



Genetic Testing Update

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CDC

Atlanta, GA

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SAFER • HEALTHIER • PEOPLE™

Developing



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



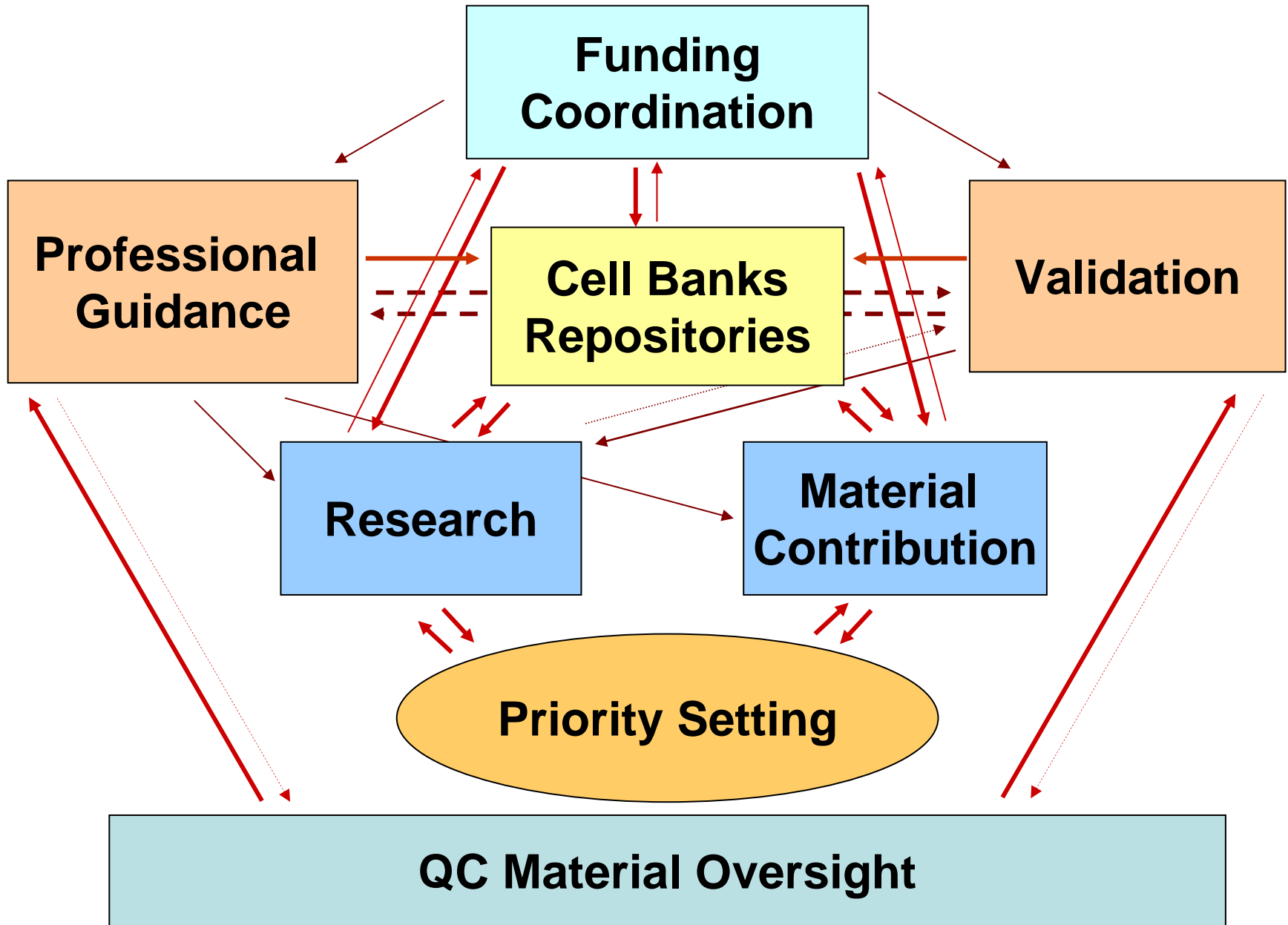
QC I – September 2003

- 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**

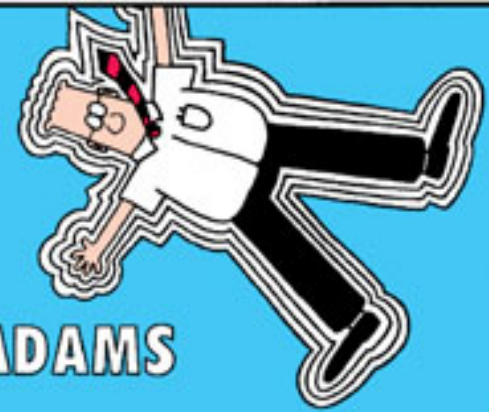


QC - steep climb – but achievable goal





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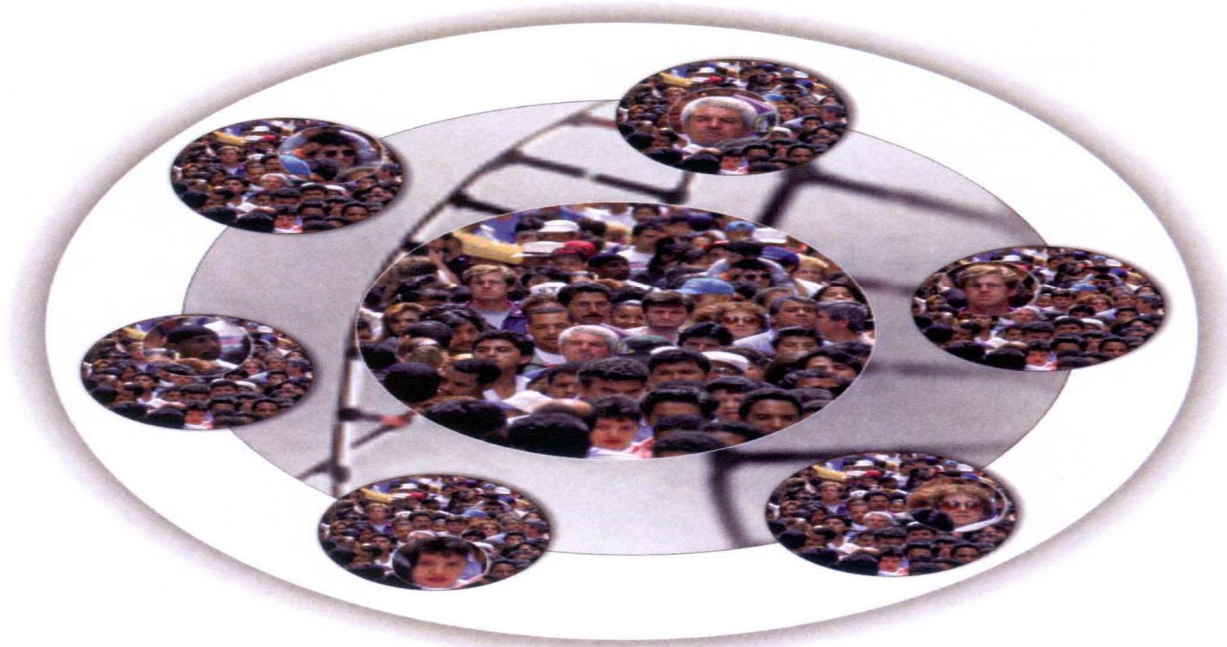


BY
SCOTT ADAMS

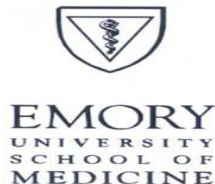


Promoting Quality

Laboratory Testing for Rare Diseases: Keys to Ensuring Quality Genetic Testing



May 19-21, 2004
Atlanta, GA





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
 - $< 1 / 1,000 / \text{yr}$ ($< 4000 / \text{y}$ 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

