P-54

Prognostic factors in paediatric restrictive cardiomyopathy

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Introduction:
Restrictive cardiomyopathy (RCM) is the rarest and most serious form of heart muscle disease, accounting for 2-5% of pediatric cardiomyopathies. Identification of prognostic factors is essential to identify high-risk patients that need to be addressed for early heart transplantation.

Methods:
We conducted a single-center retrospective study. We enrolled 47 patients that had a diagnosis of RCM between 1996 and 2017. The clinical, ultrasonography, and hemodynamic data were collected at the time of diagnosis, as well as genetic analysis in order to identify early predictors of poor prognosis (transplantation or death). We then compared the evolution according to the type of RCM (pure or hypertrophic cardiomyopathy with restrictive phenotype HRCM), and to the age group (<5 years, 5-11 years, > 11 years).

Results:
47 patients were included, with a gender ratio of 1.76 (F/M). The mean age at diagnosis was 5.9 years. 57.5% had a pure RCM and 42.5% an HRCM. The median follow-up was 10.2 months. Overall survival at 1, 2 and 5 years was 43% 35% and 15% respectively. There was no statistically significant difference in terms of survival between RCM and HCMR (p=0.651) and between age groups (p=0,582). Clinical findings at diagnosis were dominated by congestive signs (63.9%), which were related to a poor outcome (p=0,041). The relationship between a mildly altered left ventricular ejection fraction (LVEF 45-55%) and mortality was statistically significant (p=0.01). Imaging findings didn't show a late gadolinium enhancement and cavities dilatation were note associated with mortality. Regarding catheterism data, neither cardiac output nor pulmonary vascular resistance nor pulmonary capillary pressure were related to a poor outcome. The rentability of family screening was 60%: we identified one homozygous and 4 heterozygous mutations on the TNNI3 gene including a neomutation, and 1 mutation on MYH7 gene.

Conclusion:
Adverse prognostic factors were dominated by the presence of congestive signs at diagnosis and mild level of left ventricular dysfunction. Patient with these factors should be addressed for Heart transplantation as soon as diagnosed. Autosomic dominant mutation in TNNI3 was the most frequent genetic finding in this population. Family screening and genetic counseling is thus important.