**CLINICAL GENETICS OF DILATED CARDIOMYOPATHY- TESTING WISELY**

**J.D. Marsh**

University of Arkansas for Medical Sciences, Little Rock, AR, USA

Dilated cardiomyopathies (DCM) account for 10,000 deaths/year in the US; African-Americans have a 3-fold increased risk. There has been little solid information available on or the ability to make a precise genetic diagnosis in DCM. Moreover, little has been known about the prognostic or potential therapeutic advantage of genetic testing. Recent work indicates that mutations in the gene encoding the Titin protein account for up to 20% of DCM cases, with mutations in an additional 30 genes accounting for 15% of additional cases. At this point, genetic testing must be applied selectively. Several clinical factors can identify patients for whom the yield on testing will be higher, including absence of hypertension, positive family history of DCM, sudden cardiac death, elevated CK, associated muscular dystrophy, and conduction abnormalities on ECG. There are 5 – 10% of patients with DCM who have two or more likely pathogenic mutations, which may account for variance in penetrance and phenotype. High risk mutations for premature death include mutations in LMNA, DES and RBM20 genes. Truncating mutations in the Titin gene appear to account for 15% of peripartum cardiomyopathies. Presence of Titin mutations has prognostic importance. Selective testing with targeted gene panels is appropriate for patients with DCM who have increased likelihood of a pathogenic mutation. There are important therapeutic implications (ICD’s) and also potential for effective disease modifying therapy.