Consanguinity and Blood Group Distribution in an Amish Isolate

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Having noted many patients with an autosomal recessive type of muscular dystrophy (Jackson and Carey, 1961) among the Amish of Adams County, Indiana, we undertook in 1964 a genetic survey of the 1,386 Amish in this county (Fig. 1). Data are presented on the consanguinity and blood group distribution in this population. These data are compared with those from a similar study of another local Swiss population in which hereditary primary amyloidosis had been observed (Rukavina *et al.*, 1956).

METHODS

Amish individuals were interviewed by family groups in the home. Only the Old Order Amish living within the county were included in this report. Genealogical information was obtained from individuals, family records compiled by family members, Swiss and French community records, and from the Mormon Genealogical Library in Salt Lake City, Utah. Blood, drawn in the home, was brought to the Caylor-Nickel Research Foundation Laboratory for testing. For Xg^a, Duffy, and Kell typing, specimens were collected in ACD solution and sent to Butterworth Hospital, Grand Rapids, Michigan.

The other Swiss population, studied in 1959–1964, consisted of members of an Anabaptist church in Adams County and adjacent areas in Wells County, Indiana. Genealogical information was obtained from family members, funeral home records, and Swiss community records.

In order to ascertain degree of consanguinity, the coefficient of inbreeding was calculated for each mating taking into account *all* possible relationships of the parents of the individuals. Wright (1922) used this approach for the study of inbreeding in domestic animals; an excellent explanation of its application to human populations is described by Neel and Schull (1954). Examples of the type of complicated pedigrees encountered have been published in the report on limb-girdle muscular dystrophy in this population (Jackson and Strehler, 1968).

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RESULTS

Table 1 tabulates the average number of live births for the mothers with completed families in the two populations. Table 2 summarizes the consanguinity data on the two populations. Nine simple first cousin matings were encountered among the Amish; however, no marriage partners were obviously more closely related than first cousins. The values of the average coefficients of inbreeding were increased to the level shown in this table by the frequency of multiple connections between ancestors. Table 3 lists the most common surnames of the Amish families. Table 4 provides a comparison of blood group distribution between the Amish population, the other Swiss population, and random Anglo-Saxon populations.

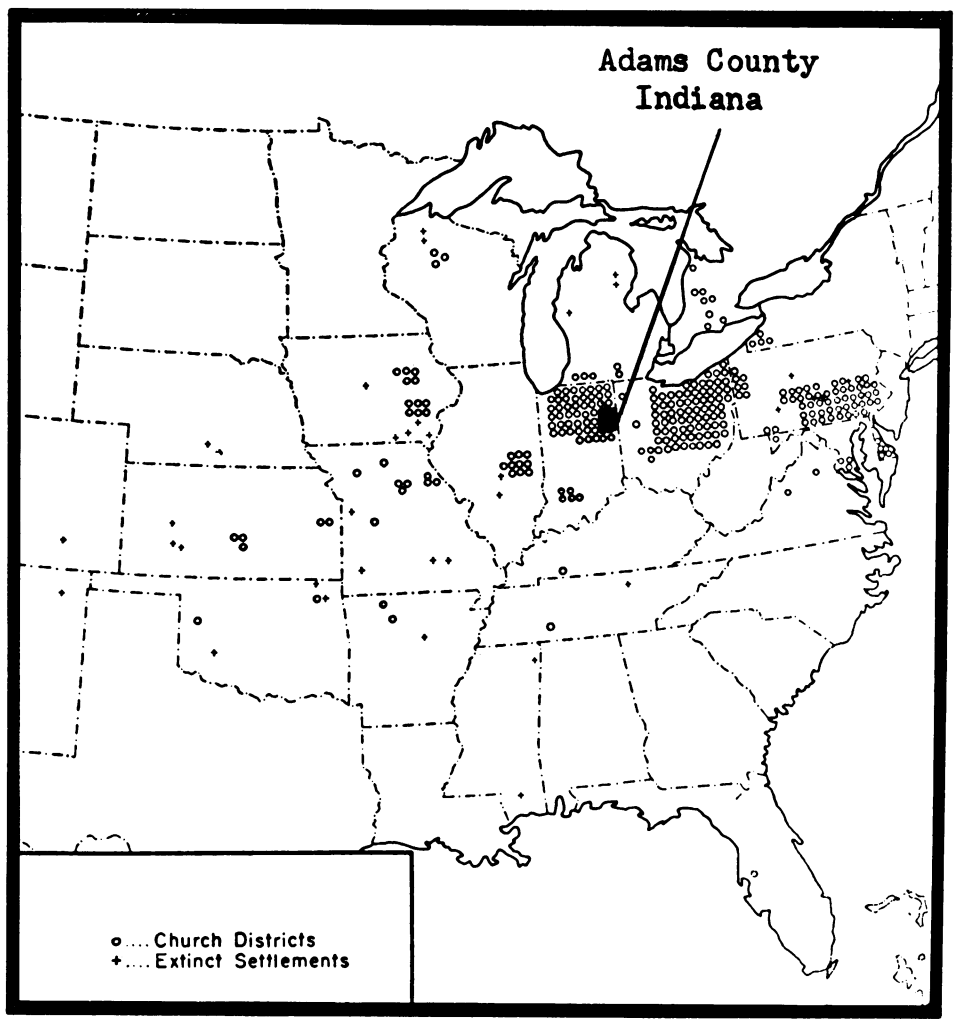

FIG. 1.—Map of Old Order Amish church districts in the United States illustrating the location of Adams County (Hostetler, 1963).

TABLE 1

NUMBER OF LIVE BIRTHS FOR COMPLETED FAMILIES*

	Mothers	Children	Average
Amish isolate	59	590	10.0
Other Swiss isolate	133	623	4.7

* Mothers over 45 years of age.

TABLE 2

CONSANGUINITY DATA FOR TWO POPULATIONS

RELATIONSHIP OF PARENTS	AMISH ISOLATE*			OTHER SWISS POPULATION†		
	Matings	Offspring	%	Matings	Offspring	%
1st cousins or closer	14	86	6.2	1	3	0.2
Between 1st and 2d cousins	109	589	42.5	43	152	12.4
Between 2d and 3d cousins.	35	187	13.5	40	126	10.4
Between 3d and 4th cousins	12	59	4.3	10	26	2.0
Parents unrelated	106	465	33.5	372	923	75.0
Total	276	1,386	.....	466	1,230	.....

* Average coefficient of inbreeding for entire population is .0195 (parents between first and second cousins).
† Average coefficient of inbreeding for entire population is .0028 (parents between third and fourth cousins).

TABLE 3

MOST COMMON FAMILY SURNAMES IN THE AMISH OF ADAMS COUNTY LISTED AS PERCENTAGE OF THE 243 FAMILIES*

Surname	%
Schwartz	50
Hilty	10
Wickey	9
Eicher	7
Girod	4
Shetler	3
Graber	2
Christner	2

* The remaining 13% is composed of only 10 additional surnames.

DISCUSSION

McKusick *et al.* (1964) have discussed the characteristics of the Amish which make them useful for genetic studies and have emphasized the types of genetically significant information obtainable from such a population. Among the advantageous features mentioned is that of large family size. It can be noted in Table 1 that the Adams County Amish have an average of 10 offspring for completed families. In 33 of the 59 families tabulated, the parents were related as third cousins or closer, and the average number of live births was 11.2. In the remaining 26 families, the average number of births was 8.5. Therefore, the degree of consanguinity appeared to have no major effect on fertility.

TABLE 4

BLOOD-GROUP DISTRIBUTION IN THREE POPULATIONS*

Blood Group	Amish Isolate	Random Anglo-Saxon Population†	Other Swiss Population‡
O	15.6%	45%	60.5%
A	64.6	41	31.5
B	11.4	10	5.9
AB	8.4	4	2.1
M	25.4	28.3	14.5
N	25.1	21.9	9.0
MN	49.5	49.8	76.5
Rh(D)+	82.2	85.1	88.9
Rh(D)−	17.8	14.9	11.1
C^w variant	10.7	1.3	.....
Fy(a+)	79.4	66.5	.....
Fy(a−)	20.6	33.5	.....
K−	99.7	91.1	.....
Xg(a+) Males	63	63.1	.....
Xg(a+) Females	82.5	89.3	.....
Xg(a−) Males	37	36.9	.....
Xg(a−) Females	17.5	10.7	.....

* For the ABO, MN, Rh, Fy, and K systems 892 Amish and 619 of the Other Swiss Population were tested. For the Xg^a system, 838 Amish were tested.

† Race and Sanger, 1962.

‡ Authors, unpublished data.

The consanguinity noted in Amish populations is also an evident advantage in genetic studies. The degree of consanguinity in the Amish population reported here (Table 2) is extremely high for a North American population. The coefficient of inbreeding probably exceeds that of the triracial isolate reported by Witkop *et al.* (1966) and approaches the .0211 and .0255 reported in Hutterite subisolates by Steinberg and his group (1967). In a previous consanguinity study of the Amish population (Hammond and Jackson, 1958), 4.4% of 627 marriages had been found to be between individuals related as first cousins or closer and 17.4% between individuals related between first and second cousins. The higher values for consanguinity in the present report are related to the fact that the tabulation was restricted only to those who remained in the church and in the county. The previous report also listed marriages from 1850 to 1949, and since 1949 the marriage couples have been more closely related

to each other. The increasing percentage of consanguineous marriages most likely accounts for the increasing number of cases of limb-girdle muscular dystrophy in recent generations (Jackson and Strehler, 1968).

The most common family surnames (Table 3) show no overlap with those tabulated by McKusick *et al.* (1964) for Lancaster County and Mifflin County, Pennsylvania, and Holmes County, Ohio. Since the Amish in various localities tend to be distinct isolates and since they tend to marry only within their sect, the increasing frequency of recessively inherited disease can probably best be prevented by Amish marrying others of their religion from separate isolates.

The cause of the disparity between blood group distribution of each of the populations and the random population (Table 4) is evident in the data on consanguinity in the populations. Gene frequencies are not presented in this blood group table because the Amish population is essentially one large kindred—only 64 of the 1,386 Amish individuals in the county are unrelated to one Swiss couple who emigrated here in 1853. Wall *et al.* (1967), in reporting blood group frequencies in an Ohio Amish population, relate the distorted blood group distribution to the founder effect of colonization of unpopulated areas by a small number of couples. The relative deficiency of Xg(a+) females apparent in Table 4 deserves further study, since it may be related to an interaction of Xg^a and the sex ratio suggested by Dewey *et al.* (1965).

SUMMARY

In a genetic study of an Amish isolate in northeastern Indiana, blood group distribution was found to differ greatly from that in random populations. These deviations are interpreted as being related to the high degree of inbreeding in the population. The parents of 6.2% of the 1,386 Amish in the county were related as first cousins or closer with a total of 49% related as second cousins or closer. The average coefficient of inbreeding was .0195 (parents between first and second cousins).

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