

Persistence of Mediterranean anaemia in Sicily

Maria Antonietta Romeo, Felicia Di Gregorio, Giuseppe Russo

Abstract

We report 40 cases of homozygous β thalassaemia, aged between 3 and 24 months, who were observed between January 1990 and June 1996 at the Thalassaemia Centre, Paediatric Department, Catania University. A questionnaire was used to find out the parents' knowledge of their risk before the birth of the affected children and showed that the persistence of Mediterranean anaemia in Sicily was mainly because of the following reasons: (1) poor information (62.5%), (2) laboratory error (12.5%), (3) difficulty in the differential diagnosis of β thalassaemia trait (10%), and (4) not performing prenatal testing or selective abortion of an affected fetus (15%).

We conclude that improved preventive measures at various medical and social levels can remove risk factors and so further reduce the incidence of Mediterranean anaemia in Sicily.

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Keywords: Mediterranean anaemia; β thalassaemia; Sicily

Clinica Pediatrica
Università, University
of Catania, Viale
Andrea Doria 6, 95125
Catania, Italy
 M A Romeo
 F Di Gregorio
 G Russo

Correspondence to:
 Dr Romeo.

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In the last few years the improvement in health education as a result of premarriage information from Family Health Services and doctors, and genetic counselling to couples at risk from gynaecologists and paediatricians, together with the possibility of prenatal diagnosis, has prevented the birth of more than 50% of the 47 or so yearly expected cases of Mediterranean anaemia in the Sicilian population.¹ According to Maggio *et al*,² the incidence of Cooley's anaemia has stabilised since 1985

because of the heterogeneous distribution of the β thalassaemia gene and the reduced efficacy of the screening programmes in peripheral centres.

Between January 1990 and June 1996, we observed 40 cases of homozygous β thalassaemia, aged between 3 and 24 months, at the Thalassaemia Centre, Paediatric Department, Catania University. The distribution of our patients per birth year is shown in fig 1. The sample represents a large proportion of the homozygous β thalassaemia children born in east Sicily in this period, because about 75% of thalassaemic subjects in this area are diagnosed in our department. However, the exact number of affected newborns per year is difficult to estimate, because the last regional report was in 1990.^{1,2}

A questionnaire was used to find out the parents' knowledge of their risk before the birth of the affected children and showed that the persistence of Mediterranean anaemia in Sicily was mainly because of the following reasons.

(1) Even today poor information is the main cause of failure in prevention programmes. In fact, 25 (62.5%) parent couples did not know about thalassaemia or related problems before conceiving the child. Excluding two couples who had immigrated from countries with a high incidence of β thalassaemia (Albania and Mauritius), the couples in this group lived in peripheral towns and had received genetic counselling only in the last months of pregnancy or after the birth of the affected child. As a result of retrospective information, six of these couples had prenatal tests performed during subsequent pregnancies.

(2) In five cases (12.5%) the birth of a child with thalassaemia major was the result of laboratory error. In fact, the β thalassaemia trait had been diagnosed in only one parent before conception of the child. Laboratory investigations carried out in our department after the birth of the affected child showed typical β thalassaemia trait with microcytosis and high Hb A₂ levels in both parents. This finding suggests the need to carry out screening for thalassaemia and haemoglobinopathies only in experienced laboratories.

(3) The parents of four other children (10%) had also undergone screening for β thalassaemia before pregnancy. However, the heterozygous state had been detected in one and ruled out in the other parent, who had mild microcytosis and normal or borderline Hb A₂ levels. These mild cases were interpreted as α thalassaemia carriers, but postnatal DNA analysis of the β gene showed the presence of the IVSI-nt6 mutation, associated with α thalassaemia trait in two cases and with mild

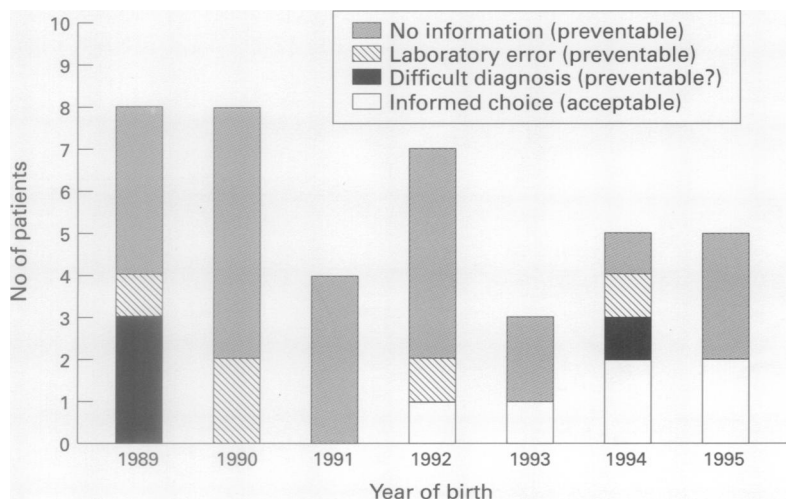


Figure 1 Distribution of the thalassaemic patients per birth year and reasons for the persistence of Mediterranean anaemia in Sicily.

Table 1 Preventive measures to reduce the incidence of Mediterranean anaemia

Risk factors	Measures to remove risk
No or poor information	Intensive educational programmes aimed at all Sicilian people to inform about thalassaemia and detect couples at risk
Non-detection of typical β thalassaemia carrier	Screening in experienced laboratory
Non-detection of atypical β thalassaemia carrier	Special laboratory investigations

iron deficiency in another. Atypical forms linked to mild β thalassaemia gene expression are frequent in the Sicilian population and can hinder differential diagnosis of β thalassaemia trait. Moreover, the identification of β thalassaemia carriers can prove more difficult because of the interaction of genetic or acquired conditions, such as α thalassaemia, δ thalassaemia, or iron deficiency anaemia, capable of reducing Hb A₂ to normal or borderline levels.³ Therefore, when a carrier's partner shows some haematological abnormality the couple should be referred to a specialist laboratory⁴ for additional diagnostic investigations, such as determination of α /non- α ratio by globin chain synthesis in vitro or DNA analysis of the β gene.

(4) Six couples (15%) knew their risk before beginning pregnancy. Three of them did not request prenatal diagnosis while the other three couples chose to continue the pregnancy of the homozygous fetus diagnosed by prenatal test-

ing. These reproductive choices of informed couples (one of them had already had a child with thalassaemia major) are obviously comprehensible and justified when genetic counselling is, as it must be, non-coercive.

We conclude that improved preventive measures at various medical and social levels can remove the risk factors reported in table 1 and so further reduce the incidence of Mediterranean anaemia in Sicily, as has occurred in other Italian regions.⁵

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