

Medical Genetics of Nevada LLC

Susanna Sorrentino, MD, FACMG, FAAP 2538 Anthem Village Drive, Suite 110 Henderson, Nevada 89052 Phone: 702-732-6800 Fax: 702-932-9611

When Does a Child Need a Genetics Consult? (Not an exhaustive list)

-Autism

-Birth Defects

-Blindness/Unusual presentation for age e.g., cataracts, coloboma, glaucoma, optic nerve atrophy

-Brain anomalies

-Cancer-if rare, multiple types in the same child, unusually aggressive, family history

-Carrier of a genetic condition

-Chromosomal Abnormality

-Congenital heart disease

-Cardiomyopathy, Arrythmia

-Degenerative symptoms

-Dilated/dissected thoracic aorta

-Failure to thrive/poor growth

-Hearing loss

-Intellectual disability/Learning Disability

-In utero exposure to alcohol, illicit drugs, tobacco, medications

-Metabolic Disorder/Inborn Error of Metabolism e.g., PKU, Biotinadase Deficiency etc.

-Newborn Screen-positive

-Precocious or Delayed Puberty

-Psychiatric disease (severe)

-Recognizable/Already diagnosed genetic disorder/suspicion for a genetic disorder e.g., Down Syndrome, Turner Syndrome etc.

-Seizures

-Tall Stature

-Short Stature/Skeletal Dysplasia

Family history of a genetic disorder or family history appears to indicate a genetic disorder. Some examples include: -Birth defects/Spina Bifida -Blindness -Cancer, especially multiple family members, early onset (less than 50 years of age), unusual or rare cancers, multiple cancer clusters (e.g., colon, breast, ovarian) -Cleft lip/palate -Congenital heart disease -Consanguinity -Hemophilia -Huntington's Disease -Infant or childhood death -Intellectual Disability -Muscular Dystrophy -Polycystic Kidney Disease -Sickle Cell Disease/Trait/Variant

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