

Health related quality of life in children and adults with Hypophosphatasia: Results from a European patient survey

Parthenaki I, Balvanyos J, Palkmets O, Hutchings A¹.
 1. Dolon Ltd., London, UK. adam.hutchings@dolon.com

Background and Objective

- Hypophosphatasia (HPP) is an ultra-rare, progressive disease characterised by a variety of serious symptoms.⁽¹⁾
- HPP is classified into 3 categories based on age of manifestation (at infancy, childhood or adulthood).⁽¹⁾
- Due to its clinically progressive nature, HPP is thought to impact the Health Related Quality of Life (HRQoL) of patients, and that of their families or caregivers, throughout the patient's lifetime.
- As there is a lack of published literature describing the impact of HPP on the HRQoL of patients and their families, a survey was conducted to evaluate the impact of HPP on the physical, emotional, social and educational/professional aspects of the lives of patients and their carers.

Methods

- Between April and May 2015, patients with HPP and their families were invited to complete an online or script questionnaire to assess the impact of HPP on their HRQoL. The questionnaire included questions from validated HRQoL instruments (EQ-5D-3L), supplemented with questions specifically aimed at understanding the impact of HPP on patient's / carers everyday life.
- The survey was completed directly by adult patients. For children <18 years, responses were submitted on their behalf by their parent or with parental assistance.
- Adults, children and families were mainly recruited from European countries; however, patients from the US and Tunisia were also included (see table 1).

Results

- 35 patients participated in the survey, of whom 22% were children.

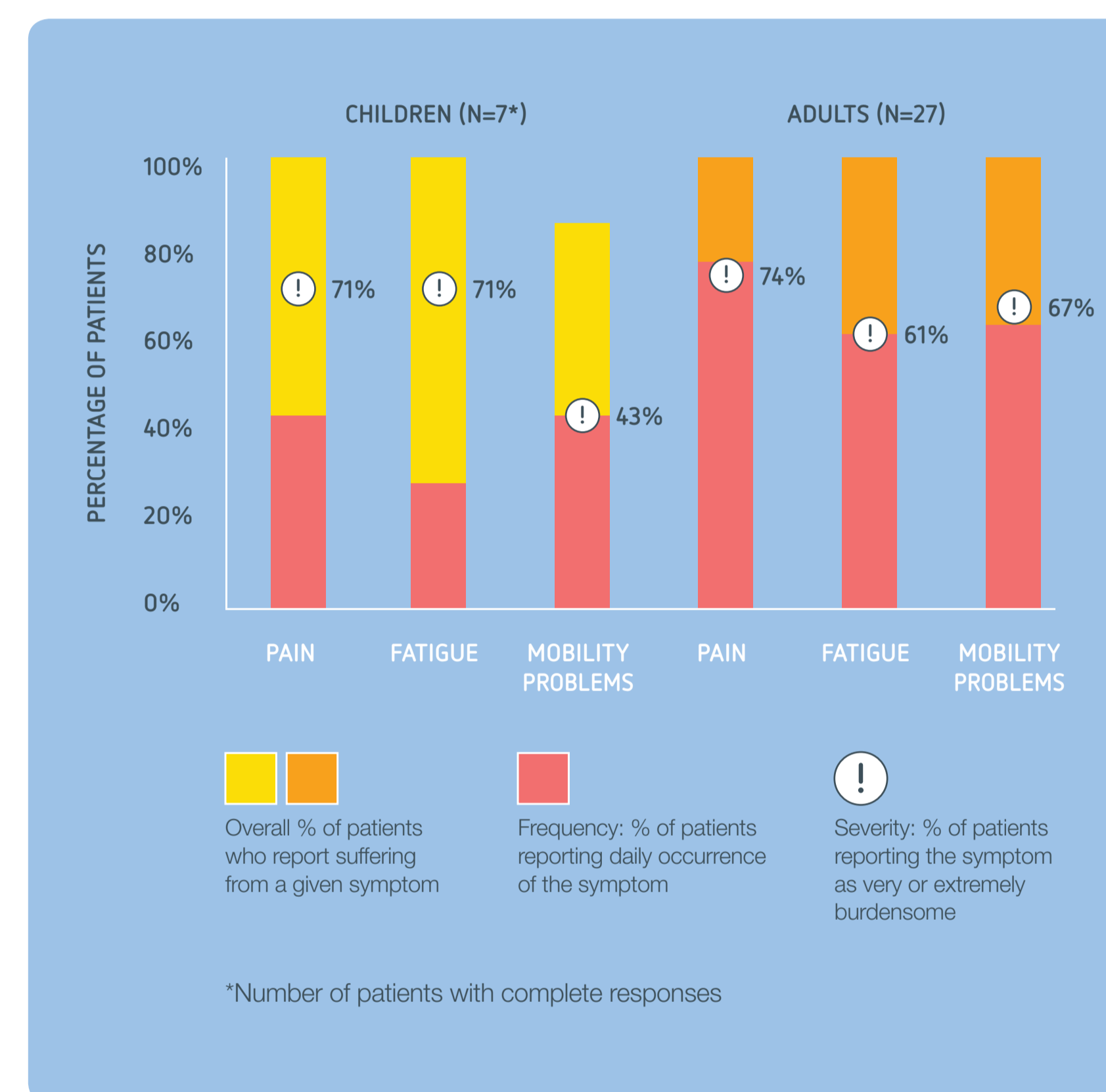
Table 1: Demographics of survey participants

Number of participants	35
Children	8
Adults	27
Female / Male	26 / 9
Nationality	(no. of patients)
German	15
British	8
French	9
American	1
Swiss	1
Tunisian	1
Median age (years) – children	7.5 (Range 3 months–14 years)
Median age (years) – adults	51 (Range 18–72 years)

Physical impact of HPP

Across responder patients (n=34) the most widely reported and impactful HPP symptoms were pain, fatigue and mobility problems (Figure 1). Patients also reported dental problems, fractures, gastrointestinal and respiratory problems; however, these occurred less frequently and were less severe than pain, fatigue and mobility problems.

Figure 1: Frequency and severity of the most commonly reported symptoms across all patients with HPP

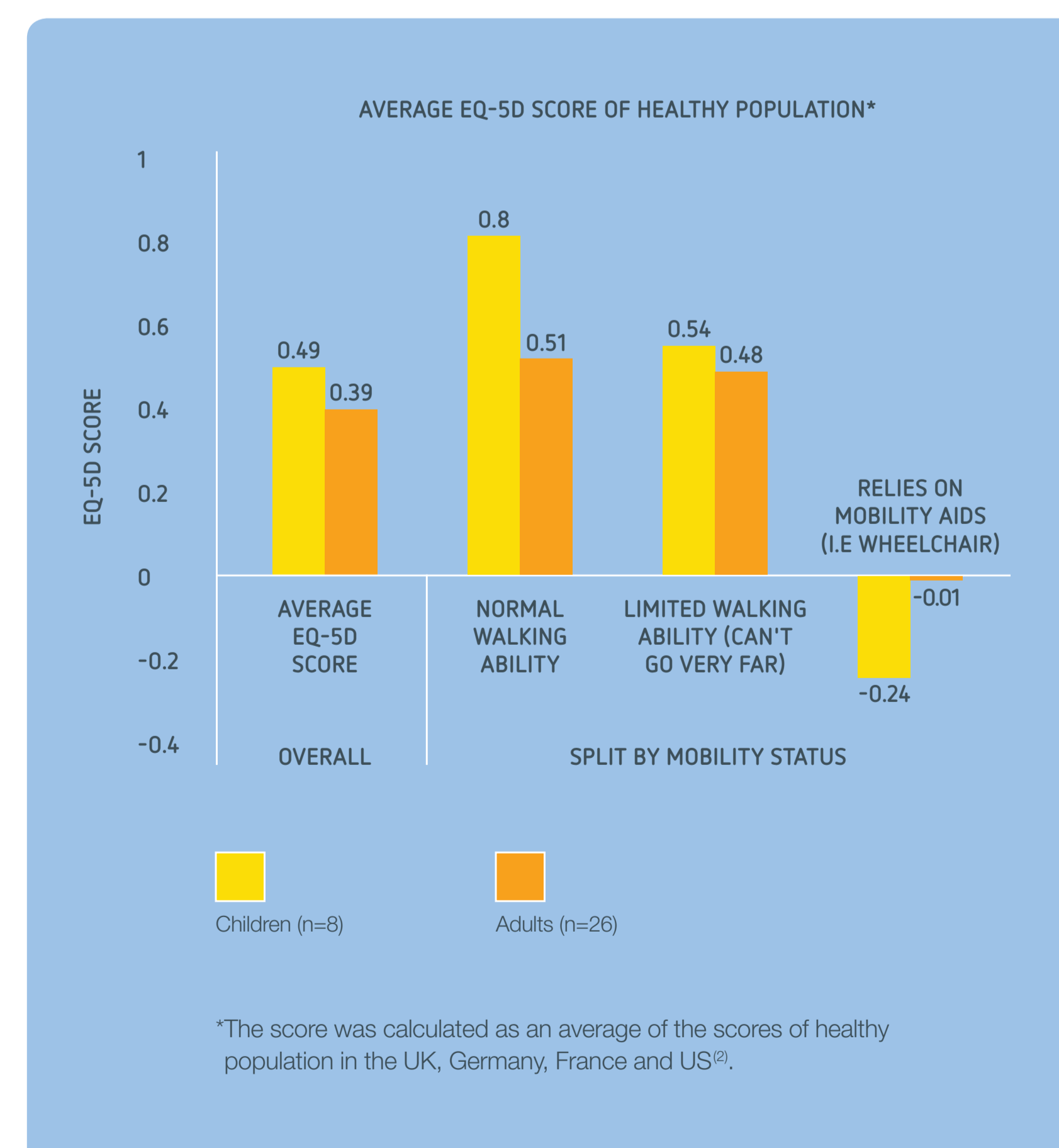


HRQoL impact of HPP

Patients with HPP (n=34) reported significantly impaired HRQoL scores when compared with the average score of the healthy population (0.86) (Figure 2)⁽²⁾.

- Mobility was one of the most important parameters affecting patients HRQoL. Patients with severe mobility problems (i.e. reliant on mobility aids) reported substantially lower EQ-5D scores compared with patients able to walk normally.

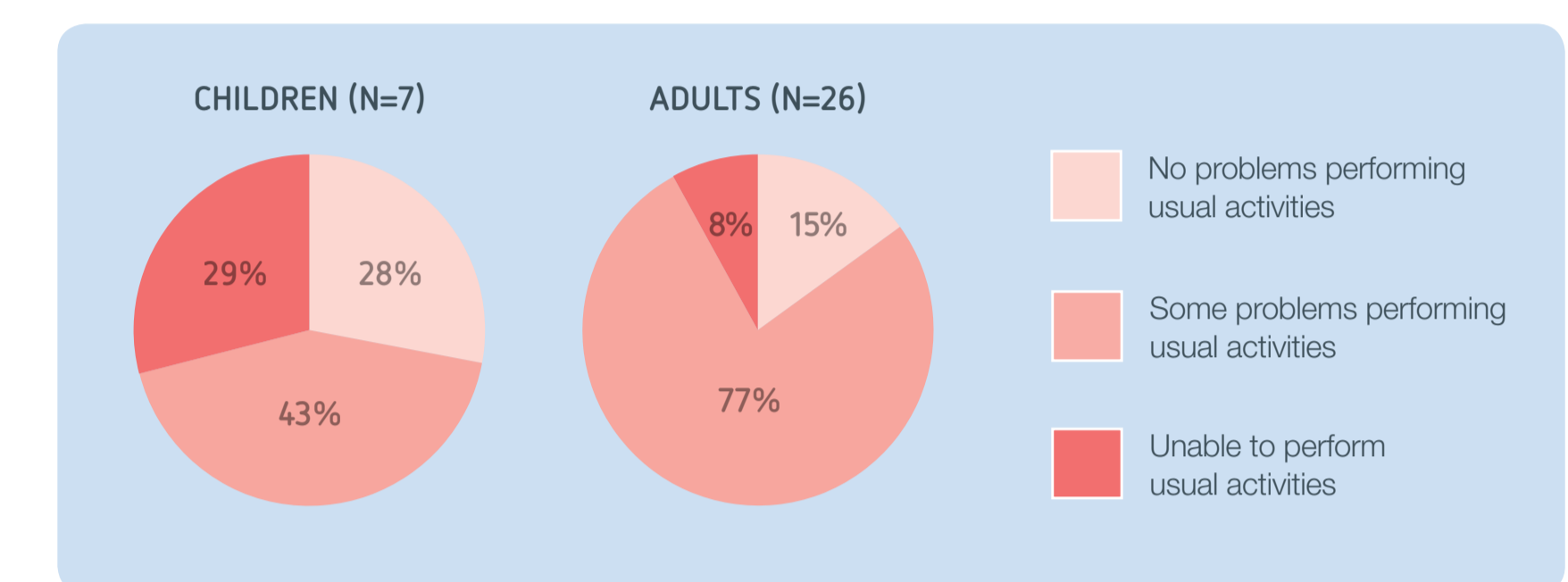
Figure 2: Average EQ-5D scores across all patients with HPP



Impact of HPP on usual activities

72% of children and 85% of adults, reported some or complete impairment of their usual activities due to HPP.

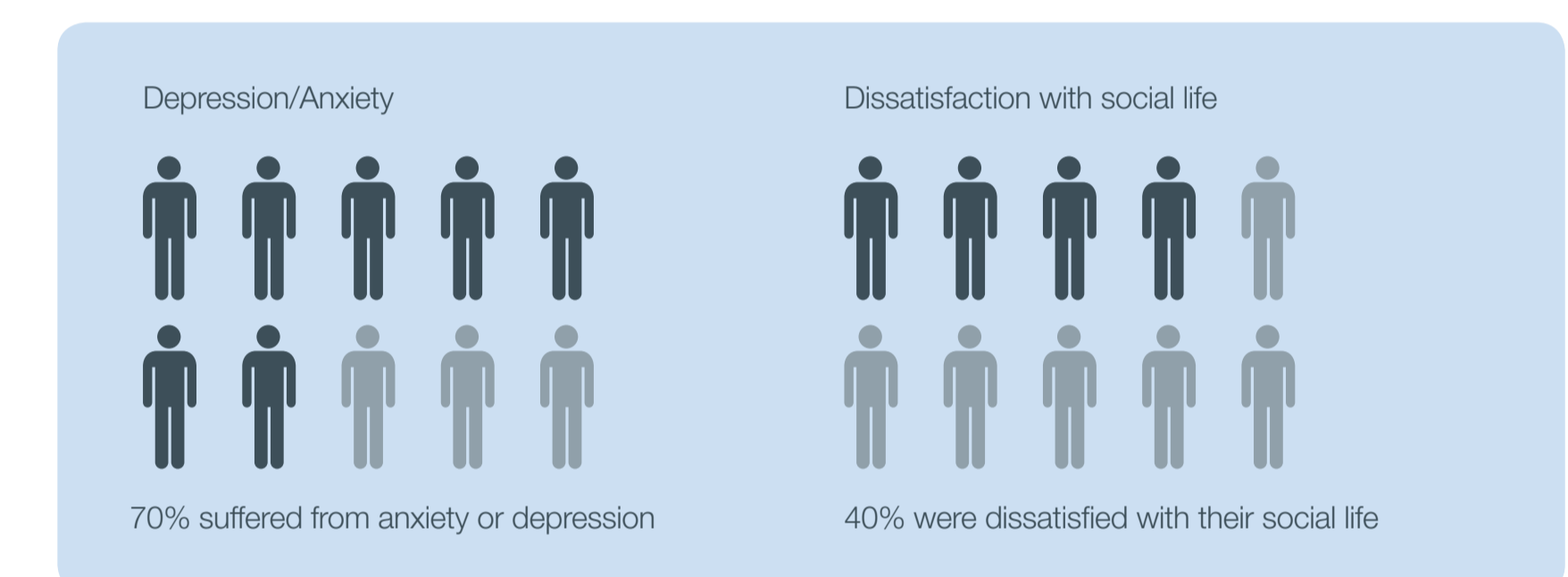
Figure 3: Impact of HPP on usual activities



Impact of HPP on mental wellbeing and social life

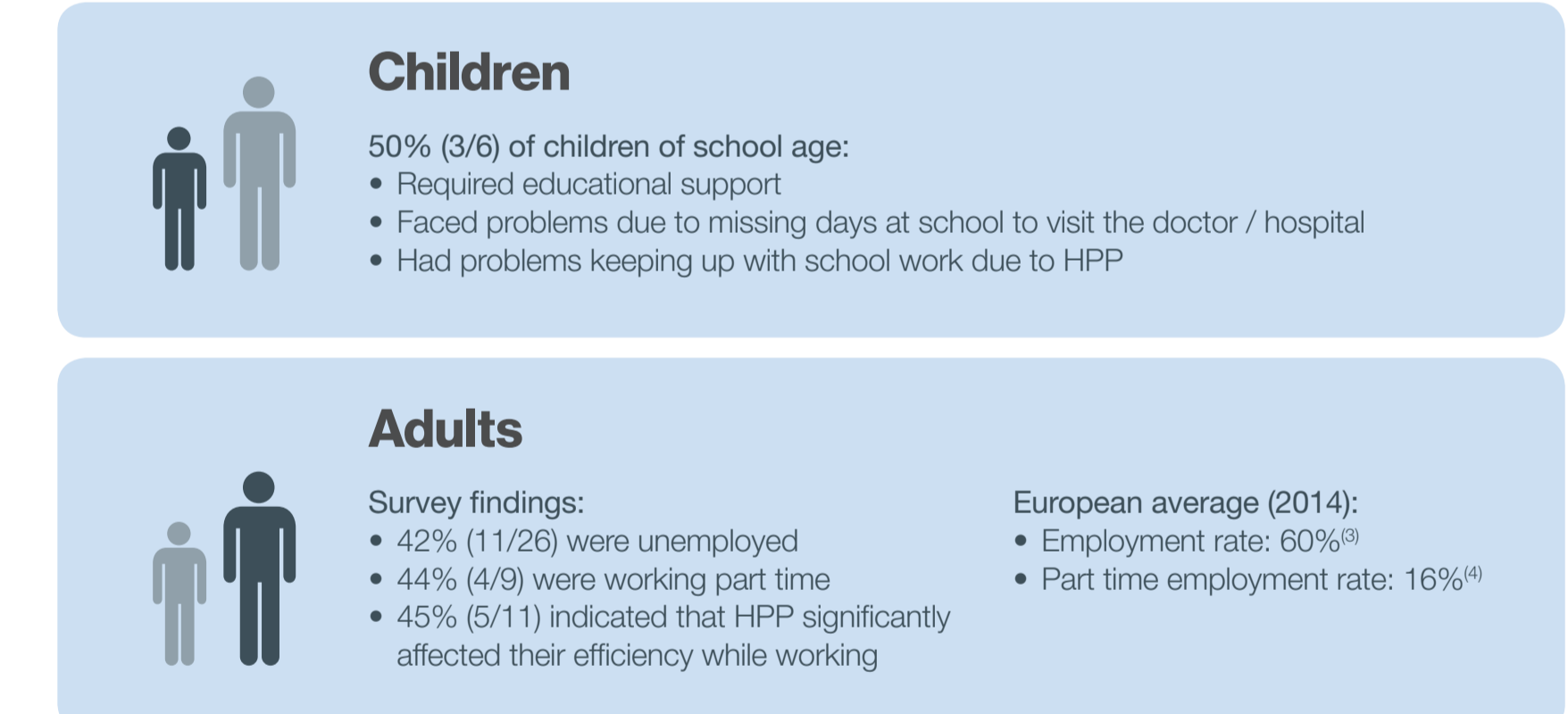
HPP considerably affected patients' emotional wellbeing & social lives.

Figure 4: Impact of HPP on mental health and social life of patients



The self-esteem of children with HPP was also affected by the disease; half of the parents completing the questionnaire (4/8) reported their child's self-esteem was a lot (n=1) or even extremely (n=3) affected by the disease.

Impact of HPP on education and employment



Impact of HPP on carers

Caring for patients with HPP was found to have an impact on carers QoL, affecting their mental health, impacting family life and employment status.

- Mental Health: All carers that responded (n=8) reported experiencing mental health problems such as mental exhaustion (n=7), stress (n=8) and anxiety or depression (n=7).
- 40% of mothers ALWAYS suffered from mental health problems i.e. exhaustion, stress and anxiety
- Family cohesion: 50% (4/8) of parents reported they took fewer holidays because of the restrictions imposed by their child's disease. In addition, 77% (7/9) of parents had to reduce the time they spent with their other children and family members in order to take care of their child with HPP.
- Employment: Two non-working carers/parents indicated they had given up their jobs because of their child's HPP.
- One carer/parent (working part-time) reduced her working hours by 14 hours per week due to her child's HPP.
- Another carer/parent retired early to take care of her child.

Conclusion

- The survey results indicated that HPP has a profound impact on patients' HRQoL, impacting the physical and emotional wellbeing of those affected and resulting in impaired social and working lives.
- In addition, caring for patients with HPP has a considerable impact on the mental wellbeing, family life & employment of carers.
- There remains a need for more in-depth research on the impact of HPP on the HRQoL of patients and carers to better understand the impact of the disease.