Open Peer Review on Qeios

Sialuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Sialuria. ORPHA:3166

Sialuria is an extremely rare metabolic disorder described in fewer than 10 patients to date and characterized by variable signs and symptoms, mostly in infancy, including transient failure to thrive, slightly prolonged neonatal jaundice, equivocal or mild hepatomegaly, microcytic anemia, frequent upper respiratory infections, gastroenteritis, dehydration and flat and coarse facies. Learning difficulties and seizures may occur in childhood.