Case Report

Congenital Erythropoietic Porphyria – Gunther’s Disease Basis for Werewolves and Vampires

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Porphyrias are a group of hereditary disorders of heme biosynthesis due to deficiency of one of any eight enzymes involved in biosynthetic pathways. Porphyrias are classified as neurovisceral, cutaneous and mixed on the basis of clinical symptoms as well as on defective enzyme. The word Porphyria was derived from Greek root for “purple” and the name was coined by German medical student Schultz in 1874. The first clinical description of acute porphyria was given by B. G. Stokvis in 1889. In 1930, Hans Fisher identified heme as chromopigment for red color of blood and green of grass. In 1960s, all the known types of porphyria were identified and environmental factor have been shown to affect the disease course. Famous historical figures like King George III of England and Vincent von Gogh are thought to be porphyria victims. Porphyrias were also linked to myths of vampires and werewolves and one of the porphyrias known as Congenital Erythropoietic Porphyria (CEP) or Gunthers disease is having manifestations linked to werewolves and vampires i.e. photosensitivity, taut skin, aversion to garlic and desire to drink blood as treatment for the disease.1-4.

Case History

A young male of 25 years resident of Rawalpindi presented to general practitioner for septic wound over both the hands and scalp. The fingers were deformed and skin was very taut and tight over them. Rest of the skin over the forearm and arm was normal. There was extreme hypertrichosis over face, limbs and back. The teeth were yellowish brown and the urine was reddish brown in color (Fig 1-5). The parents of the patient were first cousins and he had three brothers suffering from same illness.

Discussion

Congenital Erythropoietic Porphyria or Gunthers disease is a rare form of cutaneous porphyria. It is autosomal recessive in nature and very few cases have been described in medical literature. This porphyria is due to deficiency of uroporphyrinogen III cosynthase and accumulation of HMV which is converted non enzymatically to uroporphyrinogen I. In most of the cases severe photosensitivity is seen as friability and blistering over the sun exposed part of the skin. Severe hypertrichosis is present and teeth become reddish brown due to the accumulation of erythrodentia. The gene UROS responsible for formation of enzyme uroporphyrinogen III synthase is at chromosome No. 10 and cytogenetic location is 10q 25.2 – q 26.3. The most common gene mutation seen in 1/3rd of all cases is replacement of one amino acid cystein with arginine at position No. 73 (cys 73 Arg). There are other mutations which occur in nearby region of DNA regulating the UROS gene. 5-8
Reduced level of uroporphyrinogen III synthase allows accumulation of toxic substances in Red blood cells and these substances will leak into other tissues like skin, liver, bone marrow and teeth and cause over sensitivity to sunlight and other features of congenital erythropoietic porphyria.

References