Open Peer Review on Qeios

Aase-Smith syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Aase-</u> <u>Smith syndrome</u>. ORPHA:916

Aase-Smith syndrome type I is a very rare genetic disorder characterised by the following congenital malformations: hydrocephalus (due to Dandy-Walker anomaly), cleft palate, and severe joint contractures.