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Poster
Parole chiave: delta 16, African origin, thalassemia, alpha globin

Homozygosity for α"3.7" deletion in association with a delta globin variant in a young pregnant migrant.

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HBD:c.49G>C (p.Gly16Arg) (delta 16; rs34012192) is a delta chain variant identified in several populations of African origin and in few unrelated families in Sicily, as further evidence of the genetic admixture between African and Sicilian populations.

All known delta variants are clinically and hematologically silent. However, misdetection of delta chains can lead to understimate the absolute amounts of HbA2. This can then lead to misdiagnosis of β-thalassemia traits.

We report the identification of a heterozygous delta 16 variant as co-inherited with α-thalassemia gene deletion α"3.7" in homozygosis, in an nigerian pregnant woman immigrated in Italy.

The patient showed a slight increase in red blood cells and reduced mean corpuscular volume (MCV 70fL). Ferritin value was in the lower limit of the normal range, which we qualified as due to pregnancy. Identification of Hb fractions was performed using high-performance liquid chromatography with VARIANTII system (Bio-Rad Laboratories, Hercules, CA, USA).

HbA2 was 1.4%, HbF was 0.5%, and we additionally detected an abnormal peak (1.5%) in the S-window (retention time: 4.40 minutes).

Sequence analysis of the whole delta globin gene showed heterozygosity for the delta 16 variant. The α"3.7" deletion in homozygosis was identified by reverse dot blot analysis. No beta globin mutations was detected by direct sequencing of entire gene.

We noticed that the presence of the abnormal delta 16 peak was not readily identifiable using routine laboratory settings. Thus, we call for attention on methodologies, that may be prone to errors in the presence of such occurrences that are easily predicted to become more and more frequent in the future.