Our Dermatology Online

Case Letter

Is there the remote luckyness that microbic extracts may represent a godsend to combat Eritropoietic Porphyria in mammals, together with some special fats?

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Sir,

Erythropoietic protoporphyria (EPP) is an inherited porphyria resulting in the accumulation of protoporphyrins in red blood cells that causes acute, painful photosensitivity and potential liver disease. It typically presents in early childhood with immediate pain and crying upon exposure to bright sunlight. This activity reviews the evaluation and treatment of erythropoietic protoporphyria and highlights the role of the interprofessional team in the care of patients with this condition.

Erythropoietic protoporphyria (EPP) is an inherited condition resulting in the accumulation of protoporphyrins in red blood cells that causes acute, painful, non-blistering photosensitivity and potential liver disease [1]. It typically presents in early childhood with immediate pain and crying upon exposure to bright sunlight. It is seasonal in nature with symptoms principally occurring in the spring and summer season.

EPP is a lifelong disease, and repeated phototoxic reactions eventually lead to thickening of the skin and wax-like scarring on the face. In a small number of patients, the accumulation of protoporphyrins in the liver leads to cirrhosis and liver failure. Onset in adulthood is rare, but an acquired form has been identified, in which clones of cells with mutated ferrochelatase expand in the setting of the myelodysplastic or myeloproliferative syndrome [2].

EPP is caused by a mutation of ferrochelatase, which is the final enzyme in the heme biosynthesis cycle, adding Fe+2 with protoporphyrin IX (PPIX) to make heme. In patients with PP, PPIX significantly accumulates in red blood cells, plasma, liver, and skin due to ferrochelatase deficiency [3,4].

PPIX excites and releases its energy to oxygen, which may create free radicals that result in skin damages when the patients with EPP are exposed to light [4].

In 1961 erythropoietic protoporphyria was first comprehensively explained, and after that, it has been reported worldwide. It is recognized as the third most common porphyria in adults and the commonest porphyria in children. EPP equally affects both males and females. It is more common in east Asian people than in Whites and is rare in Africa. The incidence in Europian countries varies [5].

There is no definitive treatment for EPP in order to decrease the circulating porphyrin levels, but patients should avoid sunlight or fluorescent light. Parents should also learn about how to protect their children from sunlight. When going outside, patients must wear protective clothes and broad-brimmed hats and stay in shaded areas. The use of broad-spectrum

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sunscreen is also important. Cars and house windows should be equipped with protective tinted glass. Phototoxic reactions may occur as a result of visible light and ultraviolet A (UVA) light in operating rooms. Light filters should be used in operating rooms to protect the patient, especially in case of long surgical procedures like liver transplantation in a patient with hepatopathy [6].

Afamelanotide, a synthetic analog of alpha-melanocyte-stimulating hormone (MSH), normally increases the skin pigmentation and decreases the production of cytokines and free radicals [7]. It is given subcutaneously as a controlled released implant (16mg), especially in the summer season. Another agent which increases the sunlight tolerance is beta-carotene with oral doses of 30 to 300 mg/day for adults and 30 to 150 mg/day for children. It maintains plasma carotene levels in the range between 600 to 800 mcg/dL, and the serum carotene levels should be checked every three to four weeks. The risk of lung carcinoma is increased with high doses of beta-carotene. Other photoprotection measures with variable success include narrow-band UVB phototherapy and oral cysteine. Calcium and vitamin-D (daily intake of 1000mg of calcium with 800 IU of vitamin D) supplementation is necessary for patients with EPP because of sunlight avoidance. Nonsteroidal anti-inflammatory drugs (NSAIDs) or even opioids may give relief in acute painful episodes along with cold compresses.

Rapidly progressive and severe hepatopathy can be treated with a regimen that includes intravenous hemin (to reduce plasma porphyrin level), plasmapheresis, ursodeoxycholic acid, cholestyramine, vitamin-E (400IU), and correction of anemia. Liver transplantation is the treatment choice for severe protoporphyric hepatopathy or for patients who develop liver cirrhosis [8]. Bone marrow transplant is a newer treatment that may be done in conjunction wit hliver transplantation to prevent the recurrence of EPP, and reports show that it may be able to reverse or treat both photosensitivity and protoporphyric hepatopathy [9]. The presumed mechanism is related to a majority of heme synthesis in the bone marrow, thus allowing for a possibility of curative treatment in some case reports, particularly as some case reports show recrudescence of disease activity leading to fatal outcomes rapidly [8].

EPP results from a deficiency of the ferrochelatase enzyme, which is involved in the final step of heme biosynthesis. Insufficient ferrochelatase enzyme activity results in increased accumulation of protoporphyrin that lacks Fe+2 or other metals, particularly zinc (i.e., metal-free protoporphyrin). In EPP, the main source of production of increased red cell and plasma protoporphyrin is by bone marrow reticulocytes. Conditions that increase the erythropoiesis may result in increased formation of protoporphyrins by the bone marrow [10].

Swelling, burning, itching, and redness of the skin may appear during or after exposure to sunlight, including sunlight that passes through window glass. This can cause mild to severe burning pain on sun-exposed areas of the skin. Usually, these symptoms subside in 12 to 24 hours and heal without significant scarring. Blistering and scarring are characteristic of other types of cutaneous Porphyria but are unusual in EPP. Skin manifestations generally begin early childhood and are more severe in the summer.

The most common symptom of Erythropoietic Protoporphyria is hypersensitivity of the skin to sunlight and some types of artificial light (photosensitivity), with pain, itching, and/or burning of the skin occurring after exposure to sunlight and occasionally to fluorescent light. Affected individuals may also exhibit abnormal accumulations of body fluid under affected areas (edema) and/or persistent redness or inflammation of the skin (erythema). In rare cases, affected areas of the skin may develop sac-like lesions (vesicles or bullae), scar, and/or become discolored (hyperpigmentation) if exposure to sunlight is prolonged. However, scarring and/or discoloring of the skin is uncommon and rarely severe. These affected areas of skin may become abnormally thick. In addition, in some cases, affected individuals may also exhibit malformations of the nails. The severity and degree of photosensitivity is different from case to case. Photosensitivity is often seen during infancy; however, in some cases, it may not occur until adolescence or adulthood.

The authors has ideated a novel cosmetic formula which could keep on account a biomolecular sun protecting factor (generated by lysosomial estracts of bacterial colonies significative to prevent excellently sunburn especially in childhood and moreover two valuable greases that are capable to manitain softness and smoothness in damaged skin areas, idest Butyrospermum Parkii butter (Karitè butter, a fair trade oil or butter native to Ethiopian higlands that presents the same physical chemical characteristic of jojoba oil that can be buttery or oily, depending of the temperature

storage and therefore allowing a perfect pseudoplastic behaviour onto the inflamed skin and Cocos nucifera oil apt to increase the expression of Aquaporin3, filaggrin and involucrin, the maximum of the skin barrier defense in all warm blood animals [11,12].

This pommade has shown optimal and enthusiastic performances in one of the A (the oldest) who really suffers from EPP and cannot go outdoor if it is not raining or it is cloudy (frightening the occurence of severe blisters on his face and hands after the eventual exposure to actinic rays).

Very suggestive is also the fact that wjhen the A ears this pommade, his eyes do not burn or tear if whenever whichever source of artificial light could hit his body as well.

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The endeavour of the usage of this emulsion onto one of the A had been possible owing to a novel remedy called Bioidra, that is going to hit the European and Asian market.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

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