GENETICS AND GENODERMATOSES

"CARVAJAL SYNDROME: A CARDIOCUTANEOUS SYNDROME"

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Background: Carvajal syndrome also known as 'Striate palmoplantar keratoderma is a familial cardiocutaneous syndrome that consists of wooly hair, palmoplantar keratoderma and cardiomyopathy, inherited in an autosomal recessive pattern due to a defect in desmoplakin gene which is an intracellular protein that links desmosomal adhesion molecules to intermediate filaments of cytoskeleton.The skin disease presents as a striate palmoplantar keratoderma particularly at sites of pressure, Altered protein-protein interactions at intercalated discs cause both contractile and electrical dysfunction of heart and the patient is at risk of sudden cardiac death due to dilated cardiomyopathy associated with this entity.

Observation: Here we present a case of A 10-year-old male child, born of second degree consanguineous marriage presented with wooly hair since birth and thickening of palms and soles since 6 years. On examination, Pulse rate-98 beats per minute, Regular Rhythm Patient had fine brittle lustreless wooly scalp hair, Eyelashes were also curled, Linear band of Hyperkeratotic skin present over palms, Focal areas of hyperkeratotic skin over the soles.Echo cardiography-Dilated left ventricle Mildly reduced global LV function(EF-45%), Histopathology was suggestive of epidermolytic keratoderma showing hyperkeratosis and papillomatosis .A diagnosis of Carvajal syndrome was made as there was wolly hair,left heart invovlement and palmo plantar keratoderma.

Key message: Whenever a child presents with wooly hair palmoplantar keratoderma and cardiac symptoms ,Naxos syndrome and Carvajal syndrome should be considered. The primary goal of the management is to prevent sudden cardiac death. Implantation of automatic cardioverter defibrillator, antiarrhythmic drugs and management of heart failure are the recommended treatment modalities.