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## Proteus syndrome

Proteus syndrome is also known as elattoproteus syndrome and elephant man disease. The syndrome became widely recognised after the release of the movie “The Elephant Man”, a screenplay depicting the life of Joseph Merrick who was thought to have neurofibromatosis, but is now believed to actually have had severe Proteus syndrome.

A German paediatrician named the syndrome in 1983 after the Greek god Proteus, who had the ability to change his body into different shapes.

### What is Proteus syndrome?

Proteus syndrome is an extremely rare genetic disorder that is characterised by the abnormal growth of bones and blood vessels, and various skin lesions including lipomas, epidermal naevi and café au lait macules. The gene involved has yet to be identified but the mutation to the gene leading to this syndrome is thought to occur soon after conception. However, the condition is usually not evident until late infancy or early childhood when signs and symptoms start to appear.

### What are the signs and symptoms of Proteus syndrome?

#### Abnormal bone and soft tissue development

- Overgrowth of limbs, digits (fingers and toes) and skull. Overgrowth is usually disproportionate and asymmetric.
- Scoliosis (curvature of the spine) from abnormal vertebral growth
- Disproportionate overgrowth and focal atrophy (wasting away) of the upper arm muscles and neck, leading to the “hunched-over” look

#### Abnormal skin and connective tissue development

- Raised rough skin (verrucous [epidermal naevi](#)) or flat, soft variety of epidermal naevi
- Deep lines and overgrowth of soft tissue on the soles of the feet (cerebriform [connective tissue naevi](#))
- Local overgrowth of fat ([lipomas](#)) or invasive lipomas that produce large intra-abdominal or intrathoracic lesions that can be medically dangerous
- Abnormal growth of blood or lymphatic vessels ([vascular malformations](#)) can create vascular lesions such as port wine stains or [lymphatic malformations](#).
- [Café au lait macules](#) are often present

#### Common facial features include:

- Prominent occiput (enlarged back of the head)
- Drooping eyelids
- Low nasal bridge and wide, upturned nostrils
- Long, narrow face
- Open mouth at rest

Internal organs affected include the spleen and thymus, which can become enlarged. The condition also causes quite significant physical disfigurement and learning disabilities or developmental delays.

## What is the treatment for Proteus syndrome?

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There is no specific treatment for Proteus syndrome. The condition should be managed by identifying early any serious medical problems and providing prophylactic and symptomatic treatment. A multidisciplinary team of doctors is required to manage Proteus syndrome. This should include and orthopaedist, craniofacial surgeon, plastic surgeon, dermatologist, dentist, neurosurgeon, geneticist, and psychologist.

Management of skin abnormalities may include:

- Periodic evaluation of lesions to ensure lipomas and vascular malformations do not become a problem locally or internally
- Removal of cutaneous vascular markings such as port wine stains and capillary haemangiomas using [laser treatment](#)
- Surgical [excision](#) of lesions if they interfere with functional activities or for cosmetic purposes.

### Related information

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#### References:

- [Proteus Syndrome](#) – emedicine, the online textbook

#### On DermNet NZ:

#### Other websites:

- [Proteus Syndrome Foundation UK](#)

#### Books about skin diseases:

See the [DermNet NZ bookstore](#)

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DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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