

Ask a Genetic Counselor

LFS Awareness Day-March 20, 2025

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Living LFS Medical Advisory Chair

What is a genetic counselor?

Masters degree trained healthcare provider who works as a member of a healthcare team providing risk assessment, education and support to individuals and families at risk for, or diagnosed with, a variety of inherited conditions.

What does a genetic counselor do?

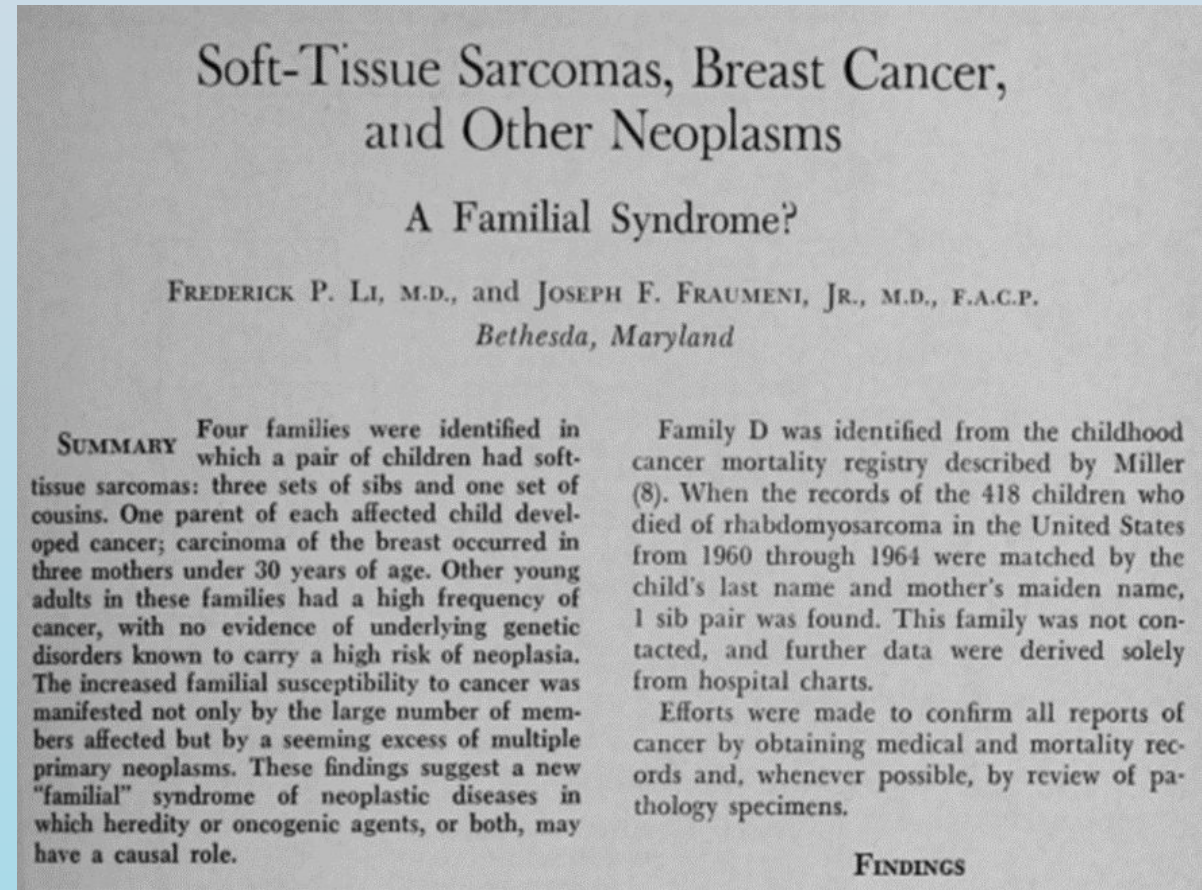
- Explains benefits and common concerns of genetic testing
- Evaluates family history and facilitates genetic testing
- Reviews latest info on health risks, and medical management recommendations
- Coordinates care and communicates with specialists
- Supports you with talking to children and family members
- Provides informational and supportive resources and referrals



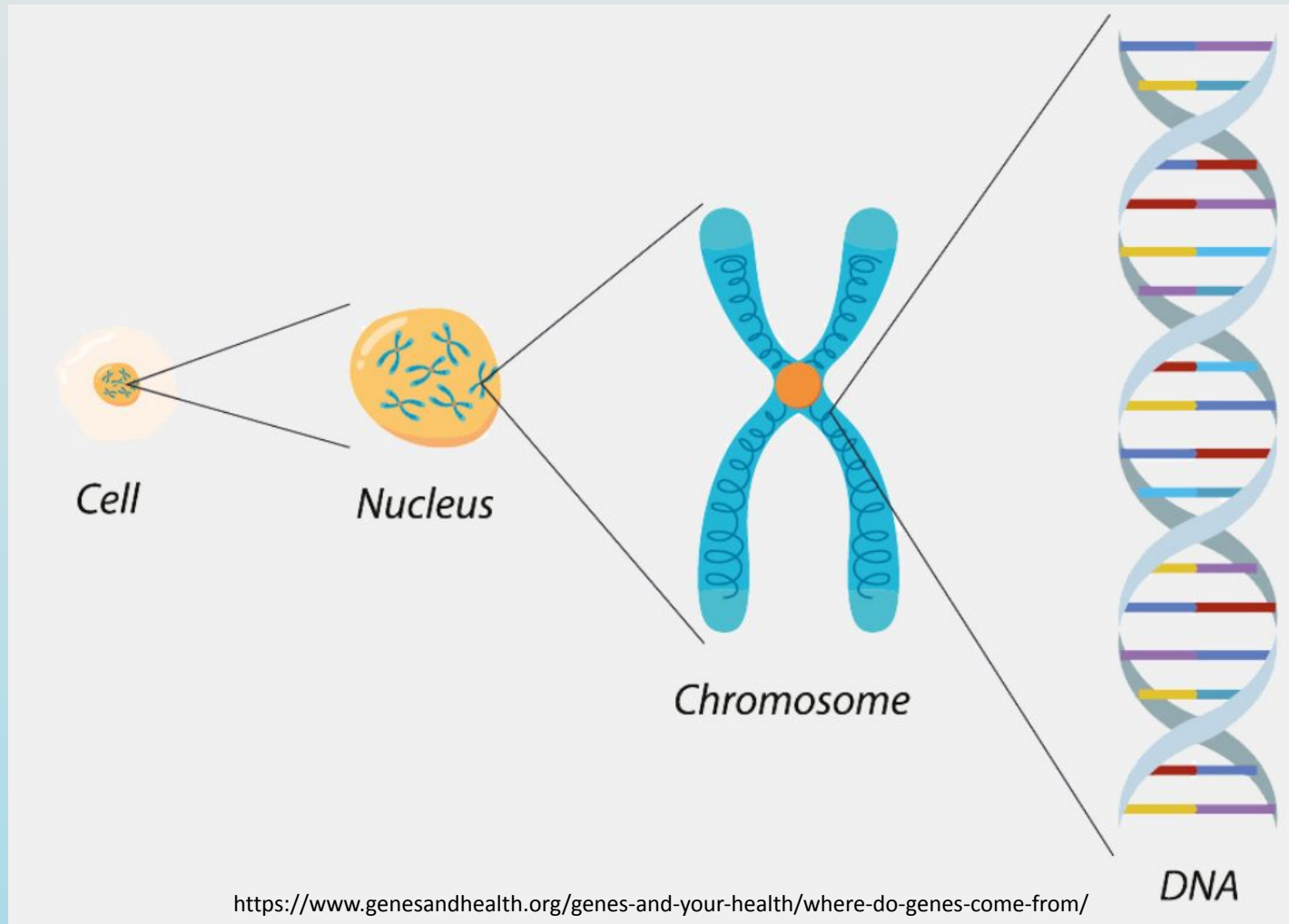
Genetic counselor is like the 'point person' for families with LFS

LFS history

- First described in 1969
- Link to *TP53* gene made in 1990
- Core tumors: bone and soft tissue sarcomas, breast cancer, adrenal cortical carcinoma, choroid plexus carcinoma, brain tumors, leukemias
- Evolving understanding of LFS in recent years-LFS as a Spectrum



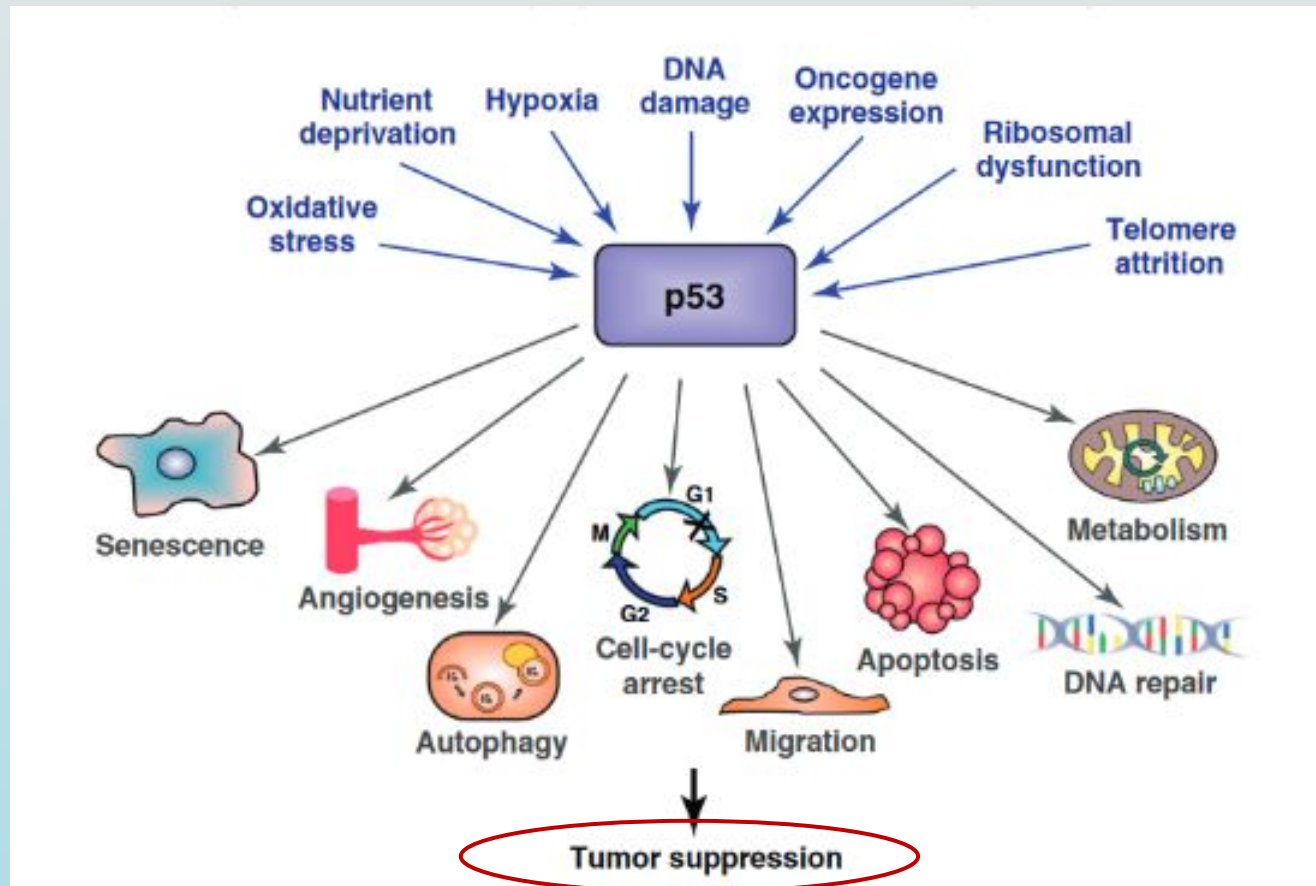
Genetics 101



A gene is a small piece of DNA with a specific job

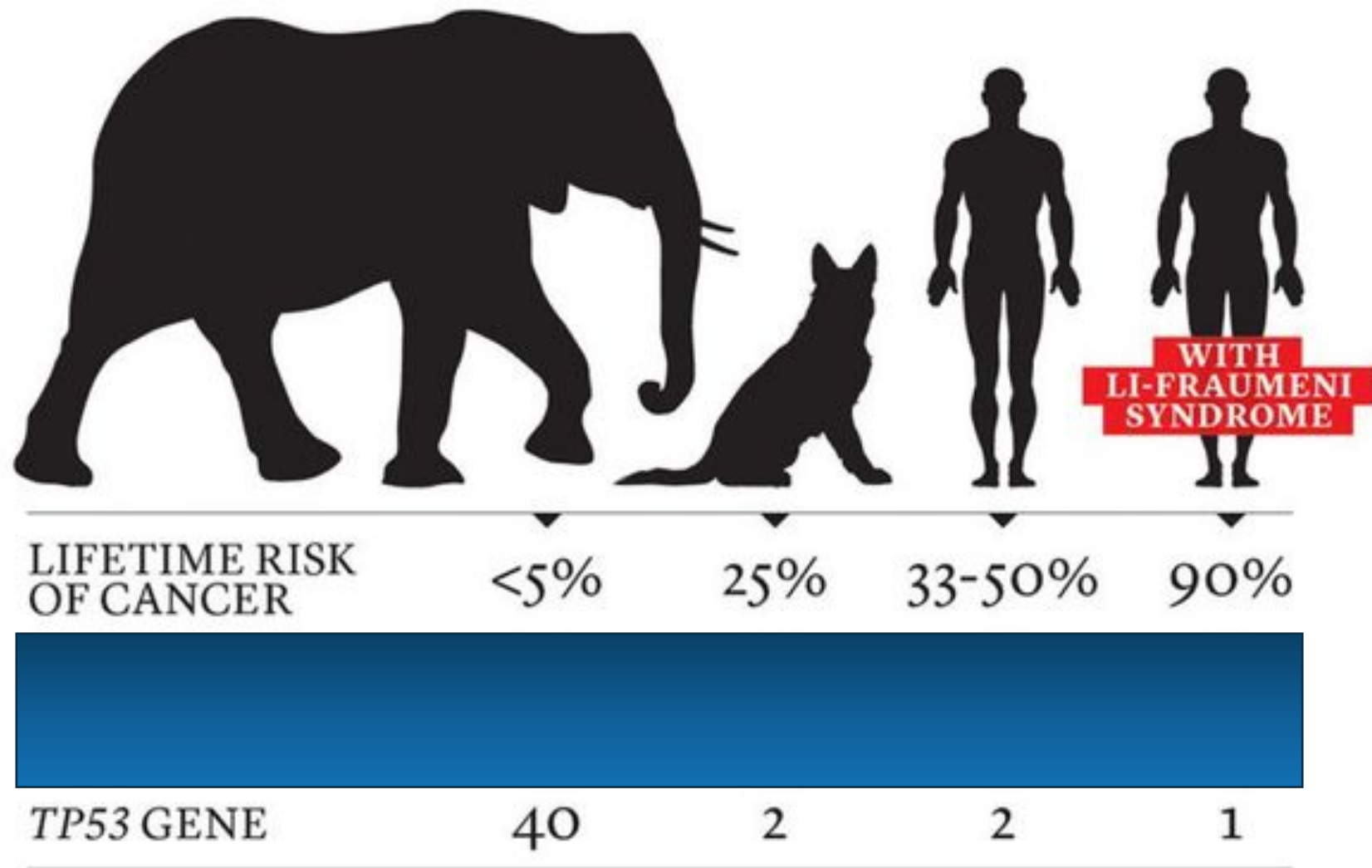
We have about 20, 000 genes and two copies of each gene, one from each parent

TP53 gene: A tumor suppressor gene “Guardian of the genome”



Biegging, K. T., & Attardi, L. D. (2012). Deconstructing p53 transcriptional networks in tumor suppression. Trends in cell biology, 22(2), 97-106.

The TP53 gene helps to prevent cancer from starting in many different ways



*Not Available /// SOURCE: Joshua Schiffman



Your questions.....

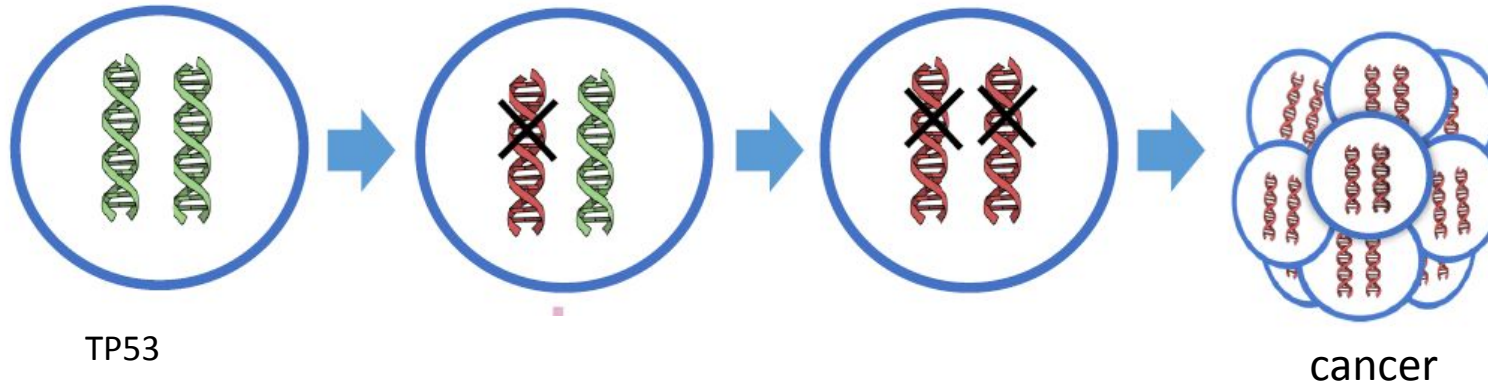
Living
LIFS



“In LFS, why does cancer suddenly happen or occur very young in some and older in others?”

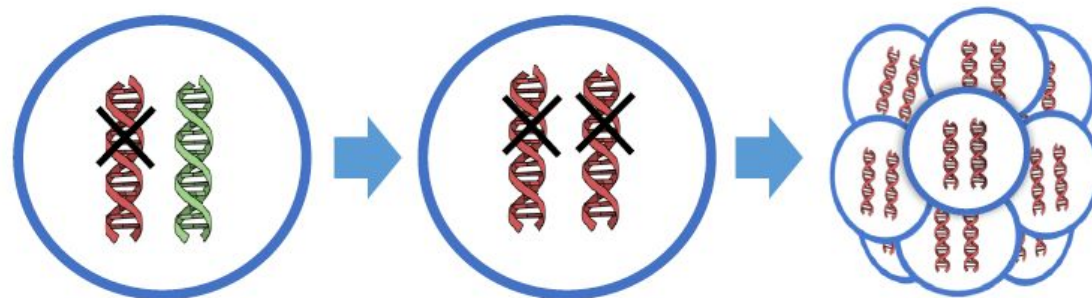
People with LFS get cancer more easily than others

People who do NOT have LFS



This process takes decades

People with LFS



This process is much shorter

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What should I be doing to protect my children with LFS?

Pediatric screening guidelines in LFS



National
Comprehensive
Cancer
Network®

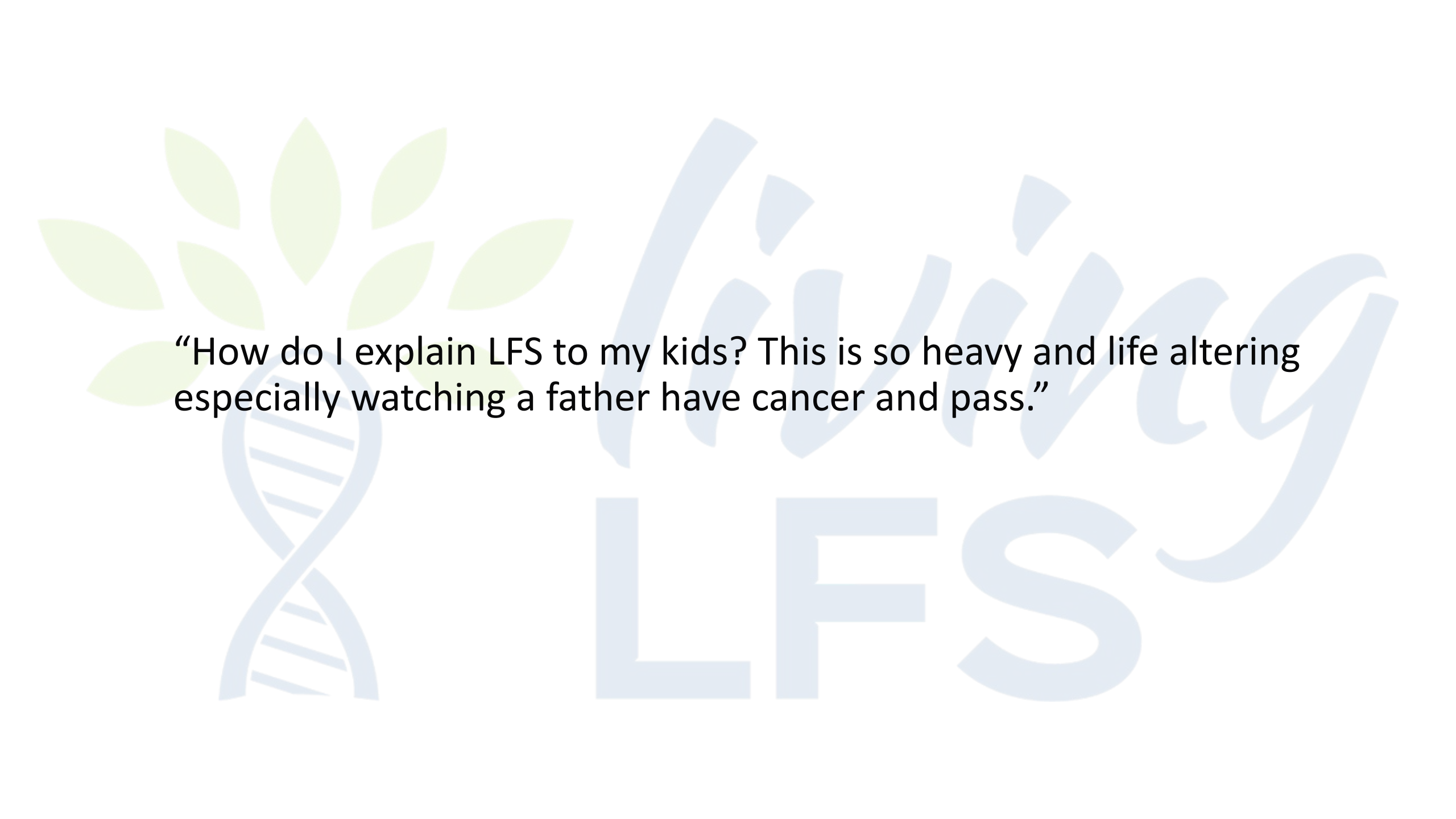
NCCN Guidelines Version 3.2025 Li-Fraumeni Syndrome Management

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LI-FRAUMENI SYNDROME: PEDIATRIC SURVEILLANCE

	Screening/Surveillance Procedure and Interval
Cancer risks	<ul style="list-style-type: none">• Comprehensive physical exam including neurologic examination with high index of suspicion for rare cancers and second malignancies in cancer survivors every 6–12 mo beginning in infancy• Annual whole body MRI^{j,k,l} beginning in infancy• Annual brain MRI may be performed as part of the whole body MRI or as a separate exam beginning in infancy• For ACC, ultrasound every 3–4 mo beginning in infancy

*Screening may vary among individuals/institutions



“How do I explain LFS to my kids? This is so heavy and life altering especially watching a father have cancer and pass.”

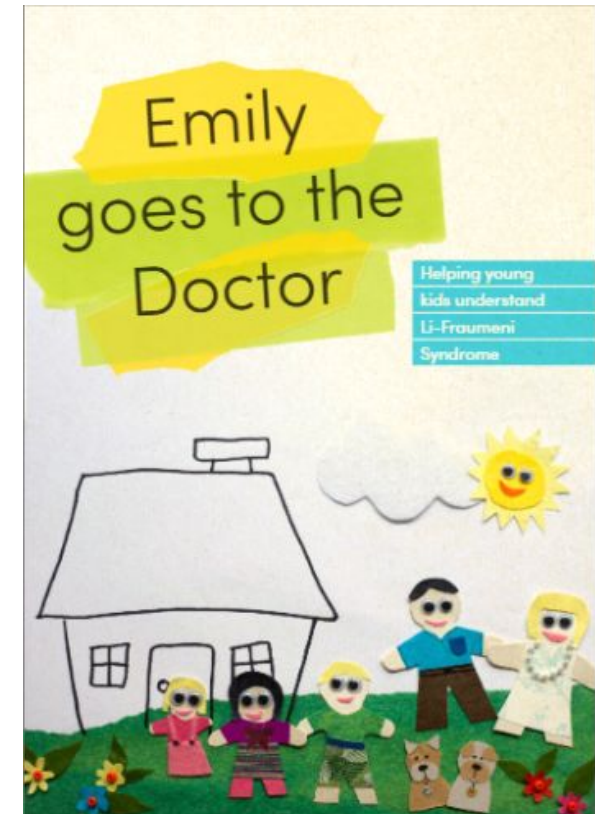
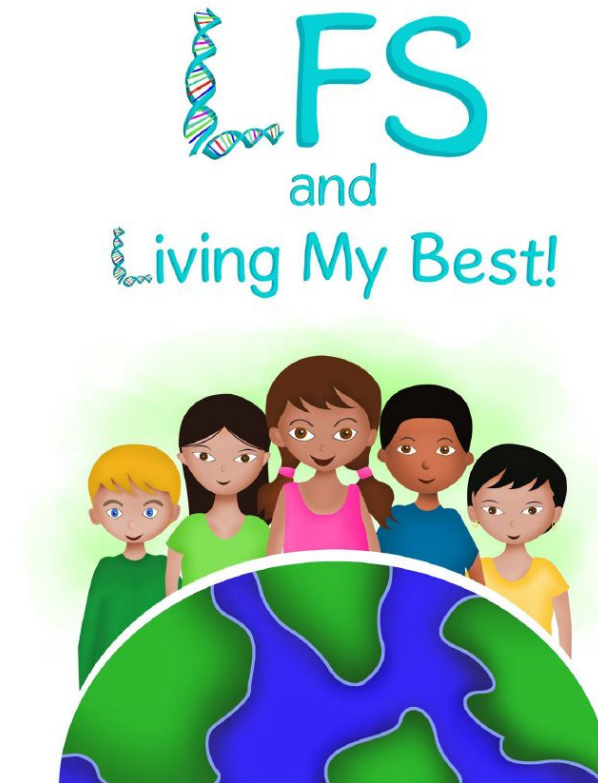
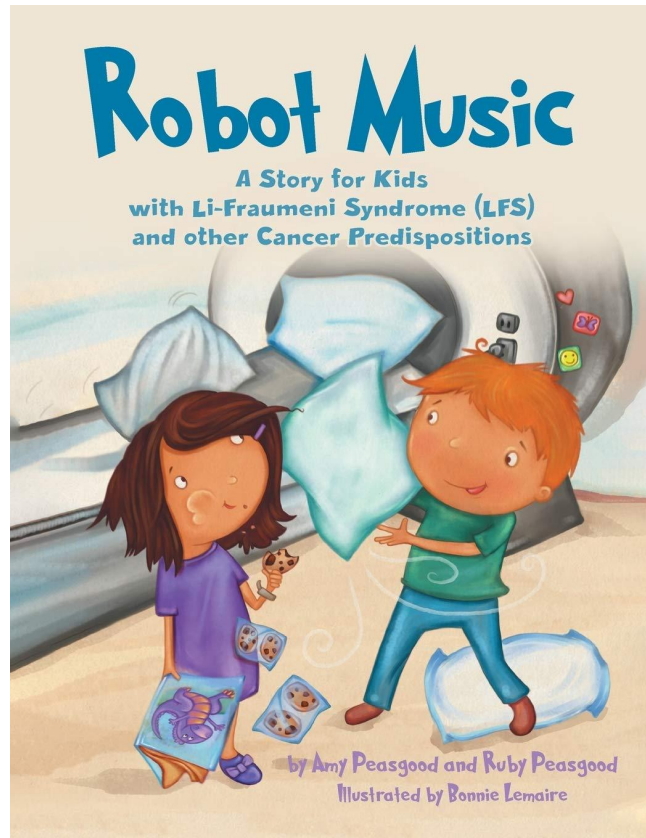
Tips on Talking to Kids about LFS

- ✓ Make sure you are taking care of yourself mentally and emotionally first
- ✓ Draw from previous experiences and trust yourself
- ✓ Take your time-may require multiple conversations over time
- ✓ Let your child(ren)'s questions and reactions guide you
- ✓ Normalize ex. "From time to time you will go to the doctor for some tests just like Uncle Joe does"
- ✓ Frame information in a positive way
 - ✓ Ex. "We're making lots of doctors visits in order to keep you healthy"
- ✓ Team approach with medical providers




Helpful website: <http://www.apa.org/helpcenter/talking-to-children.aspx>

Books/booklets for kids



by Georgina Schlub, PhD, Genetic Counselor
and Team Familial Cancer Service


<https://livinglfs.org/resources/#onlineresources>

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- “I was advised to have radiotherapy by my team but I know this is not advisable for LFS. Is Proton beam therapy a better alternative or should I just forego any treatment involving radiation?”

Radiation sensitivity in LFS

People with LFS are extra sensitive to the carcinogenic effects of radiation, however....

- If radiation is the most effective treatment for cancer, it should not be avoided
- Proton beam radiation is a more focused/precise way to deliver radiation, but whether it is recommended depends on cancer type and location and other factors to be discussed with medical team
- The levels of radiation from most routine screenings (like dental x-rays) are generally very low and probably don't need to be avoided, but can discuss doing only when necessary

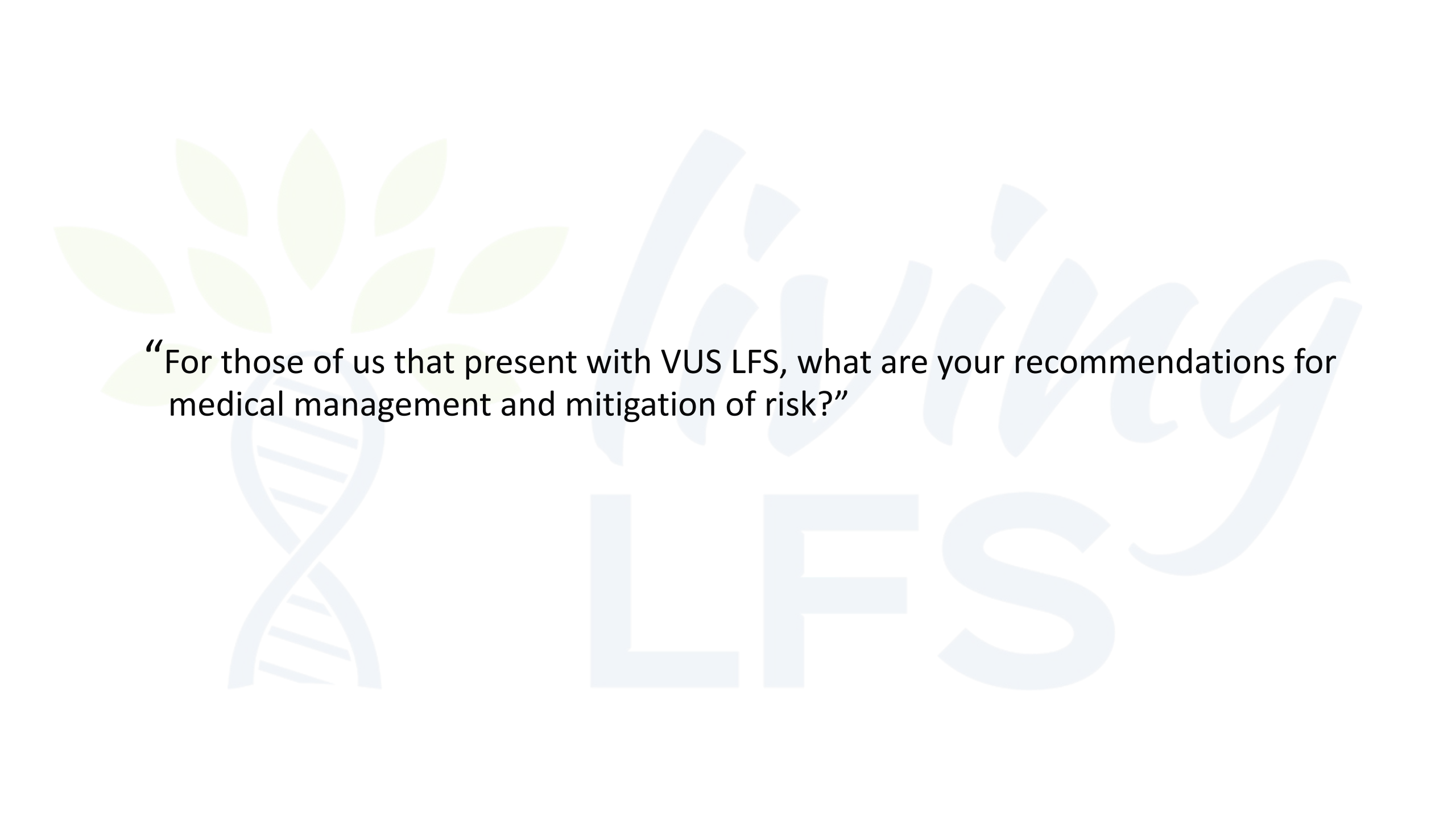


“What is the recommended dermatology skin checks for people with LFS? Is it yearly? I have several suspicious looking moles that I've had for years but don't know if they are dangerous or just weird looking.”

Adult screening guidelines in LFS

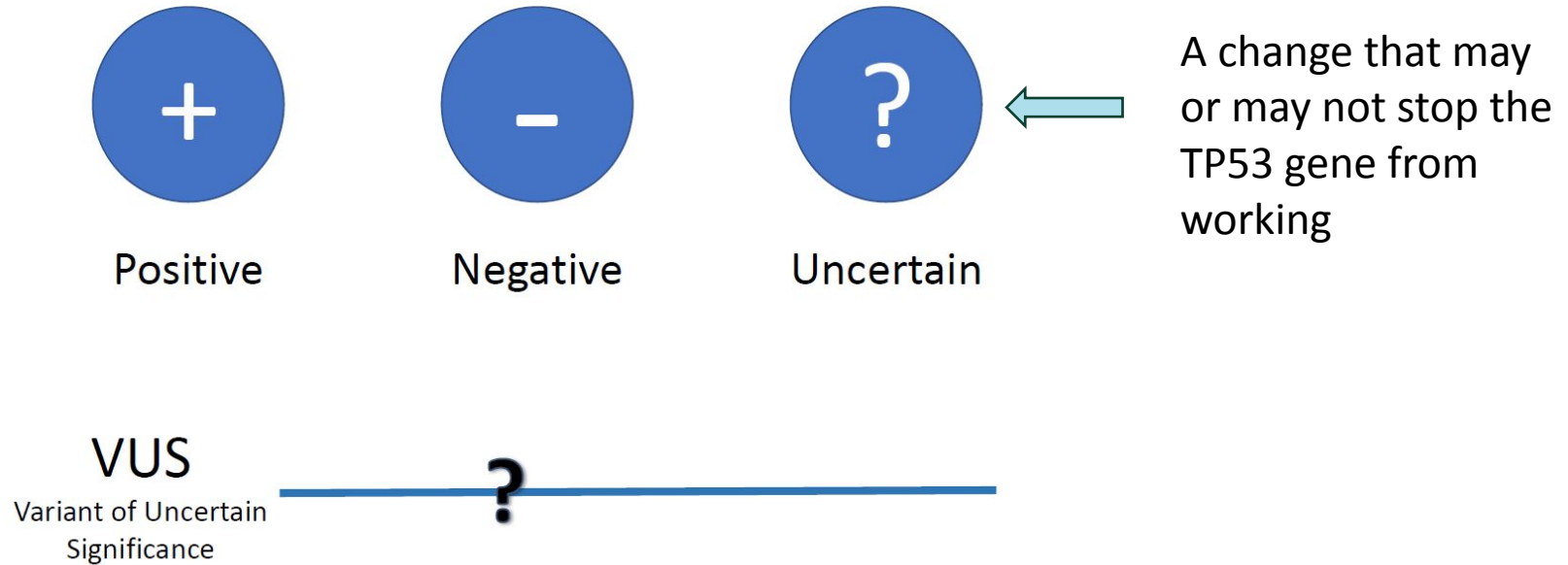
Cancer type	ADULTS: Surveillance recommendations
Breast cancer (assigned female at birth)	Breast awareness beginning at age 18 and report changes to your health care provider Clinical breast exams every 6-12 months, beginning at age 20 (or earlier, based on family history) Age 20-29 (or earlier, based on family history): Yearly breast MRI screening with and without contrast Age 30-75: Yearly breast MRI screening with and without contrast, and mammogram Age >75: Management should be considered on an individual basis
Gastrointestinal cancer	Colonoscopy and upper endoscopy every 2-5 years, beginning at age 25 (or earlier, based on family history)
Other cancer risks	Comprehensive physical exam every 6-12 months to include neurological and skin exam Awareness of signs and symptoms of rare cancers is critical Yearly dermatologic examination beginning at age 18 Yearly whole-body MRI, if available Yearly brain MRI (may be performed as part of whole body MRI or as separate exam) Yearly prostate-specific antigen (PSA) beginning at age 40 → Education regarding signs and symptoms of cancer with prompt response if concerned

Adapted from the National Comprehensive Cancer Network Guidelines Version 2.2025 Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. *Please note that these are general guidelines. Specific guidelines for individual patients and families may differ.*



“For those of us that present with VUS LFS, what are your recommendations for medical management and mitigation of risk?”

Possible Genetic Testing Results



“The laboratory does not have enough information to determine whether this VUS is a benign (harmless) genetic variant or if it is associated with an increased risk of cancer. As more information becomes available some VUS are eventually reclassified, and research shows that if a VUS is reclassified, the odds are high that it will turn out to be benign. If we receive updated information on the meaning of this VUS, we will attempt to share this with you. **This VUS should not impact your medical care plan.**”

Be sure to check in with the provider who ordered your genetic testing about their recontact plan in the case of a VUS reclassification.



Other questions

- “I would like to know what other conditions (than cancer) are linked to this.”
- “I would like to know what preventative surgeries are recommended for males.”
- “Why would squamous keep on coming repeatedly?”

Questions?



Helpful resources:



NSGC 'Find a Genetic Counselor:

<https://findageneticcounselor.nsgc.org/?reload=timezone>

Talking to kids: <http://www.apa.org/helpcenter/talking-to-children.aspx>

Carly's email: carly@livinglfs.org

Happy LFS Awareness Day!