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Medical treatment of children with spinal muscular atrophy - An investigation of parents' experiences of hopes, worries and need for rehabilitation for their child

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Results

Figure 1: Understanding how Danish parents whose children with SMA have been offered medical treatment handle hopes and worries in relation to disease progression

THE REALITY OF THE DIAGNOSIS - GRIEF AND HOPE
- THE TRAJECTORY TOWARD THE DIAGNOSIS
- HOPE RELATED TO MEDICAL TREATMENT
- GRIEF RELATED TO UNCERTAINTY

THE GLIMMER OF NORMALCY - FUTURE AND HOPE
- HOPING FOR A NORMAL LIFE
- ACCEPT OF AN UNEXPECTED TURN IN LIFE
- CONCERNS ABOUT THE PROSPECT OF DEPENDENDY

MANAGEMENT OF EVERYDAY LIFE
- ROLE MODELS AND KNOWLEDGE SHARING
- BALANCING PROFESSIONAL SUPPORT AND COUNSELLING
- AFFECTED SOCIAL RELATIONS AND SPONTANEITY

Aim

To gain knowledge about how Danish parents whose children with SMA have been offered medical treatment handle hopes and worries in relation to disease progression. And to investigate the families' needs for information, advice, and rehabilitation initiatives.

Results

Eighty-two parents of 40 children with SMA aged <14 were invited to participate.
Twenty-six parents representing19 couples and 20 children (one couple had two children with SMA) agreed to participate (table 1).

The initial analysis (figure 1) showed that the parents were affected by the reality of the SMA diagnosis, the trajectory toward the diagnosis, the hope related to the medical treatment, and grief related to uncertainty. The medical treatment carried a glimmer of hope for a 'normal' life for the child, acceptance of the unexpected turn of life, but also concerns about the prospect of dependency. The families struggled with managing everyday life which affected their social life, and they expressed needs for balanced support and counselling, and for role models. Analysis is ongoing.

Conclusion

Families of children with SMA need support with adjusting to their new life situations after the SMA diagnosis and start-up on medical treatment. It is important that professional support and counselling is balanced to the individual family's hopes, needs, and wishes for information and knowledge. The findings will be used to target rehabilitation services to families with children with SMA in medical treatment.

Background

- In Denmark, newborn screening for 5q spinal muscular atrophy (SMA) was introduced from 2023
- Per July 2023, the Danish Medicines Council extended the possibilities of medical treatment to persons ≤25 years of age.
- Most children with SMA are referred to RCFM where the families receive guidance and advice from a multiprofessional rehabilitation team
- Medical advances bring hope for both children, parents, and health professionals for a new life with SMA
- There is currently scant knowledge about how parents of a child who receive medical treatment experience their child's illness, their contact with health professionals, and their needs for information and advice

Design and methods

The study was designed as a qualitative interview study guided by the interpretive description methodology and Joyce Travelbee's theory of interpersonal aspects such as suffering, meaning, hope and communication. The method was semi-structured couple interviews with parents of children with SMA type 1, 2 and 3 aged 14 and younger. In all, 41 couples with children registered at RCFM were invited to participate.

Table 1: Demographic data on parents and their children with SMA

Parents to children with SMA (19 couples and 20 children)		N=26 (%)
Age of parents	Mean years (range years)	39,3 (26-63)
Sex of parent	Female	15 (58)
	Male	11 (42)
Cohabitation of parents	Married/partner	25 (96)
	Divorced/single/widowed	1 (4)
Children with SMA		**N = 20 (%) S/W***
Age of child	Mean years (range years)	6 (0-13)
Age at diagnosis	Mean yeas (range years)	1.8 (0-8)
Sex of child	Female	12 (60)
	Male	8 (40)
Diagnosis, copies, ambulation	SMA1, 2SMN copies	5 (x) 5/0*
	SMA1, 3SMN copies	1 (x) 0/1
	SMA1 - 4SMN copies	0 (x) 0/0
	SMA2 - 2SMN copies	0 (x) 0/0
	SMA2 - 3SMN copies	10 (x) 8/2
	SMA2 - 4SMN copies	0 (x) 0/0
	SMA3 with 2SMN copies	0 (x) 0/0
	SMA3 with 3SMN copies, S/W	1 (x) 0/1
	SMA3 with 4SMN copies, S/W	3 (x) 1/2
Ambulation	Walkers	6 (30)
	Sitters	14 (70)
First choice of medical treatment	Onasemnogene abeparvovec	3 (15)
	Nusinersen	11 (55)
	Evrysdi	6 (30)
Present medical treatment	Onasemnogene abeparvovec	2 (15)
	Nusinersen	5 (25)
	Evrysdi	12 (60)

* S=Sitter, W=walker

Clinical and patient implications

Multiprofessional teams should:
- support families with a newly diagnosed child with SMA
- balance professional support, information and knowledge sharing to the individual family
- support the parents in tackling grief and concerns and promote hope for the future
- support the parents and children in maintaining social relations and friendships
- ensure the possibility for SMA role models and mentorships for parents and children

The authors declare no potential conflict of interest. There is no funding to report.

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