RubiconMD

Using eConsults to Assess Genetic Risks

eConsults are an efficient way to get genetics-related insights and address patient questions and concerns without a referral to a genetic counselor or clinic.

In a genetics setting, eConsults can be used to address scenarios when:

Patient family history

Your patient has a family history of problems that might place them at risk for a genetic condition

Concern about a genetic disorder

You or your patient are concerned that they may currently or potentially have a genetic disorder

Guidance on the necessity of testing

You need guidance around determining whether genetic testing is necessary given a patient's individual history, presentation, and family history

Genetic test result support

Your patient has received a genetic test result and you need to provide guidance, follow-up recommendations, additional testing, or surveillance

To get the most out of a genetics-oriented eConsult, include:

- Any personal and/or family history of features or conditions related to your eConsult questions
- For hereditary cancer questions, the site of origin, age at diagnosis, and relationship to the patient for each affected relative
- For hereditary cardiac conditions, known cardiac conditions or events (such as arrhythmias, cardiomyopathy, congenital heart disease, high cholesterol, sudden cardiac death), age at diagnosis, and relationship to the patient for each affected relative
- Reproductive history, especially infertility or multiple pregnancy losses for the patient or their family members
- Any abnormal physical features or congenital defects
- Any genetic test result for the patient or family member that pertains to the eConsult
- Any imaging, pathology, or other results related to the eConsult

Examples of primary care eConsult submissions for genetics:

37 y/o female with a strong family history of cancer: maternal grandfather with colon cancer at 82, maternal grandmother with pancreatic cancer at 81, non-smoking maternal aunt with lung cancer in 40s, and father with prostate cancer at 66.

Question: Is there a genetics referral screening tool I can use? Would you recommend genetic testing for this patient?

47 y/o male with history of PKU but stopped diet after age 10. He maintained activity until 20s with worsening contractures involving upper and lower extremity.

Question: Can you give me some advice to minimize peripheral neuropathies in patients with untreated genetic disorders?

45 y/o female with history of malodorous skin over the last several years.

Question: What are some alternative testing options for diagnosis of trimethylaminuria and possible approaches to symptom management?

28 day old male baby born to 38 y/o female at full term via repeat c-section. Three siblings normal health and development. Prenatal complications included preeclampsia and gestational diabetes. Preterm labs found child to potentially have XYY Syndrome. This has been confirmed with karyotype. Child is having difficulty with breast feeding/weight gain but otherwise appears normal phenotypically. He has regained back to appropriate curve.

Question: Is there any connection with feeding difficulty/failure to thrive in infants with XYY?

46 y/o female has a family hx of ruptured aortic root. It has affected her dad, 2 paternal uncles, paternal grandmother, paternal great aunt. She has requested an echo for evaluation. States the men in her family are 6'5". Her son is 6'6". She has a hx of 3 spontaneous pneumothorax. She is otherwise healthy, normal VS, BMI normal.

Question: Do I need to check for Marfan's syndrome with genetic testing?

34 y/o male with family history of hypertrophic cardiomyopathy (maternal uncle) with newborn son. Patient is healthy and active without cardiac symptoms.

Question: Can you give me advice on possible genetic components of this disease and the need for any testing or screening for patient and patient's son?

69 y/o female with history of seizures, well controlled, who's daughter was recently diagnosed with LQTS.

Question: Can you advise on the possible connection between seizures and LQTS and the utility of genetic testing for this patient?

35 y/o female with postpartum stroke found to have carotid dissection bilaterally. Patient can place both palms the ground in forward fold position and has a 4 cm skin stretch on forearm, otherwise no past medical history or symptoms.

Question: How can I optimize the workup to evaluate for Ehlers-Danlos Syndrome or other genetic disorder predisposing patient to fragile vascular vessels?