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Chronological change of the interventricular septal thickness in hypertrophic cardiomyopathy associated with inborn errors of metabolism.

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Introduction:
It has been demonstrated that hypertrophic cardiomyopathy (HCM) associated with inborn errors of metabolism has poor outcome with the mortality rate exceeding 50% at 2 years. However, the clinical characteristics of patients with HCM and inborn errors of metabolism who survive their infancy are not well described.

Methods:
The study included 54 patients with inborn errors of metabolism who underwent echocardiography at our institution from January 2009 to August 2017. The end-diastolic interventricular septal thickness at the latest examination was retrospectively collected and standardized using z-scores based on body surface area. The diagnosis of HCM was made if the septal thickness z-score exceeded +2. For patients with HCM, the chronological changes of the septal thickness were also evaluated.

Results:
Of 54 patients, 45 patients were diagnosed with mucopolysaccharidosis, 5 with mucolipidosis (including 2 with mucolipidosis type II), and 4 with glycogen storage diseases. Patients were 13.7 (range, 0.3 to 52.3) years at the latest examination. Overall, HCM was present in 18 (33%) patients. The frequency of HCM was 26% in mucopolysaccharidosis, 80% in mucolipidosis, and 50% in glycogen storage diseases. Chronological changes of the septal thickness z-scores were available in 10 patients with mucopolysaccharidosis, 3 with mucolipidosis, and 2 with glycogen storage diseases, which were presented in Figure. The annual increase rates of septal thickness z-scores were 0.3 (range, –0.2 to 0.7) in mucopolysaccharidosis, 1.4 (range, 1.3 to 1.7) in mucolipidosis, and 0 (range, –0.1 to 0.1) in glycogen storage diseases ($p < 0.05$ by Kruskal-Wallis test). Of note, rapidly progressive HCM was observed in both patients with mucolipidosis type II.

Conclusions:
Left ventricular hypertrophy was prominent and progressed rapidly in patients with mucolipidosis type II. In other forms of inborn errors of metabolism, left ventricular hypertrophy progressed more slowly. The study demonstrates that there are distinct differences in terms of frequency and severity of HCM among patients with inborn errors of metabolism. The plan for follow-up echocardiography would need to be optimized according to the diagnosis.