BCM
Baylor College of Medicine

Disorder Table

Regions tested by Baylor version 5.0 microarray

Condition	
1p36 deletion syndrome	Holoprosencephaly 4
2q22.3-q23.3 deletion syndrome	Holoprosencephaly 5
3q29 deletion syndrome	Holoprosencephaly 7 / basal cell nevus
7q11.23 duplication; reciprocal *WBS*	Hypoparathyroidism, sensorineural deafness, and renal
8p23 deletion/duplication syndrome	Jacobsen (11q25 deletion) syndrome
9q34.3 deletion syndrome	Kallmann syndrome 1
15q11.2-q12 duplication; reciprocal *PWS/AS*	Langer-Giedion syndrome
15q21 deletion syndrome	Leri-Weill dyschondrosteosis
17p11.2 duplication; reciprocal *SMS*	Leukodystrophy
22q11.2 duplication; reciprocal *DGS1*	Mental retardation X-linked with growth hormone def.
Xp22.31 duplication; reciprocal *STS*; unclear if benign	Micophthalmia with linear skin defects
Alagille syndrome	Miller-Dieker lissencephaly syndrome
Angelman syndrome (*AS*); see 15q11.2-q12 dup	Nail-patella syndrome
Aniridia II	Nephronophthisis
Autistic features	Neurofibromatosis I
Autistic features, X-linked, susceptibility to, 2	Neurofibromatosis 2
Basal cell nevus syndrome	Noonan syndrome
Beckwith-Wiedemann syndrome	Pelizaeus-Merzbacher disease
Bruton agammaglobulinemia	Polycystic kidney disease / tuberous sclerosis 2
Campomelic dysplasia	Potocki-Shaffer syndrome
Charcot-Marie-Tooth disease type 1A	Prader–Willi syndrome (*PWS*) see 15q11.2-q12 dup
Cat eye syndrome	Retinoblastoma
CHARGE syndrome	Rett syndrome
Cleidocranial dysplasia	Rieger syndrome, type 1
Congenital adrenal hypoplasia	Rubinstein-Taybi syndrome
Cornelia de Lange syndrome	Saethre-Chotzen syndrome
Cri-du-chat syndrome	Sex reversal X/Y translocations
Down syndrome c ritical region	Smith-Magenis syndrome (*SMS); see 17p11.2 dup*
Dandy-Walker syndrome	Sotos syndrome
DiGeorge syndrome 1 (*DGS1*), *VCFS* see 22q11.2 dup	Split-hand/foot malformation-3
DiGeorge syndrome 2	Steroid sulfatase deficiency (*STS*); see Xp22.31 dup
Dosage sensitive sex reversal	Trichorhinophalangeal syndrome I
Feingold syndrome	Tuberous sclerosis 2
Glycerol kinase deficiency	Tuberous sclerosis 1
Greig cephalo-polysyndactyly syndrome	WAGR syndrome
Hereditary neuropathy with pressure palsies	Williams-Beuren syndrome (*WBS*); see 7q11.23 dup
Hernia, congenital diaphragmatic	Wilms tumor
Holoprosencephaly 1	Wolf-Hirschhorn syndrome
Holoprosencephaly 2	X-linked heterotaxy
Holoprosencephaly 3	
All clinically relevant subtelomeric and pericentromeric regions	

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