

Understanding Your Positive SDHD Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things to know

1	SDHD mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>SDHD</i> gene.
2	Non-cancerous tumor and cancer risks	If you inherited the <i>SDHD</i> mutation from your father, you have an increased chance to develop paragangliomas (PGLs). You may also have an increased chance to develop pheochromocytomas (PCCs), gastrointestinal stromal tumors (GISTs), or kidney cancer.
3	Other Medical Concerns	Individuals with <i>SDHD</i> mutations may have an increased risk to have a child with mitochondrial complex II deficiency, but only if their partner also carries a mutation in the <i>SDHD</i> gene. Mitochondrial complex II deficiency is a rare, highly variable autosomal recessive condition that can can affect many different parts of the body, including the brain, heart, and muscles.
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may also be at risk – they can be tested for the <i>SDHD</i> mutation that was found in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

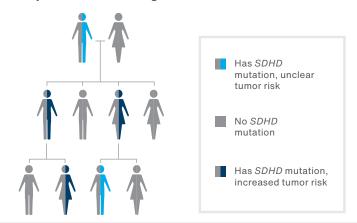
SDHD Mutations in the Family

There is a 50/50 random chance to pass on a *SDHD* mutation to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations.



SDHD Tumor Risk in the Family

SDHD mutations are equally inherited from your mother or father. Except for in rare cases, your risk for tumors is clearly known to be increased when the *SDHD* mutation is inherited from your father. The image below demonstrates this.



Reach Out

RESOURCES

- Pheo Para Alliance pheopara.org
- Genetic Information Nondiscrimination Act (GINA) ginahelp.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *SDHD* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.