Angiokeratoma corporis diffusum Fabry

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In Holland 29 patients suffering from Fabry's disease are known, belonging to six families. In one pedigree the diagnosis is certain in seven cases, in another in sixteen cases. Both pedigrees are in accordance with X-chromosomal localization of the responsible gene.

Two findings never fail: it is always possible to demonstrate the pathological lipid in the vessels of the skin, even when visible skin lesions are absent. Furthermore it is always possible to detect by split-lamp examination a very slight corneal opacity (Velzeboer).

The clinical manifestations are extremely variable. The angiomas were almost absent in a 19-year-old man. He only showed some tiny angiomas on his thigh. A 9-year-old boy however had a great many tiny angiomas on his back. A 12- and a 15-year-old boy both had tiny dilatations of the scrotal vessels.

In the female patients too the number and distribution of the angiomas differ greatly amongst the individuals.

Three male patients showed a peculiar beard growth. The hair grew slowly and remained restricted to the chin.

The systemic symptoms are as variable. In female patients they tend to be less serious than in the males, but some affected women suffer from serious painful paresthesia in the hands already in their youth.

In the male patients this symptom also shows much diversity. In most cases it is serious, in others it is slight and occurs only during fever or in extremely hot weather.

Neurological symptoms are predominant in some patients, but are absent in others. A 31-year-old man had never had any complaint. He suddenly had a paralysis of one leg. This subsided, but after several relapses he is now paraplegic. A 55-year-old woman died of a cerebellar hemorrhagia. The vessels of the cerebellum were stuffed with the lipid. The hemorrhagia probably resulted from this storage. A male patient developed an internal deafness when 35 years old.

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I think that the Dutch cases can be reduced to one or two mutations. The ancestors of two of the six families B and C lived in 1690 in the same hamlet in an isolated district near the German border. A third family (A) lived at the same time in the same vicinity. This area was thinly populated, so probably these three families are connected. This part of Holland was Roman-Catholic territory. The adjacent Dutch provinces were protestant except the region to the south.

The different religions formed a major impediment for social contacts, whereas the rivers between the two catholic areas did not raise an important obstacle. Now the fourth family (E) lived just over the Rhine, the fifth (F) a bit further on over the river Maas, all in the same catholic diocese.

The ancestors of an American pedigree described by Opitz et al. had emigrated from the same vicinity in 1856. It is highly probable that these families are connected.

M. Many old documents having been lost I cannot prove this hypothesis. The northern area where family A

B

C and E lived had close relationships with the adjacent German district. So maybe a connection can be found in Germany.