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Differential effect of consanguinity on both sexes

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RESUMO

Os resultados foram obtidos na população duma pequena aldeia isolada. De forma a estudar o efeito da consanguinidade, considerámos um período longo (1900--1950), que permitiu o levantamento de todos os fenómenos e de alguns efeitos a longo prazo. O nosso estudo abrange 76 famílias (22 consanguíneas) com período reprodutivo completo, totalizando 313 gestações.

Para este estudo tivémos em conta os seguintes parâmetros: abortos espotâneos, nados-mortos, mortalidade infantil e mortalidade até à maturidade sexual, taxa de emigração e número de pessoas com enfermidades óbvias.

Uma primeira observação é que a percentagem de sobreviventes é maior no interior do grupo consanguíneo, devido ao maior número de abortos espontâneos e à maior mortalidade neonatal.

Uma outra observação é que a mortalidade total é bastante maior entre os homens, quer no grupo consanguíneo, quer no grupo de controle.

Palavras chave: Consanguinidade; Abortos espontâneos; Mortalidade infantil; Mortalidade juvenil.

ABSTRACT

Results were collected in the population of a small isolated village. In order to study the effect of inbreeding, we considered a longer period (1900-1950) which permitted the survey of all phenomena and some long-term or late-occuring effects. Our study surveys 76 families (22 consanguineous) with a closed reproduction period, totalizing 313 pregnancies. For this study we took into account the following parameters: spontaneous abortion, stillbirths, infant mortality and mortality up to sexual maturity, emigration rate and the number of people with obvious affections.

A first observation is that the percent of survivors, within the consanguineous group is lower, because of the higher number of spontaneous abortions and of higher neonatal mortality.

A second observation is that mortality is far higher amongst the men, both in the consanguineous group and in the control group.

Key-words: Consanguinity; Spontaneous abortion; Infant mortality; Juvenile mortality.

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INTRODUCTION

The effects of inbreeding in man considered under various angles have been discussed in numerous papers, especially these last 20 to 30 years some of these papers also deal with the effect of inbreeding in raising the incidence of some anomalies and of pre and postnatal mortality.

The present work deals especially with the differentiation of this action with two sexes and with the analysis of some factors which might lead to considerable differences between the two sexes as concerns prenatal and infant mortality.

MATERIAL AND METHOD

Results were collected from a small isolated village, whose population display a high consanguinity (SCHMIDT, 1978; 1979).

In order to study the effect of inbreeding we resorted to a longer period (1900-1950), which permitted the survey of all phenomena and of some long-term or late occurring effects. Our study surveys 76 families with a closed reproduction period, having 313 pregancies. Of these 22 families with a total of 68 pregnancies (3.1 per family) from the group of consanguineous families. Within this group, we included all marriages between relatives up to the kinship of 3rd degree cousins. In a quarter of these couples, the males are related twice or even thrice by different common ancestors. The average inbreeding coefficient of this group is 0.027876. The other families, with an average of 4.2 pregnancies per family, represent the control group. Though inbreeding is absent we assume, however, that there is a high rate of homozygotes since 80% of these families are formed of local mates (marriage radius = 0); the so-called exogamous families of this period are actually formed of mates from other 2 villages with population of the same origin. Both groups belong to the same population, 90% of the studied families originate in the founding families and they live in the same milieu. These facts remove, from the very beginning, some differences caused by different anthropological structures, by different environment or culture factors: all families are submitted to the same environmental conditions.

For this study, account was taken of the following parameters: spontaneous abortion, still births, infant mortality and mortality up to sex maturity.

RESULTS

Table 1 presents an overall picture of the inbreeding effects on the whole population. A first observation is that the percent of survivors, within the consanguineous group, is lower, because of the higher number of spontaneous abortions and of higher neonatal mortality. Within the two other groups of mortality, differences are absent. Consequently it seems that genetic factors

play a certain role within total mortality up to sex maturity, especially during intrauterine life and in the first month after birth. In later stages, environment factors are largely responsable for the death rate.

Yet it is surprising that great differences were obtained for these parameters when dividing the material per sexes (Table 2). The influence of inbreeding is noticed again and it occurs especially during intrauterine life and in the first month of life. This differentiation permits us to notice also the influence in the females; however differences are higher with males. Moreover the frequencies of total mortality are considerably higher with males, both in the consanguineous and in the control group.

DISCUSSION

MULLER (1950) specified that every one has at least a lethal gene, on an average. In a homozygote condition it might induce the death of the individual, between birth and maturity. These calculation of the genetic load are based on differences concerning the frequency of some anomalies and especially differences on the frequencies of mortality from different periods of pre and postnatal development, between control and consanguineous groups. Differences in favour of the consanguineous group may be considered as an effect of inbreeding, which facilitate the appearance of the effect of some recessive detrimental genes by the increase of the number of homozygotes.

Table 3 shows these differences within the studied population both as a whole and separately on two sexes. The action of inbreeding during embryonic life and in the first month of life is obvious again. But differences are clearly in favour of men.

A first cause might be the existence of some genes or groups of lethal recessive X-linked genes. Thus in the case of marriage between a female carrier and a healthy male, the female offspring will be half healthy half carrier; the males ones — half healthy half carrier of a detrimental gene — being hemizygotes, will die. In this case, adult men will always be healthy, women will never reach a homozygote condition, being at most carrier, in exchange 50% of the male individuals will be ruled out.

A detailed reanalysis of the material and families took into consideration only families with more than 3 pregnancies, with both girls and boys. We found 19 families with a total of 96 pregnancies (54 males and females), where 29 males (i.e. 53.7%) and only one female (2.45%) died in the last month of pregnancy or in the first year of life (Table 4).

The fact that in these families 50% of the boys died, leads us to believe that here too, X-linked genes were active. Consanguinity is of unimportance, hence the similar percentage in both groups.

The disadvantageous alleles of the X chromosome are rapidly eliminated in the population, because they are exposed to male zygotic selection. Theoretically, if the frequency of these alleles is to remain constant from generation to generation, then a large number of X chromosomes would continually have

TABLE 1. *Survey on the frequency of mortality in the offspring of studied families*

Group	N.º of families	N.º of pregnancies		Spontaneous abortions of know sex	Infant mortality	Mortality between 1 year and sexual maturity	Survivors
Consanguineous group	22	68	N	68	64	53	68
			n (Affect.)	4	11	5	48
			%	5.9	17.1	9.7	70.6
Non-consanguineous group	54	245	N	245	242	209	245
			n Affect.)	3	33	20	189
			%	1.2	13.6	9.6	77.1
Total	76	313	N	313	306	262	313
			n (Affect.)	7	44	25	237
			%	2.2	14.0	9.6	75.7

TABLE 2. *Distribution of mortality up to sexual maturity depending on sex and inbreeding*

	Consanguineous group						Non-consanguineous group					
	♂			♀			♂			♀		
	N	Affect.	%	N	Affect.	%	N	Affect.	%	N	Affect.	%
Spontaneous abortions	34	3	8,8	34	1	2.9	117	2	1,7	128	1	0,8
Infant mortality	31	8	25,8	33	3	9,1	115	24	20,9	127	9	7,1
Mortality between 1 year and sexual maturity	23	3	13,0	30	2	6,6	91	8	8,8	118	12	10,1
Total	34	14	41,2	34	6	17,6	117	34	29,1	128	22	17,2

TABLE 3. *Difference concerning the frequency of mortality up to sexual maturity betwen consanguineous and non-consanguineous group*

	Sex	Consanguineous group			Non-consanguineous group			Difference
		Total	Affected	Frequence	Total	Affected	Frequence	
Spontaneous abortions	Total	68	4	0.058	245	3	0.012	0.046
	♂	34	3	0.088	117	2	0.017	0.071
	♀	34	1	0.029	128	1	0.008	0.021
Infant mortality	Total	64	7	0.109	242	17	0.070	0.039
	♂	31	5	0.161	115	13	0.113	0.048
	♀	33	2	0.060	127	4	0.031	0.029
Mortality between 1 year and sexual maturity	Total	57	9	0.158	225	36	0.160	—0.002
	♂	26	6	0.231	102	19	0.186	0.045
	♀	31	3	0.097	123	17	0.138	—0.041

TABLE 4. *Frequency of spontaneous abortion, of infant and juvenile mortality depending on detrimental possible X-linked genes and other genetic and non-genetic causes*

Group	Detrimental possible X-linked							Other genetic and non genetic causes						
	N.º of families	Sex	N.º of pregnancies	Infant mortality and spontaneous abortions		Mortality between 1 year and sexual maturity		N.º of families	Sex	N.º of pregnancies	Infant mortality and spontaneous abortions		Mortality between 1 year and sexual maturity	
				N.º	%	N.º	%				N.º	%	N.º	%
Non-consanguineous group	14	♂	40	22	55.0	1	2.5	18	♂	43	4	9.3	7	16.3
		♀	38	1	2.6	3	7.8		♀	64	9	14.1	9	14.1
Consanguineous group	5	♂	14	7	50.0			6	♂	13	4	30.7	3	23.0
		♀	4						♀	18	4	22.2	2	11.1
Total	19	♂	54	29	53.7	1	1.8	24	♂	56	8	14.3	10	17.8
		♀	42	1	2.4	3	7.1		♀	82	13	15.8	11	13.4

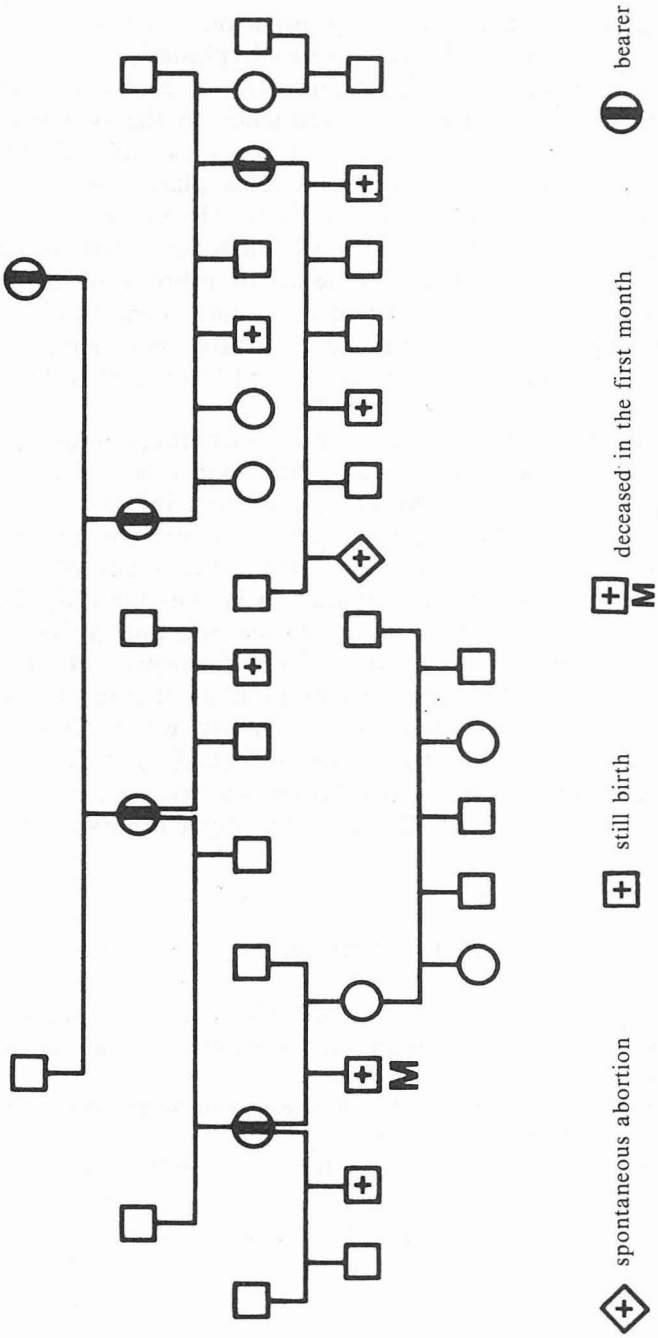

Fig. 1 — Pedigree with a possible presence of some X-linked lethal genes

to mutate from normal to abnormal. This hypothesis, however, is improbable, and can only be demonstrated with difficulty. Therefore, it is unlikely that all the cases in the aforementioned families are attributable to lethal X-linked genes. That such genes are active in the population can only be speculated. The family tree (Fig. 1) may shed light on this hyphotesis.

The rest of cases of prenatal, infant and juvenile mortality are recorded in the second part of table 4 and their frequency in the two sexes is very close (14.3%) in men and 15.8% in women). They are due either to other non-genetic causes or to some recessive lethal genes placed on the autosomes. In the latter case, inbreeding can lead to the rise of the frequency of mortality within the consanguineous group, as may be clearly seen from table 4.

Data of table 4 shows that the influence of inbreeding is higher with males (a rise in men from 9.3% to 30.7%; in woman from 14.1% to 22.2%). This fact could be expected if we think that mortality produced by X-linked recessive genes is not influenced by inbreeding and frequency is the same with both groups.

This rise of the death rate with males in the consanguineous group might also be explained by the fact that certain lethal genes act only in a certain development stage, as shown also by other authors (STERN, 1973). It often happens that the gene action has a certain degree of variableness and certain genotypic of the carriers can survive this period. The types who succeed in surviving this stage develop in a normal way. It is thus possible that certain homozygote genes behave lethally in the male sex and not in the female sex. Inbreeding will thus result in a increase of the frequency of prenatal and infant mortality with males, by the increased number of homozygotes.

We consider that the detailed study of the family, whenever possible, and the differentiation of data for the two sexes is a good method of separating the two genetic causes of prenatal and infant mortality. This permits a more accurate appreciation of the genetic load which might otherwise be underestimated.

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