
ORIGINAL ARTICLE

Thalassemia and Its Problems in the Adolescent Age

by

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Abstract

Due to better facilities and management in facing thalassemic children it is expected that there will be more surviving patients reaching the adult age. But on the other hand there appears many new problems, such as physical, emotional and social problems. Forty cases of thalassemia major and thalassemia Hb E disease who reached the adult age were discussed in this paper. Face deformity, growth disturbance and skin discoloration were seen in 82,5%, 95% and 85% of the cases respectively.

Inferiority complexes and physical disability were found in about 50% of them. Frequent mild/moderate infections were seen in 25% of the patients. No one suffered from severe or overwhelming infections. Twenty seven (27) out of 40 cases had been splenectomized at the age of more than 5 years. Only 4 cases (10%) showed signs of puberty. The growth disturbance might be due to 2 factors a) failure in maintaining the hemoglobin content at about 10 g%, and b) disturbance of gonadotropin hormone due to iron overload. It is therefore highly recommended to put the child at high transfusion regime as early as possible and administer to the child regularly iron chelating agents to prevent the iron overload. Every infection should be promptly eradicated especially in those whose spleen have been removed.

Depression which is common among them should be overcome by giving equal opportunities in educations, vocational training and employment. They should be helped to become integrated into adult society.

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Introduction

Thalassemia is a long standing hereditary hemolytic anemia which causes many hazardous alterations of body organs of the affected children. Severe anemia, failure to thrive, hepatosplenomegaly and facies Cooley are common features seen in young children under 5 years of age. In our observations most of the children died before they reached the age of 10 years. But due to the improvement of the facilities in the Hospital and Blood Bank Services many of the patients reach the adult age.

From one point of view we might be glad of this result but on the other hand there arises many new problems. These problems are mainly based on alterations of physical appearance and disability to do normal activities for their age. The problems of thalassemia are now becoming more and more serious public health problems in the area where the genes are widespread encountered. Especially after the infant mortality rates fall due to the progress in controlling malnutrition and infection, this genetic disease will pose a major challenge.

Material and Methods

Our material consisted of 40 cases, their age varied between 10 and 20 years. Twenty of them were girls and the other 20 children were boys.

Twenty one out of 40 cases were thalassemia major while the rest (19 cases) were thalassemia Hb E disease. The bo-

dy weight and height were measured by routine procedures. Psychological approach were done through intensive anamnesis of these children. Bone surveys were made on all of the patients. The diagnosis of thalassemia were confirmed by alkaline denaturation tests (Betke, 1959) and electrophoresis (starch-block and cellulose-acetate), besides the routine blood examinations. The follow-up of these patients lasted from 9-18 years.

The immunoglobulins level was examined by using immunodiffusion plate from Behring Institute, Germany.

Results and discussion

Table 1 shows that face deformity, linear growth disturbance, skin discoloration, inferiority complexes and physical disability were found in more than 50% of the cases.

Especially the first three symptoms mentioned above comprised more than 80% of the subjects. Infections were seen more in the thalassemia major than that in thalassemia Hb E disease group.

Cephalofacial deformity (CFD)

In 1964, Johnston and Krogman stated that cephalofacial manifestations of orthodontic significance seem to be concentrated mainly in maxillary alveolar bone and in the plate as well. Due to marrow hyperplasia and internal swelling of the temporal and paranasal bo-

TABLE 1: Some clinical data of the 40 cases with thalassemia syndromes

	Thal. major	Thal. Hb E disease	Total
No. cases	21	19	40
Sex	♀	9	20
	♂	10	20
Age range/year	10 — 18	10 — 20	
Face deformity	18 (85.7%)	15 (78.9%)	33 (82.5%)
Linear growth disturbance	21 (100%)	17 (89.5%)	38 (95.0%)
Skin discoloration	19 (90.5%)	15 (78.9%)	34 (85.0%)
Frequent infections	7 (33.3%)	3 (15.8%)	10 (25%)
Inferiority complex	7 (33.3%)	13 (68.4%)	20 (50%)
Physical disability	10 (47.6%)	10 (52.6%)	20 (50%)

nes, the air spaces in the temporal bones and in the paranasal sinuses are encroached on and sometimes obliterated. Swelling of the zygomas makes the cheek bones stand out and responsible for the mongoloid facies which is characteristic for the severe cases (Caffey, 1957, see Fig. 1). Failure of pneumatization of the maxillary sinuses at the usual time and considerable overgrowth of the maxillary overbite, prominence of the upper incisor teeth and separation of the or-

bits, give these patients the characteristic "rodent facies" (Baker, 1964). Logothetis et al. (1971) found that 94 out of 138 cases with thalassemia major (68.1%), presented typical cephalofacial deformities.

In this present paper 18 out of 21 cases (85.7%) with thalassemia major showed cephalofacial deformities. In the group of thalassemia Hb E disease the CFD was found in 15 out of 19 children (78.9%). This difference was due to the

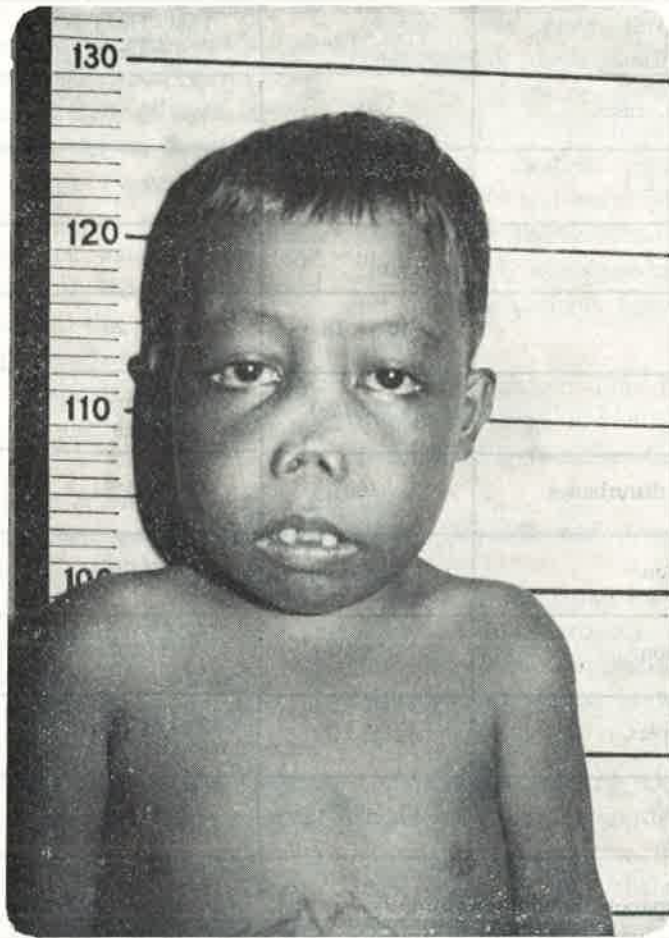


FIG 1: Typical facies Cooley of a boy 17 years of age

fact that the clinical manifestations in the latter group were not as severe as in thalassemia major. Comparing to the finding of Logothetis et al. (1971) the percentage of CFD in this study is very high. The authors are of the opinion that the high incidence of CFD in the present material were due to the inability to

keep the patients fully on the high regime transfusion at the early stage of the disease. The severity of the illness and the delay of getting transfusions, as well as the failure to maintain high hemoglobin level are important factors in developing these abnormalities.

Linear growth disturbance

Previous studies indicated that patients with Cooley's anemia (homozygous β -thalassemia) tend to fall below normal standards both for height and weight (Smith et al., 1960; Wolman, 1964; Wolman and Ortolani, 1969; Zaino et al., 1969). This growth disturbance seems to be the result of subnormal rates in growth for height, weight and skeletal maturations (Johnston and Krogman, 1964; Johnston et al., 1966).

The study of Logothetis et al. (1972) on 138 cases with thalassemia major revealed that all children above the age of 4 years showed the tendency for both height and weight to fall behind the normal average. By the age of 10 their children had an average height of one standard deviation below the mean height of the normals. Fig. 2 and 3 show that most of the patients had their weight and height below 10% tile from normal (Djumadias et al., 1966). In the thalassemia major group this growth disturbance was seen in all of the 21 cases (100%). The severity of the disease which is generally parallel to the severity of the anemia play an important role in growth retardation.

Skin discoloration

In his observation Logothetis et al. (1972) found that discoloration of the

skin secondary to hemosiderosis was noted in over half of their patients. In our study 34 out of 40 cases (85,0%) showed this picture.

Four cases with thalassemia Hb E disease and 2 cases with thalassemia major did not exhibit the discoloration of the skin. Most of the patients were unable to afford desferioxamine-B; those who could buy it were not properly treated due to their irregular visits to the Outpatient Department. It is really too expensive for our patients to have proper treatment with this iron chelating agent. This skin discoloration causes also psychological problems to the patients.

Frequent infections

It is very often that splenectomy cannot be avoided in facing thalassemic children. The indications for this procedure are based on the effect of hypersplenism in increasing the blood requirement and hence the rate of iron loading, which is the main threat to life in thalassemia. The bad general condition and the alteration of the function of the reticulo-endothelial system due to iron deposit, decrease the resistancy of the children against infections (Ellis et al., 1954); the high serum iron may be utilized by the bacteria for their multiplication in the body (Weinberg, 1974).

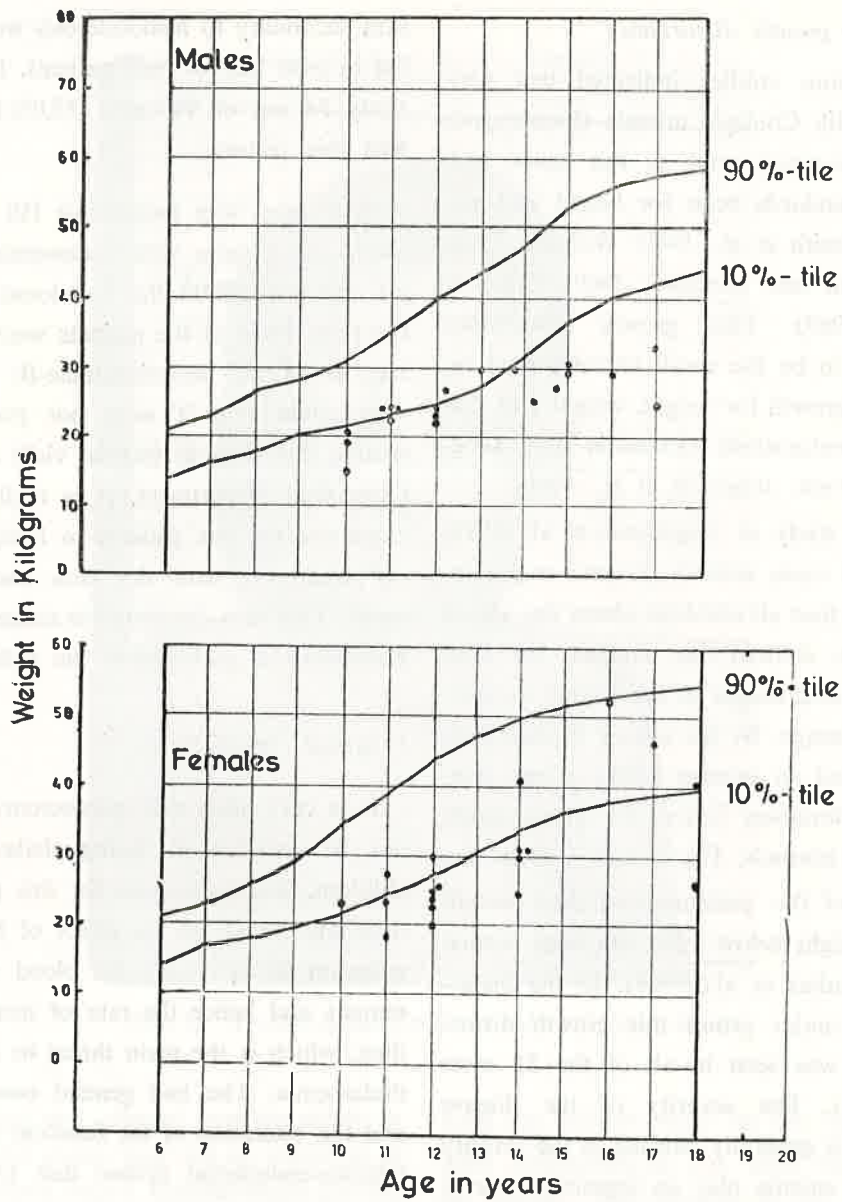


FIG 2: Distribution of weight measurements in 40 patients with thalassemia

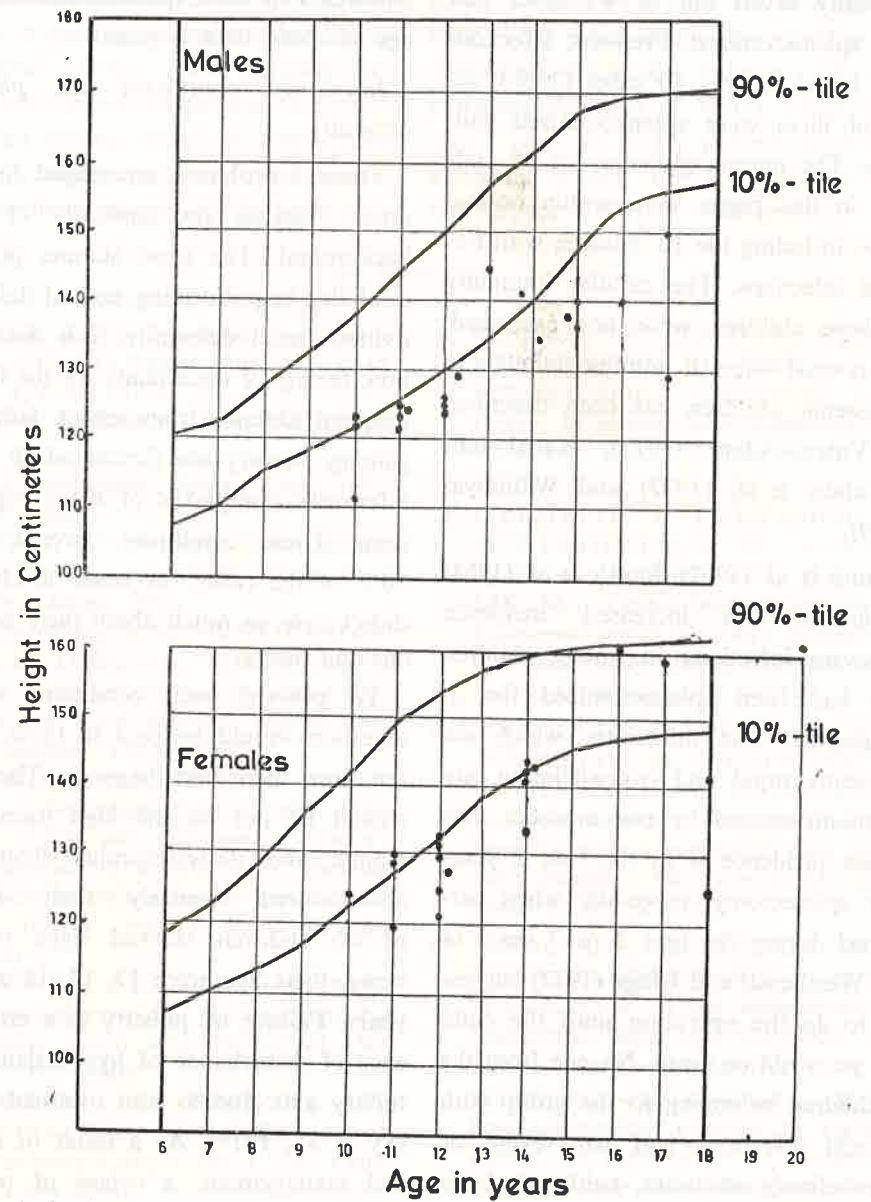


FIG 3: Distribution of height measurements in 40 patients with thalassemia

Twenty seven out of 40 cases had been splenectomized. Frequent infections were found in only 10 cases (25%); seven of them were splenectomized children. The immunoglobulins of 34 children in this paper were within normal limits, including the 10 children with frequent infections. The cellular immunity of those children were not examined. The normal value of immunoglobulins in thalassemic children has been described by Valassi-Adam (1973), Kanakoudi-Tsakalidis et al. (1977) and Wahidiyat (1979).

Smith et al. (1962), Smith et al. (1964) emphasised the increased incidence of severe infections in young children who had been splenectomized for β -thalassemia. The infections, which are frequently rapid and overwhelming, are commonly caused by pneumococci. The highest incidence is in the first 2 years after splenectomy, especially when performed during the first 2 or 3 years of life. Weatherall and Clegg (1972) suggested to do the operation until the child is 5 years old or more. No one from the 10 children belonging to the group with frequent infections had had severe or overwhelming infections, neither had the other 17 splenectomized cases. The absence of severe and overwhelming infections of the cases presented in this paper may be due to the fact that those

children had been splenectomized at the age of more than 5 years.

Inferiority complexes and physical disability

These 2 problems are related to each other, because the similarity of their background. The short stature, physical disability in performing normal daily activities, facial deformity, skin discoloration, feeling of uncertainty for the future, frequent absence from school, failure in gaining puberty are factors which cause inferiority complexes of those big children. These conditions covered about 50% of the cases, the other 20 children didn't care so much about their conditions and future.

To prevent such conditions special attention should be paid to those children since their first diagnosis. The child should be put on the high transfusion regime and desferioxamine should be administered regularly. Only 4 out of 40 children showed signs of puberty, their age were 17, 17, 16 and 20 years. Failure of puberty is a consequence of disturbance of hypothalamic/pituitary axis due to iron overload (Kletsky et al., 1979). As a result of improved management, a cohort of patients more than 10 years old is now appearing in the community where thalassemia is common. They have special physical, emotional and social problems. Many of

them became depressive. They had difficulties in gaining acceptance for higher educational courses. These patients should be encouraged to become integrated into adult society. Many of them failed to pass through puberty and need hormone replacement therapy. Orthodontic sur-

gery may help to correct the face deformity. They need equal opportunities in educations, vocational training and employment (Bulletin of WHO, 1982). Except 2 cases all of the patients were still attending school; the 2 exceptions (18 and 20 years of age) were jobless.

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