SGD BambniTest

Non-Invasive Prenatal Testing of Single Gene Disorders

Screens for pathogenic/ likely pathogenic variants (SNVs, InDels < 3bp) for 66 autosomal dominant or X-linked disorders within 34 genes

Intellectual / Developmental Disabilities (22)

Disorders	Genes	Incident/ Prevalence
CHARGE syndrome	*CHD7*	*1-9/ 100 000*
Intellectual developmental disorder, autosomal dominant 5	*SYNGAP1*	--
Bainbridge-Ropers syndrome	*ASXL3*	*<1/ 1 000 000*
Coffin-Siris syndrome 1	*ARID1B*	*<1/ 1 000 000*
Rett syndrome	*MECP2*	*1-9/ 100 000*
Developmental delay with variable intellectual impairment and behavioral abnormalities	*TCF20*	--
Kleefstra syndrome 2	*KMT2C*	*<1/ 1 000 000*
Intellectual developmental disorder, autosomal dominant 52	*ASH1L*	--
Intellectual developmental disorder, autosomal dominant 7	*DYRK1A*	*<1/ 1 000 000*
Developmental and epileptic encephalopathy 4	*STXBP1*	*1-9/ 100 000*
White-Sutton syndrome	*POGZ*	*<1/ 1 000 000*
Developmental and epileptic encephalopathy 27	*GRIN2B*	*1-9/ 100 000*
Intellectual developmental disorder, autosomal dominant 6, with or without seizures	*GRIN2B*	--
Developmental and epileptic encephalopathy 2	*CDKL5*	*1-9/ 100 000*
Developmental and epileptic encephalopathy 94	*CHD2*	--
Menke-Hennekam syndrome 1	*CREBBP*	--
Rubinstein-Taybi syndrome 1	*CREBBP*	*1-9/ 100 000*
Helsmoortel-van der Aa syndrome	*ADNP*	*1-9/ 100 000*
Rett syndrome, congenital variant	*FOXG1*	--
Cornelia de Lange syndrome 1	*NIPBL*	*1-9/ 100 000*
Intellectual developmental disorder, autosomal dominant 43	*HIVEP2*	--
Mowat-Wilson syndrome	*ZEB2*	*1-9/ 100 000*

Structural Abnormalities / Others (44)

Disorders	Genes	Incident/ Prevalence
Developmental and epileptic encephalopathy 6B, non-Dravet	*SCN1A*	--
Dravet syndrome	*SCN1A*	*1-9/ 100 000*
Generalized epilepsy with febrile seizures plus, type 2/ Febrile seizures, familial, 3A	*SCN1A*	--
Migraine, familial hemiplegic, 3	*SCN1A*	*1-5/ 10 000*
Caffey disease	*COL1A1*	*3/ 1 000*
Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1	*COL1A1*	*<1/ 1 000 000*
Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2	*COL1A2*	
Osteogenesis imperfecta, type I	*COL1A1*	*1-5/ 10 000*
Osteogenesis imperfecta, type II	*COL1A1*	
	COL1A2	
Osteogenesis imperfecta, type III	*COL1A1*	
	COL1A2	
Osteogenesis imperfecta, type IV	*COL1A1*	
	COL1A2	
Ehlers-Danlos syndrome, arthrochalasia type, 1	*COL1A1*	--
Ehlers-Danlos syndrome, arthrochalasia type, 2	*COL1A2*	
Tuberous sclerosis 1	*TSC1*	*1-9/ 100 000*
Tuberous sclerosis 2	*TSC2*	
Achondroplasia	*FGFR3*	*1-9/ 100 000*
Crouzon syndrome with acanthosis nigricans	*FGFR3*	*1-9/ 1 000 000*
Hypochondroplasia	*FGFR3*	*1-9/ 100 000*
Muenke syndrome	*FGFR3*	*1-9/ 100 000*
Achondroplasia, severe, with developmental delay and acanthosis nigricans	*FGFR3*	*<1/ 1 000 000*
Thanatophoric dysplasia, type I	*FGFR3*	*1-9/ 100 000*
Thanatophoric dysplasia, type II	*FGFR3*	
Lacrimoauriculodentodigital syndrome 1	*FGFR2*	*<1/ 1 000 000*
Lacrimoauriculodentodigital syndrome 2	*FGFR3*	
Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	*FGFR2*	--
Apert syndrome	*FGFR2*	*1-9/ 100 000*
Beare-Stevenson cutis gyrata syndrome	*FGFR2*	*<1/ 1 000 000*
Bent bone dysplasia syndrome 1	*FGFR2*	*<1/ 1 000 000*
Pfeiffer syndrome/ Craniofacial-skeletal-dermatologic dysplasia	*FGFR2*	*1-9/ 100 000*
Crouzon syndrome	*FGFR2*	*1-9/ 1 000 000*
Jackson-Weiss syndrome	*FGFR2*	--
Saethre-Chotzen syndrome	*FGFR2*	*1-9/ 100 000*
Kabuki syndrome 1	*KMT2D*	*1-9/ 100 000*
LEOPARD syndrome 1	*PTPN11*	--
LEOPARD syndrome 2	*RAF1*	
LEOPARD syndrome 3	*BRAF*	
Noonan syndrome 1	*PTPN11*	*1-5/ 10 000*
Noonan syndrome 3	*KRAS*	
Noonan syndrome 4	*SOS1*	
Noonan syndrome 5	*RAF1*	
Noonan syndrome 7	*BRAF*	
Noonan syndrome 8	*RIT1*	
Cardiofaciocutaneous syndrome 1	*BRAF*	*1-9/ 1 000 000*
Cardiofaciocutaneous syndrome 2	*KRAS*	

All incident/ prevalence are obtained from Orphanet, MedlinePlus, and others.

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