CLINICAL MEDICAL POLICY	
Policy Name:	Non-Oncologic/Congenital Anomalies Genetic Testing Panels
Policy Number:	MP-071-MD-PA
Responsible Department(s):	Medical Management
Provider Notice/Issue Date:	02/01/2025; 11/01/2024; 10/01/2023; 10/01/2022; 12/17/2021; 10/19/2020; 11/18/2019; 12/15/2018
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Products:	Highmark Wholecare℠ Medicaid
Application:	All participating hospitals and providers
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Policy History

Date	Activity
03/01/2025	Provider Effective date
01/13/2025	PARP Approval
12/18/2024	QI/UM Committee review
12/18/2024	Urgent Review: Policy name changed from "*Nononcologic Genetic Testing Panels*" to "*Nononcologic/Congenital Anomalies Genetic Testing Panels*". Added the following congenital anomaly genetic testing CPT codes: 80406, 82261, 84134, 82180, 82759, 84138, 83020, 84437, 83021, 84443, 83520, 84510, 83789 & 84030. Added Metabolic Disease list to 'Informational' section.
12/01/2024	Provider Effective date
09/09/2024	PARP Approval
08/21/2024	QI/UM Committee review
08/21/2024	Annual Review: No changes to clinical criteria. Updated 'Summary of Literature' and 'Reference Sources' sections. Updated the code Description for the following Procedure Codes: 81400, 81402, 81407, & 81408.
11/01/2023	Provider Effective date
09/22/2023	PARP Approval
08/16/2023	QI/UM Committee review

08/16/2023	Annual Review: No changes to clinical criteria. Removed deleted ICD-10 code Q85.8. Added ICD-10 code Q85.89. Updated 'Summary of Literature' and 'Reference Sources' sections.
11/01/2022	Provider Effective date
09/14/2017	Initial policy developed

Disclaimer

Highmark Wholecare℠ 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 Wholecare℠ may provide coverage under the medical-surgical benefits of the Company's Medicaid products for medically necessary non-oncologic Genetic Testing Panels.

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.

(Current applicable Pennsylvania HealthChoices Agreement Section V. Program Requirements, B. Prior Authorization of Services, 1. General Prior Authorization Requirements.)

Definitions

Prior Authorization Review Panel (PARP) – A panel of representatives from within the PA Department of Human Services who have been assigned organizational responsibility for the review, approval and denial of all PH-MCO Prior Authorization policies and procedures.

Genetic Testing Panel – A laboratory test that evaluates multiple genes simultaneously compared to sequential testing of individual genes. There are multiple uses of the testing results including but not limited to: to establish a clinical diagnosis, confirmation of a specific clinical diagnosis, the diagnosis of a hereditary disorder, to determine when a known cancer diagnosis is part of a hereditary cancer syndrome, or to assist in the identification of a cancer type/subtype and in the selection of the most appropriate treatment of a cancer type/subtype.

Germline Mutation – An alteration in the DNA that is transmissible from parent to offspring.

Panel Testing Technology – A genetic testing method that examines multiple genes or mutations simultaneously. Testing methods can include next-generation sequencing and chromosomal microarray.

Next-Generation Sequencing (NGS) – Non-Sanger-based high-throughput DNA sequencing technologies. Billions of DNA strands can be sequenced in parallel, yielding substantially more throughput and

minimizing the need for the fragment-cloning methods that are often used in Sanger sequencing of genomes.

Chromosomal Microarray Analysis (CMA) – A technique that identifies chromosomal abnormalities, including submicroscopic abnormalities that are too small to be detected by conventional karyotyping.

Variant of Unknown/Uncertain Significance (VUS) – An allele, or variant form of a gene, that has been identified via genetic testing. The significance of the finding is not established, and the connection to a human disease has not been identified.

Clinical Utility – How likely the testing is to significantly improve patient outcomes that reflect the balance between health-related benefits and/or harms that can ensue from using the information made available from the testing.

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 Counseling – A service that is provided by a Clinical Geneticist, Certified Genetic Counselor, or other approved medical provider who is independent and not employed by any clinical or genetic laboratory, who bears no conflict of interest with the entity performing the testing.

Procedures

This policy applies only if there is no other separate Highmark Wholecare℠ medical policy that addresses the criteria for a specific non-oncologic genetic test.

The ordering provider must validate the clinical utility by considering the following:

- Will the selected genetic panel test offer significant advantages compared to sequential analysis of individual genes (i.e., a genetic testing panel that addresses the disorder in question, rather than the disorder in question plus other disorders)?
- How will the genetic panel testing results be used in patient care decision making?
- Will the ancillary findings lead to further testing or care management changes?
- Is there reliable evidence in the peer-reviewed scientific literature that health outcomes will be improved as a result of treatment decisions based on the selected genetic panel testing findings?

1. Genetic testing panels may be considered medically necessary when ALL of the following criteria are met:
    A. All genetic testing panels must be performed in a Clinical Laboratory Improvement Amendment (CLIA) licensed lab; AND
    B. Genetic testing panels are to be ordered or recommended by a physician specialist such as a hematologist, a physician with expertise in the treatment of the targeted disease, or a geneticist; AND
    C. The ordering provider must not be employed or contracted by a commercial genetic testing laboratory; AND
    D. A recommendation for the genetic testing is confirmed by either:
        1) An American Board of Medical Genetics or American Board of Genetic Counselor; OR
        2) An independent Board Certified or Board eligible medical geneticist; AND

E. All components of the specific genetic testing panel must demonstrate positive clinical utility for the medical condition being evaluated, and offer substantial advantages in efficiency compared to sequential analysis of individual genes; AND
F. Genetic testing panels should be considered when clinical evaluation suggests a particular diagnosis, the disorder cannot be identified through clinical evaluation and/or other testing, and not when the diagnosis is unclear or uncertain; AND
G. The provider has had a discussion with the patient regarding the scope of the genetic testing panel being ordered and the impact of variants of unknown significance; AND
H. An informed consent must be signed by the patient prior to testing. The consent must include a statement that the patient agrees to post-test counseling, and the consent must be made available upon request; AND
I. Pre-test genetic counseling has been performed and post-test genetic counseling by an independent genetic professional is planned.

2. ALL of the following documentation requirements apply:
   A. A brief explanation of how the results of genetic testing are necessary to guide treatment decisions relevant to the patient's personal medical history for positive patient outcome (i.e., whether to perform surgery, determine chemotherapy treatment, choice between medication options, etc.); AND
   B. Medical records relevant to the testing being performed including:
      1) A thorough history and physical exam by the referring physician; AND
      2) Any previously performed conventional testing and outcomes; AND
      3) A three-generation pedigree analysis result; AND
      4) Any conservative treatments that have been provided, if applicable; AND
   C. The following information is required for a genetic or molecular diagnostic test:
      1) The specific name of the test/panel; AND
      2) Name of performing CLIA-accredited laboratory; AND
      3) The exact gene(s) and/or mutations being tested; AND
      4) Estimated cost/quote sheet for the genetic testing panel ordered.

3. When non-oncologic genetic tests are considered not medically necessary
   - Broad-based genetic testing panels are considered not medically necessary when individual components are sufficient for treatment/management of the patient. Testing for multiple genes or multiple conditions, in cases where a tiered approach/method is clinically available, will be considered medically necessary only for the number of genes or tests that are reasonable to obtain necessary therapeutic decision making, and NOT the entire panel.
   - More than one (1) multi-gene panel performed at the same time is considered not medically necessary.
   - Infant and adolescent genetic testing to predict adult onset of diseases is considered not medically necessary.
   - Genetic tests for inherited disease only need to be conducted once per lifetime of the patient.
   - If a genetic testing panel was previously performed for medically necessary indications, and a larger panel is developed and requested, only the testing for previously untested genes will be considered medically necessary.

4. Post-payment Audit Statement

   The medical record must include documentation that reflects the medical necessity criteria and is subject to audit by Highmark Wholecare℠ at any time pursuant to the terms of your provider agreement.

5. Place of Service

   The proper place of service for non-oncologic genetic testing panels is outpatient.

6. Genetic Counseling

   Pre- and post-test genetic counseling is required to be performed by an independent genetic provider (not employed by a genetic testing lab) prior to genetic testing for mutations. This service is necessary in order to inform patient being tested about the benefits and limitations of specific genetic tests. Genetic testing for mutations requires documentation of medical necessity from at least one of the following providers who has previously evaluated the patient, and intends to see the patient after genetic testing has been performed:

   - Board Eligible or Board Certified Genetic Counselor
   - Advanced Genetics Nurse
   - Genetic Clinical Nurse
   - Advanced Practice Nurse in Genetics
   - Board Eligible or Board Certified Clinical Geneticist
   - A physician of appropriate expertise or other obstetrical provider specializing in the care for the indication(s) for genetic testing

7. Related Policies

   - MP-012-MD-PA Chromosomal Microarray Analysis: Comparative Genomic Hybridization (CGH) and Single Nucleotide Polymorphism (SNP)
   - MP-003-MD-PA Fetal Aneuploidy Testing using Noninvasive Cell-Free Fetal DNA
   - MP-006-MD-PA Genetic Testing for Cystic Fibrosis
   - MP-063-MD-PA Genetic Testing for Warfarin and Clopidogrel
   - MP-010-MD-PA Testing for Genetic Disease
   - MP-013-MD-PA Whole Exome and Whole Genome Sequencing for Diagnosis of Genetic Disorders
   - MP-122-MD-PA Selected Tests for Rheumatic Diseases
   - MP-124-MD-PA Serum Biomarker Panel Testing for Systemic Lupus Erythematosus
   - MP-126-MD-PA Pharmacogenetic Testing

Operational Guidelines *Do not include on external version*

- This medical policy will be applied on a preservice, prepayment basis for both facility and professional providers.
- Claims submitted that do not match the procedure and diagnosis codes in the Coding Requirements section should deny as not medically necessary.
- Any requests for non-oncologic genetic testing panels that do not meet the guidelines listed in this policy will require an approval review by a Medical Director on a case-by-case basis.

Governing Bodies Approval

Three federal agencies play a role in the regulation of genetic tests: Centers for Medicare and Medicaid Services (CMS), the U.S. Food & Drug Administration (FDA), and the Federal Trade Commission (FTC). CMS is responsible for regulating all clinical laboratories performing genetic testing, ensuring their compliance with the Clinical Laboratory Improvement Amendments (CLIA) of 1988. The FDA has the broadest authority in terms of regulating the safety and effectiveness of genetic tests as medical devices under the Federal Food, Drug, and Cosmetic Act. Compared to the FDA and CMS, the FTC's regulatory authority is rather narrow and is limited to how tests are advertised. The FTC has the authority to regulate advertising that delivers health-related information to consumers to ensure that it is not false or misleading.

Genetic testing panels are typically laboratory derived tests that are not subject to the U.S. FDA approval.

CLIA
The genetic testing panels are offered as laboratory-developed tests under CLIA licensed laboratories. Clinical laboratories may develop and validate tests in-house and market them as a laboratory service; laboratories offering such tests as a clinical service must meet general regulatory standards of CLIA and must be licensed by CLIA for high complexity testing.

Note: This policy may not apply to multi-gene panel testing for indications that are addressed in another Highmark Wholecare test-specific medical policy. Please check to see if there is a more specific policy.

Summary of Literature

The role of genetic testing in the medical profession has continued to grow rapidly. With the completion of the Human Genome Project (HGP) and continued advances in the field of genomics, the use of genetic testing has become widespread. The World Health Organization (WHO) has published criteria to be met for any genetic test to be considered valuable: the disease is an important health problem, the risk in mutation carriers is high in the general population (not just in a high-risk group), mutations for the disease can be accurately identified, and effective interventions exist.

Genetic testing was first introduced as a clinical tool in the 1960s with chromosomal karyotyping (Satya-Murti et al. 2013). More advanced testing includes: chromosomal microarray analysis or comparative genomic hybridization (array CDG) testing, fluorescence-in-situ-hybridization (FISH), letter-by-letter sequencing of specific genes (Sanger technology), and the new technology where huge panels of genes as large as the entire exome can be sequenced (NexGen technology).

Genetic testing includes the following:

- Single gene-targeted mutation/sequence analysis
- Deletion/duplication analysis
- Multi-gene panels
- Serial testing of single genes
- Whole Exome Sequencing (WES) – sequencing of exome but interpretation focus on genes related to phenotype
- Whole Genome Sequencing (WGS) – sequences all genetic material

Genetic testing panels have been proposed to aid in the diagnosis of individuals with suspected mitochondrial disorders and may involve point mutations analysis. Genetic testing uses next-generation

sequencing (NGS) technology, massive parallel sequencing, or chromosomal microarray analysis (CMA) testing to perform genetic panels. NGS and CMA are new genetic technologies. The intended use for genetic panels is variable. Existing genetic testing panels are available for the following areas: cancer, cardiovascular disease, neurologic disease, psychiatric conditions, and for reproductive testing. In contrast to genomic testing, serial testing of single genes and multi-gene panel testing rely on the clinician developing a hypothesis about which specific gene or set of genes to test (Chinnery, 2014).

Several methods can be used for genetic testing:

- Molecular genetic tests (or gene tests) study single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder.
- Cytogenetic tests analyze whole chromosomes or long lengths of DNA to see if there are abnormalities or large genetic changes, such as an extra copy of a chromosome, that cause a genetic condition.
- Biochemical genetic tests study the amount or activity level of proteins; abnormalities in either can indicate changes to the DNA that result in a genetic disorder (Genetic Alliance, 2010)

Advantages of genetic testing panels:

- Offers greater insight, including targeting the coding part of the gene that is relevant to a particular disease. Testing involves reading a DNA sequence from start to finish to see if there are any interruptions or disruptions that stop the gene from making normal proteins
- Less chance of uncertainties, knowing that a particular mutation is absent can help ease anxiety
- Opportunity to take action and guide medical care (Joy, 2017)

Disadvantages of genetic testing panels:

- There is no standardization in the makeup of genetic panels. The panel compositions are variable with different set of genes for the same condition. This genetic panel composition is determined by the specific lab that developed the test.
- The gene selection of genetic panels is subject to change based on scientific discovery.
- Because of the large number of mutations contained in expanded panels, it is not possible to determine clinical validity for the panels as a whole.
- The risk for uncertain and incidental findings with the large numbers of genes on the panels.
- Large percentage of VUS.

Genetic testing plays a pivotal role in understanding the risk of a patient developing certain diseases while also screening and deciding on a medical treatment plan. There are various types of genetic tests performed for specific reasons:

- **Diagnostic testing** is done when symptoms of a disease are present and may be caused by mutated genes. Testing may be used to confirm or rule-out diseases such as cystic fibrosis or Huntington's disease.
- **Presymptomatic and predictive testing** can reveal if a patient is at risk for developing a genetic condition when there is a family history, for example colorectal cancer.
- **Carrier testing** may provide genetic information if a patient in a specific ethnic group has a family history of a genetic disorder (sickle cell, cystic fibrosis) and would like to be tested before having children. An expanded carrier test can detect genes associated with a wide variety of genetic diseases and mutations and identify if the patient or their partner are carriers for the same conditions.
- **Pharmacogenetic testing** may help determine what medication and dosage will be most favorable for patients' with a particular health condition or disease.

- **Prenatal testing** can detect some types of abnormalities in an unborn baby's genes. These tests screen for markers in blood or by invasive testing such as amniocentesis. Down syndrome and trisomy 18 syndrome are two genetic disorders that may be screened for as part of prenatal genetic testing. Cell-free DNA testing examines the baby's DNA using blood tests performed on the mother.
- **Newborn screening** is the most common type of genetic testing in the U.S., with all states requiring that newborns be tested for certain genetic and metabolic abnormalities. This test can reveal if there are disorders such as congenital hypothyroidism, sickle cell, or phenylketonuria (PKU).
- **Targeted gene sequencing** are focused panels that contain a select number of genes or gene regions that are known or are suspected as associates of the disease or phenotype. These panels can be designed with preselected content or custom designed. Next-generation sequencing also evaluates targeted genes of interest, however, multiple genes can be assessed.

The American Academy of Neurology (AAN) has issued recommendations for genetic testing that is "guided by the clinical phenotype, inheritance patter (if available), and electrodiagnostic features." As an example, the AAN does not support complete panels of all known Charcot-Marie Tooth genes but rather recommends a stepwise evaluation method to improve genetic screening efficiency.

The American Academy of Pediatrics (AAP) recommends that the decisions about whether to offer genetic testing and screening should be driven by the best interest of the child. Genetic testing is best offered in the context of genetic counseling. Genetic counseling can be performed by clinical geneticists, genetic counselors, or any other health care provider with appropriate training and expertise. The AAP and the American College of Medical Genetics and Genomics (ACMG) support the expansion of educational opportunities in human genomics and genetics for medical students, residents, and practicing pediatric primary care providers (AAP, 2013).

The AAP and ACMG support the mandatory offering of newborn screening for all children. After education and counseling about the substantial benefits of newborn screening, its remote risks, and the next steps in the event of a positive screening result, parents should have the option of refusing the procedure, and an informed refusal should be respected (AAP, 2013).

The American College of Obstetricians and Gynecologists (ACOG) recommends that prenatal genetic screening (serum screening with or without nuchal translucency [NT] ultrasound or cell-free DNA screening) and diagnostic testing (chorionic villus sampling [CVS] or amniocentesis) options should be discussed and offered to all pregnant patients regardless of maternal age or risk of chromosomal abnormality. After review and discussion, every patient has the right to pursue or decline prenatal genetic screening and diagnostic testing. If screening is accepted, patients should have one prenatal screening approach, and should not have multiple screening tests performed simultaneously (ACOG, 2022).

Coding Requirements

Procedure Codes

CPT Code	Description
0001U	Red blood cell antigen typing, DNA, human erythrocyte antigen gene analysis of 35 Antigens from 11 blood groups, utilizing whole blood, common RBC alleles reported
0004M	Scoliosis, DNA analysis of 53 single nucleotide polymorphisms (SNPs), using saliva, prognostic algorithm reported as a risk score
80406	ACTH stimulation panel; for 3 beta-hydroxydehydrogenase deficiency This panel must include the following: Cortisol (82533 x 2) 17 hydroxypregnenolone (84143 x 2)
81161	DMD (dystrophin) (e.g., Duchenne/Becker muscular dystrophy) deletion analysis, and duplication analysis, if performed
81240	F2 (prothrombin, coagulation factor II) (e.g., hereditary hypercoagulability) gene analysis, 20210G>A variant
81241	F5 (coagulation factor V) (e.g., hereditary hypercoagulability) gene analysis, Leiden variant
81242	FANCC (Fanconi anemia, complementation group C) (e.g., Fanconi anemia, type C) gene analysis, common variant (e.g., IVS4+4A>T)
81243	FMR1 (Fragile X mental retardation 1) (e.g., fragile X mental retardation) gene analysis; evaluation to detect abnormal (e.g., expanded) alleles
81244	FMR1 (Fragile X mental retardation 1) (e.g., fragile X mental retardation) gene analysis; characterization of alleles (e.g., expanded size and promoter methylation status)
81245	FLT3 (fms-related tyrosine kinase 3) (e.g., acute myeloid leukemia), gene analysis; internal tandem duplication (ITD) variants (i.e., exons 14, 15)
81246	FLT3 (fms-related tyrosine kinase 3) (e.g., acute myeloid leukemia), gene analysis; tyrosine kinase domain (TKD) variants (e.g., D835, I836)
81250	G6PC (glucose-6-phosphatase, catalytic subunit) (e.g., Glycogen storage disease, Type 1a, von Gierke disease) gene analysis, common variants (e.g., R83C, Q347X)
81251	GBA (glucosidase, beta, acid) (e.g., Gaucher disease) gene analysis, common variants (e.g., N370S, 84GG, L444P, IVS2+1G>A)
81255	HEXA (hexosaminidase A [alpha polypeptide]) (e.g., Tay-Sachs disease) gene analysis, common variants (egg, 1278insTATC, 1421+1G>C, G269S)
81256	HFE (hemochromatosis) (e.g., hereditary hemochromatosis) gene analysis, common variants (e.g., C282Y, H63D)
81257	HBA1/HBA2 (alpha globin 1 and alpha globin 2) (e.g., alpha thalassemia, Hb Bart hydrops fetalis syndrome, HbH disease), gene analysis, for common deletions or variant (e.g., Southeast Asian, Thai, Filipino, Mediterranean, alpha3.7, alpha4.2, alpha20.5, and Constant Spring)
81260	IKBKAP (inhibitor of kappa light polypeptide gene enhancer in B-cells, kinase complex-associated protein) (e.g., familial dysautonomia) gene analysis, common variants (e.g., 2507+6T>C, R696P)
81261	IGH@ (Immunoglobulin heavy chain locus) (e.g., leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); amplified methodology (e.g., polymerase chain reaction)
81262	IGH@ (Immunoglobulin heavy chain locus) (e.g., leukemias and lymphomas, B-cell), gene rearrangement analysis to detect abnormal clonal population(s); direct probe methodology (e.g., Southern blot)
81263	IGH@ (Immunoglobulin heavy chain locus) (e.g., leukemia and lymphoma, B-cell), variable region somatic mutation analysis

81264	IGK@ (Immunoglobulin kappa light chain locus) (e.g., leukemia and lymphoma, B-cell), gene rearrangement analysis, evaluation to detect abnormal clonal population(s)
81265	Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (e.g., pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [e.g., buccal swab or other germline tissue sample] and donor testing, twin zygosity testing, or maternal cell contamination of fetal cells)
81266	Comparative analysis using Short Tandem Repeat (STR) markers; each additional specimen (e.g., additional cord blood donor, additional fetal samples from different cultures, or additional zygosity in multiple birth pregnancies) (List separately in addition to code for primary procedure)
81267	Chimerism (engraftment) analysis, post transplantation specimen (e.g., hematopoietic stem cell), includes comparison to previously performed baseline analyses; without cell selection
81268	Chimerism (engraftment) analysis, post transplantation specimen (e.g., hematopoietic stem cell), includes comparison to previously performed baseline analyses; with cell selection (e.g., CD3, CD33), each cell type
81270	JAK2 (Janus kinase 2) (e.g., myeloproliferative disorder) gene analysis, p.Val617Phe (V617F) variant
81272	KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (e.g., gastrointestinal stromal tumor [GIST], acute myeloid leukemia, melanoma), gene analysis, targeted sequence analysis (e.g., exons 8, 11, 13, 17, 18)
81273	KIT (v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog) (e.g., mastocytosis), gene analysis, D816 variant(s)
81290	MCOLN1 (mucolipin 1) (e.g., Mucolipidosis, type IV) gene analysis, common variants (e.g., IVS3-2A>G, del6.4kb)
81291	MTHFR (5,10-methylenetetrahydrofolate reductase) (e.g., hereditary hypercoagulability) gene analysis, common variants (e.g., 677T, 1298C)
81302	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; full sequence analysis
81303	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; known familial variants
81304	MECP2 (methyl CpG binding protein 2) (e.g., Rett syndrome) gene analysis; duplication/deletion variants
81321	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; full sequence analysis
81322	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; known familial variants
81323	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; duplication/deletion variants
81324	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; duplication/deletion analysis
81325	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; full sequence analysis
81326	PMP22 (peripheral myelin protein 22) (e.g., Charcot-Marie-Tooth, hereditary neuropathy with liability to pressure palsies) gene analysis; known familial variants
81330	SMPD1(sphingomyelin phosphodiesterase 1, acid lysosomal) (e.g., Niemann-Pick disease, Type A) gene analysis, common variants (e.g., R496L, L302P, fsP330)

81331	SNRPN/UBE3A (small nuclear ribonucleoprotein polypeptide N and ubiquitin protein ligase E3A) (e.g., Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
81332	SERPINA1 (serpin peptidase inhibitor, clade A, alpha-1 antiproteinase, antitrypsin, member 1) (e.g., alpha-1-antitrypsin deficiency), gene analysis, common variants (e.g., *S and *Z)
81400	Molecular pathology procedure, Level 1 (eg, identification of single germline variant [eg, SNP] by techniques such as restriction enzyme digestion or melt curve analysis)
81402	Molecular pathology procedure, Level 3 (eg, >10 SNPs, 2-10 methylated variants, or 2-10 somatic variants [typically using non-sequencing target variant analysis], immunoglobulin and T-cell receptor gene rearrangements, duplication/deletion variants of 1 exon, loss of heterozygosity [LOH], uniparental disomy [UPD])
81407	Molecular pathology procedure, Level 8 (eg, analysis of 26-50 exons by DNA sequence analysis, mutation scanning or duplication/deletion variants of >50 exons, sequence analysis of multiple genes on one platform)
81408	Molecular pathology procedure, Level 9 (eg, analysis of >50 exons in a single gene by DNA sequence analysis)
81410	Aortic dysfunction or dilation (e.g., Marfan syndrome, Loeys Dietz syndrome, Ehler Danlos syndrome type IV, arterial tortuosity syndrome); genomic sequence analysis panel, must include sequencing of at least 9 genes, including FBN1, TGFBR1, TGFBR2, COL3A1, MYH11, ACTA2, SLC2A10, SMAD3, and MYLK
81411	Aortic dysfunction or dilation (e.g., Marfan syndrome, Loeys Dietz syndrome, Ehler Danlos syndrome type IV, arterial tortuosity syndrome);duplication/deletion analysis panel, must include analysis for GFBR1, TGFBR2, MYH11, and COL3A1
81412	Ashkenazi Jewish associated disorders (e.g., Bloom syndrome, Canavan disease, cystic fibrosis, familial dysautonomia, Fanconi anemia group C, Gaucher disease, Tay-Sachs disease), genomic sequence analysis panel, must include sequencing of at least 9 genes, including ASPA, BLM, CFTR, FANCC, GBA, HEXA, IKBKAP, MCOLN1, and SMPD1
81413	Cardiac ion channelopathies (e.g., Brugada syndrome, long QT syndrome, short QT syndrome, catecholaminergic polymorphic ventricular tachycardia); genomic sequence analysis panel, must include sequencing of at least 10 genes, including ANK2 CASQ2, CAV3M KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, AND SCNSA
81414	Cardiac ion channelopathies (e.g., Brugada syndrome, long QT syndrome, short QT syndrome, catecholaminergic polymorphic ventricular tachycardia); duplication/deletion gene analysis panel, must include analysis of at least 2 genes, including KCNH2 and KCNQ1
81434	Hereditary retinal disorders (e.g., retinitis pigmentosa, Leber congenital amaurosis, cone-rod dystrophy), genomic sequence analysis panel, must include sequencing of at least 15 genes, including ABCA4, CNGA1, CRB1, EYS, PDE6A, PDE6B, PRPF31, PRPH2, RDH12, RHO, RP1, RP2, RPE65, RPGR, and USH2A
81439	Hereditary cardiomyopathy (e.g., hypertrophic cardiomyopathy, dilated cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy), genomic sequence analysis panel, must include sequencing of at least 5 cardiomyopathy-related genes (e.g., DSG2, MYBPC3, MYH7, PKP2, TTN)
81440	Nuclear encoded mitochondrial genes (e.g., neurologic or myopathic phenotypes), genomic sequence panel, must include analysis of at least 100 genes, including BCS1L, C1orf2, COQ2, COX10, DGUOK, MPV17, OPA1, PDSS2, POLG, POLG2, RRM2B, SCO1, SCO2, SLC25A4, SUCLA2, SUCLG1, TAZ, TK2, AND TYMP

81442	Noonan spectrum disorders (e.g., Noonan syndrome, cardio-facio-cutaneous syndrome, Costello syndrome, LEOPARD syndrome, Noonan-like syndrome), genomic sequence analysis panel, must include sequencing of at least 12 genes, including BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, and SOS1
81448	Hereditary peripheral neuropathies (e.g., Charcot-Marie-Tooth, spastic paraplegia), genomic sequence analysis panel, must include sequencing of at least 5 peripheral neuropathy-related genes (e.g., BSCL2, GJB1, MFN2, MPZ, REEP1, SPAST, SPG11, SPTLC1)
81493	Coronary artery disease, mRNA, gene expression profiling by real-time RT-PCR of 23 genes, utilizing whole peripheral blood, algorithm reported as a risk score
81595	Cardiology (heart transplant), mRNA, gene expression profiling by real-time quantitative PCR of 20 genes (11 content and 9 housekeeping), utilizing subfraction of peripheral blood, algorithm reported as a rejection risk score
82180	Ascorbic acid (Vitamin C), blood
82261	Biotinidase, each specimen
82759	Galactokinase, RBC
83020	Hemoglobin fractionation and quantitation; electrophoresis (eg, A2, S, C, and/or F)
83021	Hemoglobin fractionation and quantitation; chromatography (eg, A2, S, C, and/or F)
83520	Immunoassay for analyte other than infectious agent antibody or infectious agent antigen; quantitative, not otherwise specified
83789	Mass spectrometry and tandem mass spectrometry (eg, MS, MS/MS, MALDI, MS-TOF, QTOF), non-drug analyte(s) not elsewhere specified, qualitative or quantitative, each specimen
84030	Phenylalanine (PKU), blood
84134	Prealbumin
84138	Pregnanetriol
84437	Thyroxine; requiring elution (eg, neonatal)
84443	Thyroid stimulating hormone (TSH)
84510	Tyrosine
88299	Unlisted cytogenetic study
88380	Microdissection (i.e., sample preparation of microscopically identified target); laser capture

Note: If a procedure code other than those listed above is requested, the request must be sent to a Medical Director for individual consideration. The code may also be possibly listed on another separate Highmark Wholecare medical policy.

Diagnosis Codes

ICD-10 Code	Description
D66	Hereditary factor VIII deficiency
D67	Hereditary factor IX deficiency
D68.2	Hereditary deficiency of other clotting factors
D69.42	Congenital and hereditary thrombocytopenia purpura
D70.0	Congenital agranulocytosis
D70.4	Cyclic neutropenia
E23.0	Hypopituitarism
E25.0	Congenital adrenogenital disorders associated with enzyme deficiency
E31.21	Multiple endocrine neoplasia [MEN] type I
E70.0	Classical phenylketonuria
E70.30	Albinism, unspecified
E70.310	X-linked ocular albinism
E70.311	Autosomal recessive ocular albinism
E70.318	Other ocular albinism
E70.319	Ocular albinism, unspecified
E71.0	Maple syrup-urine disease
E71.311	Medium chain acyl CoA dehydrogenase deficiency
E74.04	McArdle disease
E74.20	Disorders of galactose metabolism, unspecified
E74.21	Galactosemia
E74.29	Other disorders of galactose metabolism
E75.02	Tay-Sachs disease
E75.21	Fabry (-Anderson) disease
E75.22	Gaucher disease
E75.240	Niemann-Pick disease type A
E75.241	Niemann-Pick disease type B
E75.242	Niemann-Pick disease type C
E75.243	Niemann-Pick disease type D
E75.248	Other Niemann-Pick disease
E75.249	Niemann-Pick disease, unspecified
E75.25	Metachromatic leukodystrophy
E75.26	Sulfatase deficiency
E75.29	Other sphingolipidosis
E83.110	Hereditary hemochromatosis
E85.2	Heredofamilial amyloidosis, unspecified
E88.01	Alpha-1-antitrypsin deficiency
F70	Mild intellectual disabilities
F71	Moderate intellectual disabilities
F72	Severe intellectual disabilities
F73	Profound intellectual disabilities

F78.A1	SYNGAP1-related intellectual disability
F78.A9	Other genetic related intellectual disability
F79	Unspecified intellectual disabilities
F81.81	Disorder of written expression
F84.0	Autistic disorder
F84.3	Other childhood disintegrative disorder
F84.5	Asperger's syndrome
F84.8	Other pervasive development disorders
F84.9	Pervasive developmental disorder, unspecified
G10	Huntington's disease
G11.0	Congenital nonprogressive ataxia
G11.10	Early-onset cerebellar ataxia, unspecified
G11.19	Other early-onset cerebellar ataxia
G11.2	Late-onset cerebellar ataxia
G11.3	Cerebellar ataxia with defective DNA repair
G11.4	Hereditary spastic paraplegia
G11.8	Other hereditary ataxia
G11.9	Hereditary ataxia, unspecified
G12.0	Infantile spinal muscular atrophy, Type 1 [Werdnig-Hoffman]
G12.1	Other inherited spinal muscular atrophy
G12.20	Motor neuron disease, unspecified
G12.21	Amyotrophic lateral sclerosis
G12.22	Progressive bulbar palsy
G12.23	Primary lateral sclerosis
G12.24	Familial motor neuron disease
G12.25	Progressive spinal muscular atrophy
G12.29	Other motor neuron disease
G12.8	Other spinal muscular atrophies and related syndromes
G12.9	Spinal muscular atrophy, unspecified
G13.0	Paraneoplastic neuromyopathy and neuropathy
G13.1	Other systemic atrophy primarily affecting central nervous system in neoplastic disease
G13.2	Systemic atrophy primarily affecting central nervous system in myxedema
G13.8	Systemic atrophy primarily affecting central nervous system in other diseases classified elsewhere
G24.1	Genetic torsion dystonia
G31.82	Leigh's disease
G40.301	Generalized idiopathic epilepsy and epileptic syndromes, not intractable, with status epilepticus
G40.309	Generalized idiopathic epilepsy and epileptic syndromes, not intractable, without status epilepticus
G40.311	Generalized idiopathic epilepsy and epileptic syndromes, intractable, with status epilepticus
G40.319	Generalized idiopathic epilepsy and epileptic syndromes, intractable, without status epilepticus

G47.35	Congenital central alveolar hypoventilation syndrome
G60.0	Hereditary motor and sensory neuropathy
G71.00	Muscular dystrophy, unspecified
G71.01	Duchenne or Becker muscular dystrophy
G71.02	Facioscapulohumeral muscular dystrophy
G71.09	Other specified muscular dystrophies
G71,20	Congenital myopathies, unspecified
G71.11	Myotonic muscular dystrophy
G71.12	Myotonic congenita
G71.13	Myotonic chondrodystrophy
G71.14	Drug induced myotonic
G71.19	Other specified myotonic disorders
G72.89	Other specified myopathies
G90.1	Familial dysautonomia [Riley-Day]
H35.52	Pigmentary retinal dystrophy
H47.22	Hereditary optic atrophy
I42.1	Obstructive hypertrophic cardiomyopathy
I42.2	Other hypertrophic cardiomyopathy
I42.8	Other cardiomyopathies
I78.0	Hereditary hemorrhagic telangiectasia
N04.0	Nephrotic syndrome with minor glomerular abnormality
N04.1	Nephrotic syndrome with focal and segmental glomerular lesions
N04.2	Nephrotic syndrome with diffuse membranous glomerulonephritis
N04.3	Nephrotic syndrome with diffuse mesangial proliferative glomerulonephritis
N04.4	Nephrotic syndrome with diffuse endocapillary proliferative glomerulonephritis
N04.5	Nephrotic syndrome with diffuse mesangiocapillary glomerulonephritis
N04.6	Nephrotic syndrome with dense deposit disease
N04.7	Nephrotic syndrome with diffuse crescentic glomerulonephritis
N04.8	Nephrotic syndrome with other morphologic changes
N04.9	Nephrotic syndrome with unspecified morphologic changes
Q04.3	Other reduction deformities of brain
Q61.5	Medullary cystic kidney
Q61.9	Cystic kidney disease, unspecified
Q75.0	Craniosynostosis
Q75. 1	Craniofacial dysostosis
Q75.2	Hypertelorism
Q75.3	Macrocephaly
Q75.4	Mandibulofacial dysostosis
Q75.5	Oculomandibular dysostosis
Q75.8	Other specified congenital malformations of skull and face bones
Q75.9	Congenital malformation of skull and face bones, unspecified
Q77.1	Thanatophoric short stature
Q77.4	Achondroplasia

Q78.0	Osteogenesis imperfecta
Q79.60	Ehlers-Danlos syndrome, unspecified
Q79.61	Classical Ehlers-Danlos syndrome
Q79.62	Hypermobile Ehlers-Danlos syndrome
Q79.63	Vascular Ehlers-Danlos syndrome
Q79.69	Other Ehlers-Danlos syndromes
Q79.8	Other congenital malformations of musculoskeletal system
Q82.2	Congenital cutaneous mastocytosis
Q85.01	Neurofibromatosis, type 1
Q85.02	Neurofibromatosis, type 2
Q85.89	Other phakomatoses, not elsewhere classified
Q87.0	Congenital malformation syndrome predominantly affecting facial appearance
Q87.11	Prader-Willi syndrome
Q87.19	Other congenital malformation syndromes predominantly associated with short stature
Q87.40	Marfan's syndrome, unspecified
Q87.410	Marfan's syndrome with aortic dilation
Q87.418	Marfan's syndrome with other cardiovascular manifestations
Q87.42	Marfan's syndrome with ocular manifestations
Q87.43	Marfan's syndrome with skeletal manifestations
Q93.51	Angelman syndrome
Q93.59	Other deletions of part of a chromosome
Q99.2	Fragile X chromosome
R62.52	Short stature (child)

Informational

Examples of Genetic Testing Panels (not all inclusive)
This table is strictly informational and does not indicate medical necessity

Name of Test
ARUP Laboratories
Agammaglobulinemia Panel
Amyotrophic Lateral Sclerosis Pane
Aortopathy Panel
Ashkenazi Jewish Diseases Panel
Autism Panel
Biotinidase Deficiency (BTD) 5 Mutation
Brugada Syndrome Panel
Cardiomyopathy and Arrhythmia Panel
Cystic Fibrosis (CFTR) 32 Mutations Panel
Mitochondrial Disorders Panel
Noonan Spectrum Disorders Panel
Periodic Fever Syndromes Panel
Retinitis Pigmentosa/Leber Congenital Amaurosis Panel
Solid Tumor Mutation Panel Next-Generation Sequencing

Vascular Malformation Syndromes
Emory Genetics Laboratories
ACOG/ACMG Carrier Screen Targeted Mutation Panel
Anophthalmia/ Microphthalmia/ Anterior Segment Dysgenesis/ Anomaly: Sequencing Panel
Arrhythmias Deletion/Duplication Panel
Arrhythmias Sequencing Panel
Autism Spectrum Disorders
Cardiomyopathy Panel
Ciliopathies Panel
Congenital Glycosylation Disorders
Early Onset IBD Sequencing and Del/Dup Panels
Epilepsy
Eye Disorders
Expanded Neuromuscular Disorders
Hereditary Hemolytic Anemia Sequencing 28 Genes
Noonan Syndrome and Related Disorders
Osteogenesis Inperfecta and Osteopenia Sequencing Panel
Short Stature Panel
Sudden Cardiac Arrest Panel
X-linked Intellectual Disability
Ambry Genetics
BreastNext™
CancerNext™
ColoNext™
FHNext
HCMNext
Marfan, Aneurysm and Related Disorders Panel
OvaNext™
Pan Cardio Panel
PancNext
RenalNext
TAADNext
X-linked Intellectual Disability
Athena
Alzheimer's Disease
Amyotrophic Lateral Sclerosis Advanced Evaluation Gene Panel
Ataxia, Comprehensive Evaluation
Autosomal Recessive Ataxia Evaluation
Common Mitochondrial Disorder Evaluation
Complete Ataxia Evaluation Panel
Complete Hereditary Spastic Paraplegia Evaluation Panel
Early Infantile Epileptic Encephalopathy
Hemiplegic Migraine Profile
Hereditary Renal Tubular Disorder Panel
Intellectual Disability
Mitochondrial Disease Associated with Mitochondrial Depletion Syndrome
Myotonic Syndrome Advanced Evaluation Panel
Periodic Paralysis Advanced Sequencing Evaluation Panel

Progressive External Ophthalmoplegia Evaluation Panel
Idiopathic Hypogonadotropic Hypogonadism/Kallmann Syndrome
Baylor College of Medicine
Cobalamin Metabolism Comprehensive Panel
CoQ10 Comprehensive Panel
GeneAware
Glycogen Storage Disorders Panel
Low Bone Mass Panel
Mitochondrial Disorders Panel
Myopathy/Rhabdomyolysis Panel
Progressive External Ophthalmoplegia Panel
Pyruvate Dehydrogenase Deficiency and Mitochondrial Respiratory Chain Complex V Deficiency Panel
Retinitis Pigmentosa Panel
Usher Syndrome Panel
GeneDx
Autism/ID Xpanded Panel
Breast/Ovarian Cancer Panel
Cardiomyopathy Panel
Colorectal Cancer Panel
Combined Cardiac Panel
Combined Mito Genome Plus Mito Nuclear Gene Panel
Comprehensive Hereditary Cancer Panel
Comprehensive Arrhythmia Panel
Comprehensive Cancer Panel
Comprehensive Epilepsy Panel
Comprehensive Mitochondrial Nuclear Gene Panel
Congenital Ichthyosis XomeDxSlice Panel
Congenital Myopathy and Congenital Muscular Dystrophy Panel
Dilated Cardiomyopathy (DCM) Left Ventricular Non-Compaction (LVNC)
Endometrial Cancer Panel
EpiXpanded Panel
Heterotaxy Panel
High-Moderate Risk Panel
Hyper-IgE Syndromes Panel
Hypertrophic Cardiomyopathy (HCM) Panel
Marfan Syndrome/TAAD Sequencing Panel
Noonan RASopathies Panel
Noonan Syndrome Panel
Pancreatic Cancer Panel
Prenatal Noonan Spectrum Disorders
Prenatal Skeletal Dysplasia Panel
Progressive External Ophthalmoplegia (PEO)/Optic Atrophy Nuclear Gene Panel
Rett/Angelman Syndrome Panel
Syndromic Macrocephaly Overgrowth Panel
XomeDxPlus (whole exome sequencing [WES] + mtDNA Sequencing and Deletion Testing
Medical Neurogenetics
Leigh Disease Panel
Spastic Paraplegia Next-Generation Sequencing

Partners Healthcare
Isolated Non-syndromic Congenital Heart Defects Panel
Noonan Spectrum Panel Pan Cardiomyopathy Panel
Usher Syndrome Panel
Mayo Medical Laboratories
Arrhythmogenic Right Ventricular Cardiomyopathy Panel
Bacterial Typing by whole Genome Sequencing
Brugada Syndrome
Comprehensive Cardiomyopathy Multi-Gene Panel
Congenital Disorders Chromosome Analysis (CDCA)
Dilated Cardiomyopathy Panel
Hereditary Colon Cancer Syndromes
Hypertrophic Cardiomyopathy Panel
Long QT Syndrome
Marfan Syndrome Panel
Noonan Syndrome Panel
Signature Genomics
Signature Prenatal Microarray
Counsyl Genomics
Counsyl Panel
GoodStart Genetics
GoodStart Select

Metabolic Disorders (not all inclusive):

- 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: Arginosuccinate Lyase Deficiency

    - CIT: Arginosuccinate Synthetase Deficiency (Citrullinemia)
- Organic Acid Disorders (MS/MS)
    - GA-1: Glutaric Acidemia, Type I
    - PA: Propionic 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

Reimbursement

Participating facilities will be reimbursed per their Highmark Wholecare℠ contract.

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