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THE HISTORY OF BETA-THALASSEMIA IN TURKEY*

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SUMMARY: Aksoy M. (Department of Internal Medicine, Istanbul University Faculty of Medicine at Çapa, Istanbul, Turkey, and Department of Biology, Turkish Technical Research Council (TUBITAK), Gebze, Kocaeli, Turkey) The history of beta-thalassemia in Turkey. Turk J Pediatr 33: 195-197, 1991.

The first two patients with beta-thalassemia major in Turkey, were reported in 1941. However, the importance of beta-thalassemia as a health problem was brought to the attention of physicians only after 1950. In Turkey, the clinical and hematological pictures of beta-thalassemia were found to be mostly heterogenous. Hematological data of patients with mild disease (beta-thalassemia intermedia) severe beta-thalassemia (beta-thalassemia major) and heterozygotes indicate that patients can be grouped according to the content of Hb F, Hb A_2 and red cell indices. *Key words: thalassemia, hemoglobinopathies.*

The thalassemias and the sickle cell syndromes are the most frequent genetic disorders occurring throughout the world. Curiously, enough, the first cases of thalassemia major were diagnosed in 1925 by Cooley and Lee[1] in the United States. As rightfully pointed out by Wintrobe[2] it is difficult to comprehend why there were no cases of thalassemia major reported in Italy and Greece during those years. A possible explanation for this might have been the fact that, at that time, all unexplained hepatosplenomegalies were diagnosed as Banti's syndrome[2]. Even today, although quite uncommon, there are some patients with splenomegaly in which a diagnosis of thalassemia has not been established.

The first two cases of Cooley's anemia in Turkey were simultaneously reported by Prof. S. Tavat[3] and Prof. E. Frank[4] in 1940. The parents of Frank's patient were immigrants from Bulgaria, while the father of Tavat's patient was an immigrant from Yugoslavia. In the period that followed, numerous cases of Cooley's anemia were reported[5-9]. The first "International Symposium on Abnormal Hemoglobins and Thalassemia" sponsored by the Council for the "International Organization of Medical Sciences" and organized by Prof. T.H.J. Jonxis was held in Istanbul. At the symposium, the author of this paper presented a report entitled. "Abnormal Hemoglobins and Thalassemia in Turkey".[10] It was proposed in this presentation that considering the wide variation of clinical and hematological pictures observed in some cases of sickle cell-thalassemia disease, there must be more than one thalassemia determinant that interacts with the sickle cell gene. A short while

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later, Ingram and Stratton[11] reported that the genes for thalassemia were responsible for alpha and beta polypeptide syntheses.

In 1963, the author published the first case of homozygous Hb S and alpha-thalassemia seen in a Turkish family[12].

Incidence in Turkey: The first study to determine the incidence of beta-thalassemia in Turkey was performed by Aksoy and Lehmann[13] in which the paper electrophoresis method was used on blood samples taken from 240 Turks, mostly inhabiting the southern Mediterranean coast of Turkey. The incidence rate was found to be 0.4 percent. This figure is considered today as being too low. The most accurate data was reported by Çavdar and Arcasoy[14], who estimated it as around two percent. The incidence of beta-thalassemia, varying widely in different regions of Turkey, ranges between 0.6 and 10.8 percent. The lowest incidence was found by Kürkçüoğlu and workers[15], in the eastern province of Erzurum, and the highest, 10.8 percent, was found by the author and workers[16] to be amongst Turks who originated from Western Thrace.

Silent beta-thalassemia genes: Even though rarely, some genes for beta-thalassemia in the heterozygous state may not cause hematologic abnormalities such as the characteristic microcytosis. The author has been in favor of this assumption since 1959. In 1968, Schwartz[17] showed that some genes responsible for beta-thalassemia may not cause hematologic abnormalities, which he termed "silent genes" for beta-thalassemia. For a long period in the past, it had been generally thought that there was a large amount of fetal hemoglobin in individuals with Cooley's anemia. But, contrary to this, in 1965 the author reported four cases of Cooley's anemia with low levels of fetal hemoglobin ranging between 2 to 10 percent[18].

In addition, in 1978, the author classified 20 patients with beta-thalassemia intermedia according to the results of genetic studies[19] as follows:

1– Beta-thalassemia intermedia homozygous for beta-thalassemia with large amounts of hemoglobins A_2 and F.

2– Beta-thalassemia intermedia homozygous for beta-thalassemia with normal levels of hemoglobins A_2 and F.

3– Beta-thalassemia ihtermedia heterozygous for both beta-thalassemia with large amounts of hemoglobins A_2 and F and beta-thalassemia with normal Hb A_2 and F.

4– Beta-thalassemia intermedia heterozygous for both beta-thalassemia with a large amount of hemoglobin A_2 and "silent" beta-thalassemia.

There are two types of beta-thalassemia intermedia: One is associated with large amounts of hemoglobin F, while in the other, hemoglobin F is only slightly increased.

In recent years, further investigations using new methods have revealed the presence of "silent" abnormal hemoglobins such as "City of Hope" or "Hb Knossos" or the presence of rare haplotypes in some patients with beta-thalassemia intermedia[20, 21].

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