Desmin is a type III intermediate filament protein which is expressed in skeletal, cardiac, and smooth muscle cells. Desmin interacts with other intermediate filament proteins, as well as microfilaments and microtubules, to form the cytoskeleton of the cell. In addition, desmin and other intermediate filament proteins work together to maintain the proper connection between the contractile apparatus and the cytoskeleton. Desmin is encoded by the gene DES, which is composed of 9 exons and is located at 2q35.

Mutations in DES have been described for a spectrum of diseases. Desminopathy is typically characterized by distal muscle weakness and atrophy that usually begins in the lower limbs and spreads to the hands and arms, truncal, neck-flexor, and sometimes facial muscles. Desminopathy can also result in swallowing and respiratory impairment. The spectrum of phenotypes resulting from desmin mutations is broad ranging from skeletal myopathy with no cardiac involvement to skeletal myopathy with cardiomyopathy, to cardiomyopathy without skeletal muscle involvement. Cardiac manifestations can include dilated cardiomyopathy, arrhythmias, and conduction block. Autosomal dominant and recessive DES mutations have been identified in this disease spectrum and include missense, nonsense, splicing and small deletion or insertion mutations.

The John Welsh Cardiovascular Diagnostic Laboratory offers molecular genetic testing for DES mutations. Individuals will be tested by automatic fluorescent DNA sequencing of all 9 exons of the DES gene. We strongly recommend initial testing of an affected individual, if available, in order to provide the greatest test sensitivity and clearest interpretation of results for subsequent family members. Genetic counseling is recommended for all individuals in order to identify additional at-risk family members and to discuss reproductive issues.