Genetic Testing for Congenital Abnormality

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Approved By:	Highmark Health Options – Market Leadership
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Products:	Medicaid
Application:	All participating hospital and providers
Page Number(s):	1 of 4

Disclaimer

Highmark Health Options medical policy is intended to serve only as a general reference resource regarding coverage for the services described. This policy does not constitute medical advice and is not intended to govern or otherwise influence medical decisions.

POLICY STATEMENT

Highmark Health Options may provide coverage under medical benefits of the Company's Medicaid products for medically necessary genetic testing for congenital abnormality procedures.

This policy is designed to address medical necessity guidelines that are appropriate for the majority of individuals with a particular disease, illness, or condition. Each person's unique clinical circumstances warrant individual consideration, based upon review of applicable medical records.

The qualifications of the policy will meet the standards of the National Committee for Quality Assurance (NCQA) and the Delaware Department of Health and Social Services (DHSS) and all applicable state and federal regulations.

DEFINITIONS

Highmark Health Options (HHO) – Managed care organization serving vulnerable populations that have complex needs and qualify for Medicaid. Highmark Health Options members include individuals and families with low income, expecting mothers, children, and people with disabilities. Members pay nothing to very little for their health coverage. Highmark Health Options currently services Delaware Medicaid: Delaware Healthy Children Program (DHCP) and Diamond State Health Plan Plus members.

Congenital Anomalies – Structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.

Genetic Testing – The analysis of chromosomes, deoxyribonucleic acid (DNA), ribonucleic acid (RNA), genes or gene products to detect inherited (germline) or non-inherited (somatic) genetic variants related to disease or health.

Genetic Counseling – Provides information about how genetic conditions, birth defects and other medical conditions run in families, and how they can affect the pregnant mother and baby's health.

PROCEDURES

Prior authorization is required.

Genetic testing may be considered medically necessary to screen for inherited metabolic diseases such as:

METABOLIC DISORDERS

- Traditional Disorders
  - CH: Congenital Hypothyroidism
  - CAH: Congenital Adrenal Hyperplasia
  - GAL: Galactosemia
  - HGB: Hemoglobinopathies
    - SS Disease
    - SC Disease
    - Variant Hgb
  - BIOT: Biotinidase Deficiency
  - CF: Cystic Fibrosis
- Amino Acid/Urea Cycle Disorders (MS/MS)
  - PKU: Phenylketonuria
  - HPHE: Hyperphenylalanemia
  - MSUD: Maple Syrup Urine Disease
  - HCYS: Homocystinuria
  - HMET: Hypermethioninemia
  - TYR: Tyrosinemia, Type I
  - TYR: Tyrosinemia, Type II
  - TYR: Tyrosinemia, Type III
  - ARG: Argininemia
  - ASL: Argininosuccinate Lyase Deficiency
  - CIT: Argininosuccinate Synthetase Deficiency (Citrullinemia)
- Organic Acid Disorders (MS/MS)
  - GA-1: Glutaric Acidemia, Type I
  - PA : Proprionic Acidemia
  - MMA: Methylmalonic Acidemia
  - MCD: Multiple Carboxylase Deficiency
  - IVA : Isovaleric Acidemia
  - 2-MBCD: 2-Methylbutyryl-CoA Dehydrogenase Deficiency
  - 3-MCC: 3-Methylcrotonyl-CoA Carboxylase Deficiency
  - HMG: 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency
  - BKT: Mitochondrial Acetoacetyl-CoA Thiolase Deficiency
  - IBCD: Isobutyryl-CoA Dehydrogenase Deficiency
- Fatty Acid Oxidation Disorders (MS/MS)
  - MCAD: Medium Chain Acyl-CoA Dehydrogenase Deficiency
  - CPT II: Carnitine Palmitoyltransferase II Deficiency
  - CAT: Carnitine/Acylcarnitine Translocase Deficiency
  - GA II: Glutaric Acidemia, Type II
  - MADD: Multiple Acyl-CoA Dehydrogenase Deficiency
  - SCAD: Short-Chain Acyl-CoA Dehydrogenase Deficiency
  - LCHAD: Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency
  - TFP: Trifunctional Protein Deficiency
  - VLCAD: Very Long-Chain Acyl-CoA Dehydrogenase Deficiency
  - CUD: Carnitine Uptake Deficiency

- Other
  - NH: Newborn Hearing Screening
  - Severe Combined Immunodeficiency
  - CCHD: Critical Congenital Heart Disease (January 2013)

Genetic counseling is generally provided in conjunction with genetic testing. Counseling usually occurs when the results of the tests are provided to the patient and intervention strategies are discussed. Coverage for genetic counseling is determined according to individual or group customer benefits. When genetic testing is noncovered, the counseling performed in conjunction with the testing is also noncovered.

PLACE OF SERVICE

Genetic testing is typically an outpatient procedure which is only eligible for coverage as an inpatient procedure in special circumstances, including, but not limited to, the presence of a comorbid condition that would require monitoring in a more controlled environment such as the inpatient setting.

CODING REQUIREMENTS

GENETIC TESTING CODES

Code	Description
80406	ACTH stimulation panel; for 3 beta-hydroxydehydrogenase deficiency This panel must include the following: Cortisol (82533 x 2) 17 hydroxypregnenolone (84143 x 2).
82261	Biotinidase, each specimen.
84134	Prealbumin.
V5008	Hearing screening.
82180	Ascorbic acid (Vitamin C), blood.
82759	Galactokinase, RBC.
84138	Pregnanetriol.
83020	Hemoglobin fractionation and quantitation; electrophoresis (e.g., A2, S, C, and/or F).
84437	Thyroxine; requiring elution (e.g., neonatal).
81220	CFTR (cystic fibrosis transmembrane conductance regulator) (e.g., cystic fibrosis) gene analysis; common variants (e.g., ACMG/ACOG guidelines).
83021	Hemoglobin fractionation and quantitation; chromatography (e.g., A2, S, C, and/or F).
84443	Thyroid stimulating hormone (TSH).
83520	Immunoassay for analyte other than infectious agent antibody or infectious agent antigen; quantitative, not otherwise specified.
84510	Tyrosine.
83789	Mass spectrometry and tandem mass spectrometry (e.g., MS, MS/MS, MALDI, MS-TOF, QTOF), nondrug analyte(s) not elsewhere specified, qualitative or quantitative, each specimen.
84030	Phenylalanine (PKU), blood.

GENETIC COUNSELING CODES

Code	Description
96040	Medical genetics and genetic counseling services, each 30 minutes face-to-face with patient/family.

Reference

US National Library of Medicine, National Institutes of Health, Department of Health & Human Services. Newborn Screening Coding and Terminology Guide. Delaware.gov.

POLICY UPDATE HISTORY

08/24/2022	Approved in Medical Policy Committee
09/13/2022	Approved in QI/UM
10/10/2022	Approved in Governance