PAEDIATRIC DERMATOLOGY

BULLOUS PORPHYRIA MASKED AS EPIDERMOLYSIS BULLOSA ACQUISITA: A RARE CASE REPORT ON PORPHYRIA CUTANEA TARDA

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Background: Porphyria Cutanea Tarda (PCT) is the most common type of heme biosynthesis disorder affecting photo-distributed regions manifested by cutaneous fragility, vesicobullae and atrophic scarring. Here we report a case of PCT in a child who on histopathology and Direct Immunofluorescence studies had findings consistent with Epidermolysis Bullosa Acquisita (EBA) but was eventually diagnosed with PCT on porphyrin studies.

Observation: A 12-year-old Indian female presented with an 8-year history of spontaneous blistering with erosions on the extensor surfaces of her hands bilaterally with associated fragility, photosensitivity, photo-onycholysis and acro-osteolysis. There were areas of reticulate pigmentation with atrophic scarring over the face, pseudo sclerodermatous changes, hypertrichosis and scarring alopecia. She had no family history of PCT but there was a history of consanguinous marriage in her parents. Ocular findings revealed corneal abrasions on slit lamp microscopy and temporal pallor, peripapillary atrophy and tessellated background on Fundoscopy. Wood's lamp examination showed pink fluorescence in the lower set of incisors. Lesional and perilesional biopsy demonstrated subepidermal separation with slouged off epithelium, mixed infiltrate in the papillary dermis with admixed eosinophils and a strongly positive (3+) linear pattern of IgG along the dermo-epidermal junction on Direct Immunofluorescence respectively. These findings were consistent with EBA, but on index of clinical suspicion, High Performance Liquid Chromatography of urine uroporphyrin III was done showing elevated levels. The serum ferritin levels were also raised. A liver ultrasound demonstrated grade I hepatomegaly and Cholelithiasis. Rest of the laboratory parameters were within normal limits. The patient was then commenced on low dose Hydroxychloroquine and advised photo-protection.

Key message: In patients with undifferentiated bullous skin conditions in photo-distributed regions, a diagnosis of PCT should be kept in mind irrespective of histopathological findings and further investigated appropriately.