Type IV Hyperlipoproteinemia in a Consanguinous Family

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SUMMARY
The involvement of all five offspring of consanguinous and hypertriglyceridemic parents (first cousins) is reported.
Three sons and two daughters of this family have increased triglycerides, normal cholesterol, and normal or slightly increased phospholipids. Mother and elder daughter have abnormal glucose tolerance. Father and mother are hypertensive; father had recent development of myocardial infarction and gout. The two elder sons had myocardial infarction and repeated bouts of angina. All three male offspring of this family have gout and are being treated with xanthinoxidase inhibitor (allopurinol). Planar xanthoma was present in one son with myocardial infarction. No chylomicronemia was noted. The response to diet and Atromid-S was satisfactory.

Additional Indexing Words: Gout Triglycerides Xanthinoxidase inhibitor Atromid-S Xanthoma

It is now generally accepted that hyperlipoproteinemia and premature vascular disease are associated. This is more obvious for type II hyperlipoproteinemia, but type IV is also believed to be associated with increased likelihood of ischemic heart disease. One of the unvarying characteristics of type IV hyperlipoproteinemia is hyperglyceridemia, but cholesterol concentration is frequently normal. Hence type IV is not always appreciated when cholesterol levels alone are measured.

Because reduction in weight and sometimes carbohydrate restrictions or Atromid-S is often effective in reducing elevated lipoprotein levels in type IV,1-6 the recognition of the abnormality is important. It is not yet proved that effective treatment of this condition actually reduces the risk of vascular disease, but this seems likely. An important aspect of type IV, stressed by Fredrickson and co-workers, is the probability that it is very frequently due to genetic abnormalities. The mode of inheritance, or how many mutations might be involved, is not established.

Presented here is a most unusual example of type IV hyperlipoproteinemia, in all five offspring of consanguinous parents who also had type IV. The record of such a family should be of both interest and value in eventually unraveling this disorder, a likely genetic cause of premature vascular disease.

This family is white, muslim, and lives in the southern part of Iran where consanguinous marriages are frequently seen.

As is illustrated in figure 1, the father (M) and mother (F) are first cousins. Their offspring are three sons (A, B, and E) and two daughters (C and D). The parents and children are all hypertriglyceridemic in a different range. No chylomicon was noted in overnight storage of their blood in a cold temperature.

The parents are 70 and 60 years old, respectively, and are under therapy for hypertension. The mother had a diabetic glucose tolerance test (GTT). The father had
FAMILIAL HYPERLIPOPROTEINEMIA

Figure 1

Pedigree illustrating the members of the family (1-12) who were not studied. The parents (F and M) and five offspring (A, B, C, D, and E) are studied: F and M are first cousins. Abbreviation: H.P. = hyperlipoproteinemic.

a recent manifestation of myocardial infarction, cerebral transient ischemic attacks, and gout.

The oldest son (A) is 42 years old and has had gout for 10 years, inferior wall myocardial infarct, and repeated bouts of angina which have diminished in frequency since he is on diet and treatment.

Son B is 39 years old, is underweight for his height, and has gout, myocardial infarction, and a planar xanthoma which led us to diagnose his hyperlipemia. The related laboratory findings in these cases are shown in table 1.

The daughters, 32 (C) and 26 (D) years old, are clinically asymptomatic, save for slight obesity. The older one has abnormal GTT and like all members of the family has elevated triglycerides. The youngest of the family is a 22-year-old man (E) who has been suffering from gout since the age of 18 and is being treated with xanthinoxidase inhibitors daily.

No member of the family has hepatosplenomegaly or abdominal pain. It is possible that vascular manifestations of the two daughters are delayed due to their sex, and future follow-up would certainly clarify this. Sons A and B are married but not to any of their relatives.

A and B have followed a low-carbohydrate diet and have taken 2 g of Atromid-S daily for the last 5 months; their triglycerides are
markedly decreased from 4280 and 480 to 148 and 93 mg/100 ml, respectively.

References

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Circulation. 1972;45:988-990
doi: 10.1161/01.CIR.45.5.988

Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
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Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the World Wide Web at:
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