Characteristics of children with familial and idiopathic dilated cardiomyopathy

Dilated Cardiomyopathy (DCM) is a disease characterized by weakening of the heart muscle that becomes dilated and unable to pump blood efficiently. DCM is diagnosed in 0.57 of 100,000 children per year in North America and has a 5-year risk of death or heart transplantation of 46%.

DCM in children has a wide spectrum of causes: infection, neuromuscular disease (eg. Duchenne's muscular dystrophy), metabolic, familial, endocrine disease, cardiotoxic drugs, etc. Although many cause of DCM can be identified, nearly 70% of cases are categorized as idiopathic (no cause found). Over the past 30 years, survival in DCM has improved very little and this is due to the introduction of heart transplantation. Previous studies have shown that 5-year survival of children with DCM varied depending on the cause, with best clinical outcomes observed in children with familial DCM (FDCM) (94%). Since the outcomes of DCM is related to the pathogenesis, we hypothesized that transplant free survival is better in children with FDCM than Idiopathic DCM (IDCM).

We queried the database of the National Heart, Lung And Blood Institute funded Pediatric Cardiomyopathy Registry (PCMR) to analyse data of children with FDCM and IDCM. We included all children under 18 years of age with a DCM diagnosis: presence of left ventricle dilatation and systolic dysfunction. FDCM was defined by the presence of a clinical diagnosis of FDCM documented in the medical record or a documented family history (FH) of one or more affected family members. As of 2013, PCMR had enrolled 1834 children with DCM, 223 with FDCM and 647 with IDCM.

We found that children with FDCM were significantly older and less likely to have symptoms of heart failure (HF) at the time of diagnosis and less likely to have a FH of sudden death. Although we identify a lower pre-transplant death in children with FDCM than IDCM, after adjustment for the presence of HF at diagnosis we did not identify a significant difference. There was no significant difference between the two groups in the incidence of transplant or the combination of death/transplant. Risk factors for death and transplant included: older age at diagnosis, presence of HF symptoms at diagnosis and left ventricle dilation.

Comparing FDCM and IDCM we did not find a significant difference in outcome; we did find that presentation over one year of age increases the risk of death/transplant by 40%; also having a dilated heart and use of antiarrhythmic drugs was associated with increased risk of death/transplant. The cause of CMP, familial or idiopathic is not a significant factor in determining outcome; clinical characteristics are the major determinant.

In our study the diagnosis of FDCM was based on FH. Although an accurate FH is fundamental in the diagnosis and has to be revisited at every clinical encounter, it is imperative that immediate

relative of the affected person get screened with echocardiogram allowing for identification of asymptomatic patients. Large panel of genetic testing are also available and more affordable than in the past. Diagnosis of FDCM by genetic testing allows for identification of carriers of the disease and for proper monitoring of the subjects at risk of developing the disease.

In summary recognition of FDCM is important because of the implications for screening other family members, but not for the management of the affected individual as the outcomes are related to patient factors not whether the child has FDCM or IDCM.

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Publication

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