Developing QC Materials For Genetic Testing
Projects: 1998-2003

- Materials are needed for PT/QC and test development/research for molecular genetic testing
  Contract with DynCorp Health Research Services, 1998

- Fund research - Process to collect positive samples, conduct inter-laboratory verification, and establish stable transformed cell lines
  Contract with Duke University, 2000

- Fund research - Genetic engineering to develop positive samples and conduct inter-laboratory verification
  Contract with UCLA, 2000
 QC Materials Conferences

- Successful research leading to a reliable process for collecting and validating cell lines
- September 15-16, 2003
- March 8, 2004
- November 9, 2004

- **Purpose:**
  To develop a sustainable process to collect, store, validate, and distribute materials at a reasonable cost
QC Materials for Genetic Testing

- **Conference Organizers**
  CDC, NIH, NIST

- **Participants**
  Professional organizations
  Government agencies
  Industry
  Laboratories
  Academic institutions
Reviewed current and future needs for materials for genetic tests

Reviewed research to produce materials suitable for positive QC for genetic tests

Discussed ways to develop sustainable process to provide QC materials to genetic testing laboratories at a reasonable cost.
September 2003: Workgroups
Next Steps

- Research
- Funding
- Professional Guidance
- Oversight of QC Products
- Validation of QC Materials
- Use of Cell Banks
- QC Material Priorities
- Material Contributors
March 8, 2004: Next Steps

- Federal leadership in supporting efforts
- Public/Private partnership is essential
- National QC Material Coordinator is needed
- Expert Panel to support Coordinator
- Protocols for collection, validation and distribution
- Incentive program for contributions
- Website of resources
- Presentations and publications about materials development
November 9, 2004 Agenda

- Coriell and other cell banks as repository and distributor of materials
- National QC Material Coordinator’s role
  - Activities in Community Process
  - Priorities
- Expert Panel to support Coordinator
- Protocols/Incentives for collection, validation and distribution of materials
- National and International Cooperation
- Communications and Website development
QC - steep climb - but achievable goal
DILBERT®

BY SCOTT ADAMS

REMEMBER, QUALITY IS OUR TOP PRIORITY.

QUESTION: IS IT MORE IMPORTANT THAN SAFETY?

OOH... I FORGOT ABOUT THAT ONE.

QUESTION: IS QUALITY MORE IMPORTANT THAN OBEYING THE LAW?

WELL, PROBABLY NOT.

IF WE COULD MAXIMIZE SHAREHOLDER VALUE BY SELLING LOWER QUALITY ITEMS...

...WOULDN'T WE HAVE A FIDUCIARY RESPONSIBILITY TO DO IT?

I'M SURE IT'S IN THE TOP FOUR.

WHAT IF WE HAD TO LIE TO ACHIEVE QUALITY?
Promoting Quality
Laboratory Testing for Rare Diseases:
Keys to Ensuring Quality Genetic Testing

May 19-21, 2004
Atlanta, GA

EMORY UNIVERSITY
SCHOOL OF MEDICINE

Office of Rare Diseases

CDC
Rare Disease Testing Conference: May 19-21, 2004

- Assure access to quality laboratory testing
  - Improve oversight of research laboratories
- Expedite translation of gene findings into clinical and public health practice
- Identify data and education needs
- Promote collaboration, cooperation, partnership, and community involvement
What is a rare disease or condition?

- NIH Office of Rare Diseases
  - 1 in every 12 individuals in US

- Orphan Drug Act:
  - <1 /1,465 (affect < 200,000 in US)

- Safe Medical Devices Act of 1990
  - <1/1,000/yr ( <4000/y in US)

- Orphanet
  - <1 per 2,000 people in Europe

- 5 new diseases every week in medical literature: Over 6,000 diseases known today
Current Genetic Tests

GeneTests: April 2004

Total Tests 1,039

Clinical 694 (67%)
US: 542 (78%)
Non US Only: 152 (22%)

Research Only 354 (33%)
Testing Laboratories

GeneTests: April 2004

Total Laboratories 598

US 412 (69%)
  Clinical 247 (60%)
  Research Only 165 (40%)

Non US 186 (31%)

- “Research only” labs account for 40% of US labs listed in GeneTests
- Non US labs account for 31% of all labs listed in the directory
Testing Availability

GeneTests April 2004

Total Clinical Testing 694 Diseases

Testing available from only 1 lab 308 (44%)
Testing available from 2-5 labs 224 (32%)

Subtotal 532 (76%)

- CAP Molecular Genetics Survey: 17 tests
- EMQN: 13 tests, 1 sequencing
Summary

- Human genome: ~35,000 genes
- Genes with known sequence as of May 2004: 11,550
- New OMIM entries: 60-100 per month
- Current rare diseases: 6,000 – 7,000
- New rare diseases: ~20 per month (5/wk)
- Diseases for which clinical testing is available: 694
- New testing: <10 per month (2 in April 2004)
Rare Disease Conference Outcomes

- Formed North American Rare Disease Network
  - All network laboratories CLIA certified
  - Reports with limitations from CLIA laboratory
  - Work collectively to develop new tests
  - Foster research/clinical laboratory partnerships
  - Backup for sole source tests
Rare Disease Conference Outcomes

- American Society of Human Genetics and Office for Human Research Protections to provide education for researchers and IRBs
- NIH pilot programs to fund translation of research tests into clinically applicable tests
- 2005 meeting planned to assign responsibility for additional areas of focus – communication, coordination, roles
- Website: http://www.phppo.cdc.gov/dls/genetics
Identified Pathways to Quality for Rare Disease Testing and Testing Materials