkbox"/> Tuberous sclerosis 2 (TSC2) (OS)	191100	TSC2	16p13.3
<input checked="" type="checkbox"/> Ulnar-mammary	181450	TBX3	12q24.21
<input checked="" type="checkbox"/> Van der Woude	119300	IRF6	1q32.2
<input type="checkbox"/> Vascular endothelial growth factor (VEGFA)-related disorders	192240	VEGFA	6p21.1
<input checked="" type="checkbox"/> Waardenburg syndrome IIA	193510	MITF	3p14.1
<input checked="" type="checkbox"/> WAGR	194072	PAX6, WT1	11p13
<input checked="" type="checkbox"/> Williams-Beuren *	194050	ELN	7q11.23
<input checked="" type="checkbox"/> Wilms Tumor 1	194070	WT1	11p13
<input checked="" type="checkbox"/> Wolf-Hirschhorn	194190	(M)	4p16.3
<input checked="" type="checkbox"/> X-linked agammaglobulinemia	300300	BTK	Xq22.1
<input checked="" type="checkbox"/> X-linked heterotaxy	306955	ZIC3	Xq26.3
<input checked="" type="checkbox"/> X-linked idiopathic short stature (ISSX) (OS)	300582	SHOX	Xpter-Xp22.3 & Ypter-Yp11.32
<input type="checkbox"/> X-linked infantile spasms, ARX-related	308350	ARX	Xp21.3
<input checked="" type="checkbox"/> X-linked infantile spasms, CDKL5-related	300203	CDKL5	Xp22.13
<input type="checkbox"/> X-linked juvenile retinoschisis	312700	RS1	Xp22.13
<input type="checkbox"/> X-linked lissencephaly	300067	DCX	Xq22.3
<input type="checkbox"/> X-linked lissencephaly with ambiguous genitalia	300215	ARX	Xp21.3
<input checked="" type="checkbox"/> X-linked lymphoproliferative syndrome (XLP)	308240	SH2D1A	Xq25
<input checked="" type="checkbox"/> X-linked mental retardation 21	300143	IL1RAPL1	Xp21.3
<input type="checkbox"/> X-linked mental retardation 30	300558	PAK3	Xq22.3
<input type="checkbox"/> X-linked mental retardation 54	300419	ARX	Xp21.3
<input checked="" type="checkbox"/> X-linked mental retardation with isolated growth hormone deficiency *	300123	SOX3	Xq27.1
<input checked="" type="checkbox"/> XX male	278850	SRY	Yp11.31
<input checked="" type="checkbox"/> XY gonadal dysgenesis	306100	SRY	Yp11.31
<input type="checkbox"/> XY sex-reversal, +/- adrenal failure	184757	NR5A1	9q33.3
N/A All 41 unique subtelomeric regions		(M)	41 sites
N/A All 43 unique pericentromeric regions		(M)	43 sites
N/A Aneuploidy for 24 chromosomes		(M)	24 chromosomes

Disclaimer: The SignatureChipWG or SignatureChipOS may identify chromosome gains or losses associated with adult-onset conditions and/or cancer predisposition potentially unrelated to the current clinical findings, but that may become apparent later in life.