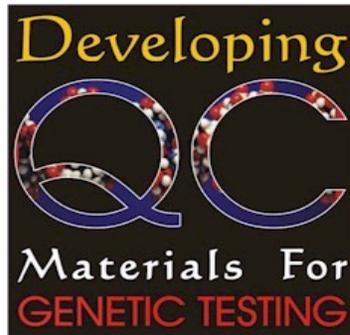


November 9, 2004

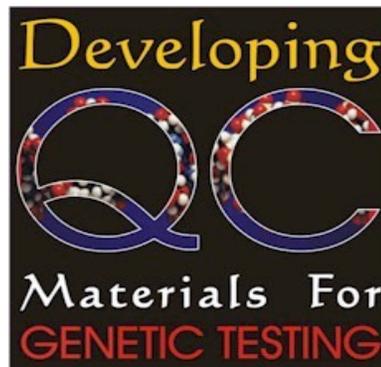
Los Angeles



**Immunoglobulin Heavy Chain Gene Analysis in Lymphomas: A Multi-Center Study Demonstrating the Heterogeneity of Performance of Polymerase Chain Reaction Assays. Bagg, et al., J. Molecular Diagnostics, 4 (#2), 2002.**

**“...19 different Association of Molecular Pathology (AMP) member Laboratories analyzed 29 blinded B cell and T cell lymphoid neoplasm samples Of extracted DNA and formalin-fixed, paraffin-embedded (FFPE) tissue.... There was, however, remarkable heterogeneity in the performance of, and the results obtained from, IgH PCR assays with diagnostic sensitivity ranging from 90% to as low as 20%, when evaluating the same specimens.... Importantly, there is a need for greater standardization to reduce the unacceptably high false rate of this crucial diagnostic assay.”**

**WE NEED TO DO BETTER THAN THIS !!!**



## **September 15-16 (2003) Meeting I - Atlanta**

- **Lack of appropriate positive materials for genetic tests:**
  - QC**
  - EQA/PT**
  - Test development**
- **Conference Organizers**
  - CDC, NIH, NIST**
- **Participants**
  - Professional organizations**
  - Government agencies**
  - Industry**
  - Laboratories**
  - Academic institutions**

# Developing QC Materials for Genetic Testing - III

## September 15-16 (2003) Meeting I - Atlanta

1. Review current efforts to develop QC materials for genetic tests
2. Identify areas of need and issues to be addressed for developing sustainable methods for provision of these materials

## Conclusions: Areas of Need

1. Develop a scheme for priorities for current and future needs
2. Develop networks of material contributors
3. Develop processes to use existing cell banks
4. Identify/facilitate QC research activities on QC material development
5. Develop validation processes for QC materials
6. Develop professional guidance on use of QC materials
7. Clarify regulatory oversight for providers and users of QC materials
8. Develop better coordination of funding sources and opportunities

# Developing QC Materials for Genetic Testing - III

**March 8 (2004) Meeting March 11 – Orlando**

- 1. Review progress of workgroups**
- 2. Develop recommendations and future directions for making QC materials available**

**Areas of need and recommendations**

- 1. Current research efforts and further research needs**
- 2. Genetic tests in urgent need of QC materials**
- 3. Establishing a process to facilitate material contribution/access**
- 4. Define validation processes for QC materials**
- 5. Professional guidance on appropriate use of QC materials**
- 6. Regulatory oversight for providers and users**
- 7. Develop better coordination of funding sources and opportunities**
- 8. Rare disease testing**
- 9. Impact of patents and licensing agreements**

# Developing QC Materials for Genetic Testing - III

## **QC Material Research Workgroup**

**Participants: Roger Lebo, David Barton, Bassem Bejjani, Wayne Grody, Elaine Lyon, Clark Rundell, James Willey**

### **Issues:**

**To review current research on QC material development**

**To identify gaps in current research and issues for further research**

**To facilitate transition of QC materials from research to public availability and distribution.**

# Developing QC Materials for Genetic Testing - III

## QC Material Priorities Workgroup

**Participants: Jean Amos, Kenneth Friedman, William Highsmith, Elaine Lyon, Walter Noll, Deborah Payne**

### **Issues:**

**To identify genetic tests in urgent need of QC materials and develop a priority listing**

**To develop criteria and key factors for ongoing needs monitoring and evaluation**

**To evaluate the identified needs and make recommendations regarding potential approaches to meet these needs**

# Developing QC Materials for Genetic Testing - III

## Material Contributors Workgroup

**Participants: C. Sue Richards, Ben Roa, Bassem Bejjani, Susan Bernacki, Nicolas Brown, Elisabeth Dequeker, Victoria Pratt**

### **Issues:**

**To develop recommendations on practical approaches to engage potential contributors**

**To identify concerns and impediments to materials contribution**

**To provide assistance to contributors in meeting institutional/ local/state/federal requirements or procedures for material contribution**

**To promote formation of networks to facilitate contribution and/or sharing of materials not available from public repositories**

# Developing QC Materials for Genetic Testing - III

## **Materials Repositories/Cell Banks Workgroup**

**Participants: Jean Beck, David Barton, Pat Charache, Carol Greene, Patrick Terry, Yvonne Reid, Ben Roa, Laurina Williams**

### **Issues:**

- To develop a submission process for material contributors to submit materials for public availability**
- To provide information to the community about the process**
- To develop consensus process to validate and provide materials at a nominal cost to the community**
- To coordinate with other cell banks or sources to disseminate information on QC material availability.**

# Developing QC Materials for Genetic Testing - III

## **QC Material Validation Workgroup**

**Participants: Catherine O'Connell, Lawrence Silverman, Jean Amos, Elisabeth Dequeker, Andrea Gonzalez, Ira Lubin, Clark Rundell, Emily Winn-Deen**

### **Issues:**

- To develop recommendations for reliable processes to validate QC materials**
- To provide information to the community about the validation processes**
- To coordinate efforts to validate QC/PT materials**

# Developing QC Materials for Genetic Testing - III

**Professional Guidance Workgroup**

**Participants: Dan Farkas, Dorothy Belloni, Wayne Grody, Daynna Wolff**

**Issues:**

**To review current professional guidelines on use of QC materials in genetic testing**

**To determine the adequacy of current guidelines and identify areas in need of guidance**

**To develop recommendations for best practices for areas where traditional QC is not feasible or QC materials are not available**

# Developing QC Materials for Genetic Testing - III

## **QC Material Oversight Workgroup**

**Participants: Maria Chan, Michele Caggana, Bin Chen, Cecelia Hinkel, Erasmus Schneider, Ann Willey**

### **Issues:**

**To clarify FDA requirements for QC materials and provide guidance to manufacturers and providers seeking compliance**

**To clarify meet CLIA and State requirements for laboratories in using QC materials**

# Developing QC Materials for Genetic Testing - III

**Funding and Coordination Workgroup**

**Participants: Joe Boone, Carol Greene, Catherine O' Connell**

**Issues:**

**To identify potential funding sources to support efforts to improve QC material availability**

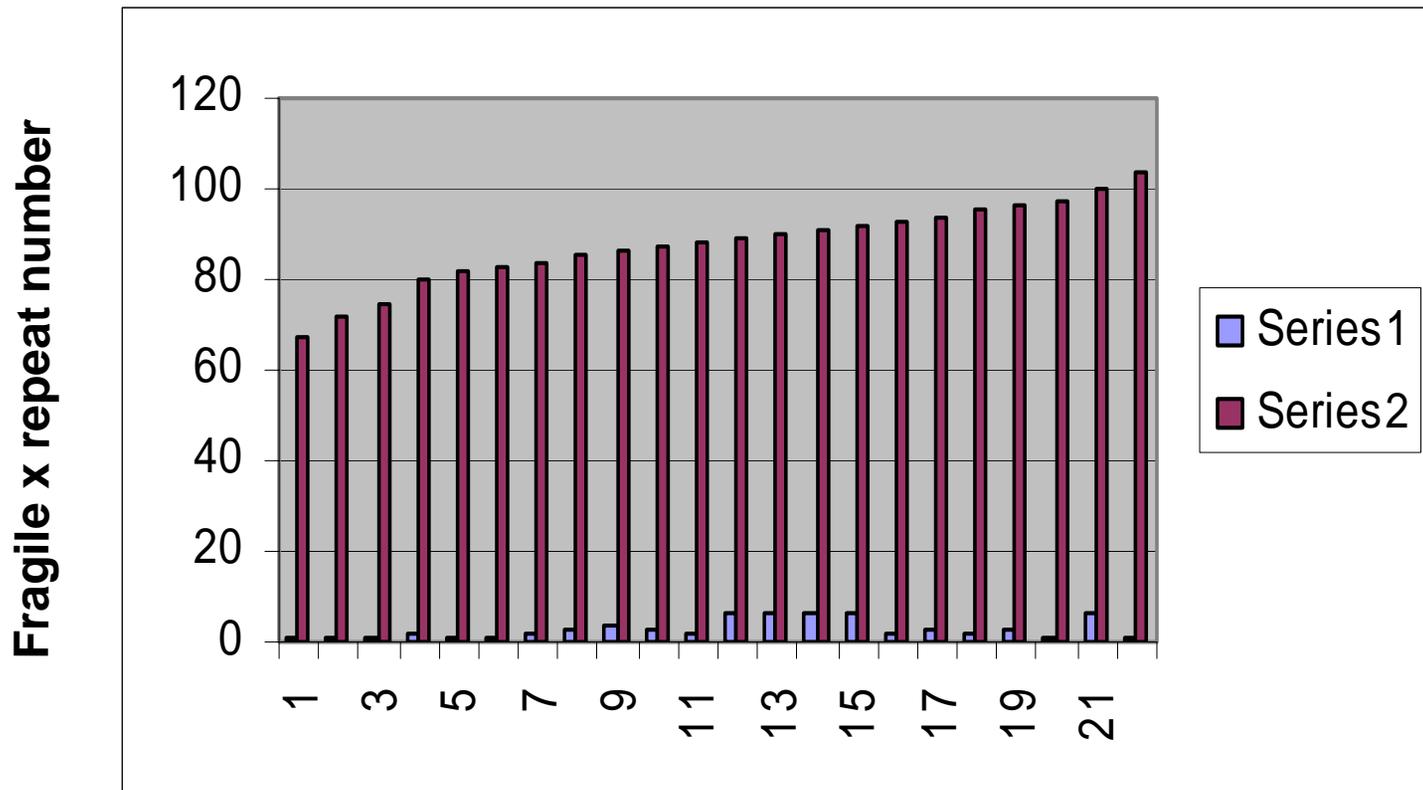
**To promote funding for QC material development, validation, and provision**

# MGL-A ACMG/CAP – Molecular Genetics – Participant Survey (Received 8/13/04)

**MGL1-04 Premutation male**

**Number of Labs Responding – 75**

**Number Reporting Repeat Sizes for Premutation - 63**



**Due to lack of participant consensus, the section was not graded.**

# Developing QC Materials for Genetic Testing - III

**NIST Updates: Development of Standard Reference Materials**  
**Barbara Levin, Kristy Richie**

**Status and Availability of Stably Transformed Cell Lines with Characterized CF Mutations - Laurina Williams and Jeanne Beck**

**Promoting Quality Laboratory Testing for Rare Diseases**  
**The Rare Disease Testing Conference and Recommendations**  
**Joe Boone**