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SPECIAL ARTICLE

Transition from pediatric care to adult care for patients with mucopolysaccharidosis☆

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Abstract Mucopolysaccharidosis are multisystem diseases that require large multidisciplinary teams for their care. Specific recommendations are therefore needed for the transition from childhood to adulthood in this patient group. To overcome the barriers that might arise during the transition, the authors consider it essential to implement a flexible plan with a coordinator for the entire process, systematizing the information through a standardized pediatric discharge report and educating the patient and their family about the disease, showing the characteristics of the healthcare system in this new stage. The final objective is that, once the transition to adulthood has been completed, the patient's autonomy and potential development are maximized and that the patient receives appropriate healthcare during this transition.

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PALABRAS CLAVE
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Transición

Transición desde la asistencia pediátrica a la adulta en pacientes con mucopolisacaridosis

Resumen Las mucopolisacaridosis son enfermedades multisistémicas que requieren para su atención equipos multidisciplinares amplios. Por ello se hacen necesarias recomendaciones específicas para la transición de la edad pediátrica a la adulta en este grupo de pacientes. Para la superación de las barreras que pudieran surgir durante la transición, los autores consideran esencial realizar un plan flexible con un coordinador de todo el proceso, sistematizar la información a través de un informe de alta pediátrico estandarizado, formar al paciente y su familia sobre la enfermedad y mostrar las características del sistema sanitario en esta nueva etapa. El objetivo final es que al concluir la transición a la edad adulta se haya maximizado la autonomía y el potencial de desarrollo del paciente y este reciba una atención sanitaria adecuada durante dicho periodo de transición.

Background

Mucopolysaccharidosis (MPS) are hereditary metabolic diseases caused by defects in a number of intralysosomal enzymes necessary for the processing of certain macromolecules called glycosaminoglycans, which accumulate in various organs and are responsible for multisystemic, chronic and progressive impairment.[1,2] The global incidence rate of MPS is estimated at 1/22,500 live births.[3]

Progress in the early diagnosis and treatment of rare hereditary metabolic diseases have increasingly helped more patients reach adulthood. Specialty departments for adults therefore need to adapt to this new situation and address diseases that have traditionally been considered pediatric.[4] This transition process has been defined as the planned step in which adolescents with chronic medical diseases continue to receive the needed services and care when changing from child-focused care to adult-focused care.[5] The transition can be especially complex for children with special healthcare needs. The objective of the transition is to maximize the individual's functioning and potential by providing high-quality healthcare services appropriate for their development that continue uninterrupted as the patient passes from adolescence to adulthood.[6]

There are general recommendations on the transition process both for the general population[7,8] and for youths with special needs,[6,9] as well as articles on the transition process for patients with chronic diseases such as diabetes,[10,11] rheumatic disease[12] and congenital metabolic diseases such as phenylketonuria[13] and other metabolic disorders.[14] However, none of the recommendations focus on the transition of patients with MPS. The objective of this consensus study, prepared by a group of pediatricians and internists from reference centers with extensive experience in MPS, is to create a number of recommendations for pediatricians, internists and other health professionals regarding the transition from pediatric to adult care for patients with MPS.

Starting from the scarce literature on the subject and from the workgroup's experience in the transition process, an initial document was developed with the available scientific evidence whose conclusions were agreed upon in 2 meetings held on October 11 and November 30, 2016. In the event of discrepancies in the criteria, the final recommendation was decided by majority. This consensus was endorsed by the Spanish Association for the Study of Inborn Errors of Metabolism and the Spanish Society of Internal Medicine.

Barriers in the transition process

Generally, the transition from pediatric to adult care involves passing from a more protective and paternal type of care (focused in good measure on the family) to a more independent, patient-focused type of care.

There are barriers that can impede the transition process (Table 1), which can arise from the people involved (patients, families, pediatricians and adult-care physicians), and these barriers need to be considered for the transition to be successful.[12,15–17]

Transition organization and facilitators

The transition to adulthood is not a specific change that occurs at a precise moment but rather a process that develops over time. Appropriate planning, preparation and implementation of this process is therefore essential.[18]

Planning

The transition should take place at 16–18 years of age, although this can vary depending on factors such as the patient's maturity and development, the disease progression and its management and the organization of healthcare services. Nevertheless, a gradual transition needs to be performed, with frequent discussions about the transition plan with the patient and family over the course of a few years. Preparations for this process should start in early adolescence (10–12 years), providing adolescents with information regarding the process so that they can begin to take