Condition: Cardiomyopathy

Inheritance:

Genetically heterogeneous.

Genetic etiology:

Multiple genes have been implicated in the etiology of the various forms of cardiomyopathy. Familial dilated cardiomyopathy is associated with mutations in genes that encode cardiac actin, desmin, g-sarcoglycan, b-sarcoglycan, cardiac troponin T, a-tropomyosin, b-myosin heavy chain, and cardiac myosin-binding protein C gene. Hypertrophic cardiomyopathy is associated with mutations in genes that encode components of the sarcromere: b-myosin heavy chain, cardiac regulatory and essential myosin light chains, myosin binding protein C, a-cardiac actin, a-tropomyosin, titan, and cardiac troponins T, I, and C.

Frequency:

Dilated cardiomyopathy: 1/3,000; Hypertrophic cardiomyopathy: 1/500.

Clinical features:

Cardiomyopathy is a disorder of the function of cardiac muscle, and is divided into dilated and hypertrophic forms. In dilated cardiomyopathy there is an increase in left ventricular end diastolic diameter with reduced function. In hypertrophic cardiomyopathy, myocytes are hypertrophic and there is cardiac fibrosis with left ventricular outflow obstruction. Each is etiologically heterogeneous, including environmental and genetic contributions. In addition, cardiomyopathy can occur in isolation or as part of a more complex syndrome.

Management:

Treatment of symptoms of heart failure; cardiac transplant.

Genetic counseling:

Based on establishment of specific genetic diagnosis; many of the causative genes can be tested at the molecular level.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.