A screening programme for the prospective prevention of Mediterranean anaemia in Latium: results of seven years' work

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SUMMARY Since 1975 the Rome Microcythaemia Centre has carried out every year, under the auspices of the health authorities of the Latium region, a screening of thalassaemias among intermediate schoolchildren of Latium. From these campaigns, knowledge about thalassaemias among the young adult population has grown which, in its turn, has resulted in screening of these young adults.

Through screening in schools between 1975 and 1982, of 289 763 students examined, 6838 thalassaemias were identified, 6045 of whom were $\alpha$- or $\delta\alpha$-thalassaemias. The total number of young thalassaemias who are identified at present in the Centre through screenings of schoolchildren and young adults is about 3300 per year. Furthermore, from January 1980 to April 1983, 110 prospective couples of child-bearing age at risk (94 of whom originated from Latium) were identified at the Centre, and five homozygous fetuses (three of which originated from Latium) were diagnosed.

These data derive from an area in which the frequency of thalassaemia is only 2.4%, and they show that the programme in Latium for the prevention of Mediterranean anaemia has been successful.

The screening of heterozygous thalassaemia carriers among intermediate school students (13 to 14 years old) started in 1975 in Latium, was continued under the auspices of the regional health authorities, and is now on a very large scale. During the last few years further publicity in the adult population, together with the schools screening, stimulated the carrying out of a second screening of young adults which has also now reached large dimensions.

Following our two previous papers published in this Journal,1,2 we present all the results of the first seven years' work.

Methods

SCREENING IN SCHOOLS

The screening and laboratory methods for the diagnosis of thalassaemia have remained the same as in the first years.1,2 They are shown in table 1 and fig 1.

APPROACHES TO THE YOUNG ADULT POPULATION

In 1983 the Centre started a new approach to the young adult population using for publicity the Local Health Units, resulting from the recent reform of the health services, and marriage registry offices. Information is disseminated through direct approaches to the public and free distribution of pamphlets and booklets. Subjects requesting examination for thalassaemia as a consequence of this information are studied with the full diagnostic procedure (steps 1 and 2, fig 1) in the Centre.

<table>
<thead>
<tr>
<th>TABLE 1</th>
<th>Obligatory stages in the schools screening.</th>
</tr>
</thead>
<tbody>
<tr>
<td>(1) Educational programme in the schools.</td>
<td></td>
</tr>
<tr>
<td>(2) Collecting of blood samples from the students and analyses in the laboratories.</td>
<td></td>
</tr>
<tr>
<td>(3) Communication of the results to thalassaemic and non-thalassaemic students. Examination of thalassaemic families and genetic counselling.</td>
<td></td>
</tr>
</tbody>
</table>
Results

The schools screening attained its present state from
the fourth year onwards. After the initial explo-
lations, the internal and mountainous areas of Rieti,
Rome, and Frosinone provinces (where the fre-
cuency of thalassaemia is about 1 %) were excluded
from the screening. However, the remaining areas
of Latium which contained the most affected were fully
screened, and the highest rates of information and
examination of students were those of the province
of Latina (table 2), where the incidence of thal-
assaemia (2.80%) is the highest of the Latium
provinces (table 3, fig 2).

In the course of the first seven campaigns 289 763
students were examined, and 6838 thalassaemias
identified, 6045 of whom are β- or δβ-thalassaemia

The screening programme was favourably accepted
by the population from its inception. Thalassaemic
families agreed to be examined in the Centre
without any prejudice or anxiety.

After only 2 to 3 years from the start of the
schools screening, and therefore before the screening
had been extended to young adults, the whole
population quickly became aware of the thalassaemia
programme due to the effect of the schools
screenings.

This publicity, in turn, produced a rapid rise in
the number of people willing to be examined in the
Centre (fig 3), which has increased from a few a day
in 1976 to 70 to 80 in 1982. Thus, a second screening
of the young adult population of child-bearing age
started, which includes at present nearly 15 000

### TABLE 2 Percentage of schoolchildren examined and in the last four mass screenings in the provinces of Latium.

<table>
<thead>
<tr>
<th>Province</th>
<th>No informed</th>
<th>No examined</th>
<th>No enrolled</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rome</td>
<td>90.5%</td>
<td>78.7%</td>
<td>71.3%</td>
</tr>
<tr>
<td>Frosinone</td>
<td>58.5%</td>
<td>82.7%</td>
<td>48.4%</td>
</tr>
<tr>
<td>Latina</td>
<td>90.8%</td>
<td>81.9%</td>
<td>74.3%</td>
</tr>
<tr>
<td>Viterbo</td>
<td>73.4%</td>
<td>84.2%</td>
<td>61.8%</td>
</tr>
</tbody>
</table>

### TABLE 3 Results of 7 years of screening for thalassaemia in intermediate schools of Latium.

<table>
<thead>
<tr>
<th>Provinces of Latium</th>
<th>No tested</th>
<th>No of thalassaemias</th>
<th>Total thalassaemia</th>
<th>% thalassaemia (SE)</th>
<th>Total non-α</th>
<th>% non-α (SE)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rome</td>
<td>219 272</td>
<td>4479 34</td>
<td>56 587</td>
<td>5156 2·35 ± 0·03</td>
<td>4569 2·08 ± 0·03</td>
<td></td>
</tr>
<tr>
<td>Latina</td>
<td>32 194</td>
<td>763 3</td>
<td>13 123</td>
<td>902 2·80 ± 0·09</td>
<td>779 2·42 ± 0·08</td>
<td></td>
</tr>
<tr>
<td>Viterbo</td>
<td>15 354</td>
<td>291 1</td>
<td>2 40</td>
<td>334 2·17 ± 0·12</td>
<td>294 1·91 ± 0·11</td>
<td></td>
</tr>
<tr>
<td>Frosinone</td>
<td>20 222</td>
<td>373 1</td>
<td>4 34</td>
<td>412 2·04 ± 0·10</td>
<td>378 1·87 ± 0·10</td>
<td></td>
</tr>
<tr>
<td>Rieti</td>
<td>2721</td>
<td>25 9</td>
<td>34</td>
<td>1·25 ± 0·21</td>
<td>25 0·92 ± 0·18</td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>289 763</td>
<td>5931 39</td>
<td>75 793</td>
<td>6838 2·36 ± 0·03</td>
<td>6045 2·08 ± 0·03</td>
<td></td>
</tr>
</tbody>
</table>
subjects a year. The pooled yield of young thalassaemias identified per year in the Centre through both screenings is now about 3300.

Finally, another outcome of the screening is the identification of couples at risk of child-bearing age, which has taken place since 1980. Some of these couples (51 out of 161) had already had an affected child (retrospective couples) and came to the Centre during or before a new pregnancy (table 4), but the majority (110 out of 161) had no affected children (prospective couples) and came for advice at the Centre before conceiving a child, as a direct or indirect consequence of the schools screening. Of the 94 prospective couples of Latial extraction, 35 (more than a third) were the same students identified in the schools screening or through one of their close relatives.

From January 1980 to April 1983, 37 of the 110 prospective couples became pregnant and six of 31 monitored pregnancies (four of which came from Latium) had a homozygous fetus which was aborted.

Discussion

The results of 7 years' work clearly show that mass screening of a young population is feasible and, above all, that it is possible to reach the final target of prospective prevention of Mediterranean anaemia. The most important data supporting this forecast are the following.

In a vast region of 17 000 km², a single Centre for thalassaemia is able to examine an average of 50 000 students per year, which is approximately 80% of all intermediate school students, and in 7 years it has examined 289 763 subjects.

In the same region (which has an average frequency of β-thalassaemia of 2.1%) the Centre identifies every year, in schools and within its catchment area, a total of about 3300 young thalassaemias and 40 prospective couples at risk. These figures show that if the screenings are continued in the future at the same rate, the goal of identifying nearly all the young thalassaemias and all the couples at risk in Latium should be attained in about 15 years. In fact, it can be calculated that there are about 80 000 young couples of child-bearing age at risk in Latium (that is, half of the 160 000 living in the region) and approximately 350 000 older adult couples at risk, that is (0.021)²·10⁻⁴·8·10⁵. If 40 couples at risk continue to be found every year, that will mean that 20 new couples (a reasonable yearly rate of substitution of the total 350 couples) and 20 couples out of those already screened will be discovered every year. Therefore, after 15 to 20 years the 'arrears' will
screening programme for the prospective prevention of Mediterranean anaemia in Latium

### Results of retrospective and prospective prevention of Mediterranean anaemia achieved by the Centre from January 1980 to April 1983.

<table>
<thead>
<tr>
<th></th>
<th>Latium</th>
<th>Other regions</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prospective prevention</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>% of couples</td>
<td>30</td>
<td>21</td>
<td>51</td>
</tr>
<tr>
<td>% of monitored pregnancies</td>
<td>9</td>
<td>4</td>
<td>13</td>
</tr>
<tr>
<td>% of interrupted pregnancies with no fetal exploration</td>
<td>2</td>
<td></td>
<td>2</td>
</tr>
<tr>
<td>% of β-thalassaemia homozygous fetuses (expected 2.25)</td>
<td>1</td>
<td>(expected 1.00)</td>
<td>4 (expected 3.25)</td>
</tr>
<tr>
<td>Retrospective prevention</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>% of couples</td>
<td>94*</td>
<td>16</td>
<td>110</td>
</tr>
<tr>
<td>% of monitored pregnancies</td>
<td>23</td>
<td>8</td>
<td>31</td>
</tr>
<tr>
<td>% of interrupted pregnancies with no fetal exploration</td>
<td>2</td>
<td></td>
<td>2</td>
</tr>
<tr>
<td>% of advanced unexplored pregnancies</td>
<td>1</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>Intanetal diagnosis refused</td>
<td>1†</td>
<td></td>
<td>1†</td>
</tr>
<tr>
<td>% of fetal losses after exploration</td>
<td>1†</td>
<td></td>
<td>1†</td>
</tr>
<tr>
<td>% of β-thalassaemia homozygous fetuses (expected 5.75)</td>
<td>4</td>
<td>(expected 2.00)</td>
<td>6 (expected 7.75)</td>
</tr>
</tbody>
</table>

* 34 of these 94 couples at least one of the partners was either the identified student him/herself or a close relative.
† The couple had two pregnancies. The first terminated after the exploration (healthy fetus). At the onset of the second both partners were diagnosed as 'normal' by another laboratory, the pregnancy was not monitored, and an affected child was born.

We have been caught up and thereafter only the 20 new retrospective couples will be identified every year.

A plan for the prevention of Mediterranean anaemia, similar to the one carried out in Latium, could therefore appear to be useful for other alassaeic areas, because it is easy, quick, and cheap and yields many positive results.

Modifications might be necessary in individual cases with respect to local situations, but schools reeining should always be included in the programme as this has proved to be a very useful means of efficient and permanent dispersal of information to the population. The intermediate school proved to be the ideal level for screening due to the age of the students (13 to 14 years), an age at which students understand and remember the information.

The authors wish to thank the health authorities of the Latium region for financial support, the school directors and teachers, and in particular the many school doctors and other health workers for their valuable collaboration.

### References


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A screening programme for the prospective prevention of Mediterranean anaemia in Latium: results of seven years' work.

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doi: 10.1136/jmg.21.4.268

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