Published on *My46* (https://www.my46.org) 11/14/2019

My46 Trait Profile

Birt-Hogg-Dube syndrome

Other Names: Hornstein-Birt-Hogg-Dubé syndrome, Hornstein-Knickenberg syndrome

Birt-Hogg-Dube syndrome is a rare genetic condition in which individuals have a higher chance to have certain cancerous and noncancerous tumors. It is caused by mutations in the FLCN gene which makes the folliculin protein.

Characteristics of Birt-Hogg-Dube syndrome

Birt-Hogg-Dubé syndrome (BHD) is a rare genetic condition that primarily affects the skin, lungs, and kidneys. Noncancerous (benign) skin tumors commonly appear on the face, neck, and chest beginning as early as 20 or 30 years of age. These tumors often become larger and greater in number with age. Individuals with BHD can also develop cysts in their lungs. These cysts generally appear in both lungs and in more than one site in each lung. Typically there are no health concerns related to the lung cysts, although there is an increased chance of experiencing a spontaneous collapsed lung (pneumothorax); this can be recurrent (i.e., happen repeatedly). BHD is also associated with an increased risk for the development of kidney tumors, both cancerous (malignant) and noncancerous (benign). Similar to the lung cysts, kidney tumors typically develop in both kidneys and in more than one site in each kidney. These tumors tend to grow slowly and generally do not appear until an individual is in their 30s or older.

Diagnosis/Testing

Most individuals with BHD have a change or mutation in a gene called FLCN that makes a protein called folliculin. The exact function of folliculin is still to be determined, but it is found in different tissues in the body, including the skin, lungs, and kidneys. Mutations in the FLCN gene are thought to interfere with proper cell growth and division.

Management/Surveillance

Management of BHD includes a complete skin exam and visualization of the lungs using specialized imaging (e.g., high-resolution computed tomography (HRCT) or CT) to determine the status of lung cysts. Individuals suspected of experiencing pneumothorax should have a chest x-ray and CT, and immediate treatment, as needed. An abdominal/pelvic CT or MRI is also recommended to monitor the kidneys. MRI is preferred by some to minimize radiation exposure.

Individuals with BHD have an increased risk of developing kidney cancer and should be monitored for tumor development. A baseline abdominal/pelvic CT or MRI should be obtained upon diagnosis, with surveillance occurring approximately every three to five years afterwards. Kidney ultrasounds may also be used, especially in cases where an individual has had two normal CT or MRI exams and has no family history of kidney cancer. It is strongly recommended that individuals with BHD do not smoke cigarettes, as it significantly increases the risk for lung disease and kidney cancer in individuals with BHD. Additionally, those with BHD should be aware of an increased risk for pneumothorax during pressure changes, such as when flying in an airplane.

Mode of inheritance

BHD is inherited in an autosomal dominant pattern. This means inheriting one FLCN mutation is enough for an individual to be affected and show signs of BHD. The mutation can be inherited from an affected parent or it can occur

brand new (de novo) in an affected child.

Risk to family members

For an individual with BHD, the chance of having a child with BHD is 50% with each pregnancy. The risk to an individual's siblings depends on whether or not the individual with BHD has a parent affected with BHD. If a parent also has BHD, then each sibling has a 50% chance to also have BHD. If a parent does not have BHD, the risk of siblings being affected is very low.

Special considerations

None

Resources

Birt-Hogg-Dubé Syndrome: Research and Support
http://www.bhdsyndrome.org/
Genetics Home Reference: Birt-Hogg-Dubé syndrome
http://ghr.nlm.nih.gov/condition/birt-hogg-dube-syndrome
Cancer.Net: Birt-Hogg-Dubé Syndrome
http://www.cancer.net/cancer-types/birt-hogg-dubé-syndrome

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