

# Understanding Your Positive *TSC2* Genetic Test Result

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

## 6 Things to Know

1	<i>TSC2</i> mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>TSC2</i> gene.
2	Tuberous sclerosis complex	People with <i>TSC2</i> mutations have tuberous sclerosis complex (TSC).
3	Cancer and noncancerous tumor risks	You have an increased chance (2-5%) to develop kidney (renal) cancer, as well as non-cancerous tumors of the skin, brain, kidneys, heart, liver, and lungs.
4	Other Medical Concerns	<p>People may also have additional signs of TSC, which can include:</p> <ul style="list-style-type: none"> <li>• Patches of lighter skin color, or patches of overly bumpy or smooth skin</li> <li>• Small bumps on the face (facial angiofibromas)</li> <li>• Learning problems or delays</li> <li>• An increased risk for seizures</li> </ul> <p>You should discuss the characteristics of TSC in more detail with your healthcare provider.</p>
5	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.
6	Family	Family members may also be at risk – they can be tested for the <i>TSC2</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.

## TSC2 Mutations in the Family

There is a 50/50 random chance to pass on a *TSC2* mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth.



### RESOURCES

- Tuberous Sclerosis Alliance [tscalliance.org](http://tscalliance.org)
- National Society of Genetic Counselors [nsgc.org](http://nsgc.org)
- Canadian Association of Genetic Counsellors [cagc-accg.ca](http://cagc-accg.ca)

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *TSC2* 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.