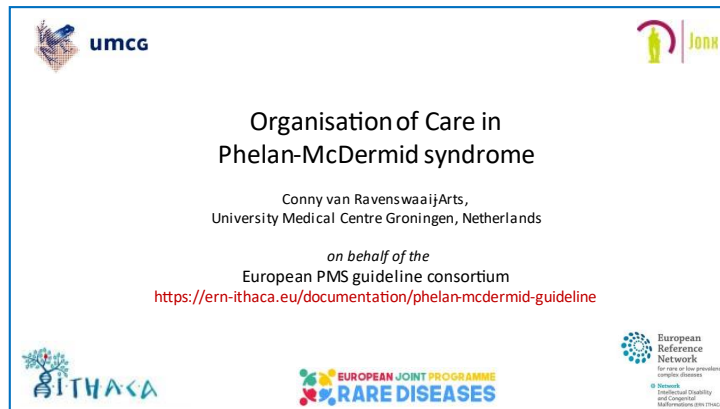
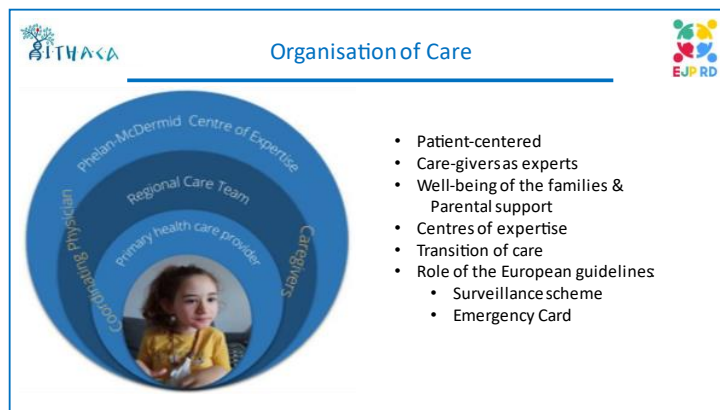


Organisation of Care in Phelan-McDermid syndrome

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This was originally one of the presentations scheduled for the PMS family meeting in Ulm on October 6, 2023. The presentation was skipped due to lack of time. In this document I briefly summarise the most important take-home messages and provide some links to important information.



Although care may be organised differently in different European countries, some main issues can be applied universally. First of all, care should be patient-centred and individualised. Everyone is unique, including individuals with Phelan-McDermid syndrome. So, care should be adjusted to their needs and abilities.

Care-givers should be considered as experts. Parents know their children best. They are able to understand their behaviour and translate it to, for instance, somatic problems. They know through experience what works for their child and what does not.

But professionals should also keep in mind that the well-being of families is crucial for the well-being of their patients. So parental support should be offered whenever needed.

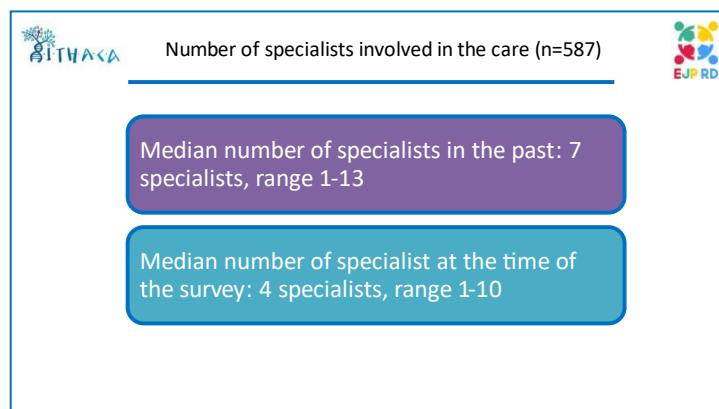
Centres of expertise should be consulted whenever needed, ideally at least once during a patient's lifetime. These centres provide cross-border care and have a predominantly advisory role: in general they do not take over care from local doctors and therapists. It is only when highly specialised care is needed that this is provided by the centre of expertise. Centres of Expertise for PMS can, amongst others, be found in Ulm (Germany), Paris (France), Madrid (Spain), Modena (Italy) and Groningen (Netherlands).

When a child grows older, the professional team organised around them changes. This transition should be arranged in good time and coordinated centrally. All medical information should be transferred to the new team, and parents should be consulted in this process.

When we talk about families, we should not forget siblings, irrespective of their age. From a young age onwards they will be involved in care and will observe the struggles that a child with special needs may give. Siblings should be supported, not only by parents but also by professionals whenever needed. A teacher at school may already be of great help. On the other hand, siblings can also be an enormous source of support. With the consent of her father, I cordially invite you to watch this video that was made following a request from Sophia, the little sister of Olivia, who has Phelan-McDermid syndrome.

https://youtu.be/blLoZiO_v-M

The next figure shows the results from a worldwide survey among 587 parents of individuals with PMS. We asked them how many different specialists they had seen during their lifetime (1 to 13) and how many they still visited (1-10).





These numbers illustrate how complicated care can be.

We also asked parents which problems they encountered in their child and could relate this information to the ages of the children:

Problem/symptom	0-4 years (n=86)	5-12 years (n=227)	13-18 years (n=119)	>18 years (n=156)
Epilepsy	9%	19%	23%	43%
Sleeping problems	41%	55%	62%	73%
Mood problems	16%	23%	42%	54%
Loss of skills	30%	44%	51%	64%
Lymphedema	6%	5%	13%	15%

The table shows that epilepsy can start at any age and that, for instance, sleeping problems still occur in adulthood and can even become worse. Mood problems are predominantly seen in teenagers and adults, while lymphedema can already start at a young age. Loss of skills is a major concern. At a young age this may be temporary, or some skills can be replaced by others. More permanent loss is seen in adulthood. Because of the age-dependent clinical features, a well-organised transition of care is important.

The information given above can also be found in the European consensus guideline for PMS, which makes the following recommendations:



Recommendations: Organisation of care

- Every person with PMS should receive PMS-specific care by a dedicated expert team.
- A coordinating professional should initiate and monitor the multidisciplinary care. The multidisciplinary team should be established based on the surveillance scheme.
- For every person with PMS, specific care needs and the responsible professionals should be recorded in the medical record and the individual care plan.
- For every teenager with PMS the transition from paediatric to adult care should be timely initiated, monitored and documented.
- Caregivers of individuals with PMS should be informed about the PMS patient registry when established

The European guideline also states that families should be informed about ongoing research and especially about clinical trials for which their child might be eligible.

Where all information can be found:

The chapter of the guideline on organisation of care can be found at: [Consensus recommendations on organization of care for individuals with Phelan-McDermid syndrome - ScienceDirect](#)

All the guideline materials can be found on the website of the European Reference Network ITHACA, which unites all the centres of expertise that have been accredited by the European Union. This material includes:

- A summary of the guideline for clinicians
- A surveillance checklist for clinicians
- An emergency card for parents (this will soon be available in multiple languages)



Link to all PMS guidelines materials

The last two documents are also attached to this handout. The emergency card (Notfallkarte) can be a handy tool when you have to visit a doctor who is not familiar with your child or with Phelan-McDermid syndrome. The card summarises the syndrome on one side, and the information on your child can be documented on the other.