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.

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 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.

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.

Contents of the paper to be published

Project team

Paul Guijt, retired policy consultant national healthcare (quality) management, co-founder Dutch Gaucher Association (1983-2008), volunteer International Gaucher Alliance, paul.guijt@outlook.com

Irena Žnidar, PhD, director International Gaucher Alliance, co-founder and vice-chair Slovenian Gaucher Association

Magy Abdelwahab, PhD, Professor of Pediatrics and Pediatric Hematology, Cairo University, Social and Preventive center, Kasralainy hospital, senior consultant Gaucher Disease Clinic Egypt, founder Egyptian Society of Inherited Paediatric Disorders of Thrombosis and Haemostasis (ESIPTH) and Gaucher disease group