Potential Predictors of Pediatric PH1*

*Based on EHR data from patient visits between January 2009 and November 2021

It is challenging to identify patients with PH1 due to its **rarity** and similar presentation to individuals with **kidney stones due to other causes**. These challenges often **lead to a delay in diagnosis**.

In a case-control study using records of 8 US pediatric health systems from PEDSnet, a national clinical research network with standardized EHR data, **37** patients with PH1 and **147** matched controls were compared*.

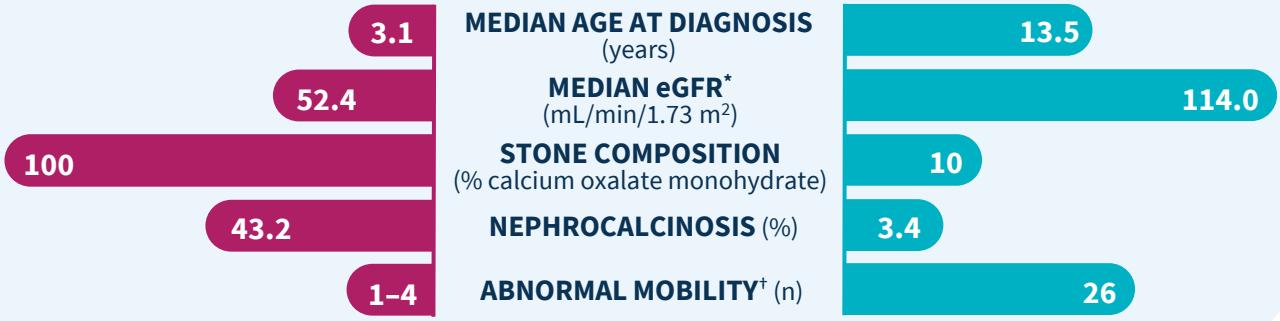
PH1 patients were differentiated by **5 clinical features**, which were more likely to be present in **combination** than in isolation. Based on these results, a **risk index** was developed.

Patients who present with:

- ☑ **Young age**
- ☑ **Low eGFR**
- ☑ **CaOx monohydrate kidney stones**
- ☑ **Nephrocalcinosis**
- ☑ **Normal mobility†**

Should warrant a physician to **consider genetic testing**.

*Individuals <21 years of age with PH1 by genetic testing were matched by sex and PEDSnet institution with up to 4 individuals <21 years of age with kidney stones not due to PH of any type.
† Abnormal mobility defined as conditions such as spina bifida or metabolic disorders resulting in reduced voluntary mobility.



All listed metrics were statistically significant ($p < 0.05$).
*From 1 year prior to 180 days after diagnosis. Calculated based on the CKiD U25 creatinine-based equation.
† Defined as conditions such as spina bifida or metabolic disorders resulting in reduced voluntary mobility.

Study limitations: most control patients did not have genetic testing; urine chemistries were not performed on all patients; diagnostic coding errors may exclude some patients with PH1.

This study offers a **potential computable phenotype for PH1** in children, which, if externally validated, **may help facilitate earlier diagnosis** of children with PH1.

This resource is intended to support scientific exchange. The information provided is not intended to serve as recommendations for clinical practice.

References: Tasian GE, et al. *J Ped Urol.* 2023;20(1):88E.1-88E.9.
CaOx, calcium oxalate; eGFR, estimated glomerular filtration rate; EHR, electronic health record; PH1, primary hyperoxaluria type 1; US, United States.

PH1 patients | Control patients