UT Southwestern
Harold C. Simmons
Comprehensive Cancer Center

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MAX Mutations

What you should know about *MAX* mutations

MAX-Associated Hereditary Paraganglioma/Pheochromocytoma (PGL/PCC) syndrome is a genetic condition caused by mutations in the *MAX* (MYC-Associated Factor X) gene. The *MAX* gene is responsible for suppressing tumor development. A change, or mutation, in the *MAX* gene increases an individual's risk for certain types of tumors of the neuroendocrine system. The types of tumors are discussed below.

Cancer risks/tumors associated with *MAX* mutations

Mutations in the *MAX* gene increase an individual's risk to develop tumors (growths of cells) called pheochromocytomas (PCCs) and paragangliomas (PGLs). PCCs and PGLs are tumors of the neuroendocrine system, which is the system of the body that releases hormones. The majority of these tumors are benign (non-cancerous). However, individuals with mutations in the *MAX* gene appear to have an increased risk for these tumors to become malignant (cancerous), so surveillance and screening are important. It is not yet known if mutations in the *MAX* gene are associated with any other types of tumors or cancer.

Risks to family members

Mutations in the *MAX* gene are inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with a *MAX* mutation have a 50% chance of having the mutation. Some studies have suggested that *MAX* mutations may only increase the risk for tumor development if inherited from the father. This is called paternal transmission.

Managing cancer risks

As the *MAX* gene was more recently discovered, we do not yet know the lifetime risk for someone with a *MAX* mutation to get PCCs/PGLs. Although there is no consensus regarding surveillance strategies for individuals with *MAX* mutations, it is reasonable to consider life-long annual biochemical and clinical surveillance which can guide imagining studies. Screening for other genetic conditions associated with PCC/PGL has started at age 10, or 10 years before the earliest diagnosis in the family. Screening may include blood or urine analysis for hormones, CT or MRI scans, and physical exams to check for signs and symptoms of tumors. Management of tumors varies and might require surgery.

Last updated 01/09/2020

UT Southwestern Harold C. Simmons Comprehensive Cancer Center | 2201 Inwood Rd., Dallas, Texas 75390-9323
Phone: (214) 645-2563 | Fax: (214) 645-2562 | utswmed.org/cancergenetics