Living with

Li-Fraumeni Syndrome

A Hereditary Cancer Syndrome

by

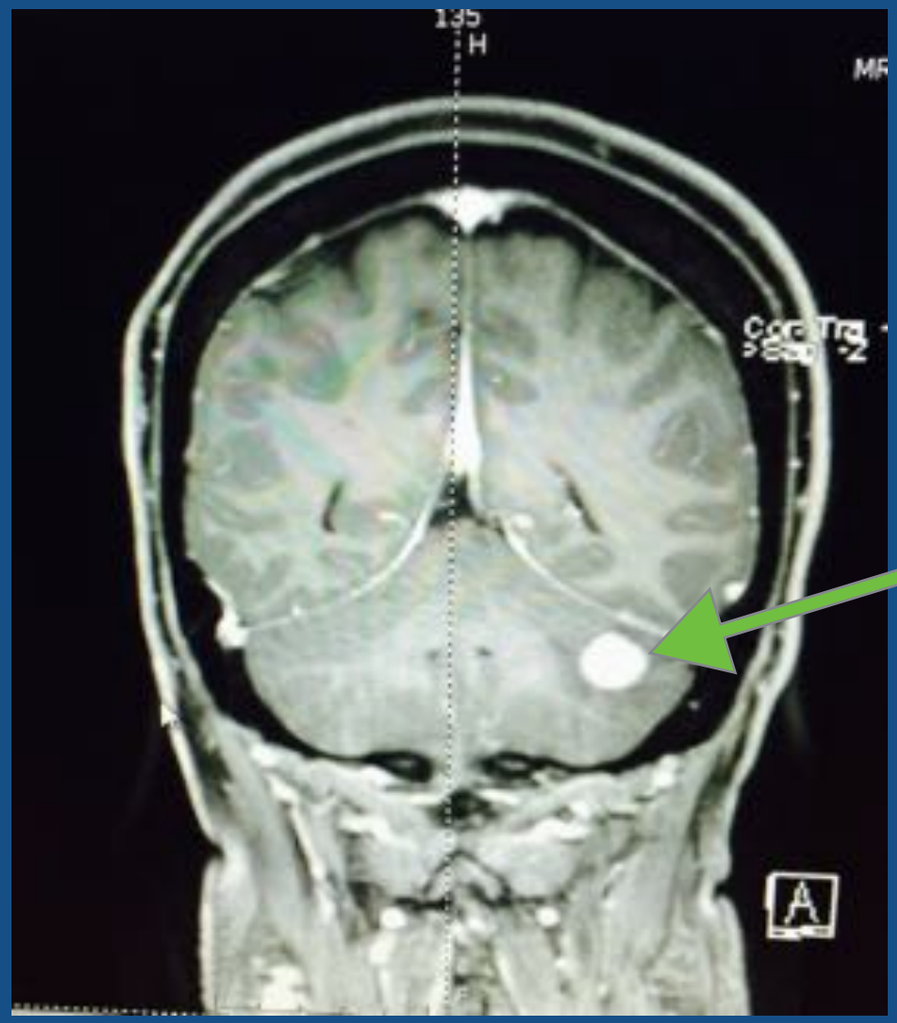
Jennifer Mallory

Cancer in the Family

Sometimes we don’t just get our hair or eye color from our parents. Sometimes we get a higher risk for cancer from them.

My name is
Jennifer.

I
have
Li-Fraumeni Syndrome



Brain mets

- I have stage IV Breast Cancer, that means breast cancer cells escaped my breast and metastasized to other places. I have little tumors called metastases in my brain, bones, and lungs.

Unfortunately, I'm not the first person in my family to have cancer.

Grandfather

Brother Brain Father

Cousin

Daughter

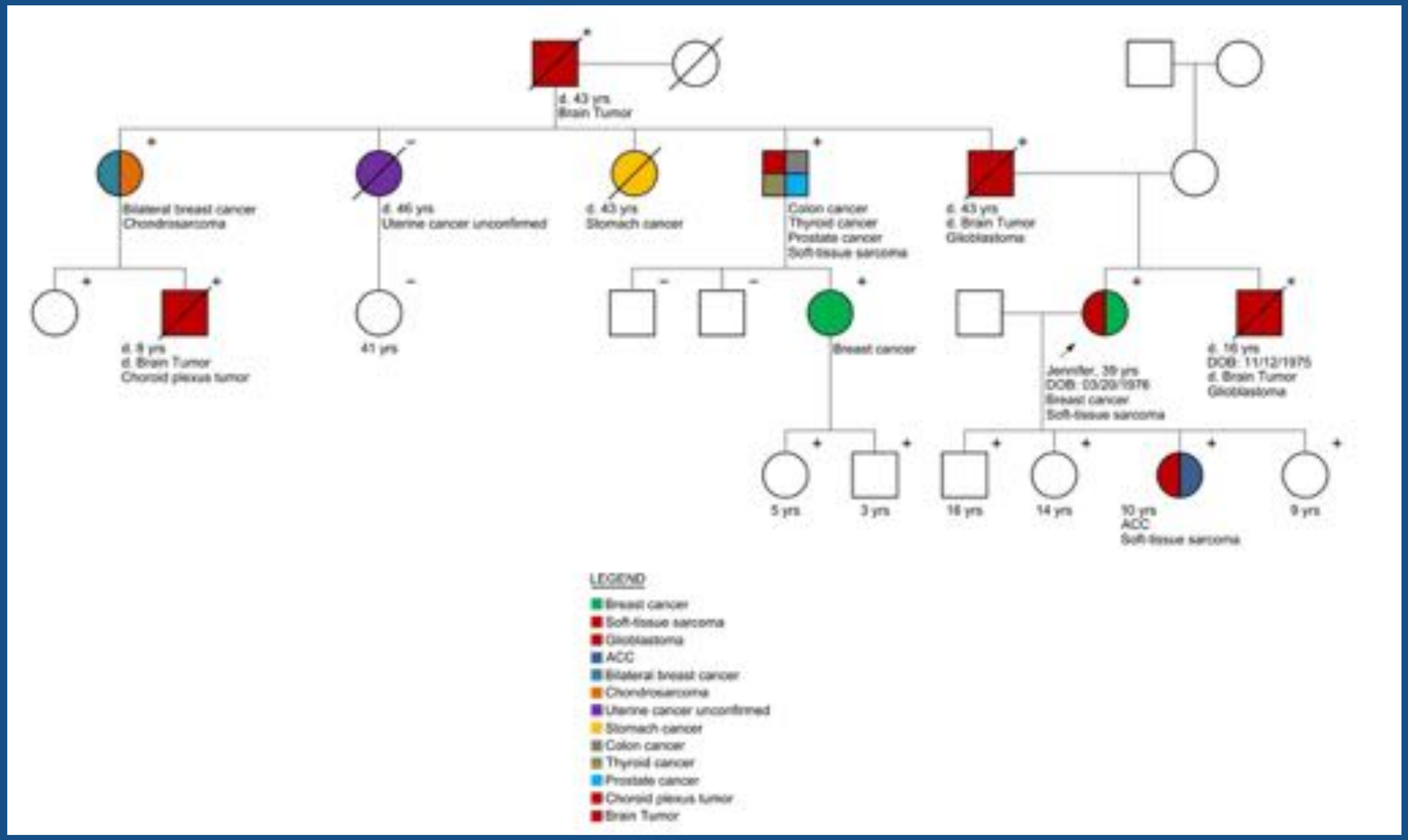
Aunts Sarcoma Adrenal

Uterine Breast

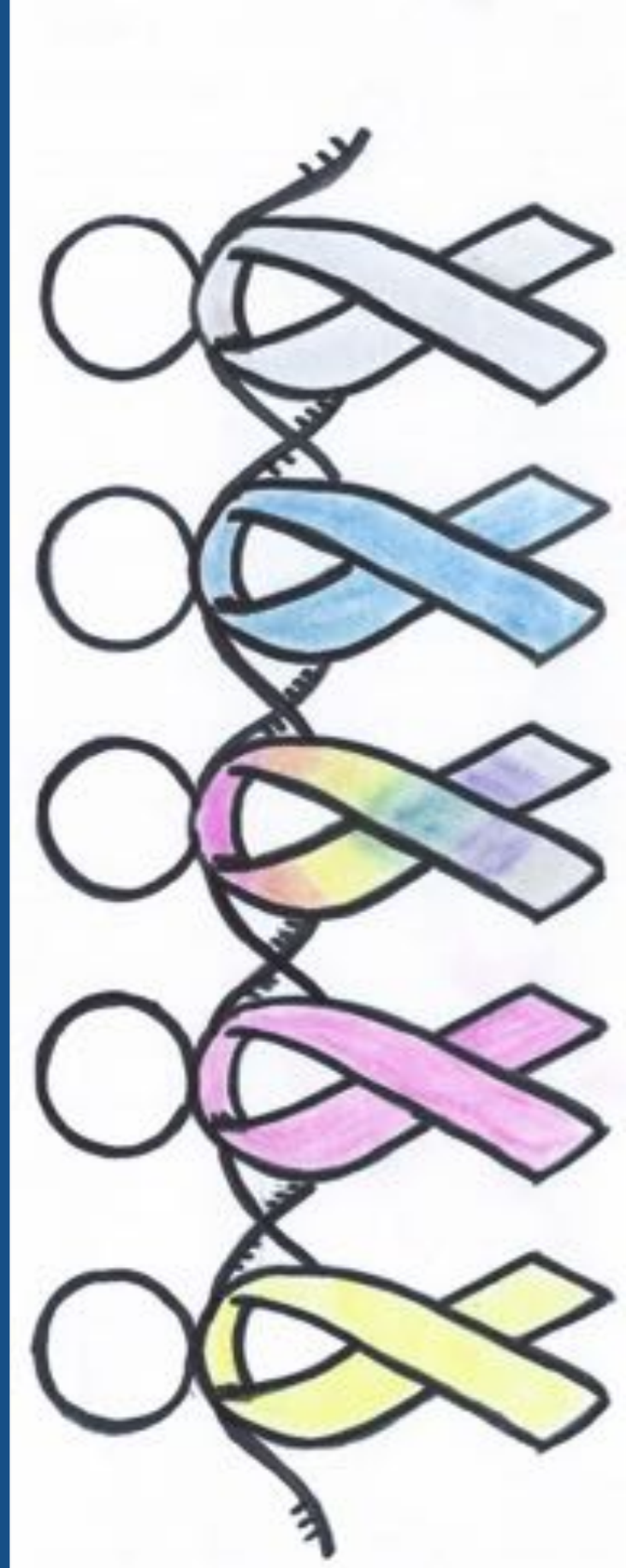
Cousin Uncle

Colon Thyroid

This is my family pedigree.



Each color represents a different cancer.



- My grandfather, father, brother and cousin all died from brain tumors.

- My daughter had adrenal cancer.

- My aunt, cousin and I all had breast cancer.

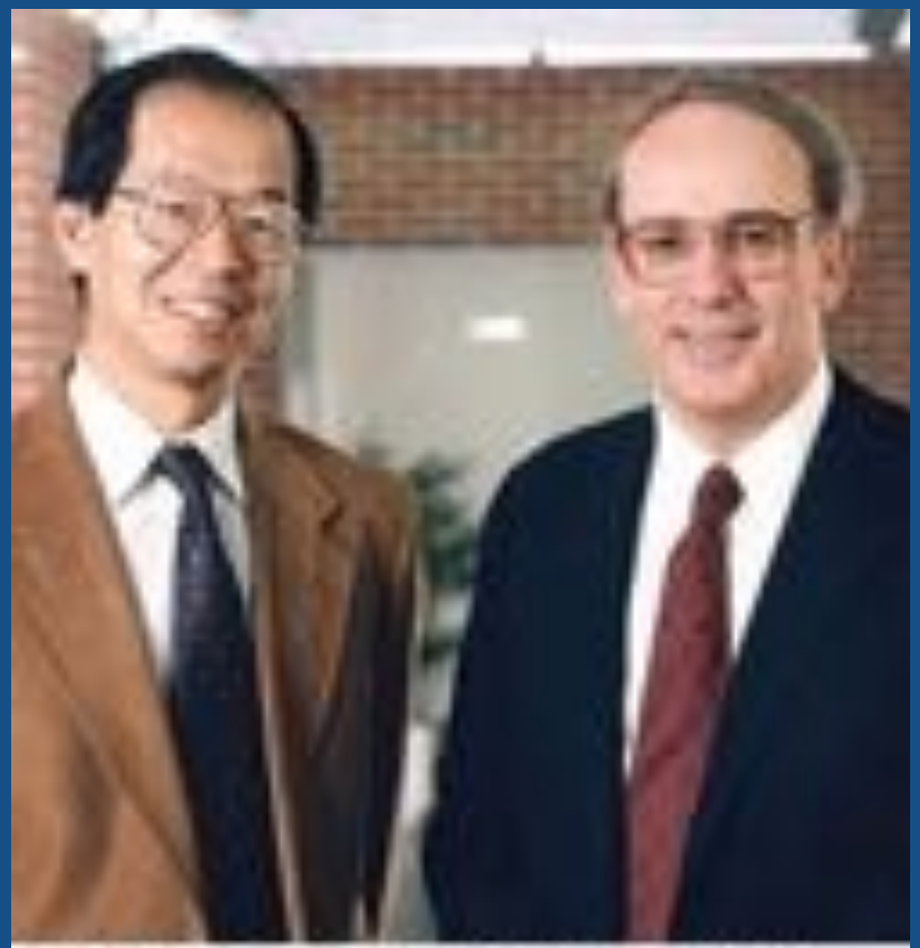
- Many of us had sarcomas.

- Some family members have multiple cancers.

For Years it felt like a FAMILY CURSE.

- In the 90's my uncle saw an article in the news and we began to wonder if we had this broken p53.
- My mom called and talked with Dr. Li about testing our family. It was still too early, there was still much to learn about the TP53 gene and cancer.

Dr. Li and Dr. Fraumeni



Frederick Li and Joseph Fraumeni, 1991.

- In 1969, Dr. Frederick Li and Dr. Joseph Fraumeni were working at the National Cancer Institute when in the course of conversation they heard about a family that had multiple children with sarcomas.
- They noticed a higher incidence of breast cancers in these families as well.

Could Cancer be Inherited?

- The notion of cancer being hereditary was not popular in the 70's and 80's.

- By scouring data records of children with sarcomas, visiting clinics, they began to see a pattern.

- They called the pattern SBLA Syndrome for the cancers seen.

Sarcoma
Breast Cancer
Leukemia
Adrenal Cancer

- In the 1990's, the same decade my family was plagued by brain tumors and other cancers, they linked the syndrome to TP53 mutations.

- TP53 mutations are found in about 50% of all tumors. But these mutations are **somatic**, meaning just in the cells of the tumor.

- **Germline** Mutations of the TP53 gene became associated with the family pattern Dr. Li and Dr. Fraumeni had observed decades before. These mutations can be inherited. It is now called

Li-Fraumeni Syndrome

Li-Fraumeni Syndrome(LFS) Cancer Risk

- 50% by age 30, 90% by age 70. (Classical tumors are breast, brain, sarcoma and adrenal, but can be anywhere.)
- About 90% of Females develop Breast Cancer by age 60.

LFS Is Suspected If….

- A child presents with a sarcoma, adrenocortical carcinoma or brain tumor

and

- a first degree family member has had cancer before age 45.

and

- Another family member has been diagnosed with cancer before age 45-60.

What if LFS is Suspected?

The patient is usually referred to genetic counseling.

Genetic Counselors are trained to take family history and help assess risk and need for genetic testing.

The National Society of Genetic Counselors can help you find the right counselor.

www.nsgc.org

Testing and Inheritance

- LFS is a DOMINANT trait.
- This means a parent carrying the mutation has a 50% chance of passing the gene along to a child.
- Some LFS carriers are De Novo, this means they did not inherit the mutation from a parent.
- Testing for LFS can be done with a blood test.
- Not all people with LFS develop cancer.

SCREENING and CONSIDERATIONS

- People with LFS are sensitive to DNA damage, this should be considered when looking at using ionizing radiation or genotoxic treatments.

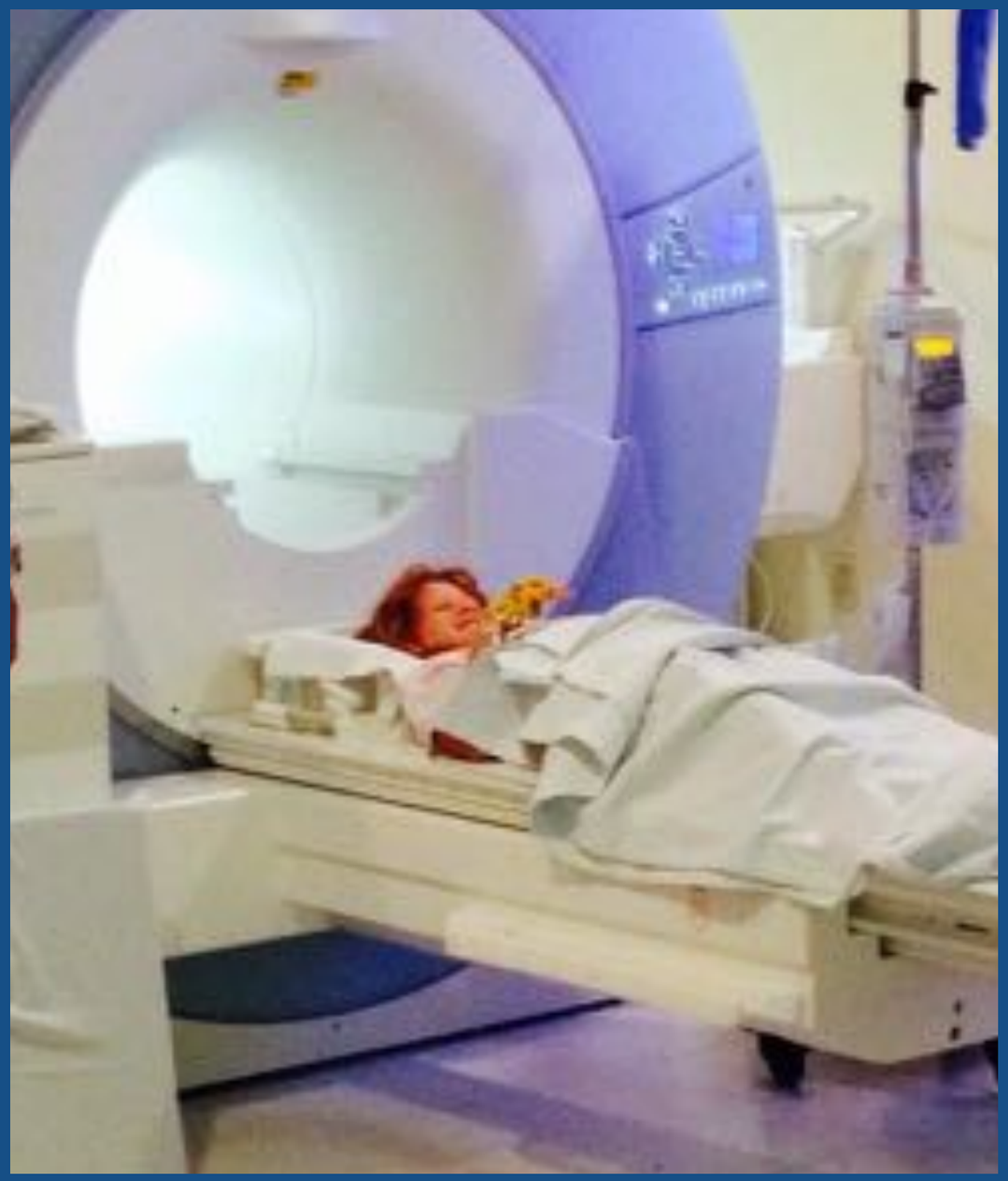
- There currently is no one protocol recommended for screening people with LFS.

- Many follow the Toronto Protocol established by Dr. David Malkin at SickKids hospital in Toronto.

- There are current trials looking into the effectiveness of rapid whole body MRI for screening.

Living with Li-Fraumeni Syndrome

- All 4 of my children have LFS.
- We work with our genetic counselor and oncologists to tailor the best screening regimen for all of us based on family history.
- Every year, they have a whole body MRI, lab work, and clinical exams to screen them for cancer.
- Sometimes it's difficult emotionally. I worry about them having to face cancer.
- But we know, you cannot treat 100% of the cancers you don't know about. Screening and early detection makes a difference in survival.



REMINDERS for LIVING with LFS

- Know Your Body-keep an eye out for changes, lumps and bumps.
- Trust Your Gut- if you feel off or that something is not right, trust that feeling. Get it checked out.
- Fight the Shark Closest to the Boat- you have to deal with the biggest threat, for example: you can not worry about potential secondary cancers if you don't survive this one.
- Make the best decision you can with the Information you have NOW. Then Make peace with that. Cancer decisions are hard, emotionally and physically. It's not a one size fits all.
- LIVE. Live well, Laugh, Cherish the moments.

FINDING SUPPORT

- When my daughter was diagnosed with cancer, I didn't know any other LFS families.
- It can be very lonely. So I created a support group on Facebook. Others wanted support too. It's been such a positive resource.
- LFS is rare. Many doctors have never heard of it. Yet, knowing LFS status could impact treatment and care decisions.
- Finding the right support and care helps us live better.

For More Information

living LFS

www.LivingLFS.org