

Meeting Report for ORDR

Li-Fraumeni Syndrome: A Multidisciplinary State-of-the-Science Conference

Supported by:

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Summary

The Li-Fraumeni Syndrome (LFS) workshop was held at the NIH on November 2, 2010. This workshop was the first to bring physicians, scientists, and families with LFS together at the same time. LFS is a highly penetrant, autosomal dominant cancer predisposition syndrome, most commonly caused by mutations in the *TP53* tumor suppressor gene. Carriers of *TP53* mutations are at very high risk of cancer, including bone and soft tissue sarcomas, pre-menopausal breast cancer, brain tumors, adrenocortical carcinomas, and others.

Approximately 200 extramural and intramural people attended the workshop. This included approximately 80 individuals from 40 different LFS families.

The speakers included 21 researchers from 14 institutions from the U.S., Canada, U.K., France, the Netherlands, and Brazil. Individuals from 3 families with LFS also spoke on their experiences with the disorder. The scientific talks included clinical findings in LFS, options for cancer screening, and updates on the biology of *TP53*. After the talks, the families met for 2 hours with representatives from other family support groups in order to learn how to set up a LFS family group. The researchers also met to discuss specific topics and develop collaborations. LFS researchers began to develop a research consortium in 2009. This was the first scientific meeting focused on reviewing scientific advances as well as consortium building.

Outcomes

1. An international research consortium of LFS investigators has been created. The group is developing collaborative studies and plans to meet again in the next few months.
2. A manuscript summarizing the topics discussed at the workshop, including the goals of the LFS research consortium is in preparation.
3. The families who attended the workshop are creating a family support group. The president was elected and 9 other individuals will be on the board of directors.
4. We are optimistic that a future meeting on LFS will be organized by the family support group within the next 2 to 3 years. This meeting would include larger representation from physicians and scientists in the LFS and *TP53* fields as well as family members and support staff. This could potentially be held in conjunction with the *TP53* research meetings.