

The experience of being diagnosed with hypertrophic cardiomyopathy through family screening in childhood and adolescence

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Introduction: Hypertrophic cardiomyopathy (HCM) is a hereditary disease and the most common medical cause of sudden death in childhood and adolescence. This is the reason for recommending screening in children with an affected first degree relative. A diagnosis of HCM implies lifestyle modifications, restrictions that may bring profound changes to the daily life of the affected individual. The aim was to describe the experiences of children and adolescents at being screened positive for HCM and how this impacts on daily life.

Methods: Descriptive qualitative interview study. Thirteen asymptomatic children or adolescents diagnosed with HCM through family screening were interviewed 12–24 months after the diagnosis. Analysis was conducted with qualitative content analysis.

Results: Children described an involuntary change, which affected their daily life with limitations and restrictions in life, both in the individual and social context. Life-style recommendations had the most severe impact on daily life and affected their social context. They tried to navigate in a world with new references and after reorientation they felt hope and had faith in the future. **Conclusions:** Children diagnosed with HCM through family screening went through an involuntary change resulting in limitations and restrictions in life. This study indicates that there is a need for support and health professionals have to consider the specific needs in these families. Our findings thus give guidance in how best to improve support to the patients and their family. Diagnosis in asymptomatic children should be accompanied by ideally multiprofessional follow-up, focusing not only on medical issues.