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Screening of thalassaemia carriers in intermediate schools in Latium: results of four years' work

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SUMMARY From 1975 to 1979 the Rome Microcythaemia Centre carried out four health education campaigns and thalassaemia screenings among students of the intermediate schools throughout Latium, under the auspices and with the financial support of the Health Authorities of the Latium region.

This project is aimed at avoiding reproduction by pairs of thalassaemia carriers and the birth of homozygous children.

During the four campaigns 138 501 students were examined, that is, 70 to 76% of those enrolled. Of these 3343 were found to be thalassaemic. Thus the overall prevalence of thalassaemia in Latium is 2.41%, with minor fluctuations from one province to another and, above all, a slight, though definite, trend towards higher values in the southern part of Latium bordering on Campania.

The screening was welcomed by the population and the thalassaemic families, there were no detectable negative side-effects, and it resulted in an increased awareness of the problem of the thalassaemias.

In a previous paper the results were presented of the first screening of thalassaemia carriers among third year intermediate students (about 13 years old) carried out in Latium during the school year 1975 to 1976.¹ In addition to the examination of students to detect thalassaemics, this work included a health education programme and was aimed at avoiding reproduction by pairs of thalassaemics and thus the birth of homozygous offspring.

From 1975 to 1979 the same work was repeated yearly, still supported by the Health Authorities of the Latium region. In this paper we report the total results of the first four screening campaigns.

Methods of screening campaigns

The methods and screening stages are the same as those already presented in our earlier paper,¹ with minor modifications. Particular care was devoted to obtaining the informed consent of students and their parents. In Rome schools this was personally dealt with by doctors from the Centre of Microcythaemia or the school doctor and, in towns lacking a school doctor, by teachers following instructions provided by the Centre.

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In communicating the results to thalassaemic students, in the first screening the words 'suspected thalassaemia' were adopted so as not to alarm the families. However, in the later campaigns, in all cases of β - or $\delta\beta$ -thalassaemia, the diagnosis was explicitly stated by the sentence 'presence of β -thalassaemia' in order to avoid uncertainty and lack of faith in the work.

A full set of investigations was carried out on the probands' families not only when the suspected thalassaemia was of the β or of the $\delta\beta$ type but also in the presumed carriers of α -thalassaemia. The unambiguous interpretation of their slight but definite erythrocytic alterations as being the result of α -thalassaemia spared them the uneasiness of further diagnostic investigations and led to a decision as to whether or not to prescribe iron treatment. It was also explained to these subjects the difference between β - and α -thalassaemia and the different implications as far as offspring were concerned.

Laboratory methods

There were two successive stages of investigation. All the samples were subjected to the first one, whereas only those selected by that preliminary

stage underwent the second set of laboratory examinations.

The first stage consisted of examination of the blood smear and of the osmotic red cell fragility by the technique of Silvestroni and Bianco.² The latter examination was further simplified. The test was done by delivering automatically 1 ml 0.36% saline into test tubes and then adding 5 μ l blood with an automatic micropipette.

The combined reliability of these two tests, red cell osmotic fragility and morphology, which was checked on 1000 samples in the first screening, was tested again on 3000 samples in the later screenings. These 4000 Hb electrophoretic analyses showed no β - or $\delta\beta$ -thalassaemic subject who had not already been detected by the two preliminary tests.

The tests of the second stage were those reported in the previous paper¹ except that from 1977 the blood tests were done with an electronic counter Coulter S. Haemoglobin studies were carried out as described in the paper of 1978.¹

Results

Table 1a,b shows the number of students examined and the number of thalassaemics detected in each of the four screenings, as well as the overall number of students examined and thalassaemics detected in the four screenings in Rome and the five provinces of Latium. It can be seen that the number of students examined rose progressively from 17 725 in the first screening, carried out in the school year 1975 to 1976, to 50 432 in the fourth screening in 1978 to 1979.

TABLE 1a Results of four screenings for thalassaemia in intermediate schools in Latium

School year	No tested	No of thalassaemia carriers	Percentage of thalassaemia*
1975-1976	17 724	429	2.42
1976-1977	26 390	713	2.70
1977-1978	43 955	1086	2.48
1978-1979	50 432	1115	2.21

TABLE 1b Results for Rome and the five provinces of Latium

Province	No tested	No of thalassaemia carriers	Percentage of thalassaemia*
Rome (City)	82 441	2036	2.47
Rome (Province)	20 893	456	1.87
Viterbo	7966	185	2.32
Latina	15 311	439	2.86
Frosinone	9780	200	2.04
Rieti	2110	27	1.27
Total	138 501	3343	2.41

*No confidence limits are given because these incidences were obtained through a census rather than from samples (see table 2).

In the four years of screenings 138 501 students were examined, 82 441 in the city of Rome alone. Of all the third year intermediate students in the Latium region, 89.6% received information on thalassaemia. In the provinces of Rome and Latina such information was given to almost all students, while it was deliberately limited for the students of Rieti and Frosinone provinces where thalassaemia is less common (table 2).

In the first screening, 70% of students were examined.¹ In the fourth screening 76% of students were examined (table 3).

At the end of the four screenings, the overall estimate of the percentage of thalassaemia in Latium was 2.41% (table 1b). There are, however, some small variations from one province to another. The highest percentage was that found in the province of Latina and the lowest that in the province of Rieti (figure, table 1b). The tables for each screening (not included for reasons of space) show a north-south gradient in the incidence of thalassaemia, with the highest frequencies towards the south of Latium bordering on Campania. There was a parallel increase in these areas of the varieties of non- β -thalassaemias (Hb Lepore, Hb F, α -thalassaemia).

This higher prevalence in southern Latium, together with the fact that in the last screenings this part of the region was preferentially examined, are the reasons for the discrepancy between the relative rate of non- β -thalassaemias with respect to all thalassaemias reported in the paper of 1978.¹ This rate was 6.3% whereas that found in the later screenings rose to 10.9%.

Even if one takes into account only the β - and $\delta\beta$ -thalassaemias when comparing the five provinces of Latium (table 4), a slightly but significantly higher frequency ($\chi^2 = 7.21$; $df = 1$; $p < 0.01$) is still found in the province of Latina (2.44%) than in the other four provinces.

As far as the investigation on families is concerned, the percentage of these families coming in each year for the check-up before 30 June (the end of the school year) rose from 50% in the first screening to

TABLE 2 Percentage of school population which received information on thalassaemia during the fourth screening campaign (1978-1979 school year)

Province	Total No of 3rd class students	No of 3rd class students informed	Percentage of students informed
Rome	53 319	51 038	95.7
Viterbo	4030	3032	75.2
Frosinone	7077	4410	62.3
Latina	7499	6998	93.3
Rieti	2141	865	40.4
Total	74 066	66 343	89.6

TABLE 3 Results of fourth school screening of thalassaemics in Latium

Place	No of 3rd class students	No tested	% tested	No of thalassaemia carriers				Percentage thalassaemia
				β	α	F	Lepore	
Rome	41 813	31 790	76	658	61	5	11	2.31
Province of Rome	9225	6857	74	104	17	1		1.77
Viterbo	958	747	78	16				2.14
Province of Viterbo	2074	1667	80	27	1		2	1.79
Latina	1858	1516	81	38	3		1	2.77
Province of Latina	5140	3869	75	80	15			2.45
Frosinone	724	538	74	16	1			3.15
Province of Frosinone	3686	2825	76	48	3			1.80
Rieti	865	623	72	6				0.96
Total	66 343	50 432	76	993	102	6	14	2.21

FIGURE Percentage frequencies of thalassaemia in Rome and the provinces of Latium: pooled data of the four screening campaigns.

TABLE 4 Frequency of β - and $\delta\beta$ -thalassaemia in Latium: pooled data of four screening campaigns

Province	No of 3rd class students tested	No of β - and $\delta\beta$ -thalassaemia carriers	Percentage of β - and $\delta\beta$ -thalassaemias
Rome (City)	82 441	1835	2.22
Rome (Province)	20 893	391	1.87
Viterbo	7966	162	2.03
Latina	15 311	374	2.44*
Frosinone	9780	189	1.93
Rieti	2110	20	0.94
Total	138 501	2971	2.14

*This value is significantly higher ($\chi^2=7.21$, $df=1$, $p<0.01$) than that found in the other provinces pooled together.

64% in the fourth. Many other families came in following years after various requests.

When the families are examined at the Centre, they are told of all the possibilities open to thalassaemics so as to avoid the birth of children with Mediterranean anaemia, that is, the choice of a

non-thalassaemic partner or the decision not to reproduce, with the possibility of adoption, or the selective termination of pregnancy after intrauterine diagnosis of thalassaemia major of the fetus.

Screening in schools did not create any anxiety or worry in the thalassaemic families. At the time of examination the parents only wanted to be reassured that there were no dangers for their thalassaemic children. Given this reassurance, they were pleased to receive the information and advice offered, and showed themselves prepared to put it into effect at the right moment. A telephone survey, begun recently, showed that even after 3 to 4 years the thalassaemic families examined at the Centre still remembered the information received. Of course it is still much too early to verify the effects of the screening and health education on the attitude to reproduction of the thalassaemics and its efficiency in reducing or preventing the birth of homozygotes.

Conclusions

The results of the first four screening campaigns for thalassaemia in intermediate schools allow the following conclusions and specifications to be made.

(1) A voluntary mass screening for thalassaemia among intermediate students is definitely possible. This at least is evident from our experience in Latium.

(2) For this screening today there are simple, quick, precise, and economical methods which have been widely tested and used, and, at least in Latium, there are the necessary social health structures available.

(3) With the methods employed the screening does not produce any appreciable inconvenient or negative side-effects.

(4) The lack of co-operation by students or families can be ascribed to ignorance of the problem of thalassaemia or the difficulty in undertaking a long trip from their home town to Rome, or both, rather than to an explicit refusal to co-operate in the investigation.

(5) Even those families who for some reason do not come for the check-up still receive the exact diagnosis concerning the child examined and all the pertinent information.

(6) The tact in communicating the results associated with the completeness of information succeeded in not disturbing the serenity of the family and not compromising the social life of young thalassaemics.

(7) The screening and health education programme in Latium leaves young thalassaemics completely free to choose the moment and the means of avoiding the birth of homozygous children, according to their moral and religious beliefs.

(8) The yearly screening in intermediate schools is the most powerful and efficient means of educating people on the problem of thalassaemia. The informa-

tion is spread from the schools and the thalassaemic families themselves to the rest of the population.

(9) A brief cost-benefits analysis shows that even in a region only slightly affected by thalassaemia like Latium, the cost of premarital screening is much lower than that of treating the patients. In the whole of Latium today there are about 150 people suffering from Cooley's anaemia. The annual cost of screening in schools over the whole region equals that of annual assistance to 20 or 25 patients. It is evident, however, that the economic benefit will be cancelled out for many years yet by the cost of patients living at present. It is also likely that in future the overall cost of premarital screening will be increased by the cost of prenatal diagnosis for thalassaemic pairs which will still be formed.

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