

HAND2 Mutation Detection in Tricuspid Atresia Patients.

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Tricuspid Atresia (TA) is a congenital heart disease in which the tricuspid valve is missing or abnormally developed. The defect blocks blood in the right atrium from flowing directly into the right ventricle. It is an uncommon form of congenital heart disease that affects about 5 in every 100,000 live births. While the cause of TA is unknown, the lab data shows that in mice loss of transcription factor Hand2 function within a population of cells that line the inside of the heart (the endocardium) results in a TA phenotype. Hand2 is a protein that belongs to the basic helix-loop-helix family of transcription factors, and has been shown to play many different roles in embryonic development. To test whether loss of Hand2 function in humans results in TA, sequencing the *HAND2* gene in 25 TA patients. Polymerase Chain Reaction (PCR) was used to amplify the TA patient Hand2 alleles. A TOPO reaction was then performed to insert the amplicons into a sequencing plasmid, followed by a transformation and minipreps to isolate individual clones. Isolated Hand2 alleles within the TOPO sequencing plasmid were sent to a sequencing core facility. In this manner the Hand2 DNA sequence for several patients was obtained and analyzed for mutations. This project will shed light on the cause of TA. Further research is currently in progress in Dr. Firulli's Lab.

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