

Open Peer Review on Qeios

Norrie Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Norrie Syndrome</u>. NCI Thesaurus. Code C118634.

A rare, X-linked recessive inherited syndrome caused by mutations in the NPD gene. It is characterized by developmental retinal abnormalities that result in blindness in male infants at birth or soon after birth. Additional manifestations include progressive hearing loss and developmental motor skills delays.

Qeios ID: YUAESR · https://doi.org/10.32388/YUAESR