Baraitser-Winter cerebrofrontofacial syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Baraitser-Winter cerebrofrontofacial syndrome. ORPHA:2995

Baraitser-Winter syndrome (BWS) is a malformation syndrome, characterized by facial dysmorphism (hypertelorism with ptosis, broad bulbous nose, ridged metopic suture, arched eyebrows, progressive coarsening of the face), ocular coloboma, pachygyria and/or band heterotopias with antero-posterior gradient, progressive joint stiffening, and intellectual deficit of variable severity, often with severe epilepsy. Pachygyria - epilepsy - intellectual disability - dysmorphism (Fryns-Aftimos syndrome (FA); see this term) corresponds to the appearance of BWS in elderly patients.