Economics of Genomic Medicine

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Views of the Diagnostic Industry

Based on Previous Workshops and Personal Opinion

- Whole Genome/Exome Sequencing is initially entering clinical practice informally via academic medical centers and biotech CLIA labs (not reimbursed). Large CLIA labs may offer WGS depending on market, reimbursement, and content of reports
- Path to approval of an FDA-approved WGS instrument / reagent system is currently unclear, with complex intended use(s), accuracy problems, no gold standard for comparison, rapid technical obsolescence (uncompetitive with LDTs), and a potential requirement for lengthy and costly prospective treatment-by-genotype clinical outcome studies
- Need a patient-oriented, medical value-based system of test reimbursement rather than a technology-based CPT system

Overall Hurdles to Translating Genomic Medicine

- Need for a national, dynamically updated, interpretative database of evidence for clinical utility of genetic variants
- A means to convey updates to patient and/or physicians
- Absent evidence of clinical utility and cost-effectiveness, private and public payers may default to non-reimbursement
- Basis for reimbursement of WGS is uncertain; at $1-8,000 per interpreted indication, single clinical indications may not be cost-effective (except perhaps for cancer indications). How to assess cost-effectiveness of WGS for multiple clinical indications over a lifetime?
- Due to inaccuracy issues, targeted confirmatory testing of some actionable variants identified by WGS may be necessary and will increase overall costs

Research Needs of CLIA Diagnostic Laboratories

- Professional organization guidelines for reporting genetic variants
- Acceptance by stakeholders of different evidentiary standards for clinical utility of tests for different medical condition; e.g., CDC's EGAPP group as a start
- A national, dynamically updated, interpretative database of evidence for clinical utility of genetic variants; e.g., NIH's Genetic Test Registry or Cancer Genome database or 23andme approach
- Government-sponsored prospective randomized clinical outcome studies; e.g., NHLBI's clinical trial (COAG) of warfarin dosing by clinical + genetic information vs clinical information alone. Studies by Patient-Centered Outcome Research Institute
- Cost-effectiveness models based on clinical outcome studies; e.g., incremental cost per QALY gained is highly sensitive to test cost (Schiffman, et al, *Clinical Therapeutics*; 34:1387-94, 2012)