Galactosialidosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Galactosialidosis. ORPHA:351

Galactosialidosis is a lysosomal storage disease characterized by coarse facial features, macular ''cherry red spot'', and dysostosis multiplex. Clinical presentation can be heterogenous ranging from a severe, early-onset, rapidly progressive infantile form to late onset, slowly progressive juvenile/adult form.