

Genetics of fetal cardiac defects

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Fetal cardiac defects are being increasingly picked up with advances in fetal imaging technology and expertise. Most severe abnormalities are being picked up in first trimester. So knowledge on basis of these abnormalities becomes important in testing and counseling families. The development of the heart is very intricate and improper switch on and off of these genes lead to errors which are generally sporadic in nature. Errors in early cell division can cause chromosomal imbalances.

Familial problems could occur due to errors in chromosomes, genes, mitochondria or can be multifactorial. Not all errors are genetic and one should consider teratogens like maternal diabetes, drugs as causative in some situations. Different case scenarios and how to work up for identifying the genetic basis will be amply illustrated in the lecture.