You have a patient clearly exhibiting the signs of a well-known genetic disorder, but you are not sure how to code the examination because the diagnosis has not been confirmed. You have a patient with signs that could indicate a genetic disorder, but it is not clear what the diagnosis is and testing has not been performed yet. Health care providers are expected to code to the highest level of specificity, but this can be challenging when a specific code for a condition does not exist or the visit could be coded several different ways. This was part of the discussion during the webinar, “Genetics and Coding: What the Primary Care Provider Needs to Know,” held on September 27, 2012. The webinar, which featured Marc S Williams, MD, FAAP, FAccMG, was part of the Time Out for Genetics webinar series hosted by the Genetics in Primary Care Institute (GPCI).

What is an ICD-9-CM Code?

Although every health care provider is familiar with International Classification of Disease (ICD) codes, not all providers necessarily understand why they were developed.

- The ICD Clinical Modification (CM) coding system was developed by the World Health Organization to classify morbidity and mortality information for statistical and recording purposes.
- ICD-CM codes justify medical necessity for physician reimbursement.
- All procedures performed in health care settings need to be coded as a diagnosis and are required for any health-related transaction in the United States.
- ICD-CM codes also can be used for quality initiatives, utilization management and research.
- All codes are maintained by the Coordination and Maintenance Committee, which consists of representatives from the Centers for Medicare and Medicaid Services and the Centers for Disease Control and Prevention’s National Center for Health Statistics.
- ICD-9-CM is the ninth revision of the ICD-CM coding system. While the United States uses this version, most of the developed countries around the world use ICD-10-CM codes.

Challenges Associated with ICD-9-CM

The ICD-9-CM coding system has significant deficiencies, including a lack of genetic codes and a limited number of five-digit numbers for new codes.

- In 2004, significant gaps in coding for metabolic disorders, chromosome anomalies, congenital anomalies, teratogens, carrier screening and family history were brought to the attention of the Coordination and Maintenance Committee. Since then, modest improvements – mostly driven by newborn screening – have been made to coding for metabolic disorders and chromosome anomalies, while significant improvements have been made to carrier screening and family history coding. However, no changes have been made to coding for congenital anomalies and teratogens.
- Although health care providers are required to code to the highest specificity, this can be challenging to do for genetic conditions because of a lack of ICD-9 genetic codes. This also can make it difficult to prove medical necessity and, therefore, obtain adequate reimbursement.
- Sometimes V codes are the best option, but these were originally intended to be used for preventive services not genetic services.
- Payers are unable to perform adequate utilization management and to determine appropriate coverage due to inexact coding.
- The Coordination and Maintenance Committee is limited in its ability to add new codes because there are few five-digit codes remaining.
- The Coordination and Maintenance Committee is hesitant to make revisions to the current version of ICD-CM because it plans to transition to ICD-10-CM in the near future.
ICD-10-CM Coding Will be an Improvement for Genetics

ICD-10-CM will be an improvement for genetics because it includes extensive coding for metabolic disorders and is superior to the ninth version in terms of coding for other genetic disorders. In addition, it uses an alphanumeric system that will significantly expand the availability of new codes. However, new genetic technologies have not been taken into account in the 10th revision. The Coordination and Maintenance Committee originally planned to introduce ICD-10-CM to the U.S. medical community in 2007, but it has been delayed until October 2014.

Case 1

- A newborn infant presents with hypotonia, a heart murmur and dysmorphic features, including a flat face, upslanting eyes and single palmar creases. The infant is clinically diagnosed with Down syndrome.

  **Coding Options for Case 1**: Option 1) Use coding for the clinical diagnosis, Down syndrome (758.0), because this is the highest level of specificity. However, the diagnosis is yet to be confirmed with testing. Option 2) Await chromosome testing and code for hypotonia (781.3 and 779.89) and a heart murmur (785.2). If genetic testing confirms Down syndrome, then the code can be changed to indicate this.

Case 2

- A newborn infant presents with hypotonia, severe feeding problems requiring gavage, and is not dysmorphic. The differential diagnosis includes: chromosome anomaly, spinal muscular atrophy, congenital muscular dystrophy, and inborn error of metabolism

  **Coding Options for Case 2**: The diagnosis is unknown so the options for coding are 1) hypotonia (781.3 and add 799.89 to indicate congenital) or 2) poor feeding (779.81). Diagnostic testing confirms Prader-Willi Syndrome and coding is changed accordingly (759.81).

Case 3

- An 18-month old presents with delays in speech and gross and fine motor development, but growth is normal as are the physical and neurologic examinations. In addition, there are no dysmorphic features.

  **Coding Options for Case 3**: The clinical diagnosis is global developmental delay. There are several options for coding: 1) delayed milestones (783.42); 2) mixed developmental disorder (315.5); 3) other specified delays in development (315.8); 4) unspecified delay in development (315.9); or 5) developmental handicaps in early childhood (V79.3).