Genetic Aspects of the Thesaurismosis Lipoidica Hereditaria Ruiter-Pompen-Wyers (Angiokeratoma Corporis Diffusum Fabry)

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Wise (1962) and Opitz (1963) assumed that this lipoidosis is inherited in an X-linked manner. Opitz states that it is incompletely recessive.

To the pedigree I published elsewhere one patient (No. 31 in the pedigree) can be added, an 8-year-old girl, whose father (No. 18) shows the fully developed picture of the disorder. The girl has no

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symptoms,

no hpoid present. F 1/8. 1

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clinical symptoms, but in a biopsy from the skin of the hip the lipoid was easily demonstrable by post-chromatic Scarlett-red or Sudan-Black staining. Moreover by slit-lamp examination the oculist (Prof. Velzeboer) detected the characteristic corneal opacity. The ophthalmo-logical symptoms of all members of this family will be published in detail.

In the pedigree:

Transmission from father to son does not occur;

as far as examined no unaffected daughters from male patients have been found;

transmission from the mother is found to some of the sons as well as to some of the daughters.

These facts are in accordance with an X-linked inheritance, but I agree that more members of the family have to be examined to conclude with certainty to this mode of inheritance.

The lipoid is demonstrable in all patients male as well as female. The same holds true for the corneal opacities. The clinical manifestations however are scanty in most women. So the lipoidosis shows a dominant inheritance, but the clinical expressivity is feeble in most women.