CTNNB1 syndrome

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CTNNB1 [OMIM *116806] encodes β-catenin, an integral part of the cadherin/catenin complex, which functions as effector of Wnt signaling. CTNNB1 is highly expressed in brain as well as in other tissues, including heart. Heterozygous CTNNB1 pathogenic variations are associated with a neurodevelopmental disorder characterized by spastic diplegia and visual defects (NEDSDV) [OMIM #615075], featuring psychomotor delay, intellectual disability, behavioral disturbances, movement disorders, visual defects and subtle facial and somatic features. We report on a new series of 19 NEDSDV patients (mean age 10.3 years), nine of whom bearing novel CTNNB1 variants. Notably, five patients showed congenital heart anomalies including absent pulmonary valve with intact ventricular septum, atrioventricular canal with hypoplastic aortic arch, tetralogy of Fallot, and mitral valve prolapse. We focused on the cardiac phenotype characterizing such cases and reviewed the congenital heart defects in previously reported NEDSDV patients. While congenital heart defects had occasionally been reported so far, the present findings configure a higher rate of cardiac anomalies, suggesting dedicated heart examination to NEDSDV clinical management.