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A *CRELD1* GENE VARIANT LEADS TO ATRIOVENTRICULAR SEPTAL DEFECTS IN DOWN SYNDROME

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Congenital heart defects (CHD) are seen in around 40% of the Down syndrome (DS) patients. Atrioventricular Septal Defect (AVSD) or endocardial cushion defect is commonest form of CHD in these children. *CRELD1* gene is implicated in causation of sporadic AVSD. In the present study, we evaluated the association and significance of *CRELD1* variants with AVSD in Down syndrome (DS) patients. Sequencing was done in blood samples from 3 groups: group I (DS with AVSD), group II (DS without AVSD) and group III (non-syndromic AVSD cases). Twenty two variants in *CRELD1* gene were identified, comprising of sixteen novel and six previously reported variants. However, on the basis of sequence, as well as structure analysis, the variant c.973G > A(p.Glu325Lys) variant was identified only in DS having AVSD group which was predicted to have significant effects on calcium binding of putative *CRELD1* protein. Since *CRELD1* gene acts as a regulator of calcineurin/NFATc1 signaling which is crucial for the regulation of cardiac development by dephosphorylation of the transcription factor, NFAT (nuclear factor of activated T cells),in cytoplasm, the variation in cb-EGF-like calcium binding domain in *CRELD1* protein is likely to have pathogenic consequences. Thus, we conclude that the *CRELD1* gene is likely to have a major role in causation of AVSD phenotype in selected DS patients.

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