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Superoxide Dismutase–A Genetic Marker in Cardiomyopathies

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ABSTRACT Cardiomyopathies are a heterogeneous group of heart muscle disorders responsible for a great deal of morbidity and mortality. Dilated, hypertrophic and restrictive are the three major categories of cardiomyopathies. Since reactive oxygen species has been implicated in wide range of genetic disorders and the role of antioxidant like superoxide dismutase as a scavenger has been highlighted, the present study envisages on identifying the specific electromorphic association of superoxide dismutase with cardiomyopathies and to delineate the genetic heterogeneity based on specific alleles at risk. Phenotyping was carried out on 8% PAGE of the red cell membrane samples following Davies (1964) and Beauchamp's (1975) protocols. Blood samples from 62 dilated cardiomyopathy, 80 hypertrophic cardiomyopathy and 86 healthy individuals were collected from CARE Hospitals and voluntary blood donor camps, Hyderabad. Significant association of homozygous SOD A*1 (c^2 = 7.58) alleles with dilated cardiomyopathy and heterozygous SOD A*2-1 (c^2 = 5.89) alleles with hypertrophic cardiomyopathy was observed, resulting in significant deviation of allelic frequency in cardiomyopathy group compared to the control group highlighting that individuals carrying SOD A*1 (c^2 = 7.588) and SOD A*2-1 (c^2 = 5.89) alleles may be at a greater risk for dilated and hypertrophic cardiomyopathy respectively. Thus SOD as a genetic marker may help in risk prediction and in delineating genetic heterogeneity of the condition and the role of superoxide dismutase with specific electromorphic association in influencing endogenous nitric oxide production and energy pathways by altering the structure and function of the cell membranes is discussed.

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