Established Risk Conditions

Aase-Smith Syndrome (Diamond-Blackfan Anemia)	Aase Syndrome
Acrocallosal Syndrome	Acrodysostosis
Acro-Fronto-Facio-Nasal Dysostosis	Adrenoleukodystrophy
Agenesis of the Corpus Callosum	Agyria
Aicardi Syndrome	Alexander's Disease
Alper's Syndrome	Amelia
Angelman Syndrome	Aniridia
Anophthalmia/Microphthalmia	Antley-Bixler Syndrome
Apert Syndrome	Arachnoid cyst with neuro-developmental delay
Arhinencephaly	Arthogryposis
Ataxia	Atelosteogenesis
Autism	Baller-Gerold Syndrome
Bannayan-Riley-Ruvalcaba Syndrome	Bardet-Biedl Syndrome
Bartoscas-Papas Syndrome	Beals Syndrome (congenital contractual arachnodactyly)
Bixler Syndrome	Blackfan-Diamond Syndrome
Bobble Head Doll Syndrome	Borjeson-Forssman-Lehmann Syndrome
Brachial Plexopathy	Brancio-Oto-Renal (BOR) Syndrome
Campomelic Dysplasia	Canavan Disease
Carbohydrate Deficient Glycoprotein Syndrome	Cardio-Facio-Cutaneous Syndrome
Carpenter Syndrome	Cataracts-Congenital
Caudal Dysplasia	Cerebro-Costo-Mandibular Syndrome
Cerebellar Aplasis/Hypoplasia/Degeneration	Cerebral Atrophy
Cerebral Palsy	Cerebro-oculo-facial-skeletal syndrome
CHARGE Association	Chediak Higashi Syndrome
Chondrodysplasia Punctata	Christian Syndrome
Chromosome Abnormality a. Unbalanced numerical (autosomal) b. Numerical trisomy (chromosomes 1-22) c. Sex chromosomes XXX; XXXX; XXXXX; XXXY; XXXXY	CNS Aneurysm with Neuro-Developmental Delay
CNS Tumor with Neuro-Developmental Delay	Cockayne Syndrome
Coffin Lowry Syndrome	Coffin Siris Syndrome
Cohen Syndrome	Cone Dystrophy
Congenital Cytomegalovirus	Congenital Herpes
Congenital Rubella	Congenital Syphilis
Congenital Toxoplasmosis	Cortical Blindness
Costello Syndrome	Cri Du Chat Syndrome
Cryotophthalmos	Cutis Laxa
Cytochrome-c Oxidase Deficiency	Dandy Walker Syndrome
DeBarsy Syndrome	DeBoquois Syndrome
Dejerine-Sottas Syndrome	DeLange Syndrome
DeSanctis Cacchione Syndrome	Diastrophic Dysplasia
DiGeorge Syndrome	Distal Arthrogryrosis
Donohue Syndrome	Down Syndrome
Dubowitz Syndrome	Dyggve Melchor-Calusen Syndrome
Dyssegmental Dysplasia	Dystonia
EEC (Ectrodactyly-ectodermal dysplasia-clefting) Syndrome	Encephalocele
Encephalo-Cranio-Cutaneous Syndrome	Encephalomalacia

Established Risk Conditions

Facio-Auriculo-Radial Dysplasia	Facio-Cardio Renal (Eastman-Bixler) Syndrome
Familial Dysautonomia (Riley-Day Syndrome)	Fanconi Anemia
Farber Syndrome	Femoral Hypoplasia
Fetal Alcohol Syndrome/Effects	Fetal Dyskinesia
Fetal Hydantoin Syndrome	Fetal Valproate Syndrome
Fetal Varicella Syndrome	FG Syndrome
Fibrochondregenesis	Floating Harbor Syndrome
Fragile X Syndrome	Freeman-Sheldon (Whistling Facies) Syndrome
Fryns Syndrome	Fucosidosis
Galactosemia	Glaucoma-Congenital
Glutaric Aciduria Type I and II	Glycogen Storage Disease
Goldberg-Shprintzen Syndrome	Grebe Syndrome
Hallermann-Streiff Syndrome	Hays-Wells Syndrome
Head Trauma with Neurological Sequelae/Developmental Delay	Hearing Loss as determined by ABR audiometry or audiometric behavioral measurements; includes bilateral or unilateral loss
Hemimegalencephaly	Hemiplegia/Hemiparesis
Hemorrhage-Intraventricular Grade III and IV	Hereditary Sensory & Autonomic Neuropathy
Hereditary Sensory Motor Neuropathy (Charcot Marie Tooth Disease)	Herrmann Syndrome
Heterotopias	Holoprosencephaly (Aprosencephaly)
Holt-Oram Syndrome	Homocystinuria
Hunter Syndrome (MPS II)	Hurler Syndrome (MPS I)
Hyalinosis	Hydranencephaly
Hydrocephalus	Hyperpipecolic Acidema
Hypomelanosis of ITO	Hypophosphotasis-Infantile
Hypoxic Ischemic Encephalopathy	I-Cell (mucolpidosis II) Disease
Incontinentia Pigmenti	Infantile Spasms
Ininencephaly	Isovaleric Acidemia
Jarcho-Levin Syndrome	Jervell Syndrome
Johanson-Blizzard Syndrome	Joubert Syndrome
Kabuki Syndrome	KBG Syndrome
Kenny-Caffey Syndrome	Klee Blattschadel
Klippel-Feil Sequence	Landau-Kleffner Syndrome
Lange-Nielsen Syndrome	Langer Giedion Syndrome
Larsen Syndrome	Laurin-Sandrow Syndrome
Leber's Amaurosis	Legal Blindness (bilateral visual acuity of 20/200 or worse corrected vision in the better eye)
Leigh Disease	Lennox-Gastaut Syndrome
Lenz Majewski Syndrome	Lenz Microophthalmia Syndrome
Levy-Hollister (LADD) Syndrome	Lesch-Nyhan Syndrome
Leukodystrophy	Lissencephaly
Lowe Syndrome	Lowry-Maclean Syndrome
Maffucci Syndrome	Mannosidosis
Maple Syrup Urine Disease	Marden Walker Syndrome
Marshall Syndrome	Marshall-Smith Syndrome
Maroteaux-Lamy Syndrome	Maternal PKU Effects
Megalencephaly	MELAS
Meningocele (cervical)	MERRF
Metachromatic Leukodystrophy	Metatropic Dysplasia

Established Risk Conditions

Methylmalonic Acidemia	Microcephaly
Microtia-Bilateral	Midas Syndrome
Miller (postaxial acrofacial-dysostosis) Syndrome	Miller-Dieker Syndrome
Mitochondrial Disorder	Mobius Syndrome
Morquio Syndrome	Moya-Moya Disease
Mucolipidosis II and III	Multiple congenital anomalies (major organ birth defects)
Multiple Pterygium Syndrome	Muscular Dystrophy
Myasthenia Gravis-Congenital	Myelocystocele
Myopathy –Congenital	Myotonic Dystrophy
Nager (Acrofacial Dysostosis) Syndrome	Nance Horan Syndrome
NARP	Neonatal Meningitis/Encephalitis
Neuronal Ceroid Lipofuscinoses	Neuronal Migration Disorder
Nonketotic Hyperglycinemia	Noonan Syndrome
Ocular Albinism	Oculocerebrocutaneous Syndrome
Oculo-Cutaneous Albinism	Optic Atrophy
Optic Nerve Hypoplasia	Oral-Facial digital Syndrome, Types I-VII
Osteogenesis Imperfecta, Types III and IV	Osteopetrosis (Autosomal Recessive)
Oto-Palato-Digital Syndrome, Types I and II	Pachygyria
Pallister Mosaic Syndrome	Pallister-Hall Syndrome
Pelizaeus-Merzbacher Disease	Pendred's Syndrome
Periventricular Leukomalacia	Pervasive Developmental Disorder
Peters Anomaly	Phocomelia
Poland Sequence	Polymicrogyria
Popliteal Pterygium Syndrome	Porencephaly
Prader-Willi Syndrome	Progeria
Propionic Acidema	Proteus Syndrome
Pyruvate Carboxylase Deficiency	Pyruvate Dehydrogenase Deficiency
Radial Aplasia/Hypoplasia	Refsum Disease
Retinoblastoma	Retinoic Acid Embryopathy
Retinopathy of Prematurity, Stages III and IV	Rett Syndrome
Rickets	Rieger Syndrome
Roberts SC Phocomelia	Robinow Syndrome
Rubinstein-Taybin Syndrome	Sanfilippo Syndrome (MPS III)
Schinzel-Giedion Syndrome	Schimmelpenning Syndrome (Epidermal Nevus Syndrome)
Schizencephaly	Schwartz-Jampel Syndrome
Seckel Syndrome	Septo-Optic Dysplasis
Severe Attachment Disorder	Shaken Baby Syndrome
Short Syndrome	Sialidosis
Simpson-Golabi-Behmel Syndrome	Sly Syndrome (MPS IV)
Smith-Fineman-Myers Syndrome	Smith_Limitz-Opitz Syndrome
Smith-Magenis Syndrome	Sotos Syndrome
Spina Bifida (Meningomyelocele)	Spinal Muscular Atrophy
Spondyloepiphyseal Dysplasia Congenita	Spondylometaphyseal Dysplasia
Stroke	Sturge-Weber Syndrome
TAR (Thrombocytopenia-Absent Radii Syndrome)	Thanatophoric Dysplasia
Tibial Aplasis (Hypoplasia)	Toriello-Carey Syndrome
Townes-Brocks Syndrome	Trecher-Collins Syndrome
Trisomy 13	Trisomy 18
Tuberous Sclerosis	Urea Cycle Defect

Established Risk Conditions

Valocardiofacial Syndrome	Wildervanck Syndrome
Walker-Warburg Syndrome	Weaver Syndrome
Wiedemann-Rautenstrauch Syndrome	Williams Syndrome
Winchester Syndrome	Wolf Hirschhorn Syndrome
Yunis-Varon Syndrome	Zellweger Syndrome