Correspondence

Rickets and dysmorphic findings in a child with abetalipoproteinemia

To the Editor

I read the interesting case report by Hasosah et al.[1] A pregnant with abetalipoproteinemia (ABLP) could deliver a baby with various dysmorphic features.[2] These features might be attributed to the critical role of fat-soluble vitamins, particularly vitamin A, D, and E, in maintaining the integrity of various aspects of fetal development.[3] These features might be the sole and/or the initial presentation of existing ABLP long time before the full-blown picture of ABLP develops. I wonder whether the mother of the studied baby was a known case of ABLP though I presume that she might be asymptomatic. The context of the study could be further expanded if the authors adopted the required laboratory tests to identify that concern but, unfortunately, that was not carried out. Discriminating mothers of babies with ABLP together with mutational analysis and genetic counseling remain critical to cut short the evolution of new cases of ABLP in the offspring.

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Reply from the Author

It was with great interest that I read Dr. Al-Mendalawi's comments and his insights regarding my recent publication.[1] Dr. Al-Mendalawi's point regarding the mother of the studied baby was a known case of ABLand he suggested mutational analysis and genetic counseling remain critical to cut short the evolution of new cases of ABL in the offspring. I described in the previous study,[1] both rickets and dysmorphic findings in a child with ABLP. Genetic testing to rule out other cause for dysmorphic features included chromosomal analysis and microsomal triglyceride transfer protein (MTP) gene were not carried out in my patient because of the following: a) The family refused these tests. b) The mother had no dysmorphic features and she had good nutrition during pregnancy. c) The patient had 2 sisters and one brother. All of them were normal.

We agreed that maternal ABLP could deliver a baby with various dysmorphic features based on some case reports. Uslu et al[4] described the MTP gene in ABLP, and he observed that some cases could be extremely severe from early postnatal life and poorly responsive to treatment. My case report emphasized that the early diagnosis and initiation of treatment regardless of ABLP in utero or postnatal offered the best chance for improved outcome.

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