Craniotelencephalic dysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Craniotelencephalic dysplasia. ORPHA:1528

Craniotelencephalic dysplasia is an extremely rare, genetic developmental defect during embryogenesis syndrome characterized by craniosynostosis with frontal encephalocele and various additional brain anomalies (severe hydrocephalus, agenesis of the corpus callosum, lissencephaly and polymicrogyria, parenchymal cysts, septo-optic dysplasia) resulting in marked cerebral dysfunction, seizures and very severe psychomotor delay. There have been no further descriptions in the literature since 1983.