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My46 Trait Profile

Carney complex

Other Names: NAME (Nevi, Atrial Myxomas, Ephelides) syndrome, LAMB (lentigines, atrial myxoma, blue nevi) syndrome

Carney complex is a rare genetic condition in which individuals have a higher chance to have certain cancerous and noncancerous tumors. It is most commonly caused by mutations in the PRKAR1A gene which makes a protein called Protein Kinase, CAMP-Dependent, Regulatory, Type I, Alpha.

Characteristics of Carney complex

Individuals with Carney complex have an increased chance of developing cancerous (malignant) and noncancerous (benign) tumors, as well as other abnormalities.

Individuals with Carney complex have an inherited tendency to develop spots on their skin, myxomas (benign tumors) in the heart, skin and bones, as well as tumors in the hormone-producing (endocrine) gland. Features of Carney complex usually become apparent in in early adulthood. The change in skin coloring may be seen on the lips, in the corners of the eyes, in the lining of the eyes, and around the genital area (e.g., blue nevi or moles, significant freckling, and coffee colored brown spots called café-au-lait macules). The myxomas are most often located in the heart, skins or. The heart myxomas can block the flow of blood through the heart, causing serious complications. The hormone-producing tumors may be in the thyroid (organ located in the neck), pituitary gland (located in the base of the brain) which can cause increased size of hands, feet, and face, or in the adrenal gland (located on top of each kidney) which can cause an overproduction of a hormone (i.e., cortisol) that when increased leads to Cushing's syndrome (a combination of weight gain, high blood pressure, diabetes, easy bruising, and other health problems).

Other characteristics that develop in some individuals with Carney complex include benign breast abnormalities, colon polyps (benign growths), tumors in the testicles (LCCST: large cell calcifying Sertoli cell tumor), tumors that grow on nerves (psammomatous melanotic schwannoma), cardiomyopathy (disease of the heart), or skin tags or fatty tumors. Although people with Carney complex have an increased risk of certain cancers (e.g., thyroid, colon, pancreatic, ovarian, testicular, adrenal, and pituitary), most tumors that develop are benign (noncancerous).

Diagnosis/Testing

Carney complex can be diagnosed either by meeting certain clinical criteria or by genetic testing for a change or mutation in a gene called PRKAR1A. The PRKAR1A gene is a tumor suppressor gene which means that it keeps cells from growing too fast. Mutations in the PRKAR1A gene do not allow the protein kinase, CAMP-dependent, regulatory, type I, alpha protein to work normally and as a result, cells may grow uncontrollably. This uncontrolled growth is what can cause the tumors and other signs and symptoms of Carney complex in affected individuals.

Management/Surveillance

Management of Carney complex depends on symptoms, but screening may include ultrasounds of the heart and/or thyroid gland (in the neck), skin examinations, certain blood and/or urine tests, and/or physical examinations of the neck (to evaluate the thyroid). For men with Carney complex, physical examination and/or ultrasound of the testicles may also be suggested.

Mode of inheritance

Carney complex is inherited in an autosomal dominant pattern. This means inheriting one PRKAR1A mutation is enough for an individual to be affected and show signs of Carney complex. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child. Most of the time (for approximately 60-80% of individuals with Carney complex), the condition is inherited from an affected parent.

Risk to family members

The risk to family members depends on whether or not the individual with Carney complex has a parent affected with Carney complex. If a parent also has Carney complex, the risk of having a child with Carney complex is 50% with each pregnancy. If a parent does not have Carney complex, the risk of other siblings being affected is very low.

Special considerations

None

Resources

Genetics Home Reference: Carney Complex
http://ghr.nlm.nih.gov/condition/carney-complex
Cancer.Net: Carney Complex
http://www.cancer.net/cancer-types/carney-complex

References

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