Retinohepatoendocrinologic syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Retinohepatoendocrinologic syndrome. ORPHA:3087

Retinohepatoendocrinologic syndrome is characterized by total colorblindness caused by progressive cone dystrophy, degenerative liver disease, and endocrine dysfunction (hypothyroidism, diabetes, repeated abortions or infertility). It has been described in six females from two sibships with a high degree of consanguinity, and in a male from another family.