Porphyria Mimic: A Note of Caution

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Received: 10 May 2021 / Accepted: 13 July 2021 / Published online: 13 August 2021
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To the Editor: A 5-y-old male child was brought for evaluation of liver disease. He was diagnosed with hereditary tyrosinemia-1 (HT-1) at the age of 3 y and started on 2-(2-nitro-4-trifluoromethylbenzoyl)-1,3-cyclohexanedione (NTBC). Since NTBC was expensive and the supply was erratic, he was not compliant with the medication. While being evaluated, he developed generalized seizures, bilateral lower limb paralysis, and systemic hypertension. Since all the features were suggestive of neurologic crisis of HT-1, NTBC was urgently restarted. Metabolic profile sent during the crisis showed elevated urinary succinylacetone and porphyrin levels with absent porphobilinogen (PBG) synthase activity. He recovered over the next 10 d without any residual neurological sequelae. The child underwent liver transplantation (LT) 1 mo later and remains well with stable graft function on 3-y-follow-up.

Neurologic crises are a hallmark of untreated tyrosinemia with high risk of morbidity and mortality [1, 2]. It is often missed and the incidence can be as high as 42% of all untreated patients [2, 3]. Clinical presentation of neurologic crisis can be mistaken as meningismus, opisthotonus, or porphyria due to an interrelationship between tyrosine catabolism and heme biosynthesis. Succinylacetone accumulates due to a deficiency of fumarylacetoacetate hydrolase (FAH) deficiency and inhibits the synthesis of porphobilinogen from δ -aminolevulinic acid (δ -ALA) by inhibiting porphyrin synthetic enzyme δ -ALA dehydratase [1, 2]. δ -ALA accumulation causes the clinical signs of neurologic crises.

In our unit, 8 children underwent LT for HT-1 from 2010 to 2019, all of whom remain well on a median follow-up of 72 mo (16–124 mo). Six (75%) were initiated on NTBC, but opted for LT since they could not afford the medications or could not ensure an adequate supply over time. Though lifesaving and helps avoid LT, NTBC is expensive and difficult to procure and should only be initiated if a constant supply is guaranteed and therapeutic drug level monitoring is possible.

Declarations

Conflict of Interest None.

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