Pediatric Genetics Referrals: When to Refer to Genetics

- Genetic risk assessment and testing can be helpful in aiding in the care of individuals with a personal or family history of genetic conditions.

- Some genetic conditions are inherited through either males or females, while others can be inherited through both males and females.

- Those with an unknown or limited family history can still be referred to genetics for evaluation.

- Genetic test results can:
  - Impact treatment and management recommendations
  - Inform unaffected individuals of possible future risks to themselves or future children
  - Guide appropriate screenings

General Referral Guidelines for Pediatric Indications

A genetics evaluation should be considered for individuals with any of the following:

- A known genetic pathogenic variant/mutation identified in an individual or family member

- A known or suspected genetic disorder, birth defect, or chromosomal abnormality

- A newborn with any of the following:
  - Abnormal newborn screening test result
  - Congenital hypotonia or hypertonia
  - Unexplained intrauterine growth retardation

- A newborn, infant, or child with any of the following:
  - A single major anomaly, or multiple major and/or minor anomalies
  - Dysmorphic features that are not familial
  - Failure to thrive
  - A known metabolic disorder or symptoms of a metabolic disorder (Note: A normal newborn screening result does not rule out all metabolic disorders)
  - Abnormal brain MRI findings
  - Unusual growth patterns
  - Evidence of a connective tissue disorder
  - Congenital eye defects or blindness
  - Significant hearing loss or deafness
  - Cardiomyopathy not secondary to a viral infection
  - > 6 café-au-lait macules at least 0.5 cm in diameter
  - Unusual skin findings such as multiple types of lesions, multiple lipomas, numerous hypo- or hyperpigmented lesions, or albinism
  - Bilateral or multifocal malignancies

- A child with:
  - Developmental delay
  - Intellectual disability
  - Autism Spectrum Disorders
  - Immunodeficiency
  - Progressive muscle weakness

*Full referral guidelines are referenced on the back of this handout. If you are concerned about a patient’s personal or family history, a genetics professional can help determine if further risk assessment and/or genetic testing is indicated.*
Take Action

If your patient meets any of these criteria:

- Talk to your patient and/or your patient’s family about recommendations for a genetics referral
- Emphasize the importance of making and keeping an appointment with a genetics provider
- For assistance locating the nearest genetics service provider, in the New York-Mid-Atlantic Consortium (NYMAC) Region, please contact the Genetic Services Referral Call Center at 1-833-545-3218 or visit our website.

Guidelines and Recommendations (Links)

The Professional Practice and Guidelines Committee of the American College of Medical Genetics and Genomics (ACMG) Indication for genetic referral: a guide for healthcare providers

American Academy of Pediatrics—Genetics in Primary Care Institute, When to Refer

Resources (Links)

- National Society of Genetic Counselors (NSGC) Find a Genetic Counselor
  A tool developed by NSGC for patients and providers to locate genetic counseling services in North America (U.S. and Canada)
- Baby’s First Test
  A website providing information about newborn screening and resources for families caring for a child with a genetic condition
- GeneReviews through the NIH
  Resource for providers about the diagnosis and management of patients with genetic conditions

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