Genetics and Coding: What the Primary Care Provider Needs to Know

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Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute
Faculty

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  - A director of the board of the American College of Medical Genetics
  - Chair on the ACMG Ad Hoc Committee on the Value of a Genetic Diagnosis
  - Founder of the American College of Medical Genetics Quality Improvement Special Interest Group
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Learning Objectives

1. Describe the purpose of the ICD coding system
2. Discuss the gaps within the current coding systems regarding genetics in primary care settings
3. Identify appropriate courses of action related to accurate coding of genetic services
Purpose of Codes

• From ICD-9-CM 2012
  – Classification of morbidity and mortality information for statistical purposes
  – Indexing of “hospital records” by disease and operations for data storage and retrieval.

• In practice the above plus
  – Use in all settings, not just hospital
  – Justification of medical necessity for reimbursement
  – Supports quality initiatives
  – Utilization management
  – Research

• Lack of purpose driven design leads to significant deficiencies
Purpose of Codes for Providers

• To get paid!
ICD-9-CM

- Worldwide clinical coding system developed by WHO
- Required to be used for any health related transaction in the United States
- Codes maintained by Coordination and Maintenance Committee
  - Limited ability to add new codes as 5 digit maximum code is reaching its capacity
  - Interaction with C&M and ACMG in 2004 to address genetic code deficiencies with limited success
ICD-9 The Most Significant Gaps in 2004

- Metabolic disorders
  - Modest improvements
- Chromosome anomalies
  - Modest improvements
- Other and unspecified congenital anomalies
- Teratogens
- Carrier screening
  - Significant improvements
- Family history
  - Significant improvements
ICD-9-CM → ICD-10-CM

- Rest of world already using ICD-10-CM
- Alpha-numeric coding system dramatically expands available codes allowing expansion
- U.S. will introduce in 2007: October 1, 2014
ICD-10 Much Improved

• Extensive coding for metabolic disease
  – Should cover most newborn screening disorders
• Improved for other genetic disorders
• ICD-10 codes have not been informed by new genetic technologies such as chromosomal microarray (in routine clinical use) and whole exome/genome sequencing (very early use)
• ICD-11 has dramatically expanded genetic codes
  – No plans for implementation in the US at this point
Practical Considerations

• Providers are required to code to the highest level of specificity
  – In genetics the deficiencies in ICD-9 impair the ability to do this

• Implications
  – Need to use more generic codes makes it difficult to determine medical necessity
  – Some conditions (e.g. family history, carrier status) are coded by V codes which are for preventive services and other issues not well covered in the numeric codes and are not reimbursed under some plans
  – Payers are unable to do utilization management to inform medical decision making and coverage
  – Providers, health systems and payers all agree that deficiencies in ICD-9 significantly impair operations
Case Based Scenarios
Case 1

• Newborn infant
  – Hypotonic
  – Heart murmur
  – Dysmorphic features
    • Flat face
    • Upslanting eyes
    • Single palmar creases
Case 1

- Clinical diagnosis—Down Syndrome
- Coding
  - Option 1: Use clinical diagnosis
    - Down’s [sic] syndrome (758.0)
      - Highest level of specificity
      - Haven’t confirmed diagnosis
  - Option 2: Await chromosomes
    - Hypotonia
      - 781.3 (description actually ataxia NOS, muscular incoordination. Index maps hypotonia to this code)
      - 779.89 (specified condition originating in the perinatal period) could be added to indicate this is congenital
    - Heart Murmur
      - 785.2 Undiagnosed cardiac murmur
    - If chromosomes show trisomy 21, then change to 758.0
Case 2

- **Newborn infant**
  - Hypotonic
  - Severe feeding problems requiring gavage
  - Not dysmorphic

- **Differential diagnosis**
  - Chromosome anomaly
  - Spinal muscular atrophy
  - Congenital muscular dystrophy
  - Inborn error of metabolism
  - Other
Case 2

- Clinical diagnosis—Unknown
- Coding
  - Option 1: Hypotonia
    - 781.3 (description actually ataxia NOS, muscular incoordination)
    - Could add 779.89 (specified condition originating in the perinatal period) to indicate this is congenital
  - Option 2: Poor feeding
    - 779.31 Feeding problems in newborn
- Diagnostic testing demonstrates abnormal methylation pattern on chromosome 15 consistent with Prader-Willi syndrome
  - 759.81 Prader-Willi syndrome
Case 2

- What if diagnostic testing demonstrates abnormal methylation pattern on chromosome 15 consistent with Angelman syndrome?
  - Index references 759.89 Other under Other specified anomalies
  - Note there is not the same specificity as there is for Prader-Willi
  - If methylation abnormality was secondary to a deletion on chromosome 15, would 758.33 (other microdeletions) or 758.39 (other autosomal deletions) be more specific?
Case 3

• 18 month old presents with delays in gross and fine motor development and speech
  – Normal growth, physical and neurologic examination.
  – No dysmorphic features
Case 3

- Clinical diagnosis - Global Developmental Delay
- Coding
  - 783.42 Delayed milestones
  - 315.5 Mixed developmental disorder
  - 315.8 Other specified delays in development
  - 315.9 Unspecified delay in development
    - NOTE 315 codes exclude that due to a neurological disorder (320.0-389.9)
  - V79.3 Developmental handicaps in early childhood
    (Subheading under Special screening for mental disorders and developmental handicaps)
      - Probably more appropriate to use in the context of a ‘special screening’ although that is not defined
      - V codes may not be reimbursed
Case 4

- 30 year old presenting for evaluation of genetic condition
  - History of repaired congenital heart disease
  - Has cleft of the soft palate
  - Learning disabilities in school
  - Mildly unusual facial features
  - Son recently diagnosed with deletion 22q11.2
Case 4

• Clinical diagnosis—Suspected velo-cardio-facial syndrome due to deletion of chromosome 22q11.2

• Coding
  – Option 1: 758.32 Velo-Cardio-Facial syndrome
    • Diagnosis strongly based on clinical findings and family history, but not confirmed by testing
  – Option 2: Use congenital malformations (740-759)
    • Congenital heart malformation (745-747)
      – Would need to code to maximal specificity, i.e. what heart lesion was repaired? (e.g. 745.4 Ventricular septal defect)
    • Soft palate cleft (no specific code for this)
      – 749.00 Cleft palate, unspecified
      – 749.02 Unilateral, incomplete (Cleft uvula is included)
      – 750.29 Other specified anomalies of pharynx (index maps velopharyngeal incompetence to this code)
Case 4

• Coding (continued)
  – NOTE Have had payers deny payment for adult with a congenital anomaly stating they were “too old”
  – Option 3 Focus on indication for testing
    • V19.5 Family history of congenital anomalies
    • V26.39 Other genetic testing of male
    • V82.79 Other genetic screening
Case 5

• 38 year old woman presents for genetic testing
  – 35 year old sister recently diagnosed with breast cancer
  – Mother died of breast cancer at age 50 (diagnosed age 42)
  – She has had a normal breast MRI 2 months ago

• Diagnosis - none
  – She has no diagnosed disease to be coded
  – V 16.3 Family history of malignant neoplasm--breast
Case 5

• The patient relates her sister with breast cancer had BRCA testing and a mutation was found
  – Could still use V 16.3
  – V26.31 Testing of female genetic disease carrier status
    • Probably implies more traditional recessive disorder testing (although carrier is not defined)
  – V26.32 Other genetic testing of female

• Patient is found to carry the same deleterious mutation as her sister
  – V 84.01 Genetic susceptibility to malignant neoplasm of breast (V 84 codes used for a “Confirmed abnormal gene”)
Conclusions

• Providers are required to code to the highest specificity
• ICD-9 is not sufficiently detailed to support the requirement requiring compromise (and creativity)
• Code definitions and explanations are not adequately explicit leading to ambiguity of code assignment
Conclusions

• Genetic codes are scattered throughout the numeric and V codes and are not logically organized
  – Index useful at times
  – Important to develop some familiarity with codes for more commonly encountered clinical conditions

• Coding resource available through GPCI
Questions?
Thank you for your participation!

For more information, please contact
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www.medicalhomeinfo.org/GPCI.aspx

Coding questions may be sent to the coding hotline at aapcodinghotline@aap.org
Time Out for Genetics

Registration is now open for

“Top 10 Genetics Resources for Pediatric Primary Care Providers”

Thursday, October 25
12:00 - 12:30pm Central

https://www2.gotomeeting.com/register/702173234