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. pigmented 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/inherit 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)