Familial Hypercholesterolemia (FH) Panels

I. Genetic testing for familial hypercholesterolemia (FH) via multigene panel to establish or confirm a diagnosis of familial hypercholesterolemia (FH) is considered **medically necessary** when:

A. The member has at least two or more elevated LDL-C measurements, including assessment after intensive lifestyle modification, **AND**

B. There is no apparent secondary cause of hypercholesterolemia (e.g., hypothyroidism, diabetes, renal disease, nephrotic syndrome, liver disease, medications), **AND**

1. The member is a child with LDL-C levels greater than or equal to 190 mg/dl, **OR**

2. The member is a child with LDL-C levels greater than or equal to 160 mg/dl with one of the following:

a) At least one first-degree relative with elevated LDL-C, **OR**

b) At least one first-degree relative with premature coronary artery disease (CAD), **OR**

c) Limited family history (e.g., adoption), **OR**

d) A family history of both hypercholesterolemia and premature coronary artery disease (CAD), **OR**

3. The member is an adult with LDL-C levels greater than or equal to 250 mg/dl, **OR**

4. The member is an adult with LDL-C levels greater than or equal to 190 mg/dl with one of the following:

a) At least one first-degree relative with elevated LDL-C, **OR**

b) At least one first-degree relative with premature coronary artery disease (CAD), **OR**

c) Limited family history (e.g. adoption), **OR**

5. The member is an adult with LDL-C levels greater than or equal to 160 mg/dl with one of the following:

    a) A family history of both hypercholesterolemia and premature coronary artery disease (CAD), **OR**

    b) A personal history of premature coronary artery disease (CAD), **OR**

C. The member is an adult with premature coronary artery disease (CAD), **AND**

1. A family history of both hypercholesterolemia and premature coronary artery disease (CAD).

II. Genetic testing for familial hypercholesterolemia (FH) via multigene panel to establish or confirm a diagnosis of familial hypercholesterolemia (FH) is considered **investigational** for all other indications.

DEFINITIONS

1. **Close relatives** include first, second, and third degree blood relatives:

    a. **First-degree relatives** are parents, siblings, and children

    b. **Second-degree relatives** are grandparents, aunts, uncles, nieces, nephews, grandchildren, and half siblings

    c. **Third-degree relatives** are great grandparents, great aunts, great uncles, great grandchildren, and first cousins

2. **Premature coronary artery disease (CAD)** is defined as male subjects at or under 55 years of age, female subjects at or under 65 years of age; adapted from the American Heart Association phenotype definition of HeFH (Sturm, et al).

REFERENCES

1. Musunuru K, Hershberger RE, Day SM, Klinedinst NJ, Landstrom AP, Parikh VN, Prakash S, Semsarian C, Sturm AC; American Heart Association Council on Genomic and Precision Medicine; Council on Arteriosclerosis, Thrombosis and Vascular Biology; Council on Cardiovascular and Stroke Nursing; and Council on Clinical Cardiology. Genetic Testing for Inherited Cardiovascular Diseases: A Scientific Statement From the American Heart Association. Circ Genom Precis Med. 2020 Aug;13(4):e000067. doi: 10.1161/HCG.0000000000000067. Epub 2020 Jul 23. PMID: 32698598.
2. Sturm, A, Knowles, J, Gidding, S. et al. Clinical Genetic Testing for Familial Hypercholesterolemia: JACC Scientific Expert Panel. J Am Coll Cardiol. 2018 Aug, 72 (6) 662–680. https://doi.org/10.1016/j.jacc.2018.05.044