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The popeye domain containing genes and tetralogy of fallot

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease in live infants. The abnormality of TOF is characterized by a gross structural abnormality of the heart development and with ~1 in 3,000 live newborns. 70% of TOF cases also occur with unknown causes. To date, a few causative genes of TOF have been identified, including GATA4. The POPDC family with highly conserved Popeye domain and three transmembrane domain consists of three members, *POPDC1* (Bves), *POPDC2* and *POPDC3*. The POPDC genes are strongly expressed in the embryonic heart, suggesting their potential function in heart development and cardiac diseases. Indeed, it has been found that the expression of *POPDC1* is altered in patients with congenital septal defects, and missense mutations of *POPDC1* and microdeletions of *POPDC3* locus have been found in TOF. Also in our preliminary test, a missense mutation of *POPDC3* was detected in sporadic TOF patients via exon sequencing. TOF is often caused by dysfunction of secondary heart field cardiac progenitor cells during early development stages or perturbation of morphogenesis of OFT, a SHF derived structure. Though POPDC has been indicated in TOF, it is an intriguing question that how the POPDC transmembrane protein regulates the important transcription factor in nucleus to play a pivotal role in the early regulation of cardiac progenitor cells. Here we will present a working model for TOF beginning with the POPDC family through GATA4.

Biography

Xiushan Wu has completed his PhD from Stockholm University during 1986-1990, Postdoctoral studies from Michigan University from 1990-1994 and worked as Scientist in Karolinska Institute from 1994-2000. He is the Director and Professor of The Center for Heart Development, Deputy Director of the Key Laboratory of the Ministry of Education of China for Developmental Biology and Protein Chemistry, Hunan Normal University. His research focuses on understanding the mechanisms by which embryonic heart is developed using *Drosophila*, zebrafish and mice as models. He has published more than 350 papers including over 110 SCI papers.

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