

Hypophosphatasia Impact Patient Survey (HIPS) Results Summary

Skrinar A, Smith J, Smith S, Landy H. Burden of
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Hypophosphatasia. 2010. ACMG, Abst. 255.



About the Hypophosphatasia Impact Patient Survey (HIPS)

The following pages outline the first results from Enobia's online Hypophosphatasia Impact Patient Survey (HIPS). HIPS was launched in September 2009 and was developed to assess the disease burden and quality of life in children and adults with hypophosphatasia (HPP).

The survey included questions about each patient's experience with HPP including symptom onset, medical and surgical history, fractures, medications and medical device use. Patient data was anonymously reported.

Enobia appreciates the participation of patients from US Soft Bones, HPP ev, Hypophosphatasie Europe, CORD, NORD, and The MAGIC Foundation.






An Overview of the Demographics of HIPS Respondents

Completed surveys were obtained from 87 patients over a five-month period and included 55 adult (≥ 18 years old) and 32 pediatric (< 18 years old) patients. Caregivers completed the survey for 36 patients, and the remainder were self-reported.

People participated in HIPS from around the world, including the United States, Germany, France, Canada and Japan.

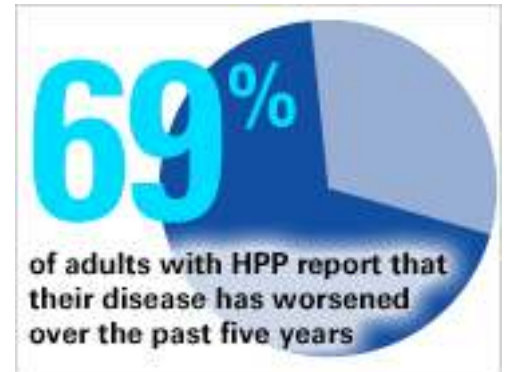
Additional details about the demographics of the survey respondents is included below.

Demographics of the 87 HIPS Respondents	
Gender	Percentage
Male	35%
Female	66%
Age Group	
Adults (≥ 18 y.o.)	63%
Children (< 18 y.o.)	37%
Age of Symptom Onset	
< 1 year old	35%
1-12 years old	46%
> 12 years old	19%

Participating Countries	
Country	Percent of Survey Respondents
 U.S.	45%
 Germany	23%
 France	10%
 Canada	9%
 Japan	5%
Other Countries	8%

Survey Suggests That HPP Is Progressive

Most patients reported worsening of their symptoms over the past five years, while no respondents reported improvement.



Developmental Symptoms and Their Impact

Adults with **early-onset** of HPP symptoms (onset before age 1) were more likely to report improper bone growth and bone formation compared to adults with later onset of symptoms, including poor weight gain, delayed walking, bowing of legs, knock knees, and short stature.

More information about the prevalence of developmental symptoms and their impact is below.

Developmental Symptoms of Adult Respondents*			
Symptom	Overall Percentage (55 Adults)	Percentage Among Patients with Onset Before Age 1 (16 Adults)	Percentage Among Adults with Onset After Age 1 (38 Adults)
Poor Weight Gain (feeding difficulty as an infant/child)	38%	81%	21%
Delayed Walking	44%	69%	34%
Bowing of Legs	36%	69%	24%
Knock Knees	22%	44%	13%
Short Stature	67%	88%	24%

Note: One patient did not report age of symptom onset.

Pain Symptoms

Patients surveyed reported pain, regardless of current age or the age of symptom onset.

- 94% of adults – and 82% of all patients – reported bone pain severe enough to limit activities of daily living.
- 76% of patients reported use of bone pain medication.
- 80% of patients reported joint pain.
- 56% of patients reported muscle pain.



HPP Impacts Mobility

Patients with HPP reported a number of functional symptoms, including 56% with abnormal gait (walking style), 60% with muscle weakness, and 36% with extreme joint flexibility.



47% of adults with early-onset of symptoms use a wheelchair as compared with 34% of children and 6% of adults with later symptom onset.

32% of patients use a walking device. Use of a walking device was not related to age of symptom onset.

Surgical Repairs and Fractures

Patients reported fractures, regardless of the age of symptom onset.

- 87% of adults reported at least one bone fracture during their lifetime.



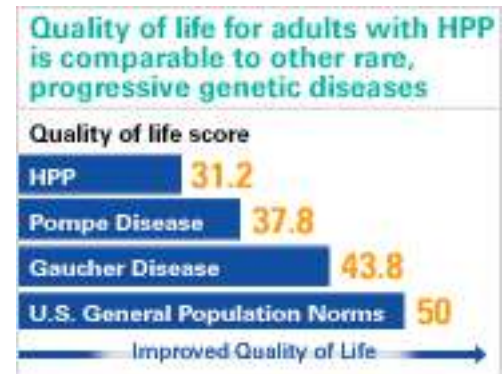
Overall, 55% of all respondents – and 75% of adults with early-onset of symptoms – reported bone fractures ***requiring repair with hardware such as rods, screws or plates.***

48% of adults experience their first fracture during adulthood (after 18 years of age).

Quality of Life

Health-related quality of life was measured using the SF-12, a health assessment questionnaire commonly used by clinicians.

When compared to historical data, health-related quality of life was lower than that of the general population and of patients with late-onset Pompe disease and Gaucher disease, two progressive genetic diseases with musculoskeletal symptoms.^{1,2}



Sources:

¹ Weinreb N, *et al.* Imiglucerase (Cerezyme) improves quality of life in patients with skeletal manifestations of Gaucher disease. *Clin Genet* 2007;71:576-588.

² Hagemans, *et al.* Late-onset Pompe disease primarily affects quality of life in physical health domains. *Neurology* 2004;63:1688-1692.

Conclusions

Among all respondents, symptom duration appears to be a significant predictor of several HPP-related symptoms, including number of fractures. This suggests that HPP is a progressive disease.

HIPS data collection is ongoing.

Enobia would like to thank all who have participated in HIPS.