Studying Cardiovascular Development in Mice that Mimic Human DiGeorge Syndrome

Sarah A. Elliott (Anne M. Moon)
Department of Pediatrics, University of Utah

American Heart Association estimates that there are over one million people in the U.S. living with congenital heart disease, and in 2002, more than four thousand Americans died from such defects. The Moon Laboratory is studying certain types of congenital heart disease in an effort to better understand normal and abnormal cardiovascular development. We believe that increasing our knowledge of heart and vessel development will enable us to better treat or even prevent congenital heart disease.

Currently, we are studying mice with mutations in the Fibroblast Growth Factor 8 (Fgf8) gene. These Fgf8 gene mutations cause birth defects, including heart malformations similar to those observed in human DiGeorge Syndrome patients. We are trying to determine how FGF8 signaling pathways contribute to heart and vessel development.

The Moon Lab has created several mouse lines as tools for studying the function of FGF8 in the developing mouse embryo. I am currently testing a new mouse line to see if it can be used to successfully "knock out" FGF8 (i.e., make it non-functional) in an embryonic tissue layer called the endoderm, which is where we believe FGF8 plays an important role in patterning the great vessels that exit the heart.

Digital Image © 2006 University of Utah. All rights reserved.