

ON THE EPIDEMIOLOGY OF JUVENILE AMAUROTIC IDIOCY IN DENMARK

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Juvenile amaurotic idiocy is an autosomal recessive disease with onset around 6 years of age. Principal symptoms are: blindness; dementia; focal and generalized seizures; cerebellar, striatic, and pyramidal deficits; and psychosis. The course is progressive, common duration 10-20 years. Epidemiological studies are very few, but important work has been done by Sjöstrand and Rayner in Sweden.

The preliminary results of the present study indicate a rather higher incidence of the disease in Denmark. It is furthermore demonstrated that there is no evidence of consanguinity between parents, and that the disease is geographically evenly distributed. These findings are in contrast with earlier epidemiological investigations.

Juvenile amaurotic idiocy is an inherited disorder characterized by onset between the ages of 3 and 9, progressive deterioration of vision, dementia, epilepsy, and other neurological signs, with death usually occurring in the third decade of life.

There is still some controversy as to the nosological entity of the disease as well as to the delineation from other conditions which seem closely related. Zeman and Dyken (1969), for example, have recently included a case with onset at the age of 23 in a series of patients with juvenile amaurotic idiocy. They may be right in doing so as we do not know the underlying metabolic defect at this stage. We are at present unable to put forward a biochemical definition of the disease.

However, I must admit that at present we are tempted to limit the term juvenile amaurotic idiocy, or Batten-Spielmeyer-Vogt's disease, to cases which fulfil the following criteria:

1. Decreasing vision, beginning in the age range 3-9 years.
2. Characteristic fundoscopic appearance: In the first stage small pigmentary changes in the macula, later narrowing of the arteries and pronounced pigmentary changes in the periphery, as well as optic nerve atrophy.
3. Progressive dementia, ataxia, rigidity, pyramidal signs, and epileptic seizures. The latter are in the beginning mostly generalized seizures, whereas myoclonic seizures are prominent in the terminal stage.
4. The presence of vacuoles in lymphocytes in peripheral blood (Rayner 1962).

In Sweden, Sjögren (1931) and later Rayner (1962) have made very considerable contributions to our knowledge of the heredity of the disease. Sjögren showed that it fulfilled the criteria for a recessive autosomal disorder. He also noted a high frequency of consanguineous marriages in the ascendance of patients (in about 25%). On the basis of this finding he de-

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TABLE 1
BIRTHPLACE OF PATIENTS WITH JUVENILE AMAUROTIC IDIOCY IN DENMARK

Birthplace	Patients (N = 28)		General population (%)
	N	%	
Copenhagen	7	25	28
Other cities and towns	10	36	40
Rural districts	11	39	30
Sealand	13	47	45
Funen	4	14	10
Jutland	11	40	45

monstrated that certain sparsely populated regions in Sweden were actually foci of the disease. In these regions the gene must have been widespread through generations.

By studying vacuolization of lymphocytes, Rayner was able to show that almost all presumed heterozygotes had a vacuolization percentage of about 1%, in contrast to unaffected, where only occasionally vacuoles could be demonstrated. He furthermore confirmed the presence of "foci" in certain regions in Sweden by using this method to detect heterozygotes in rather large population groups. Rayner noted a tendency for the foci to break up as well as a decreasing rate of consanguineous marriages. He paralleled these findings to a decline in the incidence of the disease compared to the data obtained by Sjögren 30 years earlier. This implied a hope for the gradual tendency to disappearance of the disease with increasing urbanization and mobility of the population.

Extensive epidemiological studies of this kind have not been carried out outside Sweden. However, we thought it to be of interest to put forward some data originating from our clinical study of the disease in Denmark. These data contrast markedly to the findings in Sweden.

On the basis of the criteria mentioned earlier in this communication we confirmed the diagnosis in 28 living patients in Denmark (1971). Most of the patients came to our knowledge via the National Institute for the Blind, others from the mental hospitals or from institutions for mentally handicapped. We made a survey of the places of birth of the patients, their parents and grandparents, as well as the rate of consanguineous marriages in the ascendance of the patients.

In the first place we established an incidence almost similar to that found in Sweden 40 years ago. The patients were evenly distributed throughout the country as regards their birth-

TABLE 2
INCIDENCE OF JUVENILE AMAUROTIC IDIOCY

Sweden	
Sjögren (1931)	4 per 100,000
Rayner (1962)	2 per 100,000
Denmark	
Christensen Lou and Kristensen (1973)	3.1 per 100,000

TABLE 3
JUVENILE AMAUROTIC IDIOCY AND CONSANGUINITY

Consanguinity	Patients with juvenile amaurotic idiocy			General population		
	%	Country	Reference	%	Country	Reference
Parents first cousins	15	Sweden	Sjögren 1931	0.6	Germany	Wulz 1925
	9.1	Sweden	Rayner 1962			
	0.0	Denmark	Christensen Lou and Kristensen 1973			
Parents second cousins	7	Sweden	Sjögren 1931	0.6	Germany	Wulz 1925
	0.0	Denmark	Christensen Lou and Kristensen 1973			

place compared to the distribution of the general population in Denmark (Table 1). The calculated incidence compared to the Swedish findings is shown in Table 2. On the other hand, no consanguineous marriages were found (Table 3).

In conclusion, even if we have a comparatively high incidence of juvenile amaurotic idiocy in Denmark, no isolated regions with a high rate of consanguineous marriages in the ascendance of the patients were present. Thus the epidemiology of the disease must be very different in Denmark and Sweden. We await data from other countries with great interest.

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