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Special article

A coordinated transition model for patients with cystinosis: from pediatric to adult care☆

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ABSTRACT

Introduction: Improved outcome and longer life expectancy in patients with cystinosis and the intrinsic complexity of the disease, underline the need for a guided transition of patients from pediatric to adult care. The process aims to guarantee the continuum of care and enable the empowerment of patients from guardian to self-care.

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Methods: Bibliography review, expert opinion and anonymous surveys of patients, relatives and patient advocacy groups.
Results: A new plan to support and coordinate the transition of cystinotic patients providing specific proposals for a variety of medical fields and improved treatment adherence. Nephrologists play a key role in the transition since most cystinotic patients have severe chronic kidney disease and require kidney transplantation before adulthood.
Conclusion: We present a proposal providing recommendations and a chronogram to aid the transition of adolescents and young adults with cystinosis in our area.

Transición coordinada del paciente con cistinosis desde la medicina pediátrica a la medicina del adulto

Palabras clave:
Transición
Adolescente
Cistinosis
Trasplante renal
Adherencia

RESUMEN

Introducción: El aumento de la supervivencia de los pacientes con cistinosis y la propia complejidad de la enfermedad explican la necesidad de implementar un proceso de transición guiada desde la medicina pediátrica hasta la del adulto, que permita garantizar el *continuum* asistencial y posibilite el empoderamiento del paciente desde el cuidado tutelado al autocuidado.
Métodos: Revisión bibliográfica, opinión de expertos, encuestas anónimas a pacientes, familiares y asociaciones.
Resultados: Elaboración de un documento de transición coordinada, con propuestas concretas por especialidades y de mejora de la adherencia terapéutica y del autocuidado del paciente. El nefrólogo desempeña un papel clave en la transición en la cistinosis debido a la afectación renal que domina la patología y porque la mayoría de los pacientes han recibido un trasplante renal antes de la edad adulta.
Conclusión: Se presenta un documento que establece unas recomendaciones y un cronograma para guiar la transición de los adolescentes y adultos jóvenes con cistinosis en nuestro ámbito.

Introduction

Medical advances have transformed cystinosis from a historically fatal childhood disease[1,2] into a chronic adult disease[3] with longer life expectancy.[4] Nevertheless, the transfer of young patients to adult units has led to interruptions in their medical care,[5] inadequate patient follow-up and worsening of the disease.[6] The acknowledgment of several factors underlying this situation illustrates the need to implement specific programs for the transition of adolescent cystinosis patients to adult care.[7,8]

Cystinosis is a rare (RD),[1] severe systemic disease caused by mutations in the CTNS gene (17p13) that alter cystinosin synthesis, resulting in the accumulation of lysosomal cystine crystals in all body cells.[2,9,10] With an incidence of 1/100,000–200,000 newborns,[1] cystinosis is characterized by early renal involvement that evolves to kidney failure with corneal crystal deposits and other progressive extra-renal manifestations.[11] Control of this disease requires a strict and complex therapeutic regimen[1,5,6] together with rigorous periodic monitoring of cystine concentrations in granulocytes throughout the patient's life.[9,11,12]

Cystinosis use to be an exclusively pediatric disease and patients would die prematurely from kidney failure before the end of the first decade of life.[1,4] The availability of specific treatment with cysteamine[2,13] and the success of pediatric kidney transplant (KTx) programs[14,15] now prolong patient survival to over 50 years.[4] Consequently, a growing number of adolescents and young adults with cystinosis are being admitted to adult units where the number of experts in this rare disease is extremely low.[16] Furthermore, since unexpected clinical worsening has been observed in many cases after these transfers, it is a matter of concern that the clinical progress achieved in pediatric patients tends to decline in adulthood. Numerous risk factors converge to explain this situation such as: (a) the inherent difficulty in caring for an adolescent/young adult with a chronic disease who has usually received a kidney transplant[5,6] and thus has a high risk of graft loss[7]; (b) the lack of experience and familiarity of adult patient especialists with this RD[4]; (c) extra-renal involvement progression[16]; (d) non-compliance with treatment, which is