



Proteus syndrome

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Proteus syndrome and where to get help.

What is Proteus syndrome?

A syndrome is a collection of signs and symptoms that have the same cause. Proteus syndrome is a medical condition that leads to disproportionate growth of tissues such as bone, skin, vascular and fatty tissue. It can affect almost any part of the body.

What causes Proteus syndrome?

Proteus syndrome is a genetic condition but it is not passed on from parent to child – rather it is caused by a sporadic (out of the blue) mutation or change in a gene called *AKT1*. The *AKT1* gene is responsible for the growth, division and death of cells and the mutation causes the cells to overgrow. Proteus syndrome is a 'mosaic' condition, that is, some cells contain the mutated gene but others do not, which causes the overgrowth to affect only parts of the body. It is a very rare condition.

What are the signs and symptoms of Proteus syndrome?

The main feature of Proteus syndrome is asymmetrical overgrowth of parts of the body. This overgrowth is often not present at birth but becomes more noticeable over time. In addition to overgrowth, children with Proteus syndrome may have distinctive facial features with a long face, low nasal bridge, large nostrils and downturned outer part of the eye. Some children have seizures, vision problems and learning difficulties.

How is Proteus syndrome diagnosed?

As Proteus syndrome is a rare condition, diagnosis will usually only be possible at a specialist centre with input from a multidisciplinary team. Proteus syndrome is diagnosed according to certain criteria, and must be distinguished from other overgrowth syndromes.

The diagnosis is confirmed by identification of a mutation in the *AKT1* gene in affected tissue.

How is Proteus syndrome treated?

Proteus syndrome cannot be cured but there is much that can be done to deal with the symptoms.

As Proteus syndrome can affect many areas of the body, treatment is usually managed by a multidisciplinary team including dermatologists (skin specialists), orthopaedic (bone and joint) surgeons, plastic surgeons and radiologists (imaging specialists) with other specialists brought in as needed. Regular review is advisable so that any symptoms can be monitored and managed promptly if problems occur.

Surgery may be required for scoliosis (spinal curvature) or to specific areas of overgrowth. Physiotherapy and occupational therapy can be helpful to improve movement, with adaptations to footwear where needed. Haematologists may need to be involved when it is thought there is a risk of abnormal blood clotting. Relatively recently medical treatments are in trials to modify the course of Proteus syndrome.



Psychosocial support is recommended for all children and young people with Proteus syndrome as the overgrowth affecting some areas of the body can lead to unwanted attention and self-consciousness.

What happens next?

The outlook for children and young people with Proteus syndrome is variable, depending on the proportion and type of cells in the body affected by the mutation.

Further information

The Proteus Family Network (UK) offers support and advice to anyone affected by Proteus syndrome. Call them on 01785 661263 or visit their website at www.proteus-uk.org