Indications for Referral for Genetic Services: <u>Adolescent / Adult</u>

- One or more major malformations
- Mental retardation without a definite etiology
- Visual impairment other than simple refractive errors or strabismus *(e.g. optic atrophy, rod/cone dystrophy, retinitis pigmentosa, coloboma, retinal detachment, dislocated lenses)*
- Unusual dermatologic conditions *(e.g pigmentary dysplasia, ichthyosis, bullous disorders, unusual birthmarks, hemangiomas, unusual scarring, multiple skin tumors)*
- Significant early childhood or adolescent onset hearing impairment
- Development of a degenerative disease
- A personal or family history of cancer with a known or suspected genetic predisposition *(e.g. ovarian, early-onset breast, colon, prostate, endometrial, retinoblastoma)* or multiple individual family members with cancer
- Personal or family history of a known or suspected genetic disorder *(e.g. cystic fibrosis, adult polycystic kidney disease, Marfan syndrome, myotonic dystrophy, Huntington disease, Charcot-Marie-Tooth disease, alpha-1-antitrypsin deficiency, hemochromatosis, familial hypercholesterolemia, cardiomyopathy)*
- Personal or significant family history of a common disease with possible genetic susceptibility *(e.g. early onset thrombotic events, early onset coronary artery disease, Metabolic syndrome, noninsulin-dependent diabetes in 1st degree relative or multiple family members, early-onset of or multiple close family members with Alzheimer disease)*
- Risk assessment for pregnancy planning: use <u>Preconception/Prenatal</u> referral recommendations for any adult patient of reproductive age – especially those actively considering a pregnancy

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