ABNORMAL HAEMATOLOGICAL CHARACTERISTICS IN DISPUTED PATERNITY.

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The thalassaemias, disorders of Hb globin synthesis, and the Hb globin structural variants are hereditary characters with a simple modality of trasmission. Therefore they can be usefully employed in paternity testing.

A preliminary remark of these genetic markers comes out from very simple laboratory tests:

- a blood count showing normal or elevated number of red cells along with a low M.C.V. and M.C.H. points out to a thalassaemic disorder
- the electrophoretic separation of red cells isoenzymes brings out at the same time the separation of Hb components of red cell lysate: therefore the presence of a haemoglobin variant can by easily noticed.

Further investigations are necessary only in the few cases positive at this preliminary screening.

In order to evaluate in concrete terms the use of these markers in paternity testing, along with the haematological study, we tested for 8-20 polymorphic systems the 316 members of 87 families investigated for a possible hereditary haematological disorder (Tab. I).

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Tab. I **87 FAMILIES-142 CHILDREN TESTED FOR 8-20 POLYMORPHIC SYSTEM**

45 Families-45 Children **29 Families-58 Children** **13 Families-39 Children**

18 Families - 28 Children tested for 8 - 9 polymorphisms
20 Families - 38 Children tested for 11 -14 polymorphisms
15 Families - 20 Children tested for 15 -18 polymorphisms
34 Families - 56 Children tested for 19 -20 polymorphisms

26 Families - 40 Children

L Father with an abnormal gene
observed in 26 Children
not observed in 14 Children

5 Families - 7 Children

L Father & mother with an abnormal gene
6 Children with the 2 interacting
abnormal genes

24 Families - 44 Children

Mother with an abnormal gene or
Mother & L Father with the same abnormal gene
Children etherozygous for the abnormal gene
or without the abnormal gene

32 Families - 51 Children

Mother and L Father
without abnormal genes

In 55 out of 87 families the diagnostic suspect was confirmed:the haematological abnormalities encountered are reported in Tab. II

Tab. II 55 FAMILIES WITH AN ABNORMAL GENE

Abnormal gene	Families	Children	Children with a.g.	Children without a.g.
β -thal	35	52	38	14
α-thal	2	5	4	1
Hb Lepore	1	3	1	2
Hb S	8	17	10	7
Hb C	2	2	2	-
Hb Hasharon	2	4	2	2
Other variants	3	5	2	3
Sickle-cell thal	1	2	1	1
H. spherocytosis	1	1	1	-
TOTAL	55	91	61	30

In consideration of the dispersion of values of Probability and Frequency of Exclusion observed with limited genetical typing, the contribution of an abnormal gene is calculated only for trios and duos typed for 19-20 polymorphic systems (Tab. III).

Tab. III

17 FAMILIES - 26 CHILDREN

TESTED FOR 19-20 POLYMORPHIC SYSTEMS

L Father with an abnormal gene

observed in 20 Children - not observed in 6 Children

	abnormal gene of L Father			
	not included		included	
	Pr P%	Fr Ex%	Pr P%	Fr Ex%
20 Children with a.g.	96.4	93.9	99.9	99.8
6 Children without a.g.	98.9	97.7	97.9	97.7

For comparison the values observed in the 56 trios and duos with or without paternal transmission of an abnormal gene are reported in Tab. IV.

(Tab. IV)

Mean values for 56 (C-M-LF) and (C-M)

Tested for 19-20 Polymorphic Sistems

Pr P	Fr Ex
97.1%	94.4%

No paternity exclusion due to an abnormal gene was observed in our series. In 3 families, not included in the present series, the exclusion of 1 child came out from common polymorphic systems testing.

An alleged father can be excluded from paternity of a child with a beta-thalassaemia trait, if this man and the mother do not carry the trait.

On excluding as a father a man with normal haematological characteristics, carrying the mother a beta-thalassaemia trait and showing the child beta-thalassaemia disease, one has to take into account the exceptional occurrence of a beta-thalassaemia silent gene.

Absolutely unreliable is the exclusion of a "normal" man, when the child has evidence of an alpha-thalassaemic disorder.

On the contrary Hb globin variants offer a perentory criteria for paternity exclusion.

The contribution of a gene to the value of Probability of Paternity is inversely proportional to the frequency of this gene: the abnormal haematological genes have a wide geographical variation of frequency (Tab. V).

Tab. V a. FATHER WITH A LOW FREQUENCY GENE

	Fr GENE	Pr P	I.P.	Fr Ex
Thalassaemia	0.08	86.21	6.25	84.64
	*(0.92)	35.21	0.54	0.64
	0.04	92.59	12.5	92.16
	*(0.96)	34.25	0.52	0.16
	0.013	97.47	38.5	97.42
	*(0.987)	33.62	0.51	0.02
Hb Hasharon	0.004	99.21	125	99.20
	*(0.996)	33.42	0.5	0.00
Hb S	0.00025	99.95	2000	99.95
	*(0.99975)	33.34	0.5	0.00
HbC, Hb Lepore, H.S.	0.0001	99.98	5000	99.98
	*(0.9999)	33.34	0.5	0.00

*low frequency gene absent in the Child.

The choice of the more likely frequency is a difficult one . In areas with mixed populations one has to consider the effect on matings of ethnic differences, of social stratification, and must evaluate the real opportunities of meeting for the involved persons.

A solution to the problem of frequency of the abnormal gene is that of taking the average frequency observed in the larger area in which the female can have the opportunity of finding a partner: this solution will avoid the risk of giving an excessive weight to the abnormal character in positivo or in negativo.