UNDERSTANDING**PROS**
PIK3CA-Related Overgrowth Spectrum

MACRODACTYLY

ABOUT *MACRODACTYLY*, A PROS CONDITION

What is **Macrodactyly**?

- Macrodactyly is a PROS condition that is generally characterized by an overgrowth of a limb, or part of a limb, mostly affecting the fingers or toes
- Most people with Macrodactyly are born with the condition. It is not hereditary

Different Conditions, **One Common Cause – PROS**

- PROS, or PIK3CA-Related Overgrowth Spectrum, is a wide-ranging spectrum of disorders caused by a mutation in the PIK3CA gene
- PROS conditions are rare and diverse, and are typically characterized by atypical growths and anomalies in the blood vessels and lymphatic system
- PROS conditions can look different from each other in size, shape, and type of growth or malformation based on where in the body the mutation is found

What are features of **Macrodactyly**?

Macrodactyly is different for each person and ranges in severity. Most people living with Macrodactyly do not have every sign associated with the condition.

These features may include:

- Enlargement of the affected limb or part of the limb
- Overgrowth of the skeletal structure
- Differences in growth between affected and unaffected limbs
- Loss of function in the affected limb
- Abundance of fatty cells in the affected limb

Macrodactyly Diagnosis

- Because Macrodactyly has observable features, it often can be diagnosed through a physical exam, imaging, and/or medical history
- Additionally, there are tissue tests to identify the presence of a PIK3CA mutation
- However, a negative test does not rule out having a PROS condition as mutations can be difficult to detect

Macrodactyly Management

- Managing Macrodactyly can be difficult and typically requires collaboration from a multidisciplinary team
- Like other PROS conditions, there are few management options for Macrodactyly
- Some options, such as surgery, address symptoms and manifestations rather than the root cause of the disease

Find resources and support for people with Macrodactyly and their families by visiting:

- **understandingpros.com**

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