Identifying Continuing Medical Education Needs of Physicians in the Diagnosis and Management of Patients with XLH

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1 INTRODUCTION AND PURPOSE

X-linked hypophosphatemia (XLH), or vitamin D-resistant rickets, is a rare genetic disorder affecting approximately one in 20,000 live births. Given the rarity of XLH, there is little opportunity for physicians in training or practice to become experienced in recognizing symptoms and clinical features. The goal of this study was to assess current awareness and practice patterns of US physicians in diagnosing and managing patients with XLH in order to prioritize needs that can be addressed in continuing medical education (CME) activities.

2 METHODOLOGY

A survey instrument was developed in collaboration with a clinical expert in XLH. The survey included 2 patient case scenarios designed to assess practice patterns of clinicians that manage pediatric patients and adult patients. The survey topic was initially blinded to respondents in recruitment material.

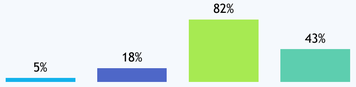
The web-based survey was randomly distributed to practicing family physicians, pediatricians, and endocrinologists – including pediatric endocrinologists – in the US during April 2018.

Statistical and qualitative analyses were conducted to understand practice patterns and perceptions by specialty, as well as the impact of physician demographics and experience in managing XLH on disease management.

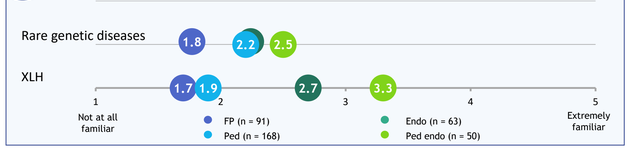
3 RESPONDENT DEMOGRAPHICS

	Family Physicians (n = 91)	Pediatricians (n = 168)	Pediatric Endocrinologists (n = 50)	Endocrinologists (n = 63)
Patients seen per week (mean)	94	88	49	87
Academic practice setting	5%	18%	70%	25%
Years in practice (mean)	29	25	23	28
Practice Setting				
Urban	20%	41%	74%	38%
Suburban	59%	53%	22%	56%
Rural	22%	6%	4%	6%

Involved in diagnosing or managing one or more patients with XLH



4 FAMILIARITY WITH RARE GENETIC DISEASES

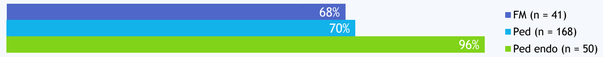


5 PEDIATRIC PATIENT

CASE: A 2-year-old boy is brought to your clinic by his mother. On exam, the boy appears happy and is interactive. His height is at the 15th percentile, and his weight at the 50th percentile for his age. His facial features are normal-appearing, and there is considerable bowing of the legs. A review of his record reveals that his height was at the 20th percentile and weight was at the 50th percentile at his 18-month visit. You also noted slight bowing of the legs at that visit, though this seems to have progressed.

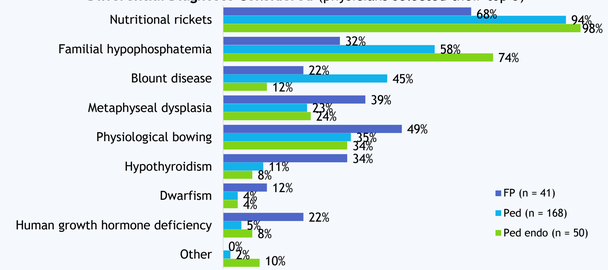
Additional information that would be gathered this visit	FP (n = 41)	Ped (n = 168)	Ped endo (n = 50)
Dietary history and nutritional intake (milk, vitamin D)	66%	85%	78%
Family history	**49%**	**43%**	**64%**
Developmental and birth history	22%	22%	20%
Sun exposure	2%	13%	14%
Medical history - medications, exams, symptoms	2%	11%	24%
Walk/gait issues	10%	10%	22%

Would order lab or diagnostic studies at this visit



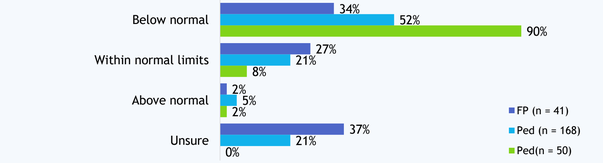
When asked about gathering additional information, less than ½ of primary care physicians would ask about family history. Just over 2/3 of family physicians and pediatricians would order additional studies at this visit.

Differential Diagnoses Considered (physicians selected their top 3)



Most physicians were considering nutritional rickets, with only 1/3 of family physicians and just over ½ of pediatricians considering familial hypophosphatemia.

Interpreting serum phosphate concentration of 2.7 mg/dL in a 2-year-old patient

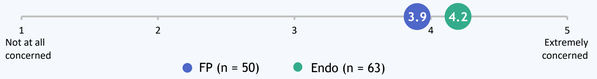


While a normal serum phosphate concentration for adults is 2.5-4.5 mg/dL, a serum phosphate concentration of 2.7 mg/dL is below normal in a 2-year-old patient. Only about 1/2 of pediatricians and 1/3 of family physicians recognized this.

6 ADULT PATIENT

CASE: A 40-year-old male presents to your clinic with complaints of pain in his shoulders, hips, legs, and back. His symptoms began about three years ago and have gotten progressively worse to the point that he is having difficulty performing the physical requirements for his job as an electrician. His past medical history is notable for a stress fracture in his leg while serving in the military in his 20s. He also reports multiple dental abscesses beginning when he was a child, leading to complete dental extraction about 10 years ago. On exam, he is shorter than average and overweight. Range of motion of the spine is limited. There is severe bowing of the legs.

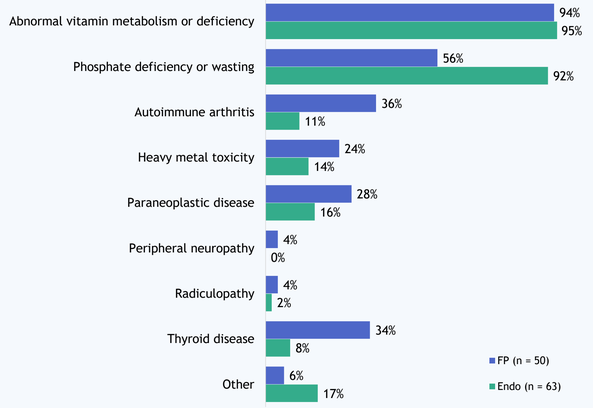
Level of concern with patient presentation



Additional information that would be gathered	FP (n = 50)	Endo (n = 63)
Dietary history and nutritional intake (milk, vitamin D)	18%	21%
Family history	**54%**	**67%**
Lab work - calcium, vitamin D, phosphate	36%	32%
Medical history - medications, exams, symptoms	68%	48%

Both family physicians and endocrinologists were generally very concerned with the patient's presentation. About 2/3 of endocrinologists would ask about family history, whereas only about half of family physicians would inquire about family history and were more likely to ask about medical history.

Differential Diagnoses Considered (physicians selected their top 3)



Almost all physicians included the potential for abnormal vitamin metabolism or deficiency in their consideration of diagnoses based on patient presentation. The vast majority of endocrinologists also considered phosphate deficiency or wasting, whereas only about 1/2 of family physicians considered this.

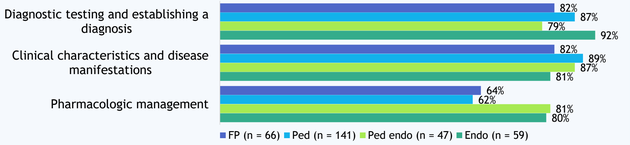
8 CONTINUING EDUCATION

Likelihood to seek out informational sources or participate in education on the topic of rare genetic disorders in the coming year

Information sources most likely to use to stay up-to-date on rare genetic disorders

FP/Ped	Ped endo/Endo
1. Clinical practice guidelines	1. Articles in peer-reviewed journals
2. CME activities	2. Clinical practice guidelines
3. Conferences, meetings, and symposia	3. Conferences, meetings, and symposia
	4. CME activities/nationally recognized expert

Topics related to rare genetic diseases would be most valuable to include in CME programs



9 CONCLUSIONS

Physicians' low familiarity with rare genetic disorders and specifically XLH is a contributing factor to the gaps identified through this survey, including recognition of the signs and symptoms of XLH and understanding of the need to ask about family history. These gaps may contribute to delays in patient diagnosis and subsequent treatment of XLH.

Additionally, study results demonstrated gaps related to lab testing, interpreting test results, and patient management.

CME can be a format for simulating practice experience as a means to address physicians' limited exposure to XLH patients.

CME that includes recognizing clinical signs and establishing a diagnosis of XLH may help close the practice gaps noted in this study and potentially shorten the time to diagnosis for patients with XLH.

Attracting physicians to educational activities will likely be an ongoing challenge, given physicians' likelihood to seek out information and education on topics that impact higher percentages of their patients than rare genetic diseases. However, if not addressed through thoughtful educational interventions, delays in XLH diagnosis and gaps in patient management will persist.

DISCLOSURES: This research was supported by Ultragenyx Corporation. Dustin Ensign and Stan Krolczyk are employees of Ultragenyx Corporation. All other authors have nothing to disclose. For more information on this study, please contact Melissa Ellis, PhD at melissa.ellis@ceoutcomes.com.