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A novel mutation in desmoplakin (DSP) gene in two siblings with Carvajal Syndrome

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Introduction: Dilated cardiomyopathy is the most common form of cardiomyopathies and it may be a component of cardiocutaneous syndromes. Carvajal syndrome is a cardiocutaneous syndrome characterized with dilated cardiomyopathy, woolly hair and keratoderma (1). Here a new homozygote frameshift mutation in *desmoplakin (DSP)* gene detected in two female siblings with diagnosis of Carvajal syndrome is presented.



Case: A five-year-old female patient, first child of parents with second-degree consanguinity, admitted with complaints of malaise and abdominal pain. She had findings of overt congestive heart failure, peculiar woolly hair, and palmar keratoderma and was diagnosed as DCM. After

being treated with pharmacological agents for congestive heart failure for two years; left ventricular assist device (LVAD) was implanted and on the 501th day of LVAD implantation, the patient had heart transplantation. The sister of the patient also had peculiar woolly hair, palmoplantar keratoderma and DCM. She is being treated with pharmacological agents for congestive heart failure. Genetic screening of both siblings were performed with suspicion of Naxos/Carvajal disease and revealed a new homozygote frameshift mutation, *c.4650_4651delTG (p. V155Efs*75)*, in *desmoplakin (DSP)* gene. (Figure)

Discussion: Cardiocutaneous syndromes are a group of syndromes associated with DCM and other types of cardiomyopathies. Dilated cardiomyopathy with woolly hair and keratoderma (DCWHK), also known as Carvajal disease, is an autosomal recessive cardiocutaneous syndrome caused by mutations in *DSP* gene which encodes desmoplakin, on chromosome 6p24 (1,2). Mutations in 10 different desmosomal proteins and more than 40 different mutations in desmoplakin leading to distinct cardiac and cutaneous features have been identified so far (3). In our case genetic screening of both siblings revealed a new homozygote frameshift mutation, *c.4650_4651delTG (p. V155Efs*75)*, in *desmoplakin (DSP)* gene.

Conclusions: Genetic screening is an important tool for early diagnosis and for predicting the presence of disease in asymptomatic family members. As hereditary diseases are an important cause of DCM, early diagnosis of diseases by genetic screening may be life saving for patients. Detection of new mutations and prenatal genetic counseling may help parents in decision of future children.

References

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