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Abstract:

Over the last few decades, the study of congenital heart disease (CHD) has benefited from various model systems and the development of molecular biological techniques enabling the analysis of single gene as well as global effects. In this chapter, we first describe different models including CHD patients and their families, animal models ranging from invertebrates to mammals, and various cell culture systems. Moreover, techniques to experimentally manipulate these models are discussed. Second, we introduce cardiac phenotyping technologies comprising the analysis of mouse and cell culture models, live imaging of cardiogenesis, and histological methods for fixed hearts. Finally, the most important and latest molecular biotechniques are described. These include genotyping technologies, different applications of next-generation sequencing, and the analysis of transcriptome, epigenome, proteome, and metabolome. In summary, the models and technologies presented in this chapter are essential to study the function and development of the heart and to understand the molecular pathways underlying CHD.