Open Peer Review on Qeios

Classic homocystinuria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Classic</u> <u>homocystinuria</u>. ORPHA:394

Classical homocystinuria due to cystathionine beta-synthase (CbS) deficiency is characterized by the multiple involvement of the eye, skeleton, central nervous system, and vascular system.