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A GOITROUS NEONATE OF A HYPOTHYROIDAL FAMILY LIVING IN NORTHERN TURKEY WHERE THIS CONDITION IS ENDEMIC*

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The advent of radioimmunoassay to measure thyroid hormones and the application of this technique in screening for congenital hypothyroidism has greatly facilitated the early diagnosis and prompt treatment of this disease in developed countries[1]. We believe that a screening program is essential in areas like the eastern Black Sea region of Turkey where hypothyroidism is endemic. We have tried to emphasize the importance of this problem through this case report.

Case Report

A.A., a ten-day-old girl infant, was admitted to the Karadeniz University Children's Hospital, the chief complaints being dyspnea and a mass in the neck. The mass had been there since birth and her breathing was dyspneic. The child had been born at home, at a gestational age of 40 weeks, to a 28 year-old mother. The family was living in a town near Trabzon, which is on the Black Sea and towards the east of Turkey. Family history revealed that the parents and the sibling had had goiter for a long time; the mother had sought no medication during pregnancy.

On physical examination the patient appeared cyanotic, the voice was hoarse and there was a stridor. The pulse rate was 180/min, temperature was 37° C, the breath was tachypneic (60/min). The weight was 4800 g, height was 53 cm, head circumference was 36 cm; the face was edematous and there was a large diffuse goiter in the neck.

Laboratory findings were as follows: Hb was 17 g/dl, WBC was 10600/mm^3 with lymphocyte predominance. Serum T_4 was 1.2 μg/dl, T_3 was 1 ng/ml, TSH was 50 μU/ml. The mother's serum T_4 was 1.3 μg/dl, T_3 was 1 ng/ml, TSH was

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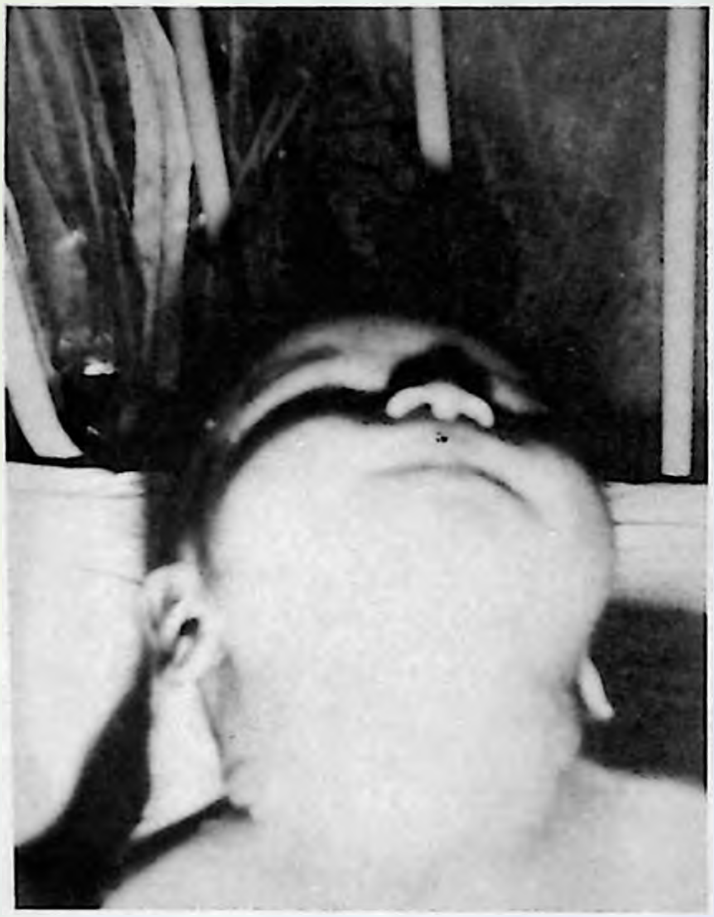

Figure 1a.
Patient.

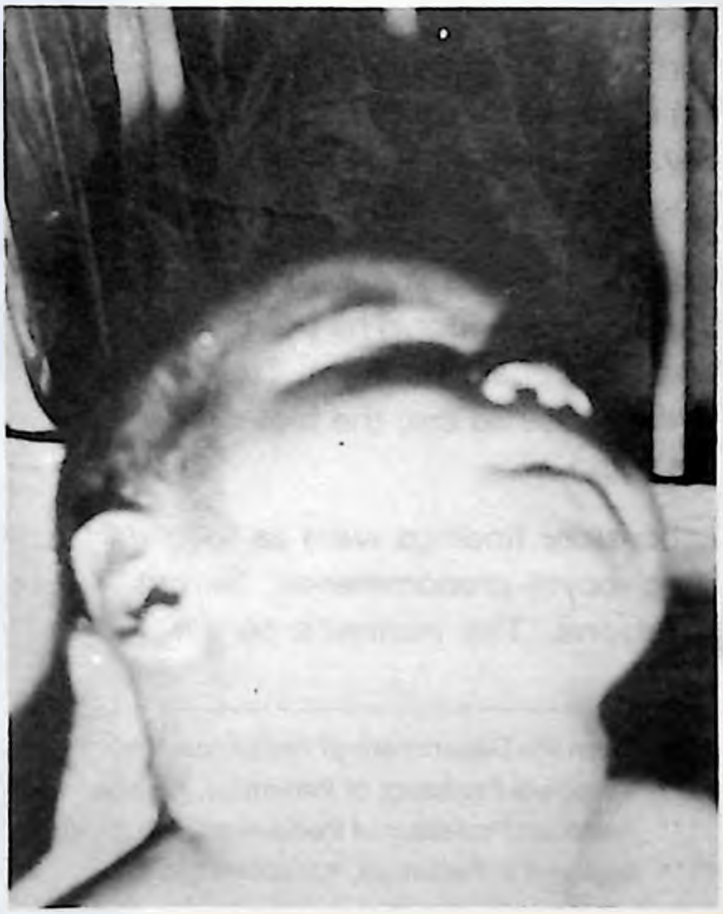

Figure 1b.
Patient.

Figure 2.
Patient's mother.

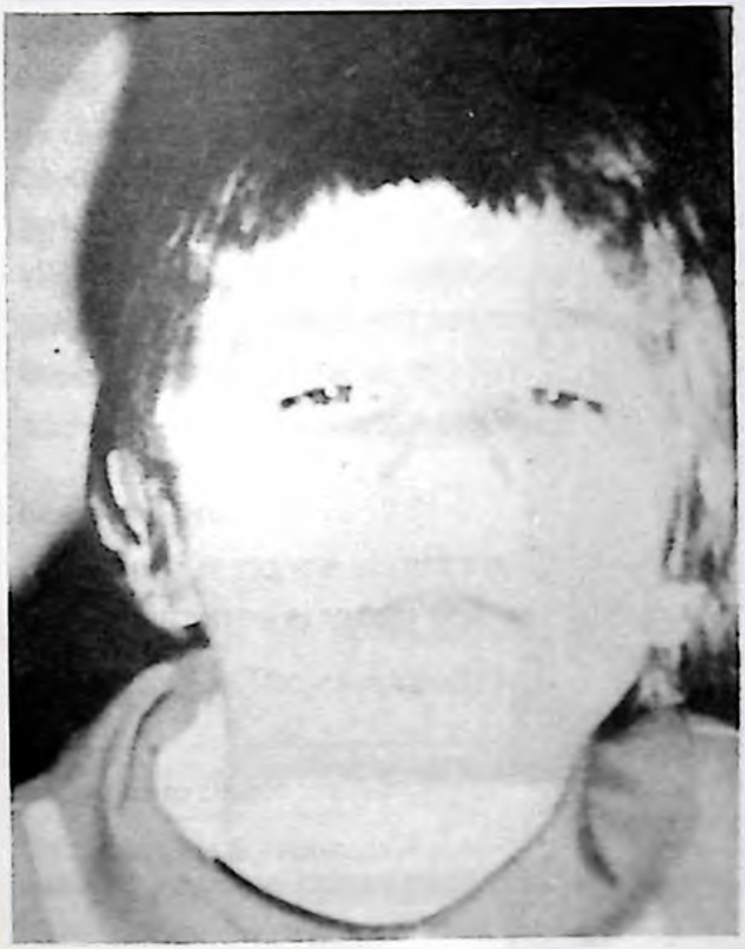

Figure 3.
Patient's brother.

15 μU/ml. There was also a diffuse goiter, constipation and hypothermia. The patient's sister was a seven-year-old girl with a diffuse goiter. Her serum T_4 was 4 μg/dl, T_3 was 1.4 ng/ml, TSH was 10 μU/ml.

The patient was put in an incubator and L-Thyroxin, 5 μg/kg, was begun immediately. On the seventh day of treatment the facial edema had disappeared, the goiter was reduced in size and she was sent home on L-Thyroxin therapy. After a month her serum T_4 was 9.8 μg/dl, T_3 was 1.75 ng/ml. The other members of the family were treated for goiter and given L-Thyroxin at a rate of 200 μg/day. At follow-up examinations their conditions were seen to have improved.

Discussion

Hypothyroidism is among the most common of endocrine disorders in childhood. Few disorders have such a drastic effect on growth and development as untreated congenital hypothyroidism. A lack of the thyroid hormone causes mental retardation but this can in large measure be prevented by prompt treatment. According to the first five screening programs to be established (Quebec, Pittsburgh, Toronto, Oregon Regional and New England Regional) the overall incidence of congenital hypothyroidism has increased from 1/6000 to 1/3684 live births[1]. The cost of screening varies from 0.7 dollars to 1.6 dollars per infant depending on which costs are included in the estimate. The cost benefit analysis of the screening program showed that the cost/benefit ratio is 1:8.9. In other words society realizes a return of 8.9 dollars for each dollar spent on the control of this disease[2].

In Turkey screening programs for congenital hypothyroidism have not yet been introduced. We think that they are especially vital for those parts of Turkey such as the eastern Black Sea region where the condition is endemic. The family in this case is a good example of the effects of iodine deficiency on both the mother and her offspring. In this case the patients were lucky for the condition was diagnosed early and treated promptly. It is also vital to iodize this region for the prevention of goiter and also to give appropriate medical treatment to people who have goiter and especially to goitrous pregnant women and their offspring.

Summary

A goitrous hypothyroidic newborn baby and her goitrous family are presented. The importance of thyroid screening programs in regions where goiter is endemic is discussed and also iodination and medical treatment.

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