

Guideline	TRANSITION CARE IN GAUCHER DISEASE
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Date of preparation	1 June 2022
Due date of review	1 June 2024 (update)
Version	1.0
Overview	The aim of this report is to provide guidance for a well-planned and coordinated transition of care from pediatric/adolescent to adult care of patients with Gaucher disease (GD). The guidance was developed by a collaboration between members of IWGGD working group and patients with GD.
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Introduction

Transition of care has been commonly described as - *“The purposeful, planned process that addresses the medical, psychosocial and educational/vocational needs for young people and young adults with chronic physical and medical conditions as they move from child-centered to adult-oriented healthcare systems”* (1). The transfer of care occurs at the last transition clinic.

The goals of a structured and coordinated transition to adult health care for young people with GD are to optimize health and empower young adults in attaining their maximum potential through the provision of high-quality, developmentally appropriate health care services that continue uninterrupted as the individual moves from adolescence to adulthood (2, 3).

Transition needs to be personalized, individualized, and family-centered, with a high level of flexibility, responsiveness, continuity, comprehensiveness, and coordination.

The transition is a gradual process starting in a young person’s clinic (from around 14-18 years of age for complex cases) and ending once the young person is successfully engaged in adult services.

Principles of transition

Currently, there is no worldwide standardized model of care for patients with GD during the transition process. In different countries, professional teams are searching to develop transition dedicated therapeutic education programs and projects for organizing the improvement of care system (4).

Transfer from childhood to adulthood care has to be anticipated and mentioned early and often during the care of a child/adolescent with GD disease. The transition process should be attended by a pediatrician, a nurse, a member of the adult team (a clinician, a nurse), and other health care professionals as applicable.

The best approach in order to improve transition in adolescents/young adults with GD seems to be to advocate for certain principles that would facilitate an effective transition, such as autonomy, adherence to the therapy, self-directed management, maintaining a relationship with the hospital staff, and ability to consent (5, 6).

Autonomy is the young adult’s right to make their own decisions and choices regarding health and lifestyle; in this context, specific consideration should be given to decisions surrounding family planning and the use of drugs or alcohol.

Adherence to the therapy is the patients' compliance with their regular treatment. It is their responsibility to continue with the therapy and minimize the number of missed medications.

Self-directed management requires some readiness to make their own decisions regarding health and full awareness of all the normal adult healthcare issues. There is some evidence that self-directed management improves the long-term outcomes of patients during the transition process. If patients receive adequate education regarding the management of their GD, they are able to self-care and able to cope with the disease complications.

Maintaining the relationship with the clinical team is essential to improve clinical outcomes, symptom control, and the overall quality of life.

Ability to consent often depends on the patient's capacity to make their own decisions. Patients may lack capacity if the underlying condition affects their ability to understand. In some individuals, their capacity may fluctuate, meaning that they are capable of making health-related decisions only during certain periods (7).

Challenges of transition of patients with GD

GD is one of the growing numbers of conditions that have survival into adulthood and often to old age due to advances in treatment (2).

In some countries, patients remain under the pediatric team follow-up for many years mainly because of a lack of adult metabolic specialists and knowledge about the condition (8). Adult patients, including those with rare diseases, cannot be hospitalized in pediatric hospitals due to legal restrictions. Adult patients with GD can develop common comorbidities, such as high blood pressure, diabetes, lipid disorders, and other conditions less familiar to pediatricians. Adults with GD may have indwelling devices such as port-a-caths for infusions (9).

The transition from pediatric to adult care is generally a concern for patients with GD and their caregivers, often associated with follow-up problems. If the process of transition is not well managed, adolescents with long-term health conditions sometimes fall into a gap in services, which can lead to deterioration in their health (10).

Previous studies have shown that the interruption of treatment for more than 6-14 months in adult patients with GD can generate worsening in biomarkers, clinical symptoms (bone pain), health status,

and general well-being (11, 12, 13) with no severe consequences. Even without serious consequences of such interruption, continuity of treatment is highly recommended.

Most stable patients with GD will have one transition clinic appointment before being transferred to adult services. However, more than one joint transition clinic appointment will be necessary for some. Psychological support during transition is recommended to empower patients in managing their condition as young adults (14, 15).

In this guideline we propose the basics to ensure a good transition for young patients with GD:

- a transition coordinator
- a healthcare plan
- elements of communication during transition
- an educational program

1. A transition coordinator

The **panel recommends** that dedicated healthcare professionals from pediatric and/or adult referent services coordinate the transition process for young patients with GD. Their role is to plan the clinic appointments during the transition process and to prepare the relevant medical and informative documents (e.g., proformas and leaflets).

Details: The role of a coordinator has been described in the guidelines on the 'Coordination of care'. The coordinator will ensure that a follow-up appointment has been arranged after the transfer of care to the adult services and that the adult group has taken over the treatment prescription.

2. Healthcare plan

The **panel recommends** that a written individualized healthcare transition plan be implemented while the patient is a teenager (e.g., by age 14 (UK), or later, between the ages of 16-18 (in most EU and non-EU countries)). The healthcare transition plan should be created by the pediatrician in cooperation with the adult clinician who will take over the care of the patient, as well as with the patients and their caregivers. The document should be reviewed and updated regularly.

Details: Children with GD are seen with their parents/caregiver but when they are older, they may ask to be seen on their own, especially to discuss more personal subjects. The emphasis of the discussion

is on the patient’s autonomy and independence. A well-structured transition clinic aims to address a young person’s clinical, educational, and vocational needs (Fig 1). This is the opportunity for adolescents/young adults to raise their concerns or clarify uncertainties regarding their future management. For patients with stable disease, these often include issues regarding port-a-caths, self-infusions and self-management of their infusions, university time and continuation of treatment, education about adult complications of GD, relationships, etc. If they receive an ERT in-home settlement, it is their responsibility to arrange the infusion on a convenient day.

The best solution for a good transition is to adapt the timing of the transfer to the individual young person’s needs depending upon an assessment of their emotional maturity, mental health, abilities and physical development, and medical and social needs.

Exceptions to this should be discussed with the young person and always be in their best interest. These should be clearly documented in the young person’s health care record, and the outcome should be communicated to the young person and their family. It is also essential for the young adult to have the opportunity to have a say in the matter and be able to share their opinion.

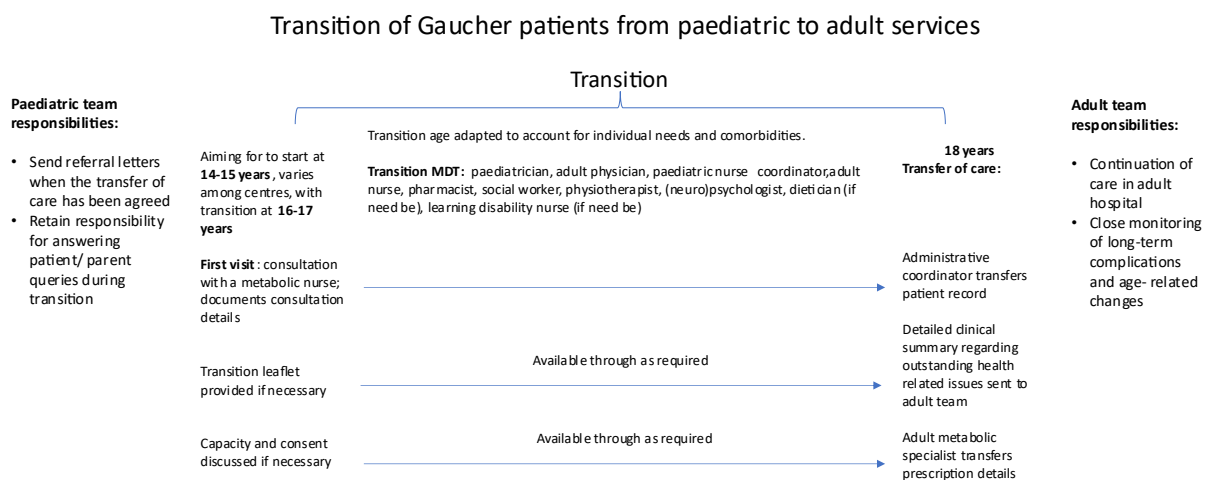


Fig 1. Transition algorithm in Gaucher disease. Patients are encouraged to understand and manage their disease and acquire skills and education to act independently. Patients may attend without their parents.

3. Communication during the transition

The **panel recommends** that both the pediatric and adult teams communicate by sharing clinical information, completing a clinic proforma, and agreeing on the patient readiness to have the care transferred to the adult services.

Details: An individualized transition passport must be prepared from the pediatric center, including information on the patient's clinical condition, eventual comorbidities, a summary of recent laboratory and imaging results, as well as the dosage of the GD-specific drug and the list of the other common drugs, risk situations, social, educational and professional considerations. The adult team takes over clinical supervision and the metabolic medication prescription. The age-related administrative and social changes are explained to the young patient. The impact of risky behavior (alcohol, tobacco, drugs) and unplanned pregnancy are discussed.

To ensure that all the relevant clinical teams involved are familiar with the clinical needs of the young adult, the clinical information should be shared among specialties involved in patients' care: internal medicine specialist, adult hematology, hepatology, neurology, ophthalmology, physiotherapy, rheumatologist, geneticist, orthopedic teams. During transition and before transfer to the adult services, a joint review with other specialties could be arranged to introduce the young adult to the adult clinicians, as required.

4. Educational program

The **panel recommends** preparing an illustrative educational program for patients and parents that addresses medical, psychosocial, and educational/vocational aspects of care.

Details: The educational programs would include online materials, transition leaflets, and eventually face-to-face discussions with patients' families, as well as digital solutions, e.g., apps and videos presenting patients and families who went through the process. It was shown that adolescents with chronic diseases are interested in online support programs providing accurate disease-related information and social support (16). This type of interaction could be encouraged in transition, too.

For instance, the 'Ready, Steady, Go' program provides different documents to support patients and their families throughout the transition process (17).

Last but not least, the role of patient organizations in the development/ implementation of a transition program at a national level is important. Young adults should be aware of the organizations supporting patients with GD (by leaflets, links to their web pages, etc.).

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