Informational

011–X-Linked Hypophosphatemia (XLH): New Knowledge for Nurses Regarding Genetics, Pathophysiology and Clinical Presentation

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Background: Over 80 years ago Fuller Albright reported a vitamin D-resistant form of rickets, which is likely the initial description of X-linked hypophosphatemia (XLH). Recently the genetic basis of the disease has been identified and a more complete understanding of the pathophysiology has emerged.

Purpose: To increase awareness of the latest published data on the genetics, clinical manifestations, differential diagnoses, biochemical findings, as well as growth and development of patients with XLH.

Description of Topic: XLH is a rare, progressive, life-long disorder and the most common form of heritable rickets. The estimated prevalence of XLH is 1:20,000 to 1:25,000. XLH is caused by loss-of-function mutations in the phosphate regulating endopeptidases on the X-chromosome (PHEX) gene, leading to high circulating levels of fibroblast growth factor 23 (FGF23). Over 300 PHEX mutations have been reported. An X-linked dominant inheritance pattern is typical; however, de novo PHEX mutations are reported in up to 20-30% of the cases. Excess FGF23 increases urinary phosphate losses with consequent hypophosphatemia, resulting in rickets and osteomalacia. Clinical manifestations may include lower limb deformities, short stature, bone and joint pain, dental abscesses, delayed walking, and gait abnormalities. Neurological features may include Chiari 1 malformation and craniosynostosis. Low serum phosphate levels, a low renal tubular threshold for phosphate reabsorption (TmP/GFR), and low or normal circulating 1,25(OH)2D are characteristic biochemical findings in patients with XLH. The symptoms of XLH vary among individuals and while there is similar pathophysiology for children and adults, clinical manifestations can differ. Debilitating consequences in adults include osteoarthritis, enthesopathy, spinal stenosis and pseudofractures. In addition, complications of conventional medical therapy (phosphate salts and active vitamin D analogs) include nephrocalcinosis and hyperparathyroidism.

Clinical Implications: Previously, XLH was considered a disorder that manifests only during growth; however, adolescent patients require attention along with a smooth transition to adult care. In addition, recognition of the complex disease features of XLH is essential for accurate diagnosis and management. Pediatric endocrine nurses are well qualified to provide the latest disease state education to patients and families, and to encourage routine clinical evaluation to assess treatment response, disease progression, and therapeutic complications.

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012–Food Literacy: Can Pediatric Endocrinology Nurses Focus Upstream?

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Background: American families are spending less time procuring, preparing and cooking meals at home. As well, there is less time dedicated to nutrition education in schools. Food literacy is defined as "a collection of inter-related knowledge, skills, and behaviors required to plan, manage, select, prepare, and eat foods to meet needs and determine food intake, as well as, the scaffolding that empowers individuals, households, communities or nations to protect diet quality through change and support dietary resilience over time." Adolescents generally lack food literacy skills and increasingly consume food away from home with fast food meals and processed snacks. Approximately 1 in 3 adolescents in the U.S. are obese, increasing their risk for diabetes and cardiovascular disease. Upstream efforts continue to be explored to combat rising obesity rates. Programs that promote food literacy in adolescents have the potential to influence healthy lifestyles.

Purpose: To inform pediatric endocrinology nurses about food literacy, its components, and existing food literacy education programs ready for implementation.

Description of Topic: A literature review on food literacy was completed. High food literacy has been associated with improved eating of a healthy diet, including more fruits and vegetables and less fast food. Food literacy programs should address adolescents' knowledge, skills, and attitudes to make healthy diet choices. Potential topics include gardening, recipe reading, food label reading, learning about healthy vs. unhealthy foods, food preparation and cooking, animal welfare, and farm to table slow food concepts. The USDA SNAP-Ed website provides well developed, user-friendly food literacy teaching ideas with accompanying learning objectives, lesson plans, and teaching strategies that could readily be implemented by pediatric endocrinology nurses.

Clinical Implications: Pediatric endocrinology nurses typically provide care to adolescents with obesity and its related comorbidities. Pediatric endocrinology nurses have an opportunity to expand their practice to primary and secondary prevention of obesity, using upstream health education to promote adolescents' food literacy. By partnering with schools, churches, Boys and Girls clubs, and Scouts troops, pediatric endocrinology nurses may offer health education, building food literacy skills in whole communities with the ultimate goal of slowing the rise in obesity rates.

Food literacy has been described as "the ability of an individual to understand food in a way that they develop a positive relationship with it, including food skills and practices across the lifespan in order to navigate, engage, and participate within a complex food system. It's the ability to make decisions to support the achievement of personal health and a sustainable food system considering environmental, social, economic, cultural, and political components.

The core of food literacy is the adolescent's ability to use food knowledge and skills to make healthy dietary choices and encompasses aspects of planning and managing, selecting, preparing, and eating healthy foods.

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013–Delivering High Quality Pediatric Diabetes Care Beyond Hospital Limits

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Background: Pediatric diabetes is a complex yet delicate condition that requires well-orchestrated interdisciplinary collaboration usually found within facilities providing high level acute care. As part of a large tertiary care centers community expansion, we were