

Genetic Testing

Reimbursement Policy ID: RPC.0098.0100

Recent review date: 06/2026

Next review date: 09/2027

Keystone First reimbursement policies and their resulting edits are based on guidelines from established industry sources, such as the Centers for Medicare and Medicaid Services (CMS), the American Medical Association (AMA), state and federal regulatory agencies, and medical specialty professional societies. Reimbursement policies are intended as a general reference and do not constitute a contract or other guarantee of payment. Keystone First may use reasonable discretion in interpreting and applying its policies to services provided in a particular case and may modify its policies at any time.

In making claim payment determinations, the health plan also uses coding terminology and methodologies based on accepted industry standards, including Current Procedural Terminology (CPT®); the Healthcare Common Procedure Coding System (HCPCS); and the International Classification of Diseases, 10th Revision, Clinical Modification (ICD-10-CM), and other relevant sources. Other factors that may affect payment include medical record documentation, legislative or regulatory mandates, a provider's contract, a member's eligibility in receiving covered services, submission of clean claims, other health plan policies, and other relevant factors. These factors may supplement, modify, or in some cases supersede reimbursement policies.

This reimbursement policy applies to all health care services billed on a CMS-1500 form or its electronic equivalent, or when billed on a UB-04 form or its electronic equivalent.

To the extent that any procedure and/or diagnosis codes are specified in this policy, such inclusion is provided for reference purposes only, may not be all inclusive, and is not intended to serve as billing instructions. Listing of a code in this policy does not imply that the service described by the code is a covered or non-covered health service. Benefit coverage for health services is determined by federal, state, or contractual requirements and applicable laws that may require coverage for a specific service. The inclusion of a code does not imply any right to reimbursement or guarantee claim payment. Other Policies and Guidelines may apply.

Policy Overview

This policy addresses reimbursement of prenatal cytogenetics and molecular pathology. Genetic testing modalities may be used in the prenatal or early post-natal period to assist in identification of the cause or confirm a diagnosis associated with developmental delay, intellectual disability, dysmorphic features, heart defects, multiple malformations, short stature, stillbirth, neonatal death, or fertility problems. Each of the genetic techniques or methods has its strengths and limitations. Different methods complement each other in trying to identify the genetic variation(s) responsible for a medical condition.

Exceptions

N/A

Reimbursement Guidelines

Prenatal Testing

Claims for prenatal cytogenetic studies (88230-88299) may be denied when the only diagnosis on the claim is one of the following:

- full-term uncomplicated delivery (ICD-10-CM code O80),
- incidental pregnant state (ICD-10-CM code Z33.1), or
- supervision of normal pregnancy (ICD-10-CM codes Z34-Z34.93).

In addition to one of the diagnosis codes above, an additional diagnosis must be on the claim to support the need for genetic testing.

Genetic Testing

Molecular pathology testing is reported with CPT codes including 81161, 81200-81383 and is used to collect clinically significant genetic variants that may guide diagnosis, risk assessment, prognosis, treatment selection, or management. Reimbursement is dependent on accurate code selection (single-gene vs. multi-gene/panel), appropriate ICD-10-CM diagnosis linkage supporting medical necessity, and complete medical record documentation (e.g., ordering provider, test methodology, and clinical indications).

Definitions

Genetic Testing

A laboratory method that looks for changes in genes, gene expression, or chromosomes in cells or tissue of a person.

Cytogenetics

Testing samples of tissue, blood, amniotic fluid or bone marrow in a laboratory to look for changes in chromosomes, including broken, missing, rearranged, or extra chromosomes.

Molecular Pathology

Molecular pathology genetic tests are used to make or confirm clinical diagnoses of Mendelian (certain patterns of how traits are passed from parents to offspring) genetic disorders, disorders of human development, immunologic conditions, infectious diseases, and malignancies.

Edit Sources

- I. Current Procedural Terminology (CPT) and associated publications and services.
- II. International Classification of Diseases, 10th Revision, Clinical Modification (ICD-10-CM).
- III. Healthcare Common Procedure Coding System (HCPCS).
- IV. Centers for Medicare and Medicaid Services (CMS).
- V. National Correct Coding Initiative (NCCI).
- VI. <https://www.ncbi.nlm.nih.gov/gtr/all/labs/?term=cytogenetics%5Borgname%5D>.
- VII. Applicable Keystone First manual reference.
- VIII. Commonwealth of Pennsylvania Medicaid Program guidance.
- IX. Commonwealth of Pennsylvania Medicaid Program fee schedule(s).

Attachments

N/A

Associated Policies

N/A

Policy History

06/2026	Reimbursement Policy Committee Approval
05/2026	Annual review Genetic testing for breast and ovarian cancer removed.
01/2026	Reimbursement Policy Committee Approval
12/2025	Annual review No major changes
06/2025	Minor updates to formatting and syntax
04/2025	Revised preamble
09/2024	Reimbursement Policy Committee Approval
04/2024	Revised preamble
08/2023	Removal of policy implemented by Keystone First from Policy History section
01/2023	Template Revised - Revised preamble - Removal of Applicable Claim Types table - Coding section renamed to Reimbursement Guidelines - Added Associated Policies section