

## PIK3CA-Related Overgrowth Spectrum (PROS)

# GLOSSARY OF COMMON TERMS AND DEFINITIONS

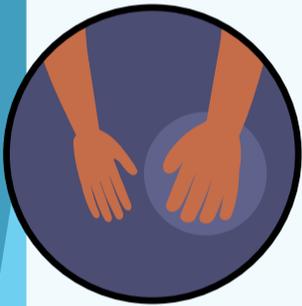
### MEDICAL TERMS

#### **PIK3CA-Related Overgrowth Spectrum (PROS)**

A group of conditions where one or more parts of the body grow at a different rate. PROS conditions are usually caused by a change in the PIK3CA gene.

#### **Angioma**

A swelling on the body made up of little blood vessels or lymph vessels under the skin that have thin, leaky walls.



#### **Overgrowth**

This means a part of the body grows extra tissue. In PROS conditions, this can happen externally on the body, meaning it is visible under the skin, as well as internally with overgrowths of deep soft tissue. An overgrowth can be in one specific area or spread over parts of the body.

#### **Congenital**

This means a condition or trait is present at birth.

#### **Vascular Malformation/Anomalies**

Growths or differences in blood vessels, like your veins.

#### **Capillary Malformation**

Very small blood vessels below the skin, called capillaries, become bigger and darker than normal. (See Port-Wine Stains)

#### **Port-Wine Stains**

A birthmark that is both red and purple resulting from capillary malformations under the skin.

#### **Fatty Truncal Mass**

A growth of fatty tissue around the stomach or chest, which is bigger than normal.

#### **Lymph**

Clear, watery fluid that contains white blood cells, which can be found under the skin circulating in lymph vessels.



#### **Vascular System**

This is also known as the circulatory system, which is made up of the bloodstream, lymph vessels, and blood vessels, like arteries (which carry blood from the heart to other parts of the body) and veins (which carry blood back to the heart). The system carries blood cells and lymph fluid to other parts of the body.

#### **Lymphatic System**

A network of vessels that collects fluid from tissue in the body and brings it back to the blood.

#### **Tissue**

A group or layer of cells that collectively perform the same function. For example, muscle tissue is composed of a group of muscle cells.

#### **Lymphatic Malformation**

Growths or differences in lymph vessels that form spongy clumps which prevent fluids from flowing in the lymph vessel system.

#### **Benign**

Something that is not harmful in effect, or results in a mild effect.

#### **Lipomatous**

A mass made up of fat cells either underneath the skin or deeply penetrating the body.

#### **Thrombus**

A blood clot that forms inside the bloodstream and blocks blood from moving forward in a vessel.

#### **Scoliosis**

Changes to the spine (bones in the back), which cause it to curve too much to one side.

# MEDICAL TOOLS

## DOCTORS' VISITS



### X-Ray

Special pictures of the human body's internal composition, like images of your bones. These relatively quick and easy pictures don't look like the ones on your camera, but are black and white showing doctors if something looks off.

### Biopsy

A procedure where your doctor removes a tiny sample of cells or tissues with surgical tools. Your doctor may do this if they are trying to confirm a diagnosis.

### CT/CAT Scan

This scan has nothing to do with cats, but CT stands for Computed Tomography. This is like an x-ray, but more time-consuming, and gives doctors a more detailed image of the soft tissue inside the body.

### Hemispherectomy

This is a special surgery where doctors remove or disconnect one side of the brain from the other. This surgery can help manage seizures, but is only used in extreme circumstances.

### Ultrasound

This is another way a doctor looks inside the body, which is different from x-rays and MRIs. The doctor uses a machine that bounces sound waves off organs to create a picture of what is happening inside the body.



### Elevation

This is when you raise a body part up, like moving one leg higher than the other, or lifting (elevating) your feet above your chest.

### Surgery

When doctors fix a problem inside the body. For example, joint replacement is a surgery where doctors can fix the places in your body where bones meet.

### Ablation

When a doctor removes a piece of body tissue during surgery.

### MRI

This is short for Magnetic Resonance Imaging. MRI machines use radiating waves to take very detailed pictures of the inside of your body to help doctors understand what's happening inside you. Taking these detailed pictures, which can include your soft tissue and organs, can be quite time-consuming because you must stay very still.

### Speech Therapy

Training to help a person talk or say words more easily.

### Occupational Therapy

Treatment by implementing mobility exercises and stretching to help a person move their small muscles better.

### Physical Therapy

Training with exercises and stretching to help a person move their large muscles better.

### Crutches

Crutches are two metal sticks that you place under your armpits to lean on when walking. They can help reduce pain or strain in legs when walking.



### Wheelchair

A wheelchair is something you can sit on that has wheels to aid mobility. It can help you move around more easily if you have a hard time moving around on your legs or need help conserving energy.

# CLOTHING



### Shoes

Sometimes standard shoes will not fit, but shoes can be customized to fit you based on your feet.

### Glasses

Glass frames can be modified in many ways to fit your specific needs.

### Leggings

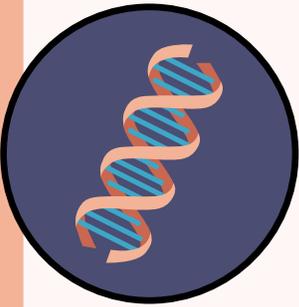
Pieces of tight cloth put on someone's legs to cover or support the body.



### Compression Garment

A very tight piece of cloth that can apply pressure to a body part, like your arms or legs. This helps to keep your blood circulating throughout your body.

# GENETIC TERMS



## DNA

This is a building block of your genes, found in all the body's cells. It carries information about how a person will look and function. DNA stands for **D**eoxyribonucleic **A**cid.

## Genes

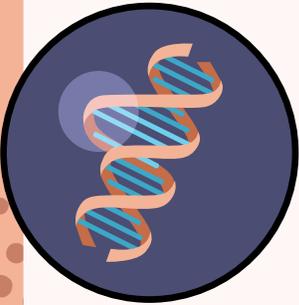
These aren't the blue pants you wear! Genes are small pieces of DNA that determine physical features, like your eye or hair color. Genes can be passed down from both your mom and dad.

## Genetics

This is the study of genes, or what makes you, you. Genetics tells us how the physical traits you have were passed down, or inherited, from your parents.

## PIK3CA Mutation

When the PIK3CA gene is affected, the wrong instructions are given to your body to increase activity of the PI3K protein. These mutations are random and sporadic. Genetics says that traits are passed down from your parents, but not in the case of sporadic mutations, which aren't passed on to you from your parents. It can present differently in cells and does not affect all your cells in the body.



## Mutation

A mutation is a change in a person's DNA sequence that can cause a cell or cells to grow or act differently. These changes can happen at birth or develop over time. In PROS conditions, the changes to DNA are something that you are born with.

## Mosaicism

Early in pregnancy, the baby, known as an embryo, develops a single mutation in a cell. The affected cell will continue to multiply alongside normal cells more than it should, creating an irregular, "mosaic" pattern. The number of affected cells in the body depends on when and where the first mutation occurs in pregnancy.

## PI3K

This is a protein called Phosphoinositide 3-Kinase. Its job is to make sure your cells grow, live, and die correctly.

## PIK3CA Gene

The PIK3CA gene gives orders to your body to make PI3K.



*This material is solely intended for educational purposes and does not depict the experiences of any specific patients. Any questions about proper care for PROS conditions should be directed to your health care provider.*