IWGGD The International Working Group on Gaucher Disease

Guideline	**COORDINATION of CARE IN GAUCHER DISEASE TYPE I**
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Overview	The aim of this report is to provide guidance for well-planned and coordinated care of pediatric/adolescent/adult patients with Gaucher disease (GD) type I. The guidance is based on a collaboration between IWGGD (clinicians) and International Gaucher Alliance (patients/patient representatives).
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Introduction

Many countries all over the world are working on the implementation of national health policies and strategies dedicated to coordinated transdisciplinary approaches to support patients with rare diseases (1, 2). Enhanced coordination is particularly beneficial for patients affected with complex chronic rare disease such as Gaucher disease (GD) (3, 4).

Care Coordination provides a well-defined framework for managing rare diseases. It addresses medical, social, developmental, behavioral, educational, and financial needs to achieve optimal health and wellness outcomes (5).

Coordination of care is already part of some national rare diseases plans and strategies with the aim to facilitate communication between specialized healthcare providers (HCPs) with expertise in diagnosing and managing these conditions, to reduce diagnostic wandering, to facilitate access to treatment, to reduce the burden of follow-up, to organize emergency situations and to reduce the burden of transition from adolescence to adulthood (2,3,4).

Coordination of care should be (6):

- Family-centered
- Planned, proactive
- Evidence-based
- With equal access irrespective of diagnosis, patient circumstances, and geographical location
- Promotion of self-care skills and independence, patient's involvement in their therapy
- Emphasis on cross-organizational relationships

Challenges of coordinated care of patients with GD

The process of coordinating care for rare diseases, including GD, can be complex since patients are frequently required to attend multiple health appointments, with various specialists, and on various days or locations (6).

Coordinated care is necessary irrespective of the patients' age, severity of symptoms and complications, related or not to GD. Patients may report unexplained tiredness, bone pain and may develop gammopathies, Parkinson disease, malignancies or common health problems. In such circumstances, patients should be able to contact the coordinator of their care at the closest reference center or to access the specialist familiar with their underlying metabolic condition. Lack of identified contact leads to delay in a referral to a specialist clinic. Reluctance of some specialists to consult a patient with an unfamiliar rare disease or to refer them to the right specialist is a common issue in many countries.

In this guideline dedicated to the coordination of care in GD, we will approach two important aspects of coordinated care:

- coordinators' roles and responsibilities, and
- communication and care delivery.

Coordinators' roles and responsibilities

There are many differences between healthcare systems regarding the care coordination according to the level of care: primary, secondary or tertiary. On a national level, establishing centers of expertise (or so-called centers of excellence) is important for creating or developing a local knowledge and a better management of the disease (2, 3, 4). Recommendations were made regarding the quality criteria for such centers of expertise for rare diseases in order to help countries in their policy development concerning the organization of healthcare pathways (7). Some countries managed to establish and develop centers of expertise, but others do not yet. Anyway, the primary care is often the first contact of the patient with the healthcare system. The role of a care coordinator depends on the specific context in which they operate, i.e. primary care or specialized center, and the level of complexity involved.

1. Primary healthcare providers (HCPs)

The primary care is often the first contact of the patient with the healthcare system. The primary HCPs, e.g., general practitioners, are usually the patients' central point of contact.

The **panel recommends** some actions to support this field of care coordination:

- Each country should establish a national diagnosis and care protocol for patients with GD, incorporating detailed recommendations for various situations such as transitions and emergencies. These guidelines should be adapted from international standards to suit the local context and should be available in the local language, specifically designed for primary HCP. In cases where local guidelines are lacking, the IWGGD guidelines can be adopted.
- Every GD specialist, including those working at referral centers, should communicate their findings and treatment recommendations to the primary HCP, typically through liaison letters.
- In instances where a local specialized center is responsible for monitoring patients with GD, clear contact information for the center's HCP should be publicly accessible.

Details:

- The primary HCPs are close to the patient.

- They have a comprehensive view of the patient's health history they oversee and manage the patient's care journey, helping to early disease detection.
- They are coordinating basic care with the other HCPs (haematologist, orthopaedist, physiotherapist, radiologist, endocrinologist, psychologist, genetic counselor), referring the patient to a specialist physician when needed.
- They play an important role in preventive care and health promotion,
- They may connect the patients with community resources and support services to address social determinants of health.

2. Specialized centres (secondary and tertiary care)

The **panel recommends** the following actions to support this field of care coordination:

- There should be individuals designated as a coordinator and a clinical lead with formal responsibilities for managing patients with GD, and their contact information should be publicly available.
- A comprehensive care plan should be established, incorporating evidence-based information, the patient's medical history, and their active participation.
- Multi-disciplinary team collaboration should be developed and clearly defined.
- Developing local expertise in GD (specific informative materials, meetings or conferences for physicians, nurses or other HCP who might take in charge such patients).
- Actions to promote education of patients and their families (online modules), empower patients to actively participate in decision-making and self-care.
- Sharing knowledge by active collaboration with other centres specialised in GD on a national level (online or hybrid multi-centric meetings, networking if more than one in a country) and abroad (establishing contact with e.g. MetabERN, IWGGD) and with the patient organisations (national GD patient organisation and/or International Gaucher Alliance).
- The development of information accessible to patients, families, caregivers, physicians and social-players in order to have an easy access to clear information, relevant medical guidance or appropriate social support (dedicated website, regular conferences).
- advocating to access to treatments and care or other interventions to support patients.

Details:

Specialised centres with expertise in GD might have broader roles and responsibilities than primary care. A clinical lead (e.g., pediatricians, adult metabolic physicians, a specialist nurse or geneticists) oversees or manage patient care, supervise HCP liaisons and interventions (often across centers), delegate and ensure accountability of responsibilities. HCPs from specialized centers have to facilitate communication between different players to improve care coordination.

These HCPs have specific roles that require a training pathway. Clinical coordinators (i.e., specialized nurses or trained personnel) are individuals with sufficient clinical expertise to coordinate complex cases working closely with the lead clinician. Coordinators help build relationships between patients and the team and supports the patients and their families. Being often the point of contact for patients and their families, they can organise various multidisciplinary care meetings. Eventually, the coordinators of care arrange the infusions in the hospital setting when the treatment cannot be administered at home.

Communication and care delivery

The **panel recommends** that each patient with GD should have a written coordinated care plan developed in keeping with evidence-based practice. Care could be better coordinated and improved by using various modes of communication among HCPs, patients, and caregivers, such as verbal and written communication, as well as the use of technology.

Details: Specialized centers, single-visit approaches, joint clinics or consultations, and specialized or condition-specific clinics could all be used to coordinate care (6). There must be one overarching specialist physician/specialist team in charge of the patient's case, who coordinates and verifies the activities of other specialists as well as the patient's pharmacotherapy. The coordinated care plan must be tailored to the patient's clinical condition, health-care system, environment, geographical location, and level of education and, more importantly, with the patient.

The care plan would also involve the frequency of regular and ad-hoc appointments and the mode of communication (face-to-face, telephone or video consultation). The geographical location of the patient influences how the care is coordinated. For patients living far away from a specialist metabolic center, a greater proportion of care could be delivered locally, online or through outreach clinic models. Models of care can be tailored to individual situations, circumstances, and resources. A shared care model could be considered to ensure a continuation of care locally for any urgent matters with a subsequent follow-up review in a specialist center. Shared care models also reduce travel and increase provision of education to local HCPs (6, 8).

The nature of patients' condition and the number of disciplines involved in their care may influence how care is organized and coordinated across several different care services (8). The specialist services ('one-stop shop') only work if services can determine exactly who a patient with GD will need to see, e.g., osteoporosis clinic with a rheumatologist and imaging studies. Collaborative working among professionals is key to provide such service model. Additionally, conditions that are more stable may require less coordination.

Charities and patient organizations are engaged in coordination by providing support to patients/carers, by collaboration with specialized or reference centers and by involvement in activities (e.g., providing information, holding support groups, providing helplines). Health care professionals from expert centers have to work closely with all the patient organizations supporting patients (e.g., participation in training of professionals and guiding coordinators) (6).

Components of care coordination

- Situational understanding: a conceptual framework is built upon the family and patients identified goals, healthcare needs, and preferences.
- Care networking: A healthcare team is formed around the patient. Individuals are identified, roles and responsibilities are clarified, and a communication process is established.
- Shared Plan of Care (SPOC): resources are organized to safely address episodes of care, contingencies, and emergencies.
- Guidelines and policies development: the importance of patient organizations in the development of health policies/protocols with expert centers for better coordinated care is key.
- The quality of services delivered and patient adherence to medical regimens need to be tracked and monitored.
- Remaining in home care or under the supervision of a primary HCP, patients with GD must regularly report to the reference center every 6 to 12 months (provided their clinical condition does not worsen) to perform comprehensive evaluation.
- Navigating the healthcare system: patients and families require logistical support to receive comprehensive and continuous care as health states and needs fluctuate.
- Learning: this is a bidirectional process that allows the health system to understand better what individual patients need to stay healthy and the patient to learn how best to access needed services.
- Psychological support: patients and their families engage better with their medical team when they are offered psychological support.

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