Ambry Genetics®

Understanding Your Positive *PTCH1* Genetic Test Result

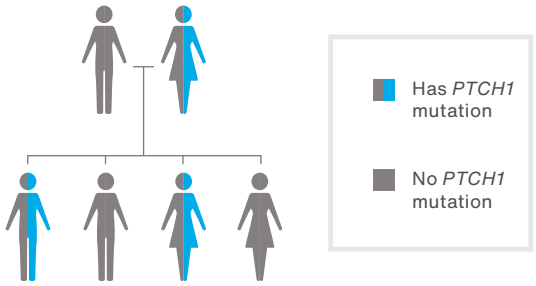
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

6 Things to Know

1	*PTCH1* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *PTCH1* gene.
2	Nevoid basal cell carcinoma syndrome (NBCCS)	People with *PTCH1* mutations have nevoid basal cell carcinoma syndrome (NBCCS), also known as Gorlin syndrome.
3	Cancer risks and other tumor risks	You have an increased chance to develop basal cell carcinoma (a type of skin cancer), noncancerous tumors called fibromas, and a type of noncancerous brain tumor (meningioma). Some children with NBCCS may develop a type of brain cancer called medulloblastoma. The risk for medulloblastoma decreases after childhood.
4	Other Medical Concerns	People with *PTCH1* mutations may have any of the following features and/or medical concerns: • noncancerous cysts in the jaw called keratocysts • facial features including a large head, broad nose, wide-set eyes, cleft lip and/or palate • pitting on the palms of hands or soles of feet • eye problems • skeletal problems including scoliosis or abnormal ribs
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 *PTCH1* 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.

PTCH1 Mutations in the Family

There is a 50/50 random chance to pass on a *PTCH1* 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	
	• Basal Cell Carcinoma Nevus Syndrome Life Support Network gorlinsyndrome.org • National Society of Genetic Counselors nsgc.org • Canadian Association 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 *PTCH1* 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.

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