



Importance of Mutational Testing in GLST

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TIMELINE

Tumor Found

- Diagnostic Imaging Scans

Tumor Tissue

- Biopsy
- Surgery

Testing

- Mutational Testing
- Genetic Testing

Treatment

- FDA Approved treatment
- Clinical Trial

BIOMARKER TESTING

Pathology Testing	Mutational Testing/ Genomic Sequencing
<ul style="list-style-type: none">• Aim: Provides a diagnosis after a histological examination of the tissue samples• Examines: Proteins• Tools: Microscope• Turnaround time: 1 to 2 days• Common Results in GIST: c-KIT, DOG1, Desmin	<ul style="list-style-type: none">• Aim: Identify the mutations that are driving the tumor development• Examines: Genes• Tools: NGS• Turnaround time: 3-6 weeks FFPE• Common results: KIT, PDGFRA

TYPES OF TESTING

- Basic Mutational Testing :

In house testing in most hospitals

Specific small range of genes (about 2-3 genes)

Tumor tissue from biopsy or surgery

- Advanced Genomic Sequencing:

Broad range of genes (600-2000+)

Specialized laboratories

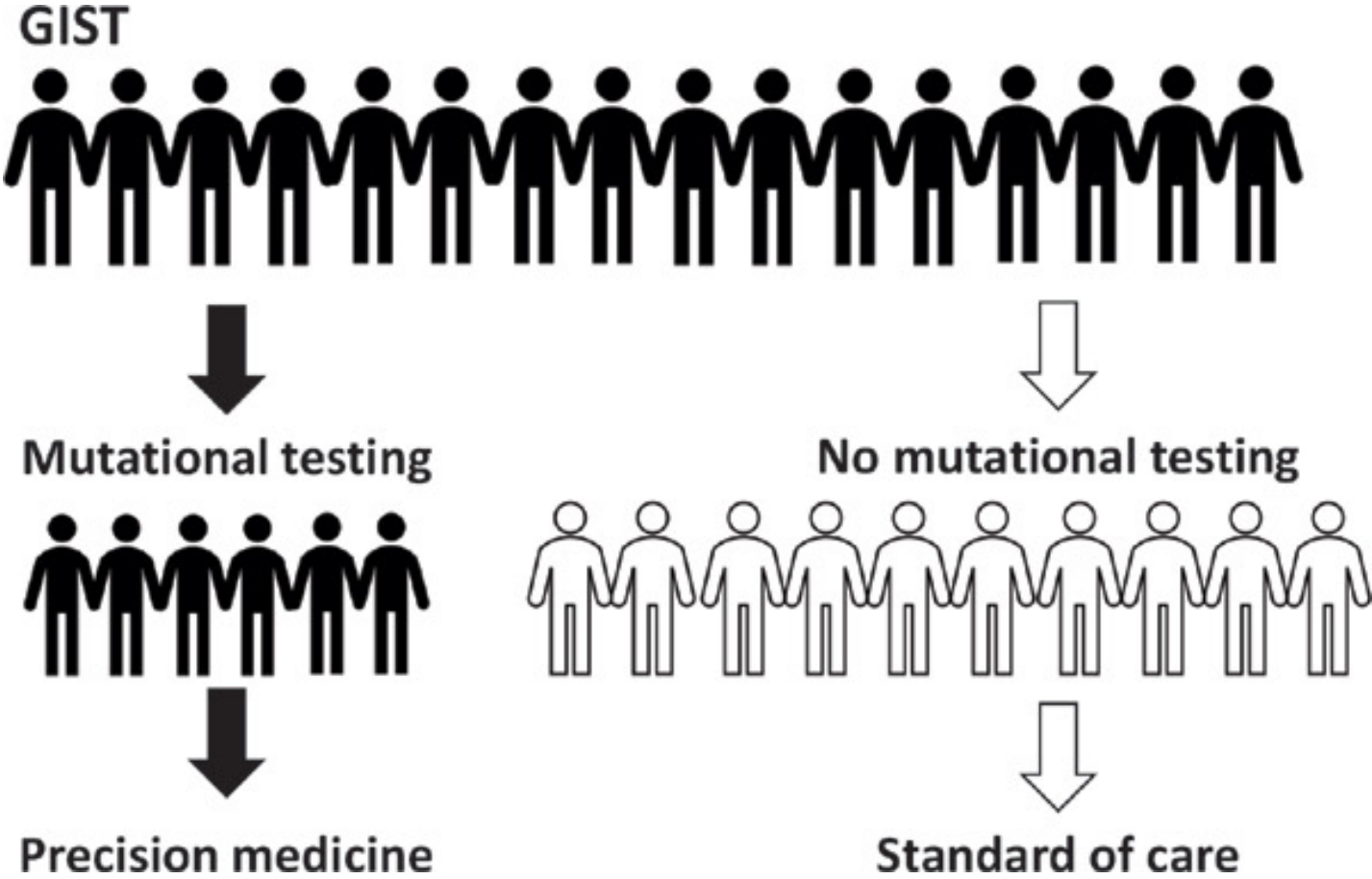
Tumor tissue from biopsy or surgery

- Genetic Testing:

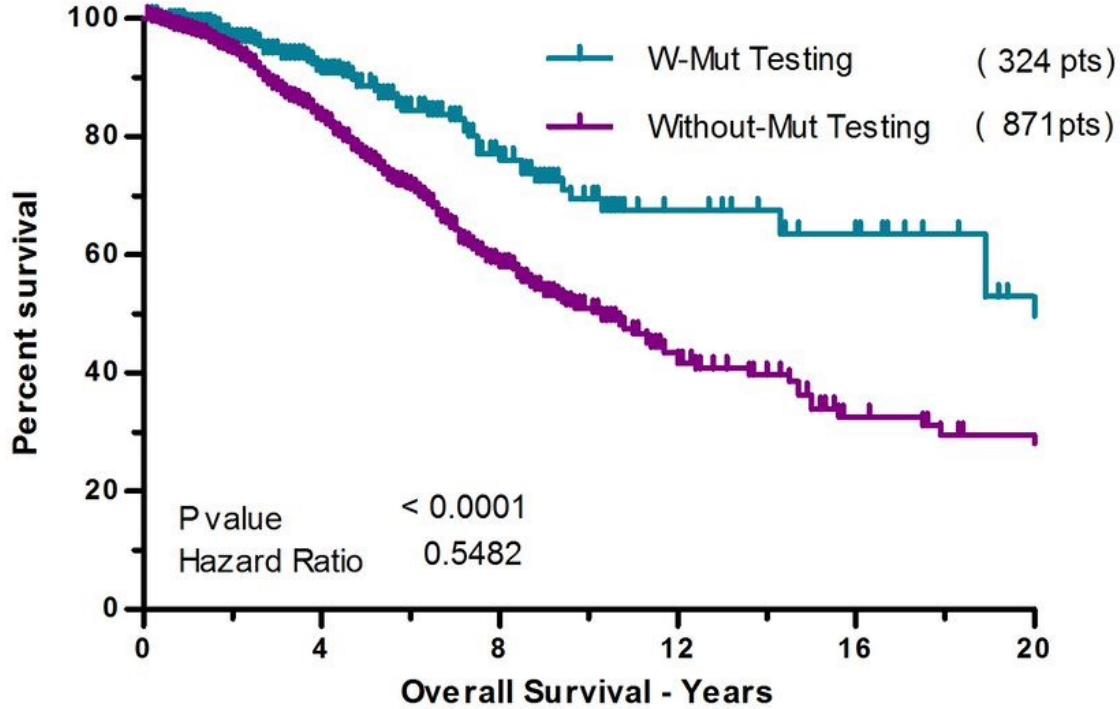
Usually done after a SDH mutation

This is performed with normal tissue (saliva or blood)

WHY IS THIS
IMPORMTANT
FOR MY GIST?



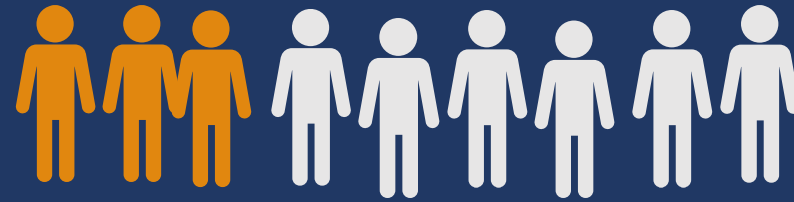
All LRG patients With or Without Mutational Testing



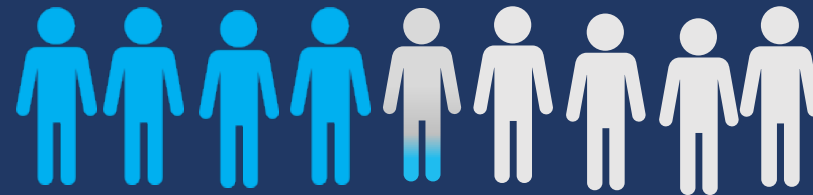
Median Overall Survival
With mutation testing 22.5 years
Without mutation testing 10.3 years

Mutational Testing Rate

About **30%** of GIST patients have received mutational testing



54% LRG patients know their mutations



CASE REPORT: PATIEN REGISTRY

Summary:

35 years old female patient; diagnosed at age 32 (2018)

Patient was pregnant and during her first trimester visit to her OBGYN a “fibroid” was observed by her doctor. The doctor recommended her to wait until her due date since the fibroid did not look suspicious

During her c-section (06/15/2018), surgeons found a 15x11x7.5 cm tumor attached to her small intestine with multiple metastasis on peritoneum measuring 5.7x5.5cm and 3.5cm

Mitotic rate: 10/50hpf

IHC stainings: CD117 positive ; Ki-67: 25%

Patient started imatinib 400mg (07/2018)

Initial mutational testing report showed (07/2018): **KIT/PDGFA WT**

Patient stopped treatment due to her WT mutation

Advanced testing was performed (09/2018) **showing KIT exon 11 C.1648-3_1673del**

Patient re-started imatinib 400mg (09/2018)

01/2019: Significant shrinkage was observed

06/2019: No tumors were observed – NED

TISSUE BANK

486

Tissue Bank Members*



957

Total Number of Tissue

* All tissue bank members are part of the LRG Patient Registry so that we are able to connect clinical data to all tissue samples in our Tissue Bank

The Life Raft Group launched a **Tissue Testing Initiative** in 2021 to improve the understanding of biomarker testing among the GIST cancer community and increase the number of patients tested by providing FREE Next-Generation Sequencing (NGS) to eligible patients.

THANK YOU!

We are grateful for all our patients, caregivers, and key stakeholders that continue to support our efforts to move GIST research forward. We are not able to do it without *you*.



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