

## Indications for Referral for Genetic Services:

### Infancy and Childhood

- One or more major congenital malformations (*cleft lip/palate, omphalocele, neural tube defect, etc.*)
- Multiple minor anomalies forming a potentially recognizable syndrome or unknown pattern of malformations.
- Unexplained hypotonia, especially in the neonatal period
- Suspected metabolic disorder
- Mental retardation, developmental delay, autism or loss in milestones without obvious etiology
- Cardiomyopathy of unknown etiology
- Abnormal newborn screening results
- Significant hearing or vision impairment of unknown etiology
- Unusual dermatologic conditions (*e.g. pigmentary dysplasia, ichthyosis, bullous disorders, unusual birthmarks, multiple hemangiomas, unusual scarring, skin tumors*)
- Tumor or malignancy with a suspected genetic predisposition (*e.g. retinoblastoma, Wilms tumor, optic glioma, adrenal carcinoma*)
- Family history of a known or suspected genetic disorder, birth defect, or chromosomal abnormality
- Abnormalities in growth (*e.g. short or tall stature inconsistent with family height, asymmetry, macrosomia, disproportionate*)
- Ambiguous genitalia or other significant genital abnormalities
- Known or suspected genetic/inherited disorder or chromosomal abnormality or family history of such (*e.g. muscular dystrophy, hemophilia, Down syndrome*)
- Newborns with prenatally suspected fetal abnormalities or growth retardation
- Stillborn or deceased infants with known or suspected external or internal abnormalities (*e.g. hydrops, fetal growth retardation, malformations, dwarfism, renal dysgenesis/agenesis, histologic findings suggestive of metabolic disease*)



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