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Truncation and micro-deletion of *EVC* accompanied by a novel *EFCAB7* missense mutation in Ellis-Van Creveld syndrome with a typical congenital heart defect

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Ellis-van Creveld syndrome (EvC) is aciliopathy with cardiac anomalies, disproportionate short stature, polydactyly, dystrophic nails and oral defects. Approximately 60% of EvC patients have severe congenital heart defects (CHD), of which more than half are atrio-ventricular septal defect and common atrium. In this study, we report one EvC Vietnamese family with an atypical CHD phenotype, short chordalis. A 32-month-old boy had a novel heterozygous *EVC* mutation (c.1717C>G-p.S572X) in exon 12, inherited from his father whose phenotype was milder than his son's. Of note, the mother without an EvC phenotype showed a lower expression of *EVC* mRNA compared with controls. SNP array analysis revealed that the patient and mother had a heterozygous 16kb deletion in *EVC*, ranging from intron 9 to intron 11. As the patient and the father had an atypical CHD, we screened *EFCAB7* and *IQCE* as the candidate for modifiers of EvC phenotype. EFCAB7 and IQCE are ciliary proteins, which positively regulate the Hh pathway and anchor the EVC-EVC2 complex in a signaling micro-domain at the base of cilia. Anovel missense mutation c.1171T>C-p.Y391H in *EFCAB7* was found in the patient and the father. This mutation located in a possible binding site of EFCAB7 and EVC2 and may have modified the EvC phenotype in this family. Our findings suggested the physiological role of EFCAB7 in cardiac development.

Biography

Tran Quynh Nhu Nguyen graduated from University of Medicine and Pharmacy, Ho Chi Minh city, Vietnam in 2007. Since 2008, she has worked as cardiologist in Children's Hospital 2, Ho Chi Minh city, Vietnam. She has completed Master course from School of Medicine, The University of Tokyo, Japan in March 2015 and at present continues PhD course in the same university.

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