Letter to Editor

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Thalassemia Major in a Patient with Hypertriglyceridemia

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Dear editor,

We reported a case titled "A Case Report of Familial Chylomicronemia Syndrome" published in the World J Peri & Neonatol 2020; 3(2): 90-3. Hypercylomicronemia is a rare autosomal recessive metabolic disorder characterized by elevated levels of triglycerides and chylomicrons in the blood and is associated with a high risk of acute pancreatitis and other complications such as cardiovascular disease.[1-3]

On the other hand, the most common inherited hemoglobin disorder around the world is thalassemia.[4] Lifelong red blood cell transfusions and iron chelation therapy are required to prevent complications of β-thalassemia major due to iron overload.[5] It is usually associated with a normal serum lipid profile. However, there are a few reports in literature stating that hypertriglyceridemia has an association with beta-thalassemia major.[6,7]

The case introduced in our article was a patient with hyperchylomicronemia. We first justified her laboratory data and clinical signs according to her hyperchylomicronemia, and treated the patient. The patient follow-up revealed triglyceride in normal range from proper control of chylomicronemia. However, the patient's severe anemia persisted and she required frequent blood transfusions. Considering the patient's condition, we tried to investigate more accurately to determine the cause of the prolonged anemia. Re-examinations and general condition of the patient raised suspicion of thalassemia major. As shown in Table 1, genetic test confirmed a definitive diagnosis of thalassemia major on finding only the thalassemia major gene.

Table 1. Genetic Examination

Major Finding					
Gene	**cDNA**	**Protein**	**Zygos.**	**Signific.**	**Phenotype**
HBB	NM_000518.5 C.92+5G > C	Splice Site	Hom	Pathogenic	Beta thalassemia (AR)
Secondary Finding					
SLC26A4	NM_000441.2 C.1226G > A	p.Arg409His	Het	Pathogenic	Deafness, autosomal recessive 4 with enlarged vestibular aqueduct (AR)
SLC26A3	NM_000111.3 C.782_783del	p.Asp261AlafsTer15	Het	Likely Pathogenic	Diarrhea 1, secretory chloride, congenital (AR)

Moreover, our previous clinical and laboratory examination were suggestive of familial chylomicronemia syndrome. There exists a controversy on the relationship between hypertriglyceridemia and thalassemia major.[8,9] We are not also sure if these two diseases are unrelated or should be noted as a syndrome.

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