COMMUNICATION:
Key to Appropriate
Genetic Test Referral, Result Reporting, Interpretation, and Use

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Public Health Practice Program Office
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Questions

How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting, and use of genetic tests and results?

1. What variability exists in the ordering and reporting of genetic tests and results?

2. What issues have arisen within the clinical practice and laboratory setting in the use of genetic testing services?
   - patient outcomes
   - other costs

3. What efforts are can be considered to assure the appropriate ordering of genetic tests and reporting of results?
   - process issues
   - an informed/educated workforce and public
Assessing Laboratory Practices

Quality Assurance in Molecular Genetic Testing Laboratories
Margaret M. McGovern, MD, PhD
Maria O. Benach
Sylvan Wallenstein, PhD
Robert J. Demick, PhD, MD
Richard Keenlyside, MD, MS

(1999)

Context: Specific regulation of laboratories
JAMA. 1999;281:835-840

Personnel Standards and Quality Assurance Practices of Biochemical Genetic Testing Laboratories in the United States
Margaret M. McGovern, MD, PhD; Maria Benach, BA; Sylvan Wallenstein, PhD; Joe Boone, PhD; Ira M. Lubin, PhD
ARCH PEDIATR-ADOLESC MED—VOL 157, JANUARY 2003

(2002)

Medical genetic test reporting for cystic fibrosis (ΔF508) and factor V Leiden in North American laboratories
Hans C. Anderson, MD
Marie A. Krousel-Wood, MD, MSPH
Kelly E. Jackson, MS
Janet Rice, PhD
Ira M. Lubin, PhD

(2002)

Physicians’ perceived usefulness of and satisfaction with test reports for cystic fibrosis (ΔF508) and factor V Leiden
Marie Krousel-Wood, MD, MSPH
Hans C. Anderson, MD
Janet Rice, PhD
Kelly E. Jackson, MS
Eunice R. Romer, DDS
Ira M. Lubin, PhD

(2003)
### Content Summary of Requisition Forms and Result Reports for Cystic Fibrosis Molecular Genetic Testing

<table>
<thead>
<tr>
<th></th>
<th>Percent (N)</th>
<th>Reports (N=28)</th>
<th>Percent (N)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Requisitions (N=17)</strong> (unpublished data (2003))</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Indications for testing</td>
<td>88% (15)</td>
<td>Clinical Indications</td>
<td>64% (18)</td>
</tr>
<tr>
<td>Clinical information</td>
<td>59% (10)</td>
<td>Detection rate</td>
<td>86% (24)</td>
</tr>
<tr>
<td>Family information</td>
<td>41% (7)</td>
<td>Adjusted risk</td>
<td>71% (20)</td>
</tr>
<tr>
<td>Ethnicity</td>
<td>94% (16)</td>
<td>Ethnicity</td>
<td>21% (6)</td>
</tr>
<tr>
<td>Pedigree</td>
<td>47% (8)</td>
<td>Interpretation</td>
<td>93% (26)</td>
</tr>
<tr>
<td>Pregnancy status</td>
<td>59% (19)</td>
<td>Genetic Counseling</td>
<td>61% (17)</td>
</tr>
</tbody>
</table>

Follow-up study:

**Physicians' Perceived Usefulness and Satisfaction with Test Reports for Cystic Fibrosis (ΔF508) and Factor V Leiden**

In this study, we found physicians desired a more comprehensive report useful for guiding clinical decision-making.

Some Recommendations / Standards Out there

CLIA
NCCLS

How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting and use of genetic tests and results?
Conference Process

1. Multi-disciplinary

2. One and a half day conference/workshop.

3. Orientation talks / panel discussion

3. Case-based discussion:
   1. Carrier testing for cystic fibrosis with a relative with CBAVD
   2. Carrier testing for cystic fibrosis (without a family history for CF)
   3. Carrier testing for cystic fibrosis (with a family history)
   4. Diagnostic testing for cystic fibrosis - infant with failure to thrive
   5. Prenatal diagnosis

4. Focus on pre- and post-analytic testing processes.
Organizational affiliation of Attendees
(attendance at the meeting does not imply endorsement)

Federal Government Agencies
Centers for Disease Control and Prevention, Department of Health and Human Services
Centers for Medicare and Medicaid Services, Department of Health and Human Services
Health Resources Services Administration, Department of Health and Human Services
Office of Science and Data Policy, Office of the Assistant Secretary for Planning and Evaluation, Department of Health and Human Services

Professional Organizations, Academics, and State entities
American Academy of Family Physicians
American Academy of Physician Assistants
American Association of Pediatrics
American College of Gynecology and Obstetricians
American College of Medical Genetics
American College of Nurse Midwives
American Medical Association
Association of Molecular Pathologists
Association of Public Health Laboratories
Association of Family Practice Residency Directors
Association of Women's Health, Obstetric, and Neonatal Nursing
Blue Cross and Blue Shield Association
GeneTests
Genetic Alliance
Genetics and Public Policy Center, Johns Hopkins University
International Society of Nurses in Genetics
March of Dimes Foundation
Minnesota Department of Health - Minnesota Children with Special Health Needs
Mount Sinai School of Medicine
National Coalition for Health Professional Education in Genetics
National Society of Genetic Counselors
New England Newborn Screening Program
St. Vincent's Hospital Cystic Fibrosis Center
Tulane University Health Sciences Center
Wadsworth Center, New York State Department of Health

International participation
Cystic Fibrosis Thematic Network
Organization for Economic Cooperation and Development
<table>
<thead>
<tr>
<th>Issue</th>
<th>Recommendation</th>
</tr>
</thead>
<tbody>
<tr>
<td>No standard format/process for requisitions and reports</td>
<td>Develop/evaluate standard practices</td>
</tr>
<tr>
<td>Limited data is available to quantify impact on patient outcomes and other costs.</td>
<td>Studies to collect and evaluate such data.</td>
</tr>
<tr>
<td>Practice/setting-specific guideline implementation and evaluation plans are virtually non-existent.</td>
<td>Develop such plans</td>
</tr>
<tr>
<td>Reimbursement issues</td>
<td>Provider/laboratory/payer forum</td>
</tr>
<tr>
<td>Role of the genetic laboratory - &quot;Consultant&quot; and/or &quot;Provider&quot; of test results</td>
<td>Develop provider/Laboratory partnerships</td>
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</tbody>
</table>
A Few Words on Roles

1. **Physicians, physician assistants, nurses, other allied health professionals** have contact with patients.

2. **Geneticists/specialists** (clinical, laboratory, counselors, etc.) are far fewer in number. (i.e. laboratory's role- "consultant" and/or "provider" of test results)

3. **Public Health** provides assessment, policy, and assurance roles that can be critically important toward assuring the appropriate use of genetic testing.

4. **Consumers** are the decision makers
Next Steps

Domestic:
1. Document findings (conference summary)
2. Quantify impact of practices on patient outcomes and other costs
3. Development of appropriate standards/guidelines
4. Partnerships with organizations (follow up conference?)
5. Identify gaps in information being provided to the professionals and the general public for making educated and informed decisions.
6. Develop efforts to provide "missing information".
   ⇒ community based
   ⇒ make use of information technology tools
   ⇒ evaluate usefulness

International:
1. Comparative international analysis of reporting practices
   (working with Cystic Fibrosis Thematic Network, Association of Molecular Pathologists, and Mt. Sinai School of Medicine)
2. Serving on the OECD* steering committee of quality assurance and proficiency schemes
   (*Organization for Economic Cooperation and Development)
International Efforts

Molecular Diagnostic Methods for Genetic Diseases; Approved Guideline

This document provides guidance for the use of molecular biological techniques for clinical detection of heritable mutations associated with genetic disease. A guideline for global application developed through the NCCLS consensus process.

Draft Best Practice Guidelines for Reporting

Payne S.
Kennedy-Galton Centre for Medical and Community Genetics, North West London Hospitals NHS Trust, London, United Kingdom.

These guidelines are an update of version 1 (issued January 1997; see CMGS website - www.cmgs.org) modified in light of experience scoring reports returned to the EQA steering committee during the 1997 and 1998 UK GA rounds. The aim has been to highlight some of the best (and worst) features of the reports returned. Guidelines prepared by Stewart Payne (stewart Payne).
How can we ensure that health-related decision making in clinical and laboratory practice is based upon the proper ordering, reporting and use of genetic tests and results?