Open Peer Review on Qeios

Fucosidosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fucosidosis. ORPHA:349

Fucosidosis is an extremely rare lysosomal storage disorder characterized by a highly variable phenotype with common manifestations including neurologic deterioration, coarse facial features, growth retardation, and recurrent sinopulmonary infections, as well as seizures, visceromegaly, angiokeratoma and dysostosis.