A Primary Care Provider View of Translating Genomic Innovation

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Key Points

- The importance of primary care
- The importance of authoritative advice to clinicians and patients
- The importance of getting it right for genomic innovations

Primary Care: The Front Line for Medical Care

FAMILY MEDICINE

GENERAL INTERNAL MEDICINE

GENERAL PEDIATRICS

(OBSTETRICS AND GYNECOLOGY)

- Account for more than half of all office visits to physicians in the US — estimated more than 500,000,000 in 2006
- Personal medical home
- First contact for most patients
- Comprehensive
- Continuous
- Community- and population-focused

Primary Care Physicians See Common Problems

- Specialize in breadth of knowledge and expertise
- Recognize patterns that suggest the unusual
  - Need information systems and decision support
- Typically high volume of patient visits means that support systems must work in time all the time
- Medical tests and interventions must be appropriate for populations in which rare conditions are rare
  - Tests with even small errors have magnified effects
  - Often most positives are false positives, requiring unproductive and expensive further testing

Primary Care Physicians are Relentlessly Practical

- **A new test or innovation must**
  - Be available, feasible, and acceptable to the patient
  - Do what it says it does
  - Be accurate
  - Be reproducible
  - Improve clinical outcomes that patients would notice and care about compared to current practice
  - Not increase adverse effects
  - Be worth it (cost-effective)

Clinicians Need Authoritative Advice

- No one can keep up
- Make sense out of voluminous literature
- Deal with complex decisions
- Improve quality of decision making
- Provide justification to patients, payers, legal system

How Are Guidelines Useful?

- Transmit medical knowledge
- Assist patient and physician decisions
- A way to set clinical norms
- Quality improvement
- Privileging and credentialing
- Payment and cost control
- Medicolegal evaluation

Hallmarks of an Evidence-Based Guideline

- Explicit
- Transparent
- Publicly accountable

General Characteristics that Should be Specified (IOM)

- Clinical condition
- Health practice
- Target population
- Health care setting
- Type of clinician
- Purpose
- Source and sponsorship

Process Characteristics (AHRQ)

- Panel selection
- Problem specification
- Literature search strategy
- Literature analysis
- Evidence summarization
- Recommendation rationale
- Clinical outcomes
- Sensitive to cost and practicality

Desirable Attributes (AHRQ)

- Valid
- Reliable
- Applicable
- Flexible
- Clear
- Multidisciplinary
- Reviewed
- Documented

What About Genomics?

- Primary care physicians are skeptical of "genetic exceptionalism"
- Many non-genomic tests in current use produce the same kinds of information promised for genetic tests:
  - Risk
  - Prognosis
  - Response to drugs and other therapies
  - Have ethical, legal, and social consequences

What About Genomic Tests?

- Thousands already available
- Little regulation — buyer beware
- Direct-to-consumer and direct-to-physician marketing
- Clinicians and consumers need reliable advice
- Precedent of the United States Preventive Services Task Force that evaluates preventive interventions — AHRQ

The EGAPP Initiative

- **E**valuation of
- **G**enomic
- **A**pplications in
- **P**ractice and
- **P**revention

EGAPP Working Group

- CDC principal sponsor, partner with AHRQ evidence centers
- Non regulatory
- Independent, non-federal, multidisciplinary
- Minimize conflicts of interest
- Evidence-based, transparent, and publicly accountable

Reviews Underway

- Testing for early detection of ovarian cancer
- Testing before placing a patient on an antidepressant drug
- Testing for family-related colon cancer
- Testing for response to treatment for colon cancer
- Genetic profiling for cardiac risk
- Breast cancer gene expression profiling

EGAPP Experience So Far

- **Quantity and quality of evidence supporting testing in typical practice settings is disappointing**
  - Weak research designs in published articles
  - Some potentially important data are proprietary
  - Scant evidence on potential benefits and harms
  - No head-to-head comparisons with current practice
  - Not tested in typical patient populations
  - Little information about cost and cost-effectiveness compared with current practice
  - No information about ethical, legal, and social implications, especially for family members

Conclusions

- Genomic innovation to assess risk or guide therapy holds great promise
- Recognize importance of appropriateness in primary care settings
- New tests and technologies must improve on what we have

Conclusions (cont.)

- There will likely be few examples of genetic tests that meet standards for common use in typical practices in the next 3-5 years
- Enormous need for more and better quality research on effects of testing on clinical outcomes (good and bad), with results publicly available