A Review on Gunther’s Disease

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Abstract

Gunther disease, also known as congenital erythropoietic porphyria (CEP), is a congenital form of erythropoietic porphyria. The word porphyria originated from the Greek word porphura. Porphura actually means “purple pigment”. It is a rare, autosomal recessive metabolic disorder affecting blood caused by deficiency of the enzyme uroporphyrinogen cosynthetase. It is extremely rare, with a prevalence estimated at 1 in 1,000,000 or less. In milder cases patients have not presented any symptoms until they have reached adulthood. In Gunther's disease, porphyrins are accumulated in the teeth and bones and an increased amount are seen in the plasma, bone marrow, feces, red blood cells, and urine.

Keywords: CEP; Porphyria; Photosensitivity; Splenectomy

Introduction

CEP is a severe and rare childhood disorder causing lifelong mutilating photosensitivity and hematological disorder. It is an uncommon autosomal passive disorder of the porphyrin metabolism created by the homozygous deformity of uroporphyrinogen III cosynthase [1-5]. High measures of uroporphyrin I aggregate in all cells and tissues, reflected by an expanded erythrocyte porphyrin concentration and discharge of high porphyrin amounts in urine and feces. Dermal stores of uroporphyrin every now and again actuate sensational phototoxic oxygen subordinate skin damage with extensive ulcerations and mutilations. Splenomegaly and hemolytic anemia are internal symptoms. Skeletal changes, for example, osteolysis and calcifications are frequent. Splenectomy, erythrocyte transfusions and bone marrow transplantation have demonstrated some helpful impact. The best treatment is the avoiding of sunlight [6-10].

Current status of knowledge

The term Porphyria refers to a group of diseases characterised by excessive production and excretion of porphyrins, porphyrin precursor or both. It is classified based on the primary site of expression of the specific enzymatic defect into erythropoietic and hepatic forms and based upon the duration of lasting, classified into acute and non-acute [11-15]. Acute porphyrin acute intermittent porphyria, variegate porphyria, hereditary copro porphyria and ALA dehydratase deficiency or Plumbo porphyria) which present with neurovisceral symptoms. Non acute (CEP and Porphyria cutanea tarda) which present with different type of cutaneous findings including mild to severe photosensitivity, increased skin fragility, vesicles and bullae, burning and stinging, edema, pruritis, hypertrichosis, scarring [16-27]. The diagnosis of different types of porphyrias
requires the analysis and differentiation of porphyrin precursors in blood, urine and stool. The pattern of changes in porphyrin metabolism is of paramount importance in labeling different types of porphyria. This in the presence of a medical history permits an exact diagnosis [28-35].

CEP is an uncommon subtype of porphyria that causes skin to be sensitive to daylight; fundamental deformity is changes of alleles for the gene encoding the enzyme Uroporphyrinogen III synthase that prompts accumulation of porphyrins of type I isomer [36-43]. The C73R mutation is the most frequent in which cysteine is substituted by arginine. The accumulated isomer I porphyrinogens are spontaneously oxidized to their corresponding porphyrins, which are naturally futile yet cause cutaneous photosensitivity described by blisters, erosions and scarring of light exposed skin. These porphyrins are discharged from the developing erythrocytes into the plasma and are excreted through urine, thereby giving a portwine colour. The interaction of abundance porphyrins in the skin and light radiation causes photo-oxidative damage, showing as mechanical delicacy and blistering [44-52].

Photosensitivity happens early in course of infection. Increased delicacy and erosions can add to mutilation, particularly on the face and hands. Hypertrichosis of the face and furthest points is common. The teeth have a reddish colour and fluoresce under Wood' light because of porphyrin deposition in dentin and enamel. Ocular manifestation incorporate blepharitis, cicatrical ectopion and conjuctivitis, subsequent bilateral corneal scarring may happen with possible blindness [53-65]. Porphyrins are also deposited in the bone, resulting in loss of bone, resulting contractures and distortions occurs in grown-ups with CEP. Radiological features incorporate calvarium and meningeal calcifications, augmenting of diploic space in frontal and occipital areas. In more genuine cases, the hemolytic anemia of CEP can bring about hypersplenism. Bright pink fluorescence of urine, teeth and bones under Wood's light enlightenment can help in diagnosis [66-79]. The bright fluorescence of nuclei in erythrocyte antecedent cells is particular for Erythropoietic porphyria. Skin biopsy indicates subepidermal blister with superficial perivascular lymphocyte infiltrate [80-86].

For management of such patients, absolute avoidance of Sun exposure is crucial and Sunscreen containing Zinc oxide is preferable. Oral photo protectants (beta carotene) may prevent tissue damage due to light exposure possibly by forming an internal light screen [87-95]. Splenectomy might be shown for unmanageable hemolytic anemia. Transfusion of erythrocytes; intravenous hematin, oral activated charcoal, bone marrow transplant and gene therapy are different choices [96-104].

Conclusion
It should never be forgotten that most people found to have porphyria are able to lead a normal healthy life. All that is required is to take the couple of basic measures to diminish the danger of sickness that is portrayed here. Indeed, even the few who do turn out to be sick as a rule make an entire recuperation and have close to maybe a couple intense attacks in early grown-up life. As one becomes older, the danger of an intense attack reduces, especially after the age of forty, however it never vanishes totally.

REFERENCE


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