

Even though many of us agree that vampires are in fact not real, there is always some fact to fiction.

The lore of “vampires” may have been real people with a rare blood disorder. Like many things that go misunderstood, people create fantasy and stories to ease the fear of the unknown. Vampire folklore (or some versions of it) date back to the first appearance of the English word vampire (as vampyre) in English from 1734. However, the modern vampire image, pale, gaunt, and sensitive to the sun, dates to the early 19th century. But like most folklore, there is a factual origin.

A new genetic study, published in **PNAS**, may be able to explain the skin-blistering “vampire” disease. Researchers report a newly discovered genetic mutation that is responsible for **erythropoietic protoporphyria** (EPP), a type of porphyria. Forms of porphyrias are caused by an abnormality in the heme production process. Heme is essential in enabling our blood cells to carry oxygen and in breaking down chemical compounds in the liver.



Left figure is urine on the first day while the right figure is urine after 3 days of sun exposures showing the classic change in color to purple.

A universal symptom of EPP is painful photosensitivity that can greatly impair quality of life. When exposed to light, even through a window, it can cause skin to swell, burn, itch, and turn red. These symptoms don't just fade either. They can last anywhere from 12 to 24 hours, but usually heal without significant scarring.

An international team of researchers, led by Dr. Barry Paw of [Dana-Farber/Boston Children's Cancer and Blood Disorders Center](#) have added one more mutation to the records with the discovery of a broken gene that usually plays a key role in changing the structure of proteins in the cell's mitochondria.

"The newly-discovered mutation really highlights the complex genetic network that underpins heme metabolism," says Barry Paw. "Loss-of-function mutations in any number of genes that are part of this network can result in devastating, disfiguring disorders."

Porphyrias is characterized by abnormally elevated levels of red blood cells and plasma, and due to sensitivity to visible light it is usually noticed in early childhood and occurs throughout life. EPP can result either from mutations of ferrochelatase gene, or less commonly the delta-aminolevulinic acid synthase-2 gene.

One component of EPP, protoporphyrin IX, is the reason for the so-called "vampirism" reaction to light. This component easily soaks up light energy, which it then passes onto oxygen to create what are called reactive oxygen species.

"People with EPP are chronically anemic, which makes them feel very tired and look very pale with increased photosensitivity because they can't come out in the daylight" says Paw.

In today's world, a patient would be advised to stay indoors during daylight hours and may be prescribed blood transfusions with sufficient heme levels to reduce symptoms. However,

back in the medieval and ancient times, they might have inspired the myths behind the nocturnal, blood-suckers known as vampires.

“Even on a cloudy day, there’s enough ultraviolet light to cause blistering and disfigurement of the exposed body parts, ears, and nose.”

The study is hoping to decipher which genes are responsible for the disease and if it could offer clues on how to treat its symptoms or even prevent it from developing. And aside from a potential origin story of vampirism, those involved in the study are hoping this insight will lead to therapies that correct the faulty genes in people with EEP.

“Although vampires aren’t real, there is a real need for innovative therapies to improve the lives of people with porphyrias,” Paw explains.

It might be too late for Count Dracula, but for those suffering from EPP, it could just be the beginning!

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Do Vampires REALLY Walk Among Us? Porphyria: The Vampire Disease



Article Name

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Description

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