Open Peer Review on Qeios

Occipital pachygyria and polymicrogyria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Occipital pachygyria and polymicrogyria. ORPHA:280640

Occipital pachygyria and polymicrogyria is a rare, genetic, cerebral malformation characterized by the presence of cortical smoothening with loss of secondary and tertiary gyri, associated with an excessive number of small, irregular gyri with increased cortical thickness, located in the occipital lobes. Patients usually present with seizures (including myoclonic-astatic, absence, atypical absence, vision loss, myoclonic-atonic, generalized tonic-clonic) and variable (absent to moderate) developmental and/or intellectual delay.