

Measuring the caregiver burden in children, adolescents and young adults with Achondroplasia: Results from a prospective, cross-sectional, multi-country study

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Background

Achondroplasia is a skeletal dysplasia and the most common cause of disproportionate short stature affecting more than 250,000 individuals globally¹

- Achondroplasia is characterized by disproportionate short stature, with standing height often below the fifth percentile¹
 - Throughout life, individuals with achondroplasia encounter a wide range of medical complications² a noticeable impacts on their health-related quality of life (HRQoL)^{3,4}
- To date, studies that explore the full extent of caring for an individual with achondroplasia have been limited and focused on qualitative insights.
- Until recently, no disease modifying treatments have been available for achondroplasia. Vosoritide (Voxzogo™), a CNP analogue, targets overactive FGFR3 to stimulate endochondral bone growth. In infants and children with achondroplasia, vosoritide significantly improves growth from birth, resulting in sustained increases in height over time⁵
- Given the ever-shifting treatment landscape, it is important to better understand the impact of caring for a child with achondroplasia on the caregiver
- This study quantitatively assessed caregiver burden and explored whether it is impacted by the relative height vs. population mean (height z-score) of the person with achondroplasia.

Table 1. Participant Demographics

Characteristic/Concept	n (%) [a]
Age of the caregiver	
n (missing)	53 (0)
Mean (SD)	41.09 (8.34)
Median	40
Min - Max	29.00 - 62.00
Have you or the other primary/co-primary caregiver (i.e., child's other parent) also been diagnosed with achondroplasia?	
No, neither of the caregivers have achondroplasia	48 (90.6%)
Yes, I have achondroplasia	2 (3.5%)
Yes, the other caregiver has achondroplasia	3 (5.7%)
What is your current marital status?	
Single	2 (3.8%)
Divorced/separated	12 (22.6%)
Partnership/Co-habiting	36 (67.6%)
Married/De facto relationship	3 (5.7%)
Age of whom you are caring	
Missing	5 (9.4%)
< 3 years	12 (22.6%)
3-8 years	20 (37.7%)
> 8 years	16 (30.2%)

[a] Missing data included in calculation of percentages

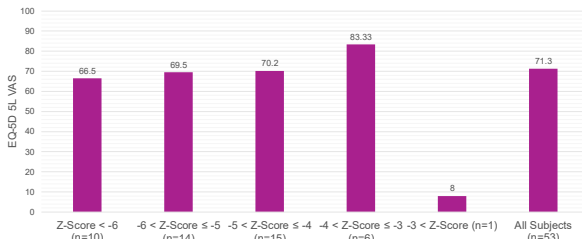
Methods

An international, prospective, cross-sectional, non-interventional study.

- Participants were recruited from Australia, Austria and Sweden via patient advocacy groups (PAGs), recruitment agencies (Austria and Sweden) or clinical sites (Australia).

- An online survey captured socio-demographics, medical history, health state (EQ-5D-5L), well-being (SF-12), work productivity (WPAI:SPH), and impacts of caregiving on the family via the PedsQL Family Impact Module (PedsQL FM).
- Data was summarized using descriptive statistics. Continuous variables are presented as n, mean, standard deviation (SD), median, interquartile range (IQR), and minimum/maximum values. Categorical variables are presented as frequencies (n) and percentages (%).

Figure 1. EQ-5D 5L mean visual analogue scale (VAS) scores by Height Z-Score



Results

- A total of 53 participants were enrolled from Austria (n=8), Australia (n=2) and Sweden (n=43) with a mean (SD) age of 41.09 (8.34).
- The majority of patients were married (36 (67.9%) with neither the participant or their spouse having a diagnosis of achondroplasia (90.6%, n=48).
- 67.9% (n=36) of participants were working full time. However, since the person they are caring for was diagnosed with achondroplasia:
 - 26.4 (n=14) have had to reduce their working hours; and
 - 35.8% stated their household income had decreased
- Work productivity was also impacted with mean (SD) WPAI:SPH overall productivity loss score of 33.91% (29.06).
 - Participants caring for a child or adolescent with a height z-score of Z-Score < -6 (n=10) reported mean (SD) absenteeism and presenteeism scores of 21.60 (22.18) and 32.86 (34.02) respectively, and overall work productivity loss of 47.36 (39.39).
- All Z-Score groups reported EQ-5D 5L UIS lower than the mean (SD) population norm of 0.91 (0.100)⁶.
- Participants reported the lowest HRQoL in the SF-12 'Role, Emotional', Mental Health and General Health domains with mean (SD) of 42.89 (12.07), 43.49 (10.04) and 43.60 (10.36) respectively.
 - Mean Mental Component Score was 40.92 (11.79). When analyzed by height z-score, all groups reported MCS means below the population norm of 49.79 (SD=9.58)⁷
- The biggest PedsQL FM decrements in mean scores vs. population norms were seen in the Communication and Worry domain where mean scores for all study subjects were respectively 24.7% and 29.5% lower than the mean for the general population.

Figure 2. SF 12 PCS and MCS by Height Z-Score vs. Population norms

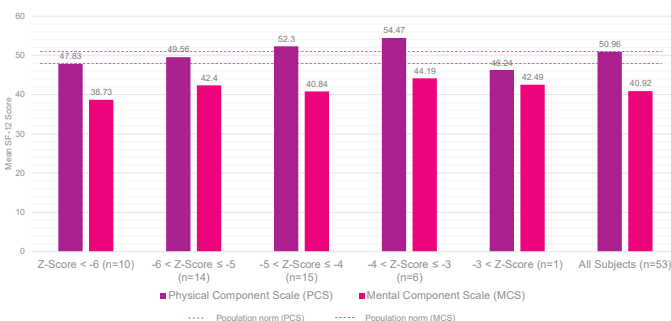
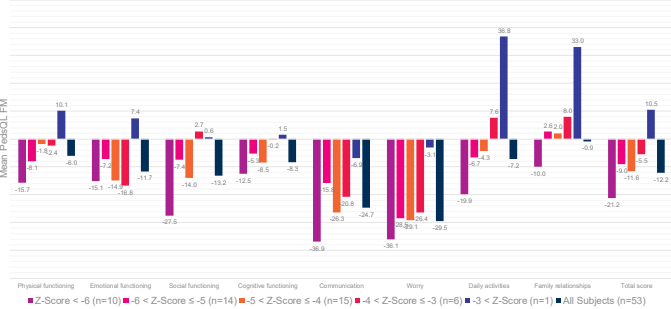


Figure 3. PedsQL FM mean score difference from population norm



Physical functioning Emotional functioning Social functioning Cognitive functioning Communication Worry Daily activities Family relationships Total score

Z-Score < -6 (n=10) -6 < Z-Score ≤ -5 (n=14) -5 < Z-Score ≤ -4 (n=15) -4 < Z-Score ≤ -3 (n=6) -3 < Z-Score ≤ -2 (n=1) All Subjects (n=53)

Conclusions

- Participants reported reduced working hours and a drop of income as a result of caregiver responsibilities.
- When grouped by height z-score, caregiver burden increased as height z-scores moved further away from population norms.
- None of the participants' children were being treated with Vosoritide (Voxzogo™) at the time of the study.
- These results describe the burden of caring for a person with achondroplasia on the caregiver's health, wellbeing, financial and employment status and demonstrate the relationship between height z-scores and caregiver outcomes.

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