MEDICAL GENETICS REFERRALS GUIDELINES

SUSPECTED GENETIC SYNDROME

1. Background
    a. Genetics is a consultative service accepting referrals for pediatric and adult patients for diagnostic evaluation for a suspected genetic disorder including consideration of genetic testing, or for genetic counseling for a personal or family history of a genetic disorder.
2. Pre-referral evaluation and treatment
    a. Please obtain relevant family history. See additional referral notes below.
3. Indications for referral
    a. Multiple congenital anomalies
        i. Referral notes:
            1. Please provide prior diagnostic testing including relevant labs and imaging in the chart or fax to the Genetics clinic 408-885-3079.
    b. Family history of known genetic disorder
        i. Referral notes:
            1. Please document the specific genetic disorder suspected.
            2. Please obtain family records of specific diagnosis including prior genetic testing if available
    c. Hearing loss
        i. Referral notes:
            1. Do not refer unless audiogram is available in the chart or faxed to the Genetics clinic 408-885-3079
    d. Heritable cardiomyopathy or heritable arrhythmia suspected
        i. Referral notes:
            1. Do not refer unless records including ECG, echocardiogram are in the chart or faxed to the Genetics clinic 408-885-3079.
    e. Marfan syndrome suspected
        i. Referral notes:
            1. Please obtain Ophthalmology and Cardiology evaluation with echocardiogram prior to referral as this is a major part of the evaluation for this condition
    f. Neurofibromatosis type 1 suspected
        i. Referral notes:
            1. Six or more café au lait macules >5 mm in greatest diameter in prepubertal individuals and >15 mm in greatest diameter in post-pubertal individuals, please refer

2. If suspected neurofibromatosis and the patient has less than 6 café au lait macules and no other findings, please contact genetics to discuss

g. Unexplained overgrowth/failure to thrive/microcephaly/macrocephaly
    i. Referral notes:
        1. Obtain Cytogenomic SNP microarray; LAB10371 (CPT code 81229) prior to referring
        2. Insurance authorization for this test should be obtain prior to ordering the test
        3. See section on autism/intellectual disability/developmental delay below if applicable
        4. Please provide developmental assessment in the chart or fax to the Genetics clinic 408-885-3079.

h. Pediatric cancer predisposition syndrome suspected
    i. Referral notes:
        1. For adult referrals, please refer to Genetics-Oncology
        2. Obtain detailed family history
        3. Please obtain family records of specific diagnosis including prior genetic testing if available and document in chart or fax to the Genetics clinic 408-885-3079
        4. Consider referring adult affected family members first to Cancer Genetics if appropriate

i. Other suspected genetic syndrome
    i. Referral notes:
        1. Please document the specific syndrome suspected. If not known, please contact genetics prior to referral.

REFERRALS NOT ACCEPTED

1. Inborn error of metabolism known or suspected
    a. Please refer to the Stanford LPCH Metabolic Center
2. Cancer predisposition syndrome known or suspected in an adult
    a. Please refer to Genetics-Oncology (see separate Cancer Genetics referral guidelines)
3. Hypermobility type Ehlers Danlos Syndrome
    a. Please see the 2017 international diagnostic criteria for hEDS for a clinical diagnosis (https://www.ehlers-danlos.com/2017-eds-international-classification/)
    b. Referrals for other suspected monogenic forms of Ehlers Danlos Syndrome are accepted

AUTISM/INTELLECTUAL DISABILITY/DEVELOPMENTAL DELAY

1. Background
    a. Evaluation for syndromic and monogenetic causes of autism, intellectual disability, or developmental day.
    b. A genetic etiology may be identified in approximately 20-25% of cases
2. Pre-referral evaluation and treatment
    a. Testing: Please obtain 1st-tier testing prior to referring to Genetics
    b. 1st-tier testing includes:
        i. Cytogenomic SNP microarray; LAB10371 (CPT code 81229)
        ii. Fragile X (FMR1) w/Reflex to Methylation; LAB8172 (CPT code 81243; if reflexed, add 81244)
        iii. For patients with Valley Health Plan, no prior authorization is required for a diagnosis of autism, developmental delay, or intellectual disability. For all other health plans, prior authorization should be obtained before ordering the test.
3. Indications for referral
    a. Abnormal results on Fragile X or microarray testing
    b. Severe intellectual disability or absent speech
    c. Episodic neurologic findings
    d. Macrocephaly/microcephaly
    e. Congenital anomalies or dysmorphic features
    f. Family history of multiple affected relatives
    g. Any other concerns, please contact genetics to discuss the referral
    Please send records of developmental evaluation to genetics fax 408-885-3079 as part of referral process or indicate where records are located at time of referral.

CHROMOSOME ABNORMALITY

1. Background
2. Pre-referral evaluation and treatment
3. Indications for referral
    a. Do not refer until chromosome report or microarray report is scanned to chart or faxed to the Genetics clinic 408-885-3079.