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Consensus statement

Transition process from paediatric to adult care in patients with inborn errors of metabolism. Consensus statement☆

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ABSTRACT

Background and objective: The transition process from paediatric to adult care is a subject of great interest in recent years, especially in chronic diseases with childhood onset, such as inborn errors of metabolism (IEM). Advances in diagnosis and treatment of these diseases have improved their prognosis, with a high number of patients with IEM who currently reach adult age and need to be attended to by non-paediatric professionals. The objective of this work is to establish action guidelines so that the specialists involved can guarantee a successful transition of these patients' healthcare.
Methodology: After carrying out a bibliographic review of the subject, the authors, beginning with their own experience, produced an initial document which was subjected to successive debates until the final document was obtained. The consensus recommendation was decided by the majority in case of criterion discrepancy.
Results: A series of recommendations are presented for the best clinical management of the transitions of care of patients with IEM from the paediatric to adult care setting in order to achieve the best results in this process given the special characteristics of this patient subgroup and the main difficulties entailed in the transition process.
Conclusions: The role of the internal medicine doctor in this transition process and correct interrelation with the paediatric and social setting is stressed. Furthermore, actions and attitudes are suggested to improve the quality of said transition.

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Proceso de transición de la asistencia pediátrica a la adulta en pacientes con errores congénitos del metabolismo. Documento de consenso

RESUMEN

Palabras clave:
Errores congénitos del metabolismo
Transición
Pediatras
Internistas
Consenso

Antecedentes y objetivo: El proceso de transición de la asistencia pediátrica a la adulta es un tema de gran interés en los últimos años, especialmente en enfermedades crónicas de inicio en la infancia, como los errores congénitos del metabolismo (ECM). Los avances en el diagnóstico y el tratamiento de estas enfermedades han mejorado su pronóstico, encontrando en la actualidad un elevado número de pacientes con ECM que alcanzan la edad adulta y necesitan ser atendidos por profesionales no pediátricos. El objetivo de este trabajo es establecer unas pautas de actuación para que los especialistas involucrados garanticen una transición exitosa de la atención sanitaria de estos pacientes.

Metodología: Tras realizar una revisión bibliográfica del tema, los autores, partiendo de su propia experiencia, elaboraron un documento inicial que fue sometido a sucesivos debates hasta obtener el documento definitivo. En caso de discrepancia de criterio, la recomendación de consenso se decidió por mayoría.

Resultados: Se presentan una serie de recomendaciones para el mejor abordaje clínico de la transición asistencial de los pacientes con ECM desde el entorno pediátrico a la asistencia de adultos, con el objetivo de conseguir los mejores resultados en este proceso, dadas las características especiales de este subgrupo de pacientes, así como las principales dificultades que conlleva el proceso de transición.

Conclusiones: Se resalta el papel del médico internista en este proceso de transición y su correcta articulación con el entorno pediátrico y social. Asimismo, se recomiendan acciones y actitudes para mejorar la calidad de dicha transición.

Introduction

Inborn errors of metabolism (IEM) are defined as biochemical abnormalities caused by a specific genetic defect in the structure or function of a protein or the lack of it.[1] They are highly disabling diseases that often occur in several members of one family, caused by their genetic load. They have a high clinical heterogeneity, depending on the degree of involvement of the gene, or the type and function of the protein that is altered. Thus, depending on whether the protein acts as an enzyme, hormone, cell membrane receptor–transporter or is part of a cell organelle, a wide range of diseases may appear. About 700 types of IEM have been defined.[2] Another essential feature of IEM is its low prevalence, it is considered part of the group of rare diseases. However, as a whole, they may affect 1:784 live births.[1]

The IEM have become increasingly important not only for its high morbidity–mortality and disability,[3] but also because advances in diagnosis and treatment have improved prognosis, finding now a large number of patients with IEM reaching adulthood.

The need for reference centres for these diseases[4,5] prompted the Ministry of Health, Social Services and Equality to recently appoint Centres, Services and Units of Reference (CSUR) for IEM within the National Health System.[6] In recent years, the progressive involvement of Internal Medicine in IEM has allowed the creation of multidisciplinary teams in these hospitals, in which the internist has become the care coordinator and interlocutor between the various professionals.[7] A growing number of young adults with these diseases need to be treated by non-paediatric professionals, so that CSUR specialists face the challenge of ensuring continuity of care from paediatric to adult age.

The transition from paediatric to adult care must be a planned and organized from a health system focused on the child to another which is adult-oriented, adapted at all times to the physical, emotional and social characteristics of an individual immersed in a process of change and adaptation to adult life. It must include normalization of the social and emotional development, and acquisition of the necessary independent living skills. However, failure in this process can lead to poor treatment adherence or even withdrawal from the circuits of care assigned to these patients.[8]

Although there is literature on the transition process in patients with chronic diseases, studies on the transition of patients with IEM are limited. This situation encouraged the drafting of this consensus document, addressed to all health professionals involved in the care of such diseases, and in order to establish action guidelines for the childhood–adulthood transition process, so that this can be successful.

Methodology

The group that drafted this document included internists and paediatricians from most IEM CSUR in the National Health System and other hospitals with extensive experience in the transition process of patients with IEM.

To make this guide, a literature search was performed in Trip Database, Cochrane Library, Cochrane, EMBASE, PubMed-Medline and ACE LOST following the PICO method-Patient/Problem, Intervention, Comparison, Outcome-recommended by clinical practice workgroup of the National Health System.[9] We used the search terms *inborn errors of metabolism*, *transition*, *adult care*, *adulthood* and *paediatric*, without specifying any date. The references that appeared on the articles found were also used. The latest and highest quality articles in terms of their scientific level were selected as sources of evidence. The *Scottish Intercollegiate Guidelines Network*[10] criteria was used to measure and establish the degree of evidence of the articles.

Based on the articles reviewed and the experience provided by each centre in the transition to adulthood with IEM patients, the authors developed an initial document which was repeatedly reviewed by all of them and whose conclusions were agreed in a working meeting held in Madrid on 17th December 2015. In case of discrepancy, the consensus recommendation was decided by majority vote.

Results

The American Society for Adolescent Health and Medicine defines transition as an intentional and planned process that addresses the move of adolescents and young adults with chronic