

MARFAN SYNDROME IN CHILDHOOD: PARENTS' PERSPECTIVES OF THE IMPACT ON THE DAILY FUNCTIONING OF CHILDREN, PARENTS AND FAMILY; A QUALITATIVE STUDY

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Background

Marfan syndrome (MFS) is a heritable connective tissue disease caused by a defect in *FBN1*. The main features involve the cardiovascular, musculoskeletal, ophthalmic and pulmonary systems. The impact of MFS on the daily functioning of children parents and family is unclear.

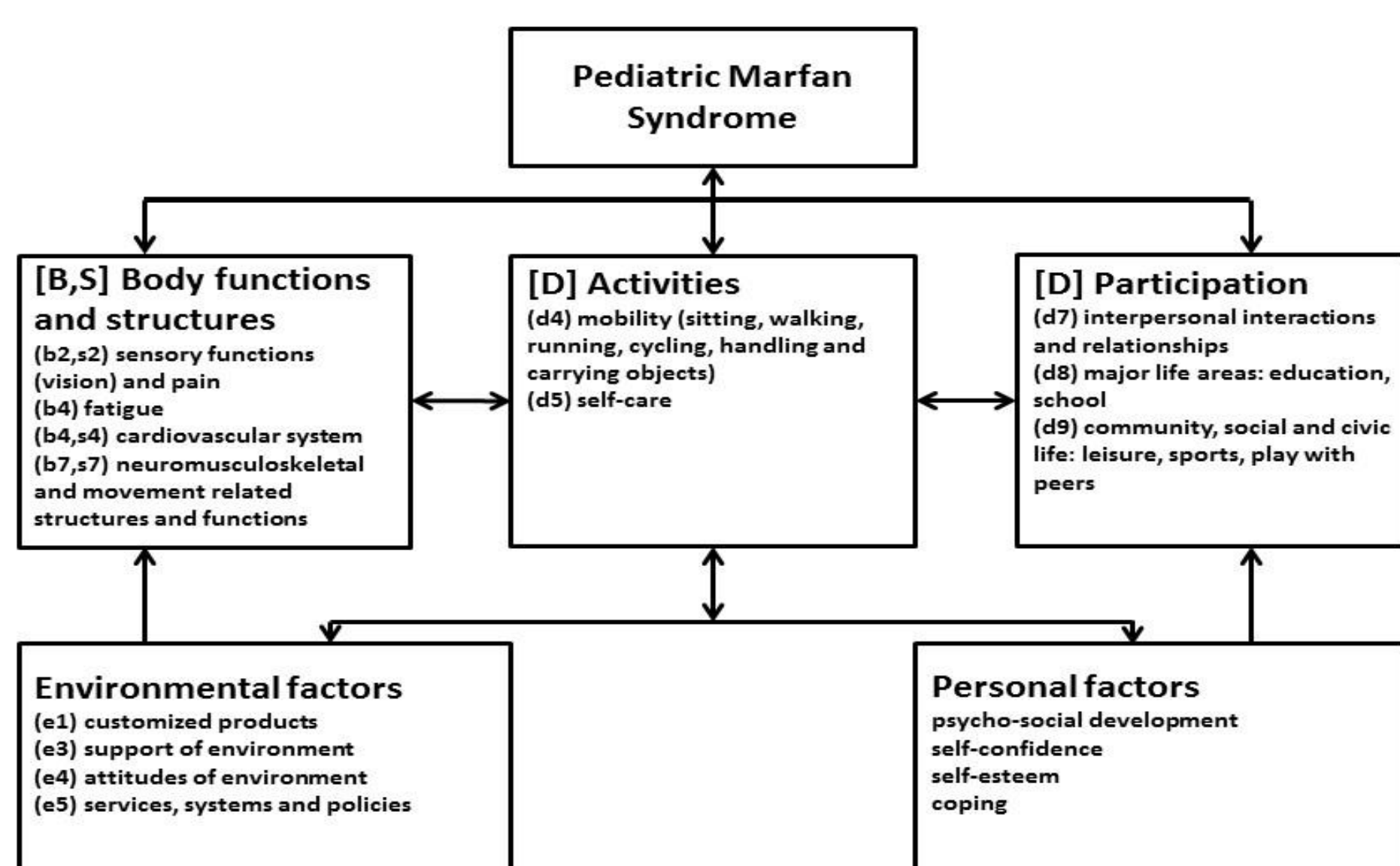
The aim

To explore parents' perspectives on the impact of MFS on the daily functioning of children with MFS aged 4-12 years, themselves and family regarding functional performance, activities, participation, personal and environmental factors, and disease burden.

Methods

Parents participated in individual semi-structured interviews (n=10) and 3 focus groups (n=5, n=5 and n=6). Meetings were transcribed, and data were analyzed using thematic analysis. Meaningful concepts were coded, and concepts concerning children with MFS were linked to the International Classification of Functioning, Disability and Health for Children and Youth. Thereafter themes were identified and interpreted.

Results



Impact of MFS on the daily functioning of

➤ Children with MFS

- children could not keep up with peers because of fatigue, pain and physical impairments.
- children experienced participation restrictions in school, sports, play and other leisure activities.
- children felt being different due to physical appearance, which provoked unsupportive attitudes.

➤ Parents

- parental burden was caused by high care needs, lack of support, a limited social life, and concerns about the child's development.

➤ Family

- family burden was caused by adjusted and complex family schedules, other family members with MFS, and reproductive planning decision-making.
- family cohesiveness and caring were positively perceived factors.

Conclusions

Parents perceived a large impact of MFS on the daily functioning of their children with MFS, themselves and their family. More awareness of the impact of MFS on the daily functioning among all professionals involved in the care of children with MFS and their families is needed so that professionals can address their support needs and provide tailored interventions, rehabilitation and/or education programs to empower children and families and improve the daily functioning of the children, parents and family.