Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome. ORPHA:2274

Ichthyosis-hepatosplenomegaly-cerebellar degeneration syndrome is characterised by ichthyosis, hepatosplenomegaly and late-onset cerebellar ataxia. It has been described in two brothers. Transmission is either autosomal recessive or X-linked.