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Brief clinical report: polymicrogyria in DiGeorge syndrome

Nadia, Fida; Caroline, Robson; Virginia, Kimonis.

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ABSTRACT

We report a girl with DiGeorge syndrome with a VSD, cleft of the soft palate, and developmental delay. Brain imaging revealed diffuse polymicrogyria, diffuse cerebral atrophy, and ventriculomegaly. A wide range of central nervous system abnormalities have been reported in DGS and VCFS, including gyral abnormalities [lissencephaly, microgyria]. The patient reported herein strengthens the association between the 22q11 deletion spectrum and cortical dysgenesis, but the underlying pathogenetic mechanism [primary neural migration vs. vascular disruption] remains unclear

Subject(s)

Humans; Female; Brain/abnormalities; Ultrasonography; Craniofacial Abnormalities; Heart Defects, Congenital