Cholestasis Familiaris Groenlandica

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In certain areas of Greenland we have found an accumulation of a rare familial progressive intrahepatic cholestatic disease, especially on the East coast (Ammassalik), but also in some districts on the West coast (Paamiut, Uummannaq and Upernavik).
We have named the disease Cholestasis Familiaris Groenlandica (CFG).
It is an autosomal recessive inherited liverdisease which causes jaundice, failure to thrive, dwarfism, severe pruritus and early death.
We have diagnosed 41 cases from 1943 until 2000, by looking in the old records, interviewing parents to children, who have died from jaundice and failure to thrive or by examining the sick children. A detailed genealogy has been of great value.
The disease cannot be cured, only symptomatic treatment can be offered and to day only 4 children are alive. Livertransplantation is a possibility in some cases.
The disease is caused by a missence mutation in region 21 on the long arm on chromosome no. 18 (18q21). A similar disease has been described in a large kindred in the Amish people and named after their founder Jacob Byler (Byler disease). The disease or variations of the disease has also been described in other families all over the world, but not in such large numbers.
Three inuit patients, from the northern part of Baffin Island, Canada have presented with the same symptomatology as the Greenlandic inuits, and in two Canadian families the same mutation was found as in the Greenlandic inuits indicating the hypothesis that the mutation was introduced by a common founder, before separation of the current East Greenlandic, West Greenlandic and Canadian inuit populations.
It is now possible to determine, whether unaffected siblings within these families are heterozygous carriers of the disease. In addition, prenatal molecular diagnosis may be offered in relevant pregnancies.