

## About *FLCN* Mutations and Birt-Hogg-Dubé Syndrome

### About Genes

Genes are in every cell in our bodies. Genes are made of DNA, which tells cells how to grow and work together.

We have two copies of each gene in each cell—one from our mother and one from our father. When genes work properly, they help stop cancer from happening.

Sometimes changes to genes can happen. We call these changes mutations. Mutations can make the genes stop working. They can raise the risk for certain types of cancer. Some mutations can be passed through families. When that happens, family members who have the mutation have a hereditary cancer syndrome.

The *FLCN* gene helps control the growth and division of cells. When *FLCN* does not work properly, cells can grow out of control. This can lead to tumors and cancer. A person who inherits a mutated copy of the *FLCN* gene has a hereditary cancer syndrome. We call this Birt-Hogg-Dubé (BHD) syndrome.

### Birt-Hogg-Dubé Syndrome (BHD)

BHD is very rare. BHD raises the risk of getting tumors that are not cancer in the skin and lungs. Your care team may call these benign tumors. BHD also raises the risk of getting kidney tumors. These kidney tumors can be benign, or they may be cancer. Not everyone with BHD gets all of these types of tumors. Signs of BHD usually develop in a person's 20s or 30s.

### BHD and Tumor Risk

**Skin.** About 9 out of 10 people with BHD get some skin bumps or tumors on the skin, head, or upper body. These tumors are not cancer. Several types of tumors can happen:

- Pimple-like bumps
- Skin tags
- Solid, reddish, or brown bumps
- Fatty lumps

**Lungs.** About 9 out of 10 people with BHD get lung cysts. They are sacs of air on the surface of the lungs. These lung cysts do not usually affect lung function. Sometimes cysts can burst and cause a collapsed lung.

About 1 in 3 people with BHD will get a collapsed lung in their lifetime.

These are signs of a collapsed lung:

- Painful breathing
- Dry cough
- Chest tightness
- Rapid heart rate
- Bluish skin color from lack of oxygen

**Kidneys.** BHD can cause sacs filled with fluid, called cysts, to happen in the kidney. By themselves, they are not harmful, but they can develop into tumors. About 1 out of 3 people with BHD get kidney cancer. BHD kidney tumors are often slow-growing and do not spread to other parts of the body.

BHD Health Concern	Risk with BHD
Skin Lesions	90 out of 100
Lung Cysts	90 out of 100
Collapsed Lung	25 out of 100
Kidney Tumors	33 out of 100

### Recommendations

No specific screening guidelines for BHD exist at this time. Due to the higher risk of tumors, physicians often recommend the following at the time of BHD diagnosis:

- Yearly MRI of the kidneys.
- Baseline CT scan of the chest. Follow up with chest CT scan if there are signs of a collapsed lung.
- Yearly full-body skin exam by a dermatologist. Follow sun safety recommendations whenever you are outside.
- Avoid scuba diving and extreme contact sports to lower the risk of a collapsed lung.

## **CHILDREN AND SIBLINGS**

Siblings and children of people with BHD have a 1 in 2 chance of also having BHD. BHD affects men and women equally.

### **For More Information**

Do you have questions about your risk for cancer? Our doctors and genetic counselors can help find the cancer screening plan you need.

Call Huntsman Cancer Institute's Family Cancer Assessment Clinic at 801-587-9555