# **The Joseph Merrick Story: The Elephant Man's Bones Reveal Mystery**

## **The Elephant Man Stumped Physicians for 100 Years—Find out Why**

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When he was just two years old, Joseph Merrick's mother noticed that some areas of his skin began to change. Some darkened, discolored skin growths were appearing, and they began to look bumpy and rough. Lumps began to grow under the boy's skin - on his neck, his chest, and the back of his head. Mary Jane Merrick began to worry about her son, Joseph, and the other boys were starting to make fun of him.

As Joseph grew older, he began to look even more strange. The right side of his head began to grow, as did his right arm and hand. By the time he was 12 years old, Joseph's hand was so deformed it became useless. The growths on his skin were now large and repulsive for most people to look at.

### How Joseph Merrick Became the "Elephant Man"

Over the next years and with the passing of his mother, Joseph left home, tried working in a factory but was abused by the workers there, and finally ended up in a freak show. By now his face was distorted by the overgrown half of his head, and the flesh around his nose had grown, too, leading the show promoter to dub Joseph "The Elephant Man."

### The Wrong Diagnosis

Most people know the rest of the story from the 1980 movie, "[The Elephant Man](http://www.amazon.com/The-Elephant-Man-John-Hurt/dp/B00003CX9S%3FSubscriptionId%3DAKIAJPGHBBSYH2CAHVXQ%26tag%3Daboutcom02allnetwork-20%26linkCode%3Dxm2%26camp%3D2025%26creative%3D165953%26creativeASIN%3DB00003CX9S/ref=as_at?tag=aboutcom02allnetwork-20&linkCode=as2&)," starring John Hurt: how, at first, a doctor, then others including royalty, came to see the intelligent, sensitive man behind the grotesque deformities.

People have been moved by the universal message of tolerance of the different found in Joseph Merrick's story. But what most people don't know is that it took 100 years for doctors to correctly identify his medical condition.

At the time Joseph Carey Merrick lived (1862-1890), leading authorities stated he suffered from elephantiasis.

This is a disorder of the lymphatic system that causes parts of the body to swell to a huge size. In 1976, a doctor postulated that Merrick suffered from neurofibromatosis, a rare disorder that causes [tumors to grow on the nervous system](https://www.verywell.com/neurofibromatosis-2860969). Photos of Merrick, however, do not show the brown skin spots characteristic of the disorder. Also, his disfigurement came not from tumors but from bone and skin overgrowth. Unfortunately, even today people still (wrongly) call neurofibromatosis the "Elephant Man disease."

It wasn't until 1996 that the answer to what affected Merrick was found. A radiologist, Amita Sharma, of the National Institutes of Health (U.S.), examined x-rays and [CT scans](https://www.verywell.com/what-is-a-ct-scan-2860786) of Merrick's skeleton (kept at the Royal London Hospital since his death). Dr. Sharma determined that Merrick had Proteus syndrome, an extremely rare disorder, itself only identified in 1979.

### Proteus Syndrome

Named for the Greek god who could change his shape, this rare hereditary disorder is characterized by:

* multiple lesions of the lymph nodes (lipolymphohemangiomas)
* overgrowth of one side of the body (hemihypertrophy)
* an abnormally large head (macrocephaly)
* partial gigantism of the feet, and darkened spots or moles (nevi) on the skin.

Merrick's appearance, and especially his skeleton, carry all the hallmarks of the disorder, although apparently an extremely severe case. His head was so large that the hat he wore measured three feet in circumference.

### How the Story Ended

More than anything, Joseph Merrick wanted to be like other people. He often wished he could lie down while sleeping, but because of the size and weight of his head he had to sleep sitting up. One morning in 1890 he was found lying down in bed on his back, dead. The immense weight of his head had dislocated his neck and crushed his windpipe, suffocating him. He was 27 years old.

**PROTEUS SYNDROME**

Proteus syndrome is a rare condition characterized by overgrowth of the bones, skin, and other tissues. Organs and tissues affected by the disease grow out of proportion to the rest of the body. The overgrowth is usually asymmetric, which means it affects the right and left sides of the body differently. Newborns with Proteus syndrome have few or no signs of the condition. Overgrowth becomes apparent between the ages of 6 and 18 months and gets more severe with age.

In people with Proteus syndrome, the pattern of overgrowth varies greatly but can affect almost any part of the body. Bones in the limbs, skull, and spine are often affected. The condition can also cause a variety of skin growths, particularly a thick, raised, and deeply grooved lesion known as a cerebriform connective tissue nevus. This type of skin growth usually occurs on the soles of the feet and is hardly ever seen in conditions other than Proteus syndrome. Blood vessels (vascular tissue) and fat (adipose tissue) can also grow abnormally in Proteus syndrome.

Other potential complications of Proteus syndrome include an increased risk of developing various types of noncancerous (benign) tumors and a type of blood clot called a [deep venous thrombosis (DVT)](https://ghr.nlm.nih.gov/art/large/deep-venous-thrombosis-dvt.jpeg). DVTs occur most often in the deep veins of the legs or arms. If these clots travel through the bloodstream, they can lodge in the lungs and cause a life-threatening complication called a [pulmonary embolism](https://ghr.nlm.nih.gov/art/large/pulmonary-embolism.jpeg). Pulmonary embolism is a common cause of death in people with Proteus syndrome.

Proteus syndrome is a rare condition with an incidence of less than 1 in 1 million people worldwide. Only a few hundred affected individuals have been reported in the medical literature.

Researchers believe that Proteus syndrome may be overdiagnosed, as some individuals with other conditions featuring asymmetric overgrowth have been mistakenly diagnosed with Proteus syndrome. To make an accurate diagnosis, most doctors and researchers now follow a set of strict guidelines that define the signs and symptoms of Proteus syndrome.

Proteus syndrome results from a mutation in the [AKT1](https://ghr.nlm.nih.gov/gene/AKT1) gene. This genetic change is not inherited from a parent; it [arises randomly in one cell](https://ghr.nlm.nih.gov/art/large/proteus-syndrome-akt1.jpeg) during the early stages of development before birth. As cells continue to grow and divide, some cells will have the mutation and other cells will not. This mixture of cells with and without a genetic mutation is known as [mosaicism](https://ghr.nlm.nih.gov/art/large/mosaicism-2.jpeg).

The AKT1 gene helps regulate cell growth and [division](https://ghr.nlm.nih.gov/art/large/mitotic-cell-division.jpeg) (proliferation) and cell death. A mutation in this gene disrupts a cell's ability to regulate its own growth, allowing it to grow and divide abnormally. Increased cell proliferation in various tissues and organs leads to the abnormal growth characteristic of Proteus syndrome. Studies suggest that an AKT1 gene mutation is more common in groups of cells that experience overgrowth than in the parts of the body that grow normally.

**QUESTIONS:**

1) Why was Joseph Merrick not properly diagnosed for a long time?

2) How is Proteus Syndrome inherited? How does this differ from other genetic disorders you have learned about thus far?

3) Using your knowledge of the cell cycle, compare Proteus Syndrome to cancer (similarities and differences).

4) Use at least FIVE of the key terms from this unit to summarize how Proteus Syndrome works. Highlight or underline your five chosen terms.