

# Li-Fraumeni Syndrome Workshop

November 2, 2010

Natcher Auditorium (Building 45), NIH Bethesda

*Sponsored by the NIH Office of Rare Diseases Research and the National Cancer Institute*

This meeting will bring together intramural and extramural clinicians and scientists to review the existing knowledge regarding Li-Fraumeni syndrome (LFS), to share current research projects, and to identify areas, such as cancer screening and risk reduction interventions, in which further research is needed. The meeting will also serve as a starting point for the creation of a LFS research consortium. Families with LFS are invited to attend in this meeting. This will be an opportunity to participate in education sessions on existing scientific knowledge, including cancer screening recommendations, and in workshops that will foster interaction and the development of a support group.

We anticipate that this meeting will be the first of several LFS-related scientific meetings. The goals of the first meeting are to bring the clinicians/scientists together to review the current knowledge and, more importantly, to foster further collaborations.

## ***DRAFT Agenda***

### **Tuesday, November 2, 2010, Natcher Auditorium, NIH**

**7:45 - 8:30 Registration**

**8:30 - 8:45 Welcoming remarks & meeting overview – Joseph Fraumeni**

**8:45 – 9:00 History and Overview of LFS – Louise Strong**  
**Chair: Sharon Savage**

**9:00 - 10:00 Perspectives from Li-Fraumeni Syndrome families**  
**Chair: Andrea Patenaude**  
Oliver and Jamie Wyss  
Luana Locke  
Von Means

**10:00 - 10:15 Coffee Break**

**10:15 - 12:00 Clinical Aspects**  
**Chair: Jeffrey Weitzel**

The Story from Brazil – Maria Isabel Achatz  
Genotype-phenotype correlations in the TP53 mutation database – Pierre Hainaut  
Chompret Criteria and experience in France – Thierry Frebourg (invited)  
Screening and Research from the UK – Gareth Evans  
Cancer Screening: Adults – Judy Garber  
Cancer Screening: Children – David Malkin

**12:00 - 1:00 Genetic Counseling and Psychosocial Aspects**  
**Chair: June Peters**  
Genetic counseling and testing for Li-Fraumeni Syndrome – Kathy Schneider  
Psychosocial Considerations – Andrea Patenaude  
Ethical concerns with testing minors – Ben Wilfond  
International perspectives on psychosocial reactions – Evelyn Bleiker

**1:00 - 2:00 Lunch - on your own**

**2:00 - 3:00 Molecular Biology of TP53**  
**Chair: David Malkin**  
Overview of TP53 function – David Malkin  
Mitochondria and TP53 – Paul Hwang  
Telomeres and LFS – Uri Tabori  
DNA Repair and TP53 – James Ford

**3:00 - 3:15 Coffee Break**

**3:15 - 5:00 Concurrent Sessions**

**A. Creating a research consortium and development of new studies**

a. Presentations on database and projects in development

**Chair:** Joshua Schiffman

Children's Oncology Group LFS registry proposal – Simona Ognjanovic

Collecting and managing biospecimens – Joshua Schiffman

Studies of metformin– Philip Dennis

Identifying a consortium-wide screening protocol – Judy Garber

b. Consortia building

Open discussion on logistics

**A. B. Family Support and Advocacy:** Chaired by patient advocates with experience in family support and advocacy groups

Kevin McQueen (Fanconi Anemia Research Fund)

David Smith (SpecialLove)

Connie Lee (Angioma Alliance, Genetic Alliance)

John Berkeley (Li-Fraumeni Syndrome Support group)

Representatives from NIH Office of Advocacy Relations

Discussion topics to include:

Fostering family communication and support

Organization of family meetings

Fundraising

Other issues

**5:00 - 5:30 Meeting summary and report from break-out sessions**

**5:30** Adjourn