The Coding and Reimbursement Landscape: An Impediment To Access

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October 12, 2022

What Genetic Testing Logistics Issues Need to be Addressed?
Realizing the Potential of Genomics across the Continuum of Precision Health Care

What are the key barriers in the logistics process for genetic testing?

- Let’s not be too hard on ourselves
- “It's supposed to be hard. If it were easy, everyone would do it.”
  — Tom Hanks in A League of Their Own
- Genetic testing is a relatively new discipline
  - It is complicated for people who don’t live and breathe it every day
  - Molecular geneticists and laboratory professionals are discovering new techniques, relationships and services faster than any other area of laboratory medicine
  - The way we report and are paid for services have not kept pace with scientific advances
- Cristi reminded us that this is a tool. Candice, Keri and Cristi made the point that this tool is only useful in the context of delivering appropriate care (e.g., making sure the calcitonin is done, and then followed)

So what are the system barriers and what should we be thinking about to bust them?

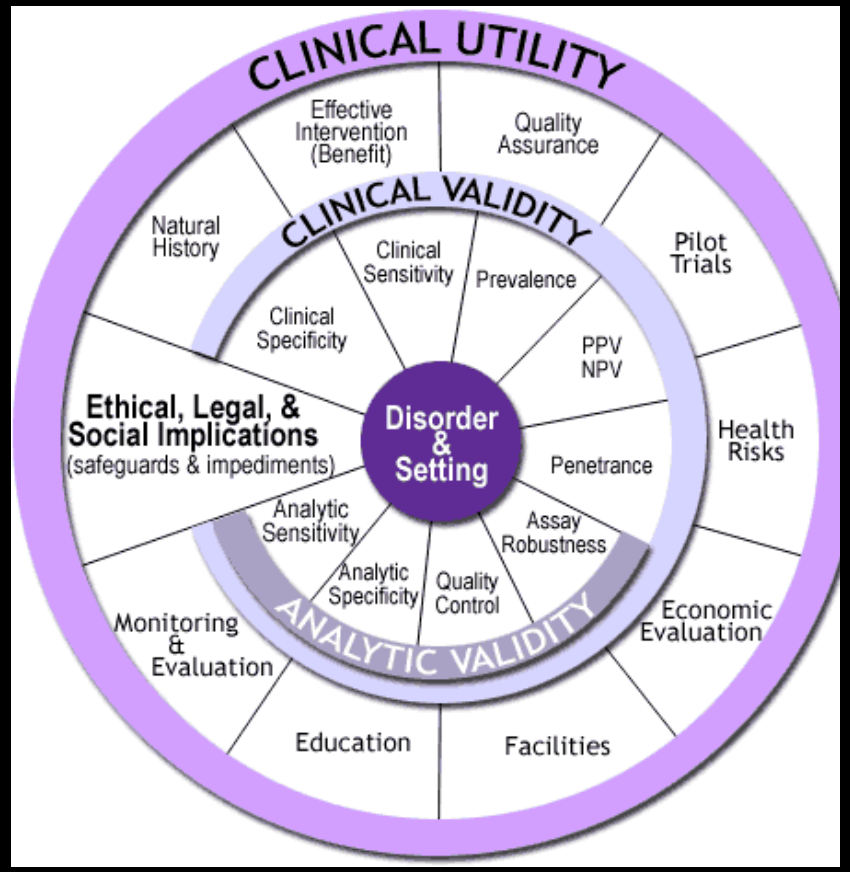
- Knowledge
  - Most have general awareness, but a limited understanding of genetics and the role in precision medicine
  - Patients, Providers (physicians, genetic counselors, laboratory professionals, etc.), Payers and employers, Public
- Language
  - Providers struggle with far less complex testing (e.g., Vitamin D)
  - ALK, BRAF, CDKN2A, CEBPA, DNMT3A, EGFR, ERBB2, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, KRAS, MLL, NPM1, NRAS, MET, NOTCH1, PDGFRA, PDGFRB, PGR, PIK3CA, PTEN, RET
  - Unless you work with these every day, this looks like some sort of cult that has been concocting a language to confuse even the most astute providers!
  - Candice talked about competency and literacy. We rely on GCs, geneticists AND mental health professionals to translate this into something meaningful to patients
- Complexity

Beyond language, there is considerable complexity in what to order and where benefits lie

- Indications for testing but this is just about getting the test
  - Diagnostic
  - Prognostic
  - Predictive testing
  - Carrier status for recessive conditions
  - Therapeutic response (e.g., pharmacogenetic testing)
- Panel variation among laboratories offering testing for the same condition
- Strong interplay between indication, history, ethnicity, known genetics, and patient preferences

The ACCE Model continues to provide a useful guidepost for examining “value”

- The laboratory is generally comfortable with analytic and clinical validity
- Addressing clinical utility has been the largest challenge for new and emerging genetics. The “so what” factor – just because you can measure it doesn’t mean you necessarily should
- Payers continue to establish high bars for clinical utility before making positive coverage determinations for most tests



CDC Office of Public Health Genomics (OPHG)

Genetic testing remains disproportionately costly compared to other laboratory diagnostics

- Medical appropriateness (NAS, 1990) "the expected health benefit [exceeds] the expected negative consequences. . . by a sufficiently wide margin that the procedure [is] worth doing“
  - A pure medical risk benefit model would exclude cost but in a constrained system a purely medically appropriate service may be more limited
  - Medical necessity subsequently emerged to try to address this
    - It would be unethical to enter a study subject into an arm that did not get the service
    - The service should be part of a basic benefit plan
- New technologies cost more, but technology drives costs down
  - Clearly it is more efficient to use an ion-selective electrode to measure electrolytes than to use a flame photometer
  - NGS technologies have generally replaced manual sequencing procedures
    - And soon the “norm” will be technical genome (exome) testing – with potential downstream reanalysis
    - Dr. Ashley reminded us that the technical cost is now $100 to $200
  - But in some areas the costs and reimbursement do not reflect technological advances

But with just about 12 minutes, let me focus on the coding and reimbursement challenges

Remember molecular diagnostics stacked codes until 2013?

Code	Descriptor
83890	Molecular isolation or extraction, each nucleic acid type (ie, DNA or RNA)
83891	Isolation or extraction of highly purified nucleic acid, each nucleic acid type (ie, DNA or RNA)
83892	enzymatic digestion, each enzyme treatment
83893	dot/slot blot production, each nucleic acid preparation
83894	separation by gel electrophoresis (eg, agarose, polyacrylamide), each nucleic acid preparation
83896	nucleic acid probe, each
83897	nucleic acid transfer (eg, Southern, Northern), each nucleic acid preparation
83898	amplification, target, each nucleic acid sequence
83900	amplification, target, multiplex, first 2 nucleic acid sequences
+83901	amplification, target, multiplex, each additional nucleic acid sequence beyond 2
83902	reverse transcription
83903	mutation scanning, by physical properties (eg, single strand conformational polymorphisms [SSCP], heteroduplex, denaturing gradient gel electrophoresis [DGGE], RNA'ase A), single segment, each

Code	Descriptor
83904	mutation identification by sequencing, single segment, each segment
83905	mutation identification by allele specific transcription, single segment, each segment
83906	mutation identification by allele specific translation, single segment, each segment
83907	lysis of cells prior to nucleic acid extraction (eg, stool specimens, paraffin embedded tissue), each specimen
83908	amplification, signal, each nucleic acid sequence
83909	separation and identification by high resolution technique (eg, capillary electrophoresis), each nucleic acid preparation
83910	interpretation and report
83913	RNA stabilization
83914	Mutation identification by enzymatic ligation or primer extension, single segment, each segment (eg, oligonucleotide ligation assay [OLA], single base chain extension [SBCE], or allele-specific primer extension [ASPE])
88384	Array-based evaluation of multiple molecular probes; 11 through 50 probes
88385	51-250 probes
88386	251-500 probes

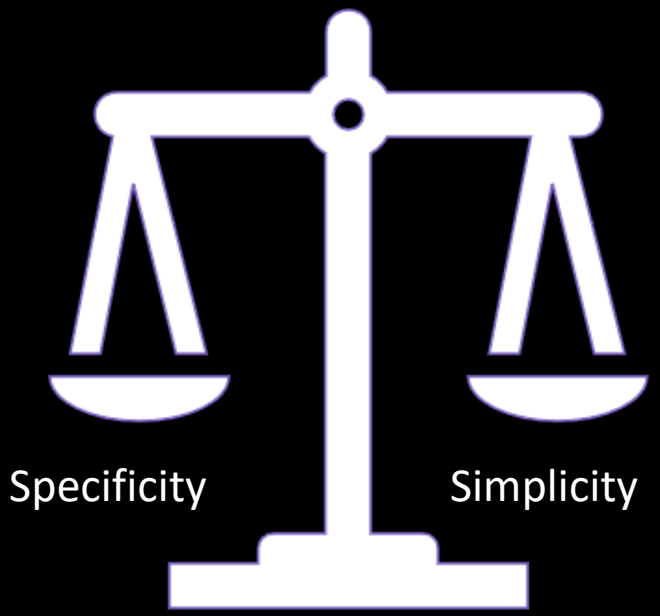
Then we moved to gene specific codes (sort of)

- Codes include all analytical services performed (eg, cell lysis, nucleic acid stabilization, extraction, digestion, amplification, and detection). Any procedures required prior to cell lysis (eg, microdissection) should be reported separately.
- Tier 1 Specific molecular pathology procedures/genes
  - Series 81161-81383
  - Initial list included services describing 90% of the services provided (using data mostly from reference labs), not 90% of the genes
  - More codes have moved to tier 1 over time based on general volume criteria
- Tier 2 Grouped procedures/genes based on work complexity
  - Series 81400-81408 (but specificity is lacking)
- Unlisted molecular pathology procedures
  - 81479 (and no specificity here)

The original pricing of these tests was based mostly on labor intensive, individual gene tests

- While this was the case, technology continued to advance
- Multianalyte assays with algorithmic analyses began emerging also in around 2013
- And the laboratory community began to develop genomic sequencing procedure coding in 2015
  - Now there are close to 50 codes in the GSP section
  - But this section is neither comprehensive nor all too specific for individual laboratory performance
  - These GSP procedures identify the indication but also do not entirely solve the ambiguity problem due to variation in procedures offered
- Proprietary laboratory analysis (PLA) codes began in 2017
  - They are usually laboratory specific, but come with some payment challenges

There's been an ongoing dialog regarding specificity and simplicity



- Simplicity: Payers and others sought a more streamlined solution to the single gene approach
  - How can there be so many codes for a molecular service and only one code for complex surgical procedures?
  - **47135** Liver allotransplantation, orthotopic, partial/whole, cadaver or living donor, any age
- Specificity: How can a payer adjudicate a claim without knowing what services were actually performed?

As complex as genetic testing is, so is the complexity of the way services are reported

- Current Procedural Terminology (AMA-CPT®)
  - The recognized HIPAA codeset for reporting laboratory procedures
- Genetic Test Registry (NIH GTR®)
- Palmetto MolDx DEX™ Registry
- Concert Genetics Coding Engine

- And services and reporting is changing quickly
  - But not quite as fast as the science

So where are we with respect to realizing the potential of genomics across the continuum of precision healthcare?

- Need clear evidence for clinical validity and most importantly clinical utility
- General agreement on what constitutes an appropriate genetic evaluation for a patient with a specific condition
  - Curb fraud, waste, and abuse that emerges from uncertainty
- Reexamine what constitutes a genetic test to prep for the future
  - Genome sequencing (technical component) with downstream analysis and reanalysis of data
  - One code to report technical component, separate codes for data analysis by laboratory professionals and geneticists.

Should people with limited genetic knowledge be expected to order the right genetic test(s)?

- In clinical medicine, for patients with complex and uncertain conditions, ordering a specialty consultation is the expected outcome
- What if the ordering clinician presents the problem to the laboratory (6 month old with hearing deficit, 26 year old with family history of breast and ovarian cancer), allowing the laboratory to perform needed testing?
  - Certainly easier if the specialty goes to whole genome technical analysis
  - Reimbursement systems are challenged by reflex testing
    - The laboratory cannot add on additional diagnosis codes
    - The original diagnosis codes may not support downstream testing
  - Consider restructuring the order form around the clinical problem rather than the diagnostic test
- Genetic test reports have improved, but are still too complex for most community practice physicians
  - Continue to refine this process that the CDC and others have championed