Genetic Testing for Rare Diseases

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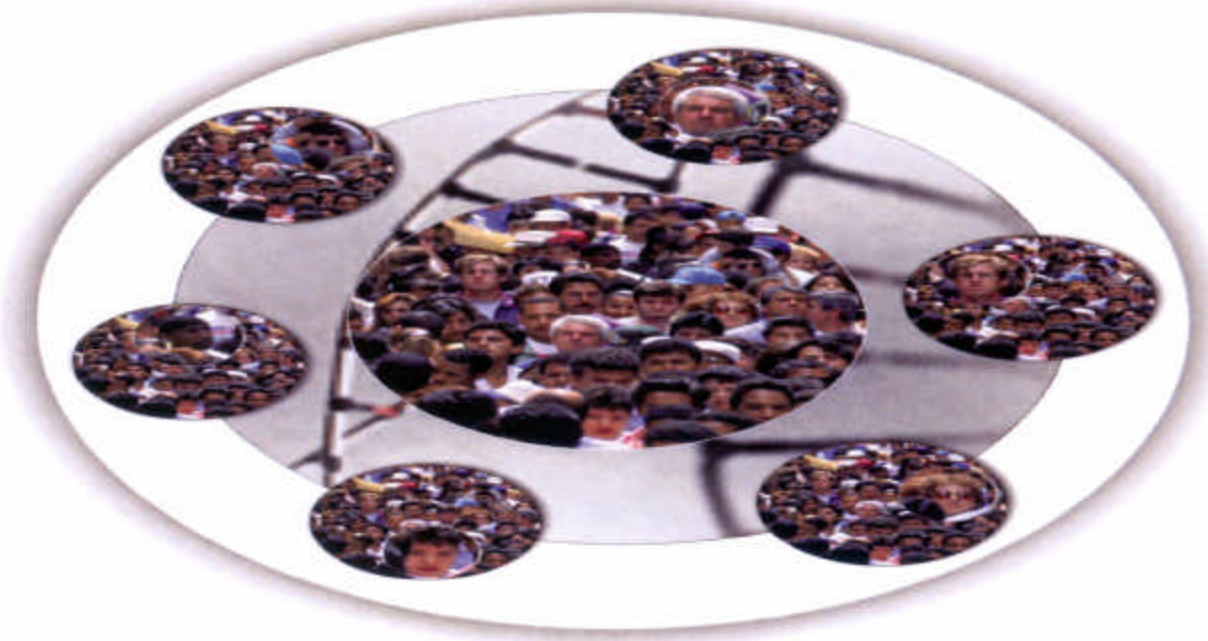
Promoting Quality

Laboratory Testing for Rare Diseases:

Keys to Ensuring Quality Genetic Testing



May 19–21, 2004

Atlanta, GA

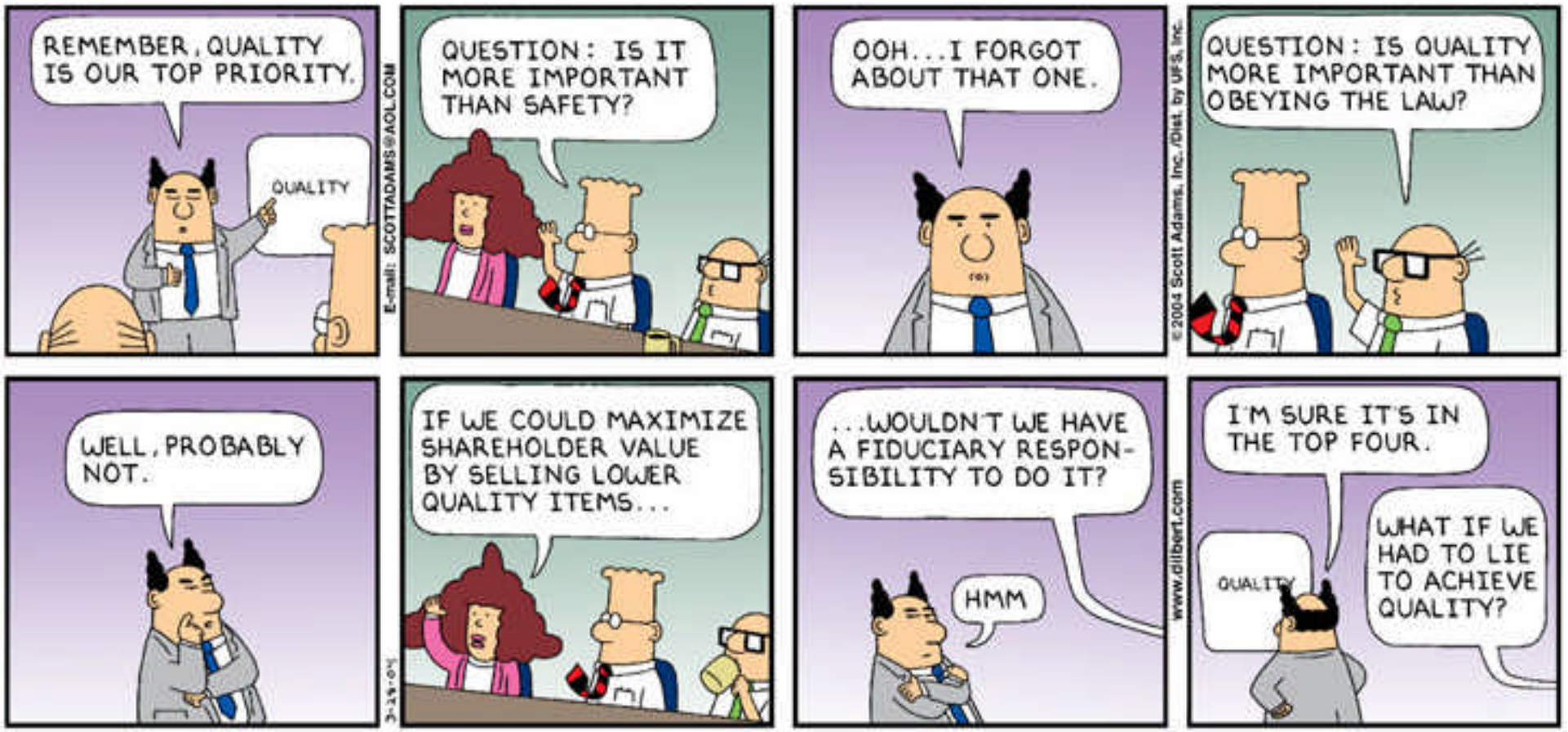
EMORY UNIVERSITY SCHOOL OF MEDICINE

CDC

Office of Rare Diseases

DILBERT®

BY SCOTT ADAMS



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

- Assure access to quality laboratory testing
  - Research laboratories providing patient testing
- Expedite translation of gene findings into clinical and public health practice
- Identify data and education needs
- Promote collaboration, cooperation, partnership, and community involvement

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%)

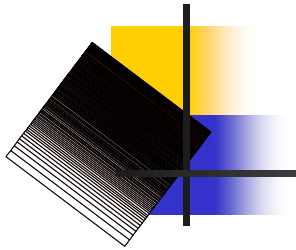
- “Research only” labs account for 40% of US labs listed in GeneTests
- Non US labs account for 31% of all labs listed in the directory

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)

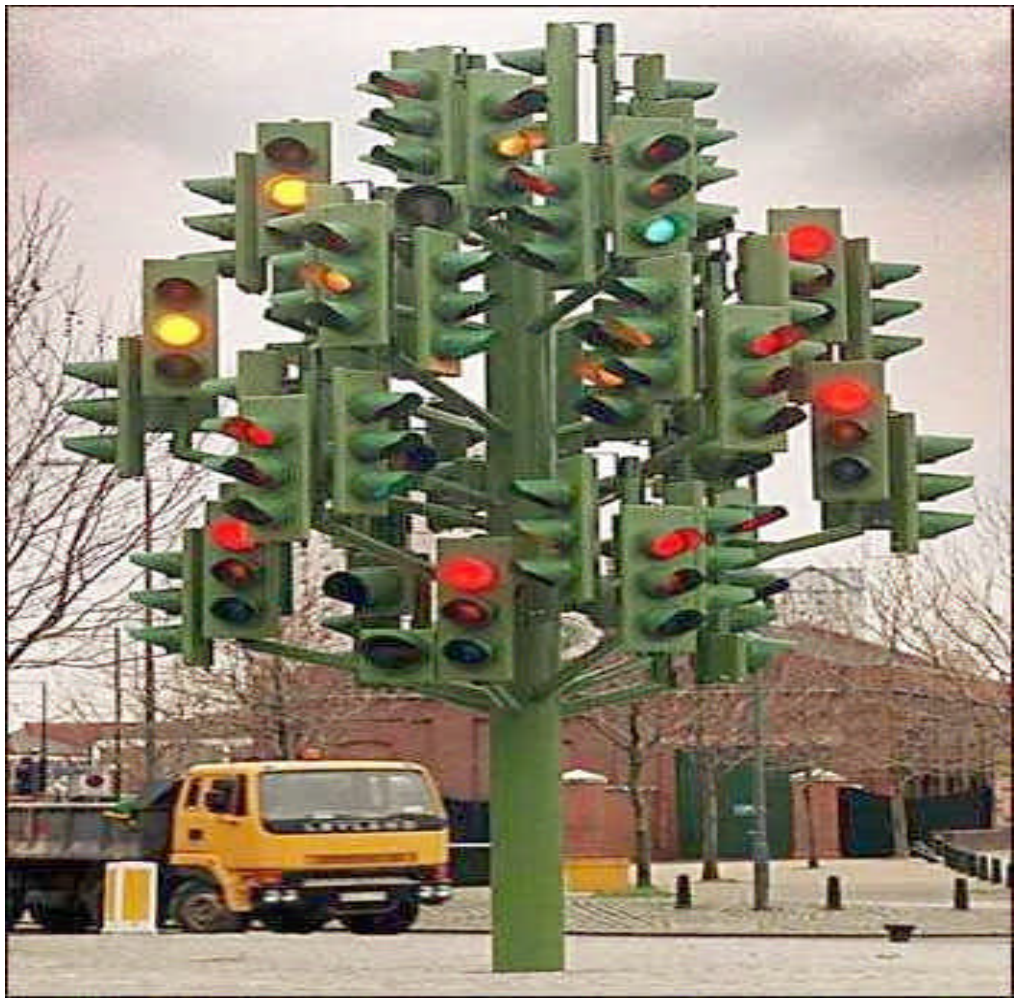
Rare Disease Conference Outcomes

- Formed North America National Network for Rare Disease Genetic Testing
- http://www.rarediseasetesting.org
  - **All network laboratories CLIA certified**
  - **Reports with limitations from CLIA laboratory**
  - **Work collectively to increase development of new tests**
  - **Foster research/clinical laboratory partnerships**
  - **Backup for sole source tests**
  - **Organizational Meeting – February 23, 2005**

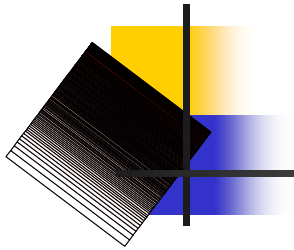
Rare Disease Conference Outcomes

- American Society of Human Genetics and Office for Human Research Protections to provide education for researchers and IRBs
- Expansion of 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

Pathways to Quality and Access for Rare Disease Testing



Genetic Testing for Rare Diseases: Building Bridges to the Future



- Workgroups formed
  - Vision – defining success
  - Infrastructure – systems and services
  - Networks – clinical laboratories and researchers
  - Quality assurance – national and international
  - Education – IRB and research communities

Genetic Testing for Rare Diseases: Building Bridges to the Future

- March 17 - Working meeting at ACMG
  - Evaluate progress of workgroups to date
  - Review plans for 2005 Rare Disease Conference
    - Identify major issues to address
    - Identify target audience and key attendees
    - Revise conference agenda, if needed
    - Assure broad-based participation

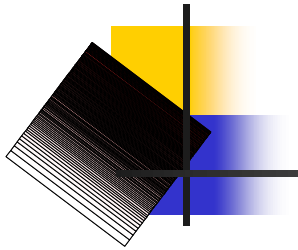
Rare Disease Testing:
Building Bridges to the Future

- Plans for September 2005 Conference
  - Title – Access to Quality Testing for Rare Diseases
  - Location – Washington DC area
  - Proposed Format –
    - Day 1 – Plenary sessions: reviews and overviews
    - Day 2 – Workgroups, Workgroup Reports, Next Steps

Rare Disease Testing: Building Bridges to the Future

- Expected Outcomes from 2005 Conference
  - Shared vision
  - Federal agency roles defined
  - Private sector roles defined
  - Defined system for moving selected tests from research to practice
  - Better definition of needs and service gaps

Rare Disease Testing:
NIH Support for Translational Research



- Diseases or Conditions - NIH Priority
- Support for transition – research to clinical
- Conditions for clinical laboratory participation in current NIH Program
  - Collaboration with researcher
  - CLIA certified
  - Offer test for 5 years at reasonable charge
  - Other?

Rare Disease Testing: Vision

- **Long-term vision:** Individual's health outcomes are better because of access to accurate rare disease tests: ease of access, usefulness of tests, follow-up and support after results

- **Short-term vision:** Rare disease tests should be accessible. Voluntary approaches to fixing the problems are most likely to succeed. This can range from:
  - completely passive approaches
  - listing anyone who says they can do something in Gene Tests
  - systems defining the minimal criteria that should be met to support the claim that a particular lab does some thing well.

Rare Disease Testing: Success

- **Patients, families and providers**: access to high quality services recognized by the health care system - private and public payers are willing and able to cover expenses.

- **Rare disease testing group**: not entrenching, not worrying about maintaining current systems, not using these problems to shore up current systems – instead examining the problems with an open mind, with a concern for the whole, not just where we each come from - more networking.

- **Systems and services –** Incentives that provide solutions – get the players to want to make necessary changes.

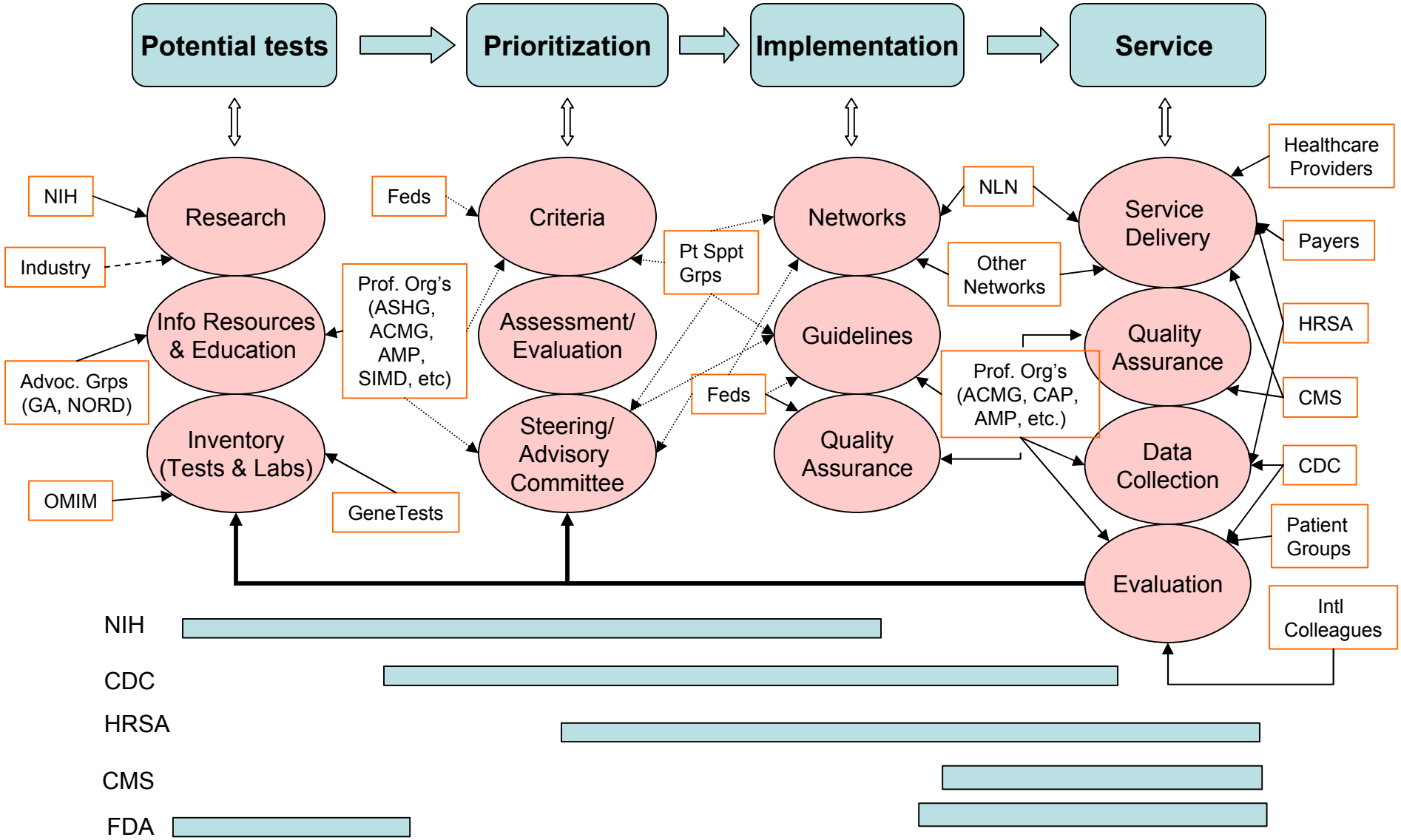
Rare Disease Testing: Evaluating Success

- **Poll advocacy groups**
  Pre and post survey of the concerns of:
  - Labs
  - Consumers/advocacy groups
  - CMS and other payers (specifically to considered accessible and what is not)
  - Monitor tests available versus known genetic cause (gene/mutation); monitor quality (adverse outcomes).

- **Removing roadblocks to success**

  - Define the roadblocks and create new models that generate the energy we need to move toward novel solutions
  - Avoid passions around people's territorial needs

Potential Process to Enhance Genetic Testing for Rare Diseases



Note: Dashed lines indicate potential roles and involvement to be discussed. Solid lines indicate current/existing roles and involvement.