Transition in X-Linked Hypophosphatemia

Subjects: Others

Contributor: Takuo Kubota

X-linked hypophosphatemia (XLH) is the most common form of inherited disorders that are characterized by renal phosphate wasting, but it is a rare chronic disease. XLH presents in multisystemic organs, not only in childhood, but also in adulthood. Transition barriers, including a fear of a new health care system and/or hospital, inadequate planning, and system difficulties, are experienced by AYAs and their families. Core elements in HCT consist of transition policy, transition tracking and monitoring, transition readiness, transition planning, transfer and/or integration into adult-centered care, and transition completion and ongoing care with adult clinicians. Regarding HCT for XLH, the timelines of transfer include transition readiness tracking, the initiation of assessments on transition readiness, transition planning, transfer of care, and post transfer.

X-linked hypophosphatemia transition team management metabolic bone diseases

1. Introduction

X-linked hypophosphatemia (XLH) is the most common form of inherited disorders that are characterized by renal phosphate wasting. XLH presents with a number of symptoms, not only in childhood, but also in adulthood. Despite the long-term and progressive disease burden continuing to adulthood, XLH is often perceived as a rare childhood disease [1][2][3]. A lack of recognition of the symptoms and signs of XLH in adulthood delays adequate intervention. Although the clinical manifestations of XLH may persist or recur in later life, standard clinical practice involves the discontinuation of conventional treatment when skeletal growth is completed. This is due to limited evidence for the benefits of continuing conventional treatment into adulthood [3][4]. The resumption of treatment based on symptoms results in gaps in care. Seamless follow-ups are needed. Moreover, since XLH presents in a number of organs, the multidisciplinary management of patients with XLH is essential to improve health outcomes. Therefore, appropriate transition is critical for patients with XLH.

2. Transition

Increased survival from a wide range of chronic illnesses has resulted in greater numbers of children with disabilities reaching 20 years of age [5]. In 2002, a consensus statement by the American Academy of Pediatrics (AAP), the American Academy of Family Physicians (AAFP), and the American College of Physicians (ACP)—American Society of Internal Medicine was published, and the statement mentioned the importance of facilitating the transition of adolescents with special health care needs into adulthood [6]. In 2011, the AAP, AAFP, and ACP with the authoring group published a clinical report entitled “Supporting the Health Care Transition (HCT) from Adolescence to Adulthood in the Medical Home” [7]. This report describes the process for transition preparation,

planning, tracking, and completion for all youths and young adults (AYAs) beginning in early adolescence and provides a structure for training and continuing education to understand the essence of adolescent transition. In 2018, the clinical report was updated to provide more practice-based guidance on key elements of the transition; however, the policy and algorithm was not changed [8]. HCT is defined as “the process of moving from a child model to an adult model of health care with or without a transfer to a new clinician”. The purpose of HCT is to decrease the numbers of patients that are lost to follow-ups and improve the quality of care through organized navigation that is provided to AYA patients and their caregivers. Patients that are lost to the follow-up do not receive appropriate practice management. Experts in XLH are commonly endocrinologists, nephrologists, and geneticists, although this depends on the countries and institutes that see patients with XLH. In general, geneticists are thought to see patients in both childhood and adulthood, rather than endocrinologists and nephrologists, who are usually either pediatric- or adult-specific. In some situations, a clinician that is familiar with pediatric and adult patients with XLH may continue to follow the patients through their life and provide them with proper medical care.

Transition barriers, including a fear of a new health care system and/or hospital, inadequate planning, and system difficulties, are experienced by AYAs and their families [8]. The greatest barrier mentioned is the difficulties that are associated with leaving their pediatric clinicians with whom they have had a long-standing relationship. Clinicians also find many transition barriers, such as communication and/or consultation gaps, training limitations, care delivery, care coordination, staff support gaps, a lack of patient knowledge and engagement, and a lack of comfort with adult care. The most common impediments are the lack of communication and coordination and the different practice behaviors between clinicians. Core elements in HCT consist of transition policy, transition tracking and monitoring, transition readiness, transition planning, transfer and/or integration into adult-centered care, and transition completion and ongoing care with adult clinicians. The process of HCT may be divided into three stages: (1) setting the stage: the initiation of HCT planning and a transition readiness assessment; (2) moving forward: the ongoing provision of HCT services; (3) reaching the goal: the transfer to adult healthcare services [9]. Based on expert opinions and limited research evidence, HCT planning needs to start at approximately 10 to 12 years of age for children with chronic conditions. Fruitful HCT requires collaborations between pediatric and adult-focused providers and settings that encourage AYA to continue to increase skills, even into their mid-20s. Effective HCT needs to be delivered in a similar culture and linguistic background based on the unique necessities of each AYA. An assessment of HCT readiness will direct interventions that lead to better outcomes and quality of life for AYAs.

3. Transition in X-Linked Hypophosphatemia

Regarding HCT for XLH, patient advocacy organizations for XLH, such as the XLH Network in the US and XLHuk in the UK, and for various rare diseases, such as the Genetic and Rare Diseases Information Center and National Organization for Rare Disorders in the US, provide patients with information on their disease. The international XLH alliance, consisting of more than 23 organizations worldwide, has been established to amplify the voices of patients with XLH and set a global multi-disciplinary standard of care and research. The XLH Network developed a toolkit on the transition from pediatric to adult care for patients and their caregivers (http://www.xlhnetwork.org/application/files/1916/0311/3210/XLH_TRANSITIONS_TOOLKIT.pdf, accessed on 4

July 2022), as well as the “Voice of the Patient Report”, about the symptoms and treatment of XLH (http://www.xlhnetwork.org/application/files/5515/9317/2550/VOP_Report.pdf, accessed on 4 July 2022). Gianni et al. emphasized that the transition to adult care is a responsibility that is shared by the pediatric and adult teams involved in XLH, because XLH involves lifetime multi-organ morbidities that are associated with age [3]. Dahir et al. provided expert recommendations on HCT for patients with XLH [1]. Three areas of competency have been described: patient foundational knowledge, information transfer, and timelines and supportive behaviors to drive engagement. The timelines of transfer include transition readiness tracking, the initiation of assessments on transition readiness, transition planning, transfer of care, and post transfer (**Table 1**). Even though ages are mentioned in the timelines, HCT plans need to be individualized. Of note, age- and sex-specific patterns in growth velocity and bone mineral acquisition are distinct between girls and boys, especially in adolescence [10]. Girls reach both peak height velocity and peak bone-mass gain at a younger age than boys. The difference of the patterns in growth and bone accretion between females and males needs to be considered in HCT. It is important to begin the transition process in early adolescence and regularly assess transition readiness. The transition documents for patients with XLH include patient information, healthcare information, disease history, XLH complications, treatment history, the support of advocacy groups, and education, such as XLH symptoms emerging in adulthood [1].

Table 1. Simple timelines of transfer [1].

12 years: transition readiness tracking	- Pediatric practice approach to transitioning to adult care - Educate patients about self-advocacy, self-care, shared decision-making, and self-sufficiency - Educate parents about guidance on encouraging children to succeed in disease ownership
14 years: initiate assessments of transition readiness	- Assess understanding of symptoms, treatment goals, lab results, making appointments, available resources, and legal and insurance age-related changes - Educate patients about disease and management - Assess transition readiness yearly from the ages of 13 to 17 years
17 years: transition planning	- Discuss the optimal time of transition - Checklist of medical, laboratory, and imaging histories for adult providers

	- Discuss potential dosing changes (pediatric to adult) - Identify adult providers - Connect with advocacy groups
18–26 years: transfer of care	- Confirm the first adult provider appointment - Establish a process to orient adolescents/young adults into practice
3–6 months post-transfer	- Confirm the transfer of care - Continue collaborations between pediatric and adult providers

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