

Self-Management and Related Concepts in Rare Diseases – Empowerment, Implementation, and Relevance

Background

- Self-management of rare diseases is of the utmost importance. Most rare diseases lack a cure, and many lack effective treatment. **The rarity of the disease comes with a rarity or even lack of expertise,** be it clinical or in living with the disease.
- Acknowledging the importance of self-management and self-management support in this field is even rarer. Our project aims to boost this acknowledgement and **facilitate** subsequent **implementation**.
- This project is a spin-off of the joint guideline-development program of the global organisations of professionals and patients involved with Gaucher disease.

Step 1: Thorough exploration of literature

This project was initiated by the discussion on Gaucher disease management guidelines by the International Working Group on Gaucher Disease.

We started out by doing a thorough literature review to get ourselves well acquainted with the field. Our extensive findings are compiled into **a comprehensive toolbox**.

We did not find any differences between Gaucher disease self-management and self-management of rare diseases in general, so our review bears on rare diseases as a whole.

We are currently formatting our findings tentitatively into an article **to be published**.

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Discussion

- Our project focuses on rare diseases that evolve into chronic illnesses, as most of them do. For the most part, **the instrumentation of self-management (support) in a specific rare disease is the same for them all**. Where specific insights lack, the body of knowledge generated in more common chronic diseases can be used in rare diseases too.
- Special attention is required on **dealing with the rarity of expertise**, also in terms of a kind of health literacy and in partnering of patient and professional as experts. And on the **autonomy of the individual or dyad living with the rare disease**, as usually being the expert outside the consultation room. Each individual living with a rare disease is unique in the combination of genetic mutations, epigenetics, environmental factors, and self-management potential.
- As most patients with rare diseases are children, paediatrics and family care play an important part. **Siblings and (potential) partners**, having their own burden, must be included in self-management support.

Step 2: Gaucher disease management guideline

Based on the review of literature we developed a draft disease-specific guideline on self-management. It will be finalised as soon as the review is accepted as an article for publication.

The guideline defines and describes self-management, as well as the role and responsibility of the healthcare providers involved. It touches upon the clinical relevance of self-management.

It also brings others around the patient into view, having a burden of disease in their own way as well as their own role in self-management.

Each discussed item will include recommendations on how to support self-management. The guideline concludes with suggestions for further research.

Project team

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