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

\[ \begin{array}{c}
\text{male} \\
\text{female} \\
\text{affected} \\
\text{normal} \\
\text{patient} \\
\text{family member} \\
\text{diagnosis certain} \\
\text{symptoms} \\
\text{no lipoid present} \\
\text{clinical symptoms} \\
\text{in biopsy from skin of hip} \\
\text{post-chromatic Scarlett-red or Sudan-Black staining} \\
\text{by slit-lamp examination} \\
\text{characteristic corneal opacity} \\
\text{ophthalmological symptoms of all members of this family will be published in detail} \\
\text{in pedigree} \\
\text{transmission from father to son does not occur} \\
\text{no unaffected daughters from male patients have been found} \\
\text{transmission from the mother is found to some of the sons as well as to some of the daughters} \\
\text{facts are in accordance with an X-linked inheritance} \\
\text{I agree that more members of the family have to be examined to conclude with certainty} \\
\text{lipoid is demonstrable in all patients male as well as female. The same holds true} \\
\text{for corneal opacities. The clinical manifestations however are scanty in most women} \\
\text{lipoidosis shows a dominant inheritance} \\
\text{but the clinical expressivity is feeble in most women}.
\end{array} \]