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

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1, Introduction

- Hypertrophic cardiomyopathy (HCM) associated with inborn errors of metabolism has poor outcome with the mortality rate exceeding 50% at 2 years since diagnosis.
- However, the clinical characteristics of HCM patients associated with inborn errors of metabolism who survive their infancy are not well described.

2, Methods

Study 1 : 54 patients have inborn errors of metabolism.

- They underwent echocardiography at our institution **from January 2009 to August 2017**.
- Their echo date were **retrospectively** collected at last follow-up visit, and standardized using z-scores based on body surface area.
- The **degree of end-diastolic interventricular septal (IVSd) thickness** were evaluated.

Study 2 : 18 patients were diagnosed as HCM.

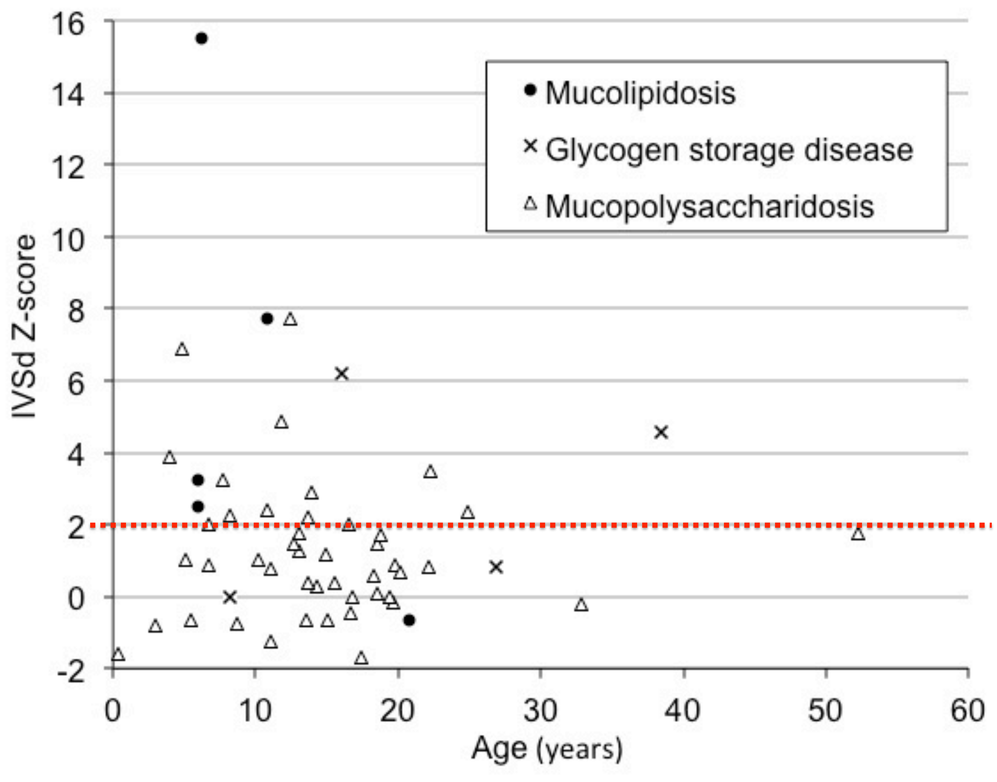
- Diagnosis of HCM is defined as IVSd Z-score > +2.
- In the HCM patients, the **chronological changes of IVSd Z-score** were also evaluated.
- The Kruskal-Wallis test was used to examine the annual increase rates of IVSd z-scores.

3, Results

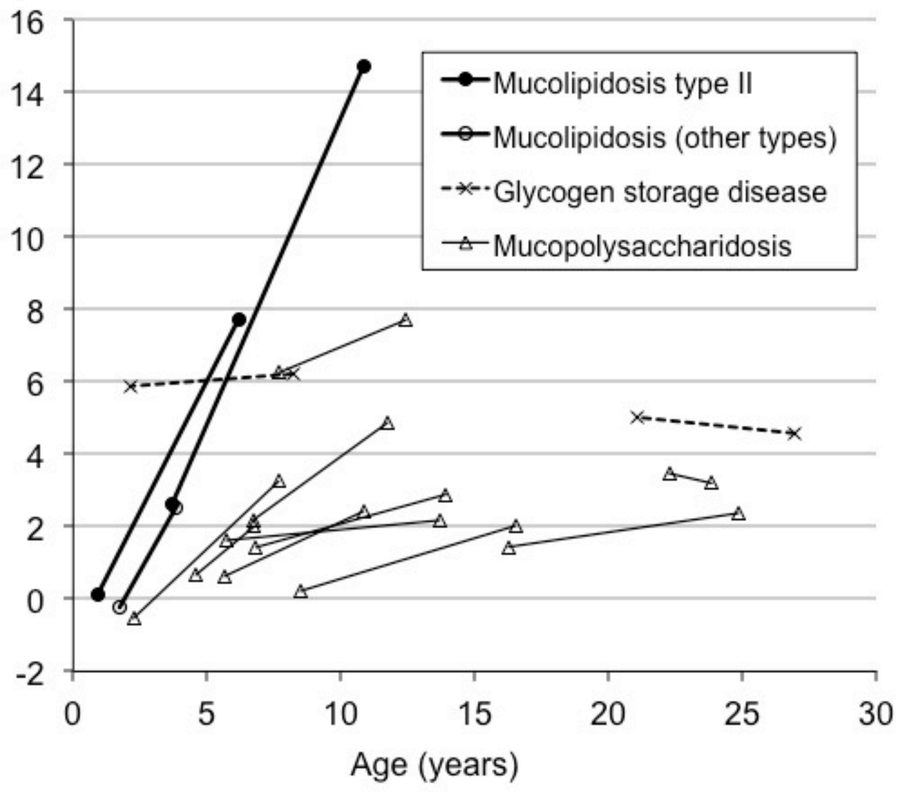
Characteristics of the patients

Diagnosis	n	age at examination(years)	IVSd Z-score	HCM patients	Annual increase rate of IVSd Z-score in HCM
Inborn errors of metabolism	54	13.6(0.2-52.3)	+1.0(-1.6 - +15.7)	18(33%)	
Mucolipidosis	5	6.2(6.0-20.7)	+3.2(-0.7 - +15.7)	4(80%)	+1.4 (+1.3 - +1.7) : (p<0.005)
Mucolipidosis Ⅱ(I-cell disease)	2	8.5(6.1-10.8)	+11.9(+7.7 - +15.7)	2(100%)	
Mucolipidosis Ⅲ	3	6(6-20.75)	+2.5(-0.6 - +3.2)	2(66%)	
Glycogen storage disease(GSD)	4	21.5(8.3-38.5)	+2.7(0 - +6.2)	2(50%)	0(-0.1 - +0.1)
GSD type Ⅱ(Pompe disease)	2	17.5(8.2-26.9)	+0.4(0 - +0.8)	0(0%)	
GSD type Ⅲ	2	27.2(16.0-38.5)	+5.3(+4.5 - +6.2)	2(100%)	
Mucopolysaccharidosis(MPS)	45	13.6(0.3-52.3)	+0.9(-1.6 - +7.7)	12(26%)	+0.3 (-0.2 - + 0.7)
MPS Ⅰ(Hurler syndrome)	6	13.3(5.5-17.4)	-0.2(-1.6 - +2.2)	1(16%)	
MPS Ⅱ(Hunter syndrome)	30	13.5(0.3-32.8)	+0.9(-1.5 - 6.9)	10(33%)	
MPSⅢ(Sanfilippo syndrome)	4	12.4(5.0-18.5)	+1.0(+0.1 - +7.7)	1(33%)	
MPSⅣ(Morquio syndrome)	6	18.7(13.6-52.3)	+1.7(+0.4 - +3.4)	1(20%)	
MPSⅥ(Maroteaux-Lamy syndrome)	1	13.5	-0.6	0(0%)	

Study 1 : Degree of IVSd Z-score in all IEM patients



Study2 : Chronological changes of IVSd Z-score In HCM patients



4, 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.**