VARIEGATE PORPHYRIA: AN UNUSUAL CASE AND PRESENTATION

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CASE PRESENTATION

Since birth, a 3-year-old female has had recurrent episodes of apnea and severe headaches. Suspected to have congenital central hypoventilation syndrome, the patient underwent genetic testing for mutations in the PHOX2B gene. This testing revealed no abnormalities, which prompted the patient to undergo whole exome sequencing testing.

RESULTS

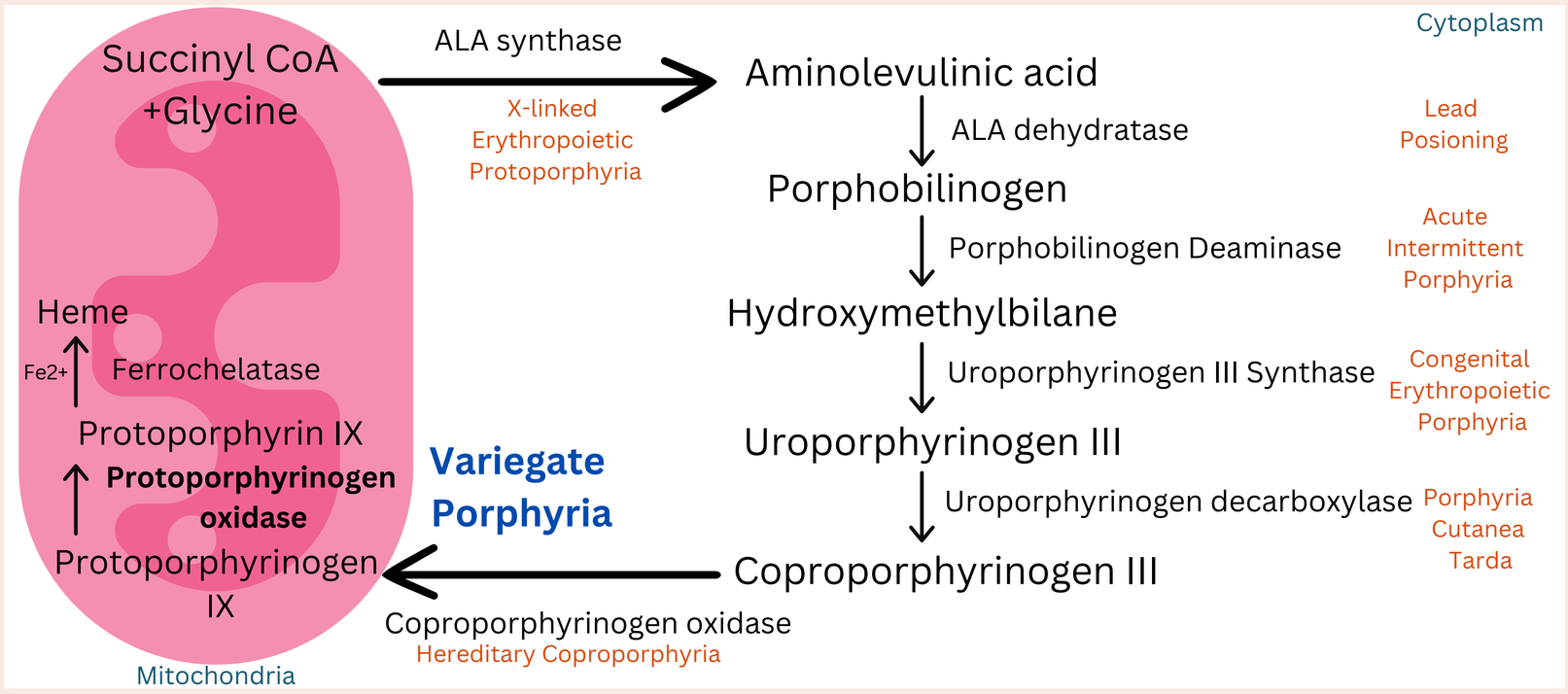
Whole exome sequencing results detected a heterozygous mutation in the PPOX gene c.1287delp (K429NFs*2) which is associated with variegate porphyria. The patient's mother also disclosed that she has experienced episodes of abdominal pain and photosensitivity since puberty.
The patient's genetic testing results prompted evaluation of the mother and other siblings which revealed that 2 of the patient's sisters (ages 7 and 4) and the mother have the same mutation, however the sisters have experienced no symptoms.

DISCUSSION

Common skin manifestations of variegate porphyria include blistering lesions such as subepidermal vesicles, bullae, and erosions on sun-exposed skin, particularly the hands and face. Other chronic skin findings include milia, scarring, and thickening with varying pigmentation levels. The treatment for variegate porphyria includes photo-protection and avoiding triggers. Although variegate porphyria typically presents after puberty with episodes of neurovisceral symptoms and photosensitivies, atypical symptoms may be seen in children including headaches and apnea.

BACKGROUND

Variegate porphyria (VP) is classified as both a cutaneous and an acute porphyria. It can present with chronic blistering cutaneous manifestations and/or acute attacks of neurovisceral manifestations that may become chronic. Chronic blistering photosensitivity, typically on the backs of the hands, is the most common manifestation of VP. The lesions result from sun exposure that activates porphyrins and makes the skin fragile and prone to blister formation. Because sun-induced damage is not acute, the role of sunlight is often not recognized. Cutaneous manifestations may improve in winter and be less prevalent in northern regions and in dark-skinned individuals. VP is characterized by a lack of the protoporphyrinogen oxidase enzyme.



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CONCULSION

Positive genetic testing in a child should prompt further testing of family members. Relatives may be diagnosed years later and may finally have an answer for their symptoms. Variegate porphyria is a disorder that can have a significant impact on quality of life, but if appropriately managed, can lead to improved outcomes.