Two Calabrian children with Hemoglobin San Diego

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In 2002 Gerli and co-workers reported a 57-year-old woman coming from the South of Italy as the first case of Hemoglobin San Diego in Italy (1). This hemoglobin is characterized by a mutation in the codon 109 of the β-chains (GTG→ATG, valine to methionine) and has an high-oxygen affinity. It was firstly reported in 1974 in a Filipino family by Nute (2).

Thereafter it has been described in subjects of different origin (1-5). All adults with Hemoglobin San Diego reported in literature showed hemoglobin levels (Hb) above 18g% with erythrocytosis and some of them were treated with phlebotomy.

(3-6) Few data are available on children with this hemoglobin (2, 5).

In the Filipino family referred by Nute there were two sisters and their four children resulted heterozygous for the hemoglobin San Diego (Table 1).

Hemoglobin San Diego.

The father and the two uncles had marked erythrocytosis. The children (2M, 12F and 15-year-old respectively) were admitted to our department for a celiac disease familial screening.

During the evaluation of these children it was independently recorded that the paternal grandmother (1), his three sons (one is the father of our patients) were heterozygous for Hemoglobin San Diego.

Than we investigated the hematological parameters of the children. Two of them resulted heterozygous for the hemoglobin San Diego (Table 1).

They showed Hb and Hct over 97°centile (6).

White blood cells and platelets were normal. Iron parameters were normal too.

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The two new patients reported by us here have hematological parameters quite similar to the four of the children published by Nute and to the one referred by Coleman; all the seven children had Hb and Hct at the upper levels of normal ranges.

On the contrary adult affected patients in our family as well as all but one reported in the literature showed evident erythrocytosis (2, 5).

These data agree to the fact that in childhood hemoglobin San Diego may induce a slight increase of hemoglobin levels just up to the normal range, thereafter in adulthood the erythrocytosis becomes evident and patients may be symptomatic.

In conclusion, children with high-oxygen affinity hemoglobin may not have true erythrocytosis but Hb and Hct parameters at upper limit of normality.

Treatment is not necessary but they must be carefully followed for hematological parameters and conditions at risk for vascular accident must be early careful investigated.

On the other hand children with Hb and Hct levels at the upper limit or slight above normal ranges might be evaluated for high affinity-oxygen hemoglobins (7, 8).

References