MARFAN SYNDROME IN ADOLESCENCE: ADOLESCENTS’ PERSPECTIVES ON (PHYSICAL) FUNCTIONING, DISABILITY, CONTEXTUAL FACTORS AND SUPPORT NEEDS

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Disclosure of speaker’s interests

I have no conflict of interest to disclose

Introduction

Marfan syndrome (MFS) - rare heritable connective tissue disease caused by FBN-1 gene
- diagnosis is based on the revised Ghent criteria

MFS affects multiple organs and systems:
- cardiovascular (aortic aneurysm, mitral valve prolapse),
- musculoskeletal (arm span/height, arachnodactyly, scoliosis, genu valgum, pes planus, pectus deformity)
- ophthalmic (ectopia lentis, severe myopia)
- pulmonary (pneumothorax) systems
- skin striae
- facial features (dolichocephaly, enophthalmos, downward slanting of the eyes, malar hypoplasia and retrognathia)

Although essential for providing optimal adolescent patient support, knowledge of the impact of Marfan syndrome (MFS) in adolescence is limited

Aim 1
- to explore adolescents’ perceived impact of MFS on (physical) functioning (activities, participation), disability (limitations, restrictions), contextual factors and support needs.

Aim 2
- development of an adolescent MFS International Classification of Functioning Disability and Health for Children and Youth (ICF-CY) model describing the impact of MFS on adolescents (physical) functioning and disability.
Patients and methods

Design
- qualitative study: semi-structured interviews

Approval
- the Medical Ethics Review Committee of the Amsterdam University Medical Centers, in the Netherlands has waived ethical approval under Dutch Law and written informed consent

Patient characteristics
- 19 adolescents (2 declined)
- 19 DNA mutation FBN-1 gene (100%)
- 12 boys (63%); 7 girls (37%)
- 14.5 years (12-18 y)
- 10 patients Ghent score >= 7 (53%)
- Amsterdam UMC

Methods
- Audio-recordings of the interviews were transcribed (30-75 minutes)
Thematic analysis approach
- Meaningful concepts were coded by 2 investigators in MAXQDA
- Codes were structured to the domains of the ICF-CY: body functions and structures, activity limitations, participation restrictions and contextual factors.

Identification of
- Themes
- Contextual factors acting as a barrier or facilitator

The adolescents’ reported support needs were categorised.

Questions:
- What do you know about MFS?
- How did you gather this information?
- Which features of MFS do you have yourself?
- What is the impact of MFS on your activities?
- What is the impact of MFS on your participation in daily life?
- What are your concerns about your daily life (related to MFS)?
- How do you manage and cope with your limitations, restrictions and concerns (related to MFS)?
- What kind of physical or emotional support do you get and what helps you to participate in daily life?
- Who and how did you tell about MFS?
- What is the attitude of others towards your disease?
- What is the impact of MFS on your family life?
- Have you thought about your future adult life (work, relationships, family life, health, leisure)?
- Which supplementary (medical) support do you need and what is your advice to optimise adolescent MFS care?

Results

Themes

1) Difficulties in keeping up with peers

“I really notice that I am not always able to participate. In sports, in particular, I often have to give up earlier. My knees ache a lot, so many times I stop before we’re done.” AB

in school, sports, leisure, friendships/relationships and they could not meet work requirements.
**Results**

**Themes**

2) Being and feeling different from peers

“It bothers me sometimes, being an exception. I am insecure because you can see that we are very tall or very thin, and people will notice the dent in my chest, and ask me “what’s wrong with you?”; It’s not always nice, having to tell them you have Marfan’s, and I have to keep telling them over and over.”

- Appearance, fatigue and pain: Adolescents described themselves as different from their peers due to their appearance, fatigue and pain problems.
- Disability: Adolescents perceived limitations in activities compared to peers.
- Feeling different from peers: Adolescents indicated feeling different from peers because of their MFS appearance and disability.

**Contextual factors reported acting as a barrier or facilitator on themes**

**Conclusions**

- Adolescents with MFS perceive limitations and restrictions in (physical) functioning (activities and participation).
- They perceive problems in keeping up with peers in school, sports, leisure, friendships/relationships and work requirements.
- They perceive to differ from peers due to their appearance and disability.
- An adolescent MFS-specific ICF-CY model of (physical) functioning and disability with its contextual factors derived from the data.
- Adolescent support needs are categorised.
Clinical implications

This perceived impact of MFS: limitations and restrictions in (physical) functioning

- warrants awareness

- tailored physical, psychosocial, educational and environmental support programmes to improve (physical) functioning and empowerment of adolescents with MFS.

Thank you for your attention, questions?

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