VENTURA COUNTY HEALTH CARE PLAN

Medical Policy: Universal Carrier Screening Testing Specific to Parents

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Universal Carrier Screening Testing Specific to Parents

Purpose:

VCHCP provides carrier screening to members identified to be at risk of having offspring with inherited recessive single-gene disorders. Carriers are usually not at risk of developing the disease but can pass pathogenic variants to their children. There are different types of Carrier testing that may be performed in prenatal or preconception periods.

Methodology:

Types of Prenatal Genetic carrier screening include the following:
-A blood test that screens for genetic variations (mutations) in parents that could potentially lead to inherited disorders in their children.
-A saliva or cheek swabs can also be used.
Test results indicate whether the individual is a carrier and can assist families with making informed decisions about family planning.

Clinical Indications:

VCHCP follows the American College of Obstetricians and Gynecologists (ACOG) and American College of Medical Genetics and Genomics Recommendations (ACMG) on targeted risk-based screening. *(1)*

Cystic Fibrosis (CF)-CFTR Gene Analysis

- Cystic fibrosis carrier screening should be offered to women considering pregnancy or are pregnant who fulfill the following:
- For couples in which both partners are unaffected but one or both has a family history of cystic fibrosis, genetic counseling and medical record review should be performed to determine if CFTR mutation analysis in the affected family member is available. CFTR gene analysis should be offered. Routine screening absent a family history is not advisable.
- If a woman's reproductive partner has cystic fibrosis or apparently isolated congenital bilateral absence of the vas deferens, the couple should be provided follow-up genetic counseling by an obstetrician–gynecologist or other health care provider with expertise in genetics for mutation analysis and consultation.
  [Click here for URL] *(1)*

Fragile X Syndrome

- Fragile X premutation carrier screening is recommended for women with a family history of fragile X-related disorders or intellectual disability suggestive of fragile X syndrome and who are considering pregnancy or are currently pregnant.
- If a woman has unexplained ovarian insufficiency or failure or an elevated follicle-stimulating hormone level before age 40 years, fragile X carrier screening is recommended to determine whether she has an FMR1 premutation.
- All identified individuals with intermediate results and carriers of a fragile X premutation or full mutation should be provided follow-up genetic counseling to discuss the risk to their offspring of inheriting an expanded full-mutation fragile X allele and to discuss fragile X-associated disorders (premature ovarian insufficiency and fragile X tremor/ataxia syndrome).
- Prenatal diagnostic testing for fragile X syndrome should be offered to known carriers of the fragile X premutation or full mutation.
- DNA-based molecular analysis (eg, Southern blot analysis and polymerase chain reaction) is the preferred method of diagnosis of fragile X syndrome and of determining FMR1 triplet repeat number (eg, premutations).

In rare cases, the size of the triplet repeat and the methylation status do not correlate, which makes it difficult to predict the clinical phenotype. In cases of this discordance, the patient should be for genetic counseling.

Hemoglobinopathies (e.g. Sickle Cell Disease; Thalassemia)

- Any woman who is currently pregnant should receive a complete blood count with red blood cell indices to assess not only their risk of anemia but also to allow assessment for risk of a hemoglobinopathy. Ideally, this testing also should be offered to women before pregnancy.
- If a red blood cell indices indicate a low mean corpuscular hemoglobin or mean corpuscular volume, hemoglobin electrophoresis also should be performed.
- A hemoglobin electrophoresis should be performed in addition to a complete blood count if there is suspicion of hemoglobinopathy based on ethnicity (African, Mediterranean, Middle Eastern, Southeast Asian, or West Indian descent).

Spinal Muscular Atrophy (SMA)

- Screening for spinal muscular atrophy should be offered to all women considering pregnancy or are pregnant.
- In members with a family history of spinal muscular atrophy, molecular testing reports of the affected individual and carrier testing of the related parent should be reviewed, if possible, before testing is offered. If the reports are not available, SMN1 deletion testing should be recommended for the low-risk partner.
- Because spinal muscular atrophy is present in all populations, carrier testing should be offered to all couples regardless of race or ethnicity.

Tay–Sachs Disease

- Screening for Tay-Sachs Disease should be offered to all women considering pregnancy or are pregnant if either member of a couple is of Ashkenazi Jewish, French Canadian, or Cajun descent.
- In members with family history of consistent with Tay–Sachs disease also should be offered screening

Coding:

Below is a table that lists services that are covered based on the medically necessary criteria for carrier testing:

Coverage Criteria Sections	Example Tests (Labs)	Common CPT Codes	Common ICD-10 Codes
Cystic Fibrosis Carrier Screening	CFTR Common Mutation Panel	81220	O09, Z13, Z31, Z34, Z36, Z84
Fragile X Syndrome Carrier Screening	FMR1 Repeat Analysis	81243, 81244	O09, Z13, Z31, Z34, Z36, Z84
Hemoglobinopathies	hemoglobin electrophoresis or HPLC, CBC, Blood smear	81257; 81259; 81361; 83020	Z13.71, D58.2, Z13.0
Sickle Cell Disease-Thalassemia	hemoglobin electrophoresis or high-performance liquid chromatography (HPLC)	81257; 81259; 81361	Z13.0; Z13.71, D57.3
Spinal Muscular Atrophy Carrier Screening	SMN1 Deletion/Duplication Analysis	81329, 81336, 81337	O09, Z13, Z31, Z34, Z36, Z84

	SMN2 Deletion/Duplication Analysis		
Tay–Sachs Disease	enzyme analysis and gene sequencing	81255; 81443; 81406	Z13.71; Z14.8, Z31.430; Z31.440

References:

(1) *American College of Obstetricians and Gynecologists (ACOG).* [2017, March 17(reaffirmed 2020)]. Carrier Screening for Genetic Conditions. Committee Opinion No. 691. https://www.acog.org/clinical/clinical-guidance/committee-opinion/articles/2017/03/carrier-screening-for-genetic-conditions

A. Attachments: None

B. History:

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