

Screening for Hypertrophic Cardiomyopathy in Asymptomatic Children and Adolescents

Psychosocial consequences and impact on quality of life and physical activity

Akademisk avhandling

Som vid avläggande av medicine doktorsexamen vid Sahlgrenska akademien vid Göteborgs universitet kommer att offentligen försvaras i föreläsningssal 1 på Drottning Silvias Barn och Ungdomssjukhus, Göteborg, fredagen 28 oktober 2011 kl 13.00.

av

Ewa-Lena Bratt

Fakultetsopponent: Professor Britt-Marie Ternestedt, Institutionen för Vårdvetenskap, Ersta Sköndal högskola, Stockholm

Avhandlingen baseras på följande arbeten:

- I. Bratt E-L, Östman-Smith I, Sparud-Lundin C, Axelsson Å B. Parents' experiences of having an asymptomatic child diagnosed with hypertrophic cardiomyopathy through family screening. *Cardiol Young* 2011 Feb; 21(1):8-14.
- II. Bratt E-L, Östman-Smith I. Selective or non-selective high-dose beta-blockade – evaluation of exercise capacity in children and adolescents with hypertrophic cardiomyopathy. Submitted for publication
- III. Bratt E-L, Östman-Smith I, Axelsson Å B, Berntsson L. Quality of life in asymptomatic children and adolescents before and after diagnosis of hypertrophic cardiomyopathy through family screening. Submitted for publication.
- IV. Bratt E-L, Sparud-Lundin C, Östman-Smith I, Axelsson Å B. Children's and adolescents experience of being diagnosed with hypertrophic cardiomyopathy through family screening. Submitted for publication.

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ABSTRACT

The aim of this thesis was to describe the consequences of being diagnosed with hypertrophic cardiomyopathy (HCM) while being asymptomatic, taking into consideration psychosocial effects and medical aspects of diagnosis and treatment.

Methods: Quality of life (QoL) was measured according to Lindström before the diagnosis, and after two years, comparing with healthy controls. Psychosocial consequences of the diagnosis were explored in interviews with children and their parents and analysed using content analysis. Exercise performance was measured at baseline and after one year in patients randomized to no pharmacological treatment or selective or non-selective high-dose beta-blocker therapy.

Results: The total QoL score was similar in both groups at baseline and at follow-up. Parents described an immediate reaction of shock, grief and feelings of injustice but were also grateful that their child had been diagnosed and was still asymptomatic. The diagnosis resulted in a change in life-style for most families due mainly to restrictions of sports activities. Parents had difficulties to adapt to the new life but after re-adjustment they regained hope and confidence. The children described an involuntary change of their daily life with limitations and restrictions because of life-style recommendations and this also affected their social context. However, after a reorientation process they felt hope and had faith in the future. There was no significant difference in exercise capacity between the groups at baseline, or after one year of observation versus beta-blocker treatment.

Conclusions: Family screening for HCM did not appear to negatively influence QoL. Children diagnosed with HCM through family screening went through an involuntary change of daily life, mainly ascribed to life-style-modifications. They strived to create a life where they could feel secure and have faith in the future, and with the support of parents and health care professionals they achieved a new state of normality. Neither selective nor non-selective beta-blockade caused significant reductions in exercise capacity in patients with HCM above that induced by life-style changes.

Keywords: adolescents, beta-blocker therapy, children, exercise performance, family screening, hypertrophic cardiomyopathy, inherited cardiac disease, life-style recommendations, parents experiences, psychosocial consequences, quality of life, transition.

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