

My Mutation Matters

UNDERSTAND • TEST • TREAT



A guide to mutational testing for people with gastrointestinal stromal tumors (GIST)

All GIST is driven by an underlying genetic mutation. This guide will help you discuss with your care team which mutation drives your GIST and the best way to treat it.

Your test results and what they mean*

One way to think of your results is like an address:



The gene is the **city**; this is the gene that is affected.



The exon is the **street**; this is the area on the gene where the mutation is located.



The alteration is the **house number**; this is the exact change within the gene that is driving your GIST.

Test results usually take around 2 weeks to come back. If you started a treatment while waiting for your results, ask if your treatment plan should be changed based on your mutation.

Use the checklist below to record your mutational test results.

KIT gene mutation

no *KIT* mutation detected

Exon (check all that apply):

9 11 13 14 17

no exon information in my report

Alteration (write in): _____

PDGFRA gene mutation

no *PDGFRA* mutation detected

Exon (check all that apply):

12 14 18

no exon information in my report

Alteration (write in): _____

Additional gene mutational testing

(if no mutation is detected in *KIT* or *PDGFRA*)

BRAF mutation detected

SDH A/B/C/D mutation detected

NF1 mutation detected

NTRK fusion detected

Alteration: _____

My mutation

What gene, exon, and alteration do I have?


Answer

Gene: _____

Exon (if included in report): _____

Alteration: _____

If your care team cannot provide the information you need to complete the gene and alteration fields, ask them to clarify why this information is not available and what other options you have for understanding your mutation.

 **Take this guide to your next appointment to help start the conversation about which mutation may be causing your GIST.**

* Your alteration result may contain letters and numbers that explain how the gene (DNA) or protein (amino acid) has changed. Always ask your care team to clearly explain your mutational testing results. Some test results may not include your exon, but results should always include information about your gene and alteration. The alteration listed in mutational test results can represent a change in the amino acid or DNA change.

Key questions for your care team

The following questions may be helpful in discussing mutational testing results with your care team and how they may affect your treatment.

Getting treatment for your GIST

Key question

Imatinib is usually the first treatment for many patients with GIST, but I have learned that it does not work for all mutations. Is imatinib effective in treating my type of mutation?

Answer

Key question

What other therapies may or may not work for my mutation?

Answer

Key question

Are any new treatments for GIST approved or available in clinical trials that may be suitable for my mutation?

Answer

Important: Once you have your test results, you may be able to join a clinical trial. Talk with your doctor about which treatment might be best for your type of GIST, including treatments available in clinical trials.

References:

- Bannon AE, Klug LR, Corless CL, Heinrich MC. Using molecular diagnostic testing to personalize the treatment of patients with gastrointestinal stromal tumors. *Expert Rev Mol Diagn.* 2017;17(5):445-457.
- Casali PG, Abecassis N, Bauer S, et al, on behalf of the ESMO Guidelines Committee and EURACAN. Gastrointestinal stromal tumours: ESMO–EURACAN Clinical Practice Guidelines for diagnosis, treatment and follow-up. *Ann Oncol.* 2018;29(suppl 4):iv68-iv78. Retrieved November 27, 2018, from the European Society for Medical Oncology website.
- Demetri GD, Benjamin RS, Blanke CD, et al. NCCN Task Force report: management of patients with gastrointestinal stromal tumor (GIST)—update of the NCCN Clinical Practice Guidelines. *J Natl Compr Cancer Network.* 2007;5(2_suppl):S-1-S-31.
- Gastrointestinal stromal tumor. Genetics Home Reference. National Institutes of Health website. <https://ghr.nlm.nih.gov/condition/gastrointestinal-stromal-tumor>. Updated January 2, 2019. Accessed January 2, 2019.
- Hameed M, Corless C, George S, et al, for the Members of the Cancer Biomarker Reporting Committee, College of American Pathologists. Template for reporting results of biomarker testing of specimens from patients with gastrointestinal stromal tumors [published online for public access]. *Arch Pathol Lab Med.* 2015;139(10):1271-1275. doi:10.5858/arpa.2014-0578-CP
- Mutation testing. GIST Support International website. <http://www.gistsupport.org/about-gist/for-new-gist-pages/mutation-testing>. Accessed November 26, 2018.
- Solid tumors GeneTrails GIST genotyping panel. Oregon Health & Science University website. <https://knightdxlabs.ohsu.edu/home/test-details?id=GeneTrails+GIST+Genotyping+Panel>. Accessed January 2, 2019.
- Understanding GIST. Life Raft Group website. <https://liferaftgroup.org/understanding-gist>. Accessed January 2, 2019.

Developed by



© 2019 Blueprint Medicines Corporation.
All rights reserved. USBP-DAA-18-004

Consider these tips when discussing GIST mutational testing with your care team

- 1 Take this guide with you, and address any questions or concerns you have noted below.
- 2 If possible, take a friend, family member, or caregiver with you to your appointment. Share your questions and concerns so that this person can help you ask questions and note your care team's answers.
- 3 Don't be afraid to tell your GIST care team if you don't understand something or if you need more information.
- 4 Ask for a copy of your mutational test report for your records.

Notes from my doctor visit

❖ **For more information about GIST and mutational testing, visit www.MutationMatters.com.**

