Endemic cretinism in Sicily

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ABSTRACT. This work reports the presence of endemic cretinism in a small district located inside an endemic goiter area in north-eastern Sicily, personally described. The study covers 19 mental defectives (11 females and 8 males, mean age 35.8 ± 15.5 yr) selected on the basis of severe mental retardation recognized by the local doctors. No systematic survey for cretinism was carried out in the total population. Marked mental retardation was evident in all subjects. Nine of them exhibited clinical and biochemical signs of hypothyroidism (myxedematous cretins). The 10 others were clinically euthyroid and had deaf-mutism and/or pyramidal tract dysfunction (neurological cretinism). Familial aggregation of cretinism was also observed. In both myxedematous and neurological cretins the urinary iodine excretion was very low, but not significantly different from that recorded in the euthyroid controls of the same area. The data available do not clarify the pathogenesis of endemic cretinism in Sicily. However, the marked height retardation, the observation of delayed bone maturation and the severity of mental deficiency suggest that thyroid failure was present in early life. The presence of endemic cretinism today in Sicily constitutes a strong argument in favour of the immediate introduction of adequate iodine prophylaxis.

INTRODUCTION

Endemic goiter is one of the most common nutritional diseases in man. Extensively reported in Europe at the term of the last century, the disease is presently considered as a vanishing problem in this continent thanks to the improvement of the nutritional and socioeconomic situations, while it remains one of the major public health problems in developing countries (1). However, endemic goiter is still common in many places in Italy, not only in the mountain belts (Alpes and Apeninnes) but also spread all over the peninsula and the main islands (2-6).

The most important complication of endemic goiter is endemic cretinism. In the absence of precise knowledge concerning etiology, pathogenesis and pathology, the condition is defined on a pure clinical basis. The term endemic cretinism refers to an individual, born and living in an endemic goiter area who also exhibits obvious mental retardation associated with either stunted growth and signs of thyroid failure (myxedematous endemic cretinism) or neuromuscular disorders and abnormalities in hearing and speech which can lead to deaf-mutism (neurological endemic cretinism) (7). These 2 types of cretinism constitute 2 extremes, since cretinism really is a continuum from the myxedematous to the neurological form (8). The presence of endemic cretinism in endemic goiter area constitutes a criterion of major severity.

We have recently described so far unrecognized areas of endemic goiter in north eastern Sicily (5). The unusual feature in this area is that it is less than 100 km from the Mediterranean Sea and that the goiter may result either from severe iodine deficiency or from the combined effect of a mild iodine deficiency and an overload of thiocyanate (5, 9), a naturally occurring goitrogen, whose role in the etiology of endemic goiter has been evidenced (8). The aim of this work is to report the presence of endemic cretinism in a small district located inside this endemic area. This observation, independent of its scientific interest, has important public health implications. It constitutes a strong argument in favour of the immediate introduction of adequate iodine prophylaxis.

MATERIALS AND METHODS

Area studied

The present study was conducted in the severe goiter endemia in the highlands of northeastern Sicily. This zone lies between the provinces of Catania and Messina on the northwest side of the Mount Etna. The area studied is shown in Figure 1. The subjects reported in this study were observed in
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Fig. 1 - Map of Sicily (scale 1:1,000,000). The shaded area indicates the endemic goiter area in northeastern Sicily.

The village of Maniace and the small town of Tortorici. Maniace (3,000 inhabitants, mainly living in country farms) is close to the town of Bronte (760 m above sea level; 21,600 inhabitants), while Tortorici (11,500 inhabitants) lies more to the north in the province of Messina, on the Nebrodi Mountains (475 m above sea level). It is of interest that people living in the Maniac area originally came from Bronte or Tortorici.

The way of life in this zone is largely agricultural and pastoral, but small factories and mechanical workshops are also present in the area. Connections with the main cities (Catania and Messina) are easy by car, local trains and buses.

Epidemiological survey of goiter

The prevalence of goiter was determined in all school children in Maniac: (217; 101 boys and 116 girls, mean age 7.5 ± 2.3, range 5-14 yr) and in Tortorici (1376; 651 boys, 795 girls, mean age 10.7 ± 6.0, range 5-18) as well as in 2 groups of randomly selected clinically euthyroid adults used as controls, 57 in Maniace (12 males and 45 females, mean age 37.3 ± 16.1 yr, range 14-68) and 180 in Catania, (50 males, 130 females, mean age 35 ± 4, range 16-65 yr). The volume of the thyroid and its nodularity were recorded according to the technique of Perez et al. (10) as modified by Querido et al. (7). Consequently, goiter was recognized as endemic when the prevalence of goiter in schoolchildren was higher than 20%.

Mental defectives studied

The study covered 19 mental defectives, 15 in Maniac and 4 in Tortorici. Eleven were females and 8 males, their ages ranging from 15 to 78 yr (mean: 35.8 ± 15.5). They were selected on the basis of severe mental retardation recognized by the local population and the local doctors. According to these sources, there were no other severe mentally retarded individuals among the 3,000 inhabitants of Maniac, but no information was available for Tortorici. In both places no systematic survey of the total population was performed in order to determine the prevalence of endemic cretinism.

Methods

The clinical examination of the patients included height measurement, goiter scoring, evaluation of clinical symptoms of thyroid failure and in 16 cases, neurological examination. A psychological examination was performed in 4 subjects using the Raven Progressive Coloured Matrices test. Finally, bone maturation was assessed in patients by means of X-rays of hands and wrists, and the degree of bone maturation was established according to the criteria of Greulich and Pyle (11). Previously described methods were used to determine the serum levels of TSH (12), T4 (13), T3 (14), reverse T3 (15), Thyroglobulin (16) and anti-thyroglobulin antibodies (17). In addition, the urinary excretion of iodine was measured (18). Since only casual samples of urines were available, the results are expressed by the urinary concentration of iodine. The iodine/creatinine ratio was not used because in 80 euthyroid controls in Catania, this ratio was 73.9 ± 4.7 µg/g (m ± SE). The coefficient of variation of this ratio (58%) was not lower than the one of the iodine urinary concentration (52.8%) (Bourdoux, unpublished).

RESULTS

i) Prevalence of goiter and urinary excretion of iodine

Table 1 shows the results obtained for the epidemiological surveys conducted in Tortorici and in Maniac as well for previous epidemiological studies conducted in schoolchildren in Catania and Bronte and for the urinary iodine concentration and excretion in these 4 places. The prevalence of goiter increased sharply from 2.2% in Catania to 66.8% in Maniac, while the iodine excretion decreased from 113.8 µg/day in the former to 18.9 µg/day in the latter. In spite of a distance of only 8 km between Maniac and Bronte the iodine excretion in Maniac was less than one half that in Bronte.

ii) Clinical and radiological data in the defectives.

Marked mental retardation was evident in all subjects. However, its degree varied considerably from one subject to another, but all of them understood simple orders and participated to some extent in family activities. An important defect of attention span and a discontinuous psychoperceptive activity was recorded in all cr(tins; answers to most stimuli were both delayed and inadequate. In verbal and/or visual stimulation tests, the phenomenon of interference was frequently observed wherein the response was referred not to the immediate stimulus, but to the preceding one. Perception was syncretic and very rudimentary. It was strongly reflected in the inability to analyze facts and to abstract (cases nos. 4, 8, Tables 2 and 1, 6 Table 3).