ICD-10-CM DOCUMENTATION AND CODING BEST PRACTICES:

Congenital Metabolic Disorders, Not Elsewhere Classified

Overview

Congenital metabolic disorder refers to a genetic disease that results in metabolism problems. Metabolism is a term that describes the biochemical processes that allow people to grow, reproduce, repair damage and respond to their environment. A metabolic disorder is a condition that impairs these processes.

Causes

Metabolism is a complex process that involves many biochemicals, tissues, and organs. This means there are many opportunities for something to go wrong and cause a metabolic disorder. Some examples of causes are:

- **Genetic:** Genes can influence metabolic processes in a variety of ways.
- **Organ Dysfunction:** Organs involved in metabolism can fail to function properly.
- **Mitochondrial dysfunction:** Mitochondria are small parts of cells that primarily produce energy.

Diagnostic Testing

Inherited metabolic disorders are present at birth, and some are detected by routine screening. All 50 states screen newborns for phenylketonuria (PKU). Most states also test newborns for galactosemia. However, no state tests babies for all known inherited metabolic disorders.

If an inherited metabolic disorder is not detected at birth, it is often not diagnosed until symptoms appear. Once symptoms develop, specific blood or DNA tests are available to diagnose most genetic metabolic disorders.

(continued)

Treatment

Treatment may include such measures as:

- Special diets that eliminate certain nutrients
- Taking enzyme replacements, or other supplements that support metabolism
- Treating the blood with chemicals to detoxify dangerous metabolic by-products

Documentation Guidance for Providers

Because treatment is limited, these inherited or congenital disorders are lifelong conditions. Current treatment or management of condition plan should be linked with the diagnosis.

The medical record should include:

- ✓ Current symptoms and complaints
- ✓ Associated physical findings
- ✓ Incorporate/document relevant findings from radiologic and laboratory testing into the progress note.
- ✓ Clear and concise treatment plan
  - Prescriptions
  - Diet changes
  - Referrals
- ✓ Include the final diagnosis in the assessment portion of the note
- ✓ Avoid documenting "history of" to describe a current disorder.

Coding Guidance for Coders

To ensure accurate code assignment:

- ✓ Review the entire medical record to verify the condition is active.
- ✓ Based on the description documented in the medical record, search the ICD-10-CM alphabetic index for that specific description and verify the code in the tabular listing; follow all instructional notes as appropriate.
- ✓ Pay close attention to the Excludes notes as they provide further specificity.

References

"ICD-10". Centers for Medicare & Medicaid Services, CMS.gov. http://www.cms.gov/medicare/coding/icd10

Medical News Today, Common metabolic disorders. https://www.medicalnewstoday.com/articles/metabolic-disorders#common-disorders

National Institute of Neurological Disorders and Stroke – Kearns-Sayre Syndrome. https://www.ninds.nih.gov/health-information/disorders/kearns-sayre-syndrome

(continued)

ICD-10-CM Information

CHAPTER 16 Certain Conditions Originating in the Perinatal Period

For coding and reporting purposes the perinatal period is defined as before birth through the 28th day following birth.

ICD-10-CM Category	Description	ICD-10-CM Notes
P70 – P74	Transitory endocrine and metabolic disorders specific to newborn	This category includes transitory endocrine and metabolic disturbances caused by the infant's response to maternal endocrine and metabolic factors, or its adjustment to extrauterine environment.

CHAPTER 17 Congenital Malformations, Deformations, and Chromosomal Abnormalities

Codes from chapter 17 may be used throughout the life of the patient. If a congenital malformation or deformity has been corrected, a personal history code should be used to identify the history of the malformation or deformity. Although present at birth, a malformation/deformation/or chromosomal abnormality may not be identified until later in life.

Q80 – Q89	Other congenital Malformations	
Q90 – Q99	Chromosomal abnormalities, not elsewhere classified	Excludes2: mitochondrial metabolic disorders (E88.4-)

Chapter 4 Endocrine, Nutritional and Metabolic Diseases

Excludes1: transitory endocrine and metabolic disorders specific to newborn (P70-P74)

Some of the more common congenital metabolic disorders include:

ICD-10-CM Code	Description	Clinical manifestations
E70.1	Phenylketonuria (PKU)	Deficiency of the enzyme PAH results in high levels of phenylalanine in the blood
E71.0	Maple syrup urine disease	Deficiency of an enzyme called BCKD causes buildup of amino acids in the body. Nerve damage may develop, and the urine smells like syrup.
E71.510	Zellweger syndrome	Abnormal facial features, enlarged liver and nerve damage in infants
E71.529	Adrenoleukodystrophy	Symptoms of nerve damage can develop in childhood and early adulthood depending on the form
E75.22	Gaucher disease	Bone pain, enlarged liver, and low platelet count, often mild, in children or adults
E74.00	Glycogen storage disease, unspecified	Problems with sugar storage lead to low blood sugar levels, muscle pain, and weakness.
E74.21	Galactosemia	Impaired breakdown of the sugar galactose leads to jaundice, vomiting and liver enlargement after breast or formula feeding by a newborn.
E75.02	Tay-Sachs disease	Progressive weakness in a months-old child, progressing to severe nerve damage; the child usually lives only until age 4 or 5.
E75.21	Fabry (-Anderson) disease	Pain in the extremities in childhood, with kidney and heart disease and strokes in adulthood; only males are affected
E75.23	Krabbe disease	Progressive nerve damage, developmental delay in your children; occasionally adults are affected. For congenital muscle hypoplasia, please refer to code Q79.8.
E75.249	Niemann-Pick disease	Babies develop liver enlargement, difficulty feeding and nerve damage
E76.02	Hurler-Scheie syndrome	Abnormal bone structure and developmental delay