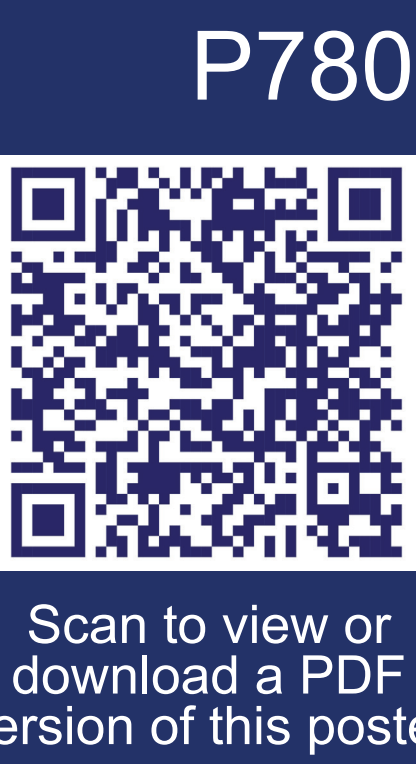


Patient and caregiver experiences with setmelanotide treatment in Bardet-Biedl syndrome – real-world evidence and a patient support program

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Background

- Bardet-Biedl syndrome (BBS) is a rare genetic disease resulting from dysfunctional cilia, and is marked by numerous symptoms, which include hyperphagia and early-onset obesity^{1,2}
 - Hyperphagia is characterised by an insatiable feeling of hunger, impaired satiety, and abnormal food-seeking behaviours
- Patients with BBS experience significant disease-related health burden that both reduces health-related quality of life (HRQoL) and negatively affects the lives of caregivers, parents, and other family members³
- The MC4R agonist setmelanotide has demonstrated clinically meaningful weight and hunger reductions and improvements in HRQoL in patients with BBS^{4,5}
- Coordination of care for patients with rare diseases is often suboptimal, and patients affected by rare diseases like BBS generally have high unmet support needs⁶
- In Germany, a specialist nurse support service was established to support patients with BBS and their caregivers at the start of treatment with daily subcutaneous injections of setmelanotide

Objective

- To evaluate real-world patient experiences before and during treatment with setmelanotide, as well as the use of the specialist nurse support service

Methods

- A one-time online survey was administered to patients with BBS who began treatment between June 2023 and December 2023, or their caregivers, to capture their real-world experiences of setmelanotide treatment and the support service
 - The support service aimed to enable patients and their caregivers to administer setmelanotide injections independently and provided daily or weekly education and support from a certified nurse specialist
 - Caregivers reported for paediatric patients who were too young or who could not self-report; ages were 6–9 years (n=4), 10–13 years (n=4), 14–18 (n=2), not reported (n=2)
- The survey, including yes/no questions, Likert scale questions (1=“does not apply” and 5=“applies to a high degree”) and free text questions, was fielded from January 2024 to May 2024
 - Topics covered were disease symptoms prior to starting treatment, changes noticed after starting treatment, treatment experience, and experience with the support service
- Anonymised survey results were analysed using descriptive statistics

Results

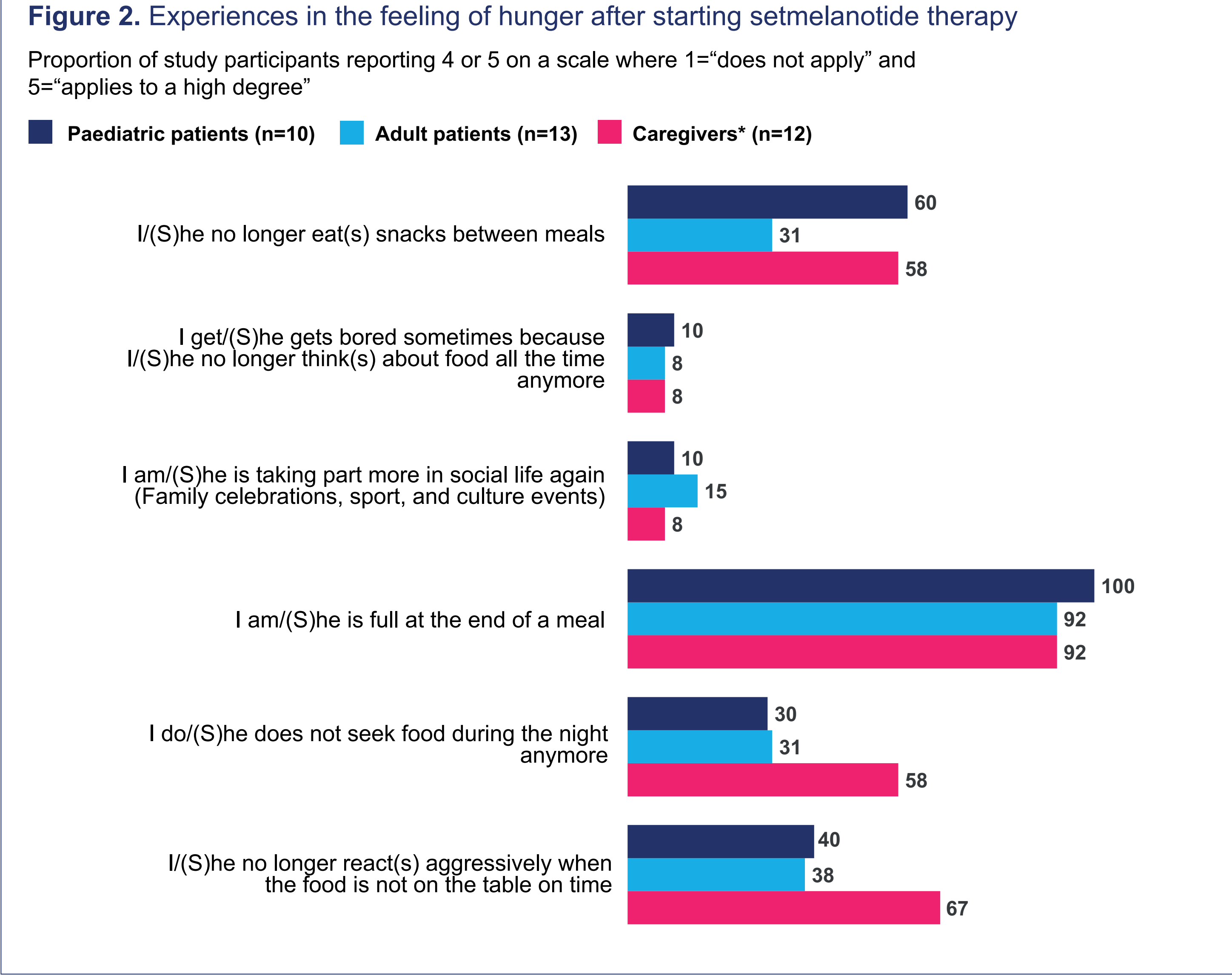
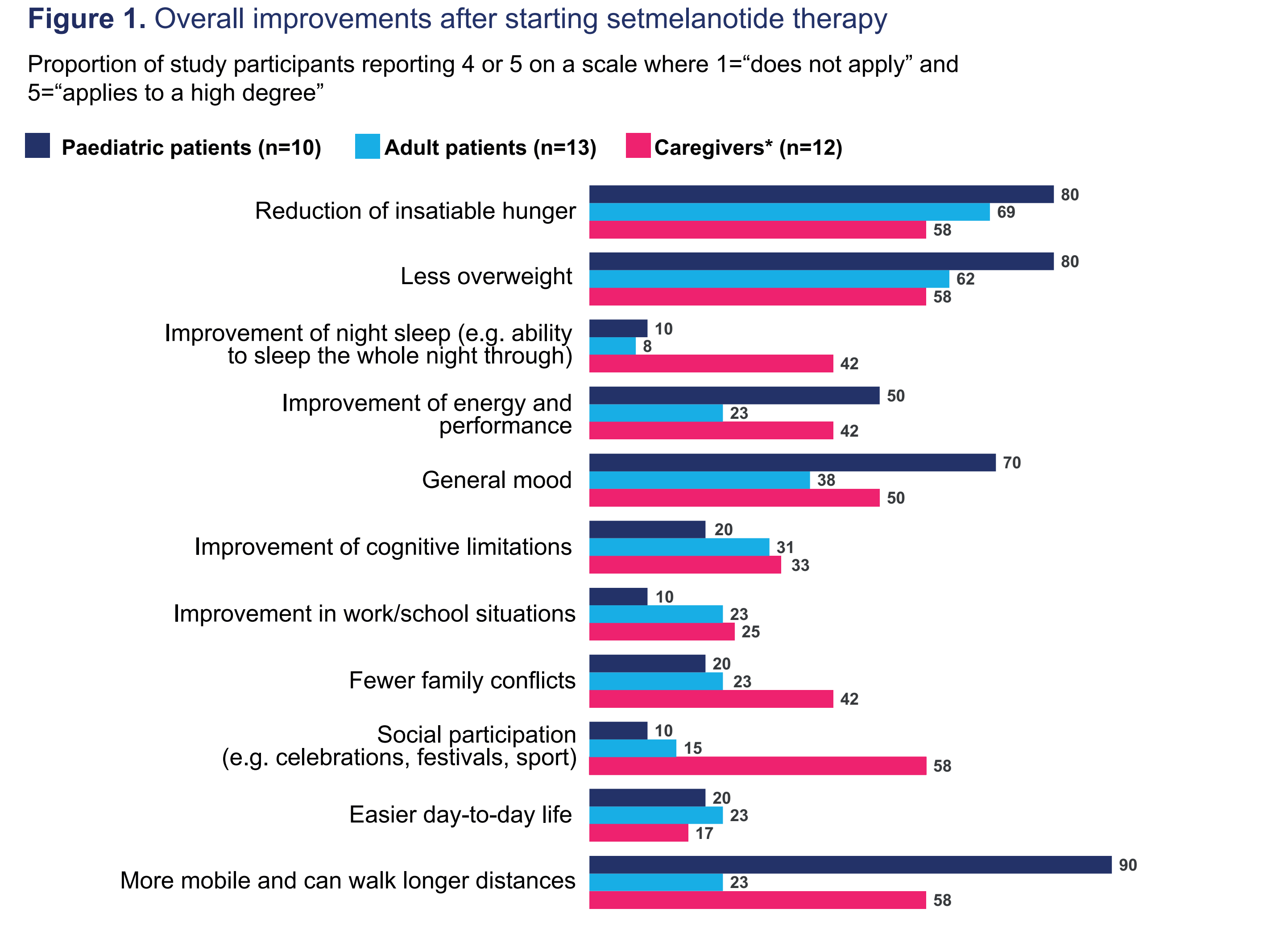
Baseline characteristics

- 10 paediatric patients, 13 adult patients, and 12 caregivers participated in the study (Table 1)
- All patients received specialist care from a nephrologist or endocrinologist; some patients received care in parallel from a general practitioner (15% of adult patients), an ophthalmologist or otolaryngologist (8% of adult patients), or an outpatient paediatrician (33% of paediatric patients)
- Prior to treatment, paediatric patients reported experiencing insatiable hunger (80%) and obesity (50%) as their most burdensome symptoms influencing the decision to start treatment; for adult patients, these were vision loss (69%) and obesity (69%); caregivers reported obesity (92%) and insatiable hunger (83%)
 - “Insatiable hunger” in adult patients prior to treatment start could not be reported due to a technical mistake in the survey

	Paediatric patients (n=10)	Adult patients (n=13)	Caregivers (n=12)
Sex			
Male, n (%)	5 (50)	5 (38)	4 (33)
Female, n (%)	5 (50)	8 (62)	8 (67)
Age			
6–9 years, n (%)	3 (30)	0	4 (33)
10–13 years, n (%)	5 (50)	0	4 (33)
14–18 years, n (%)	2 (20)	0	2 (17)
18–25 years, n (%)	0	6 (46)	0
26–30 years, n (%)	0	2 (15)	0
30–35 years, n (%)	0	1 (8)	0
36–40 years, n (%)	0	1 (8)	0
Not specified, n (%)	0	3 (23)	2 (17)
Ethnicity			
European/Eurasian, n (%)	10 (100)	13 (100)	10 (83)
African, n (%)	0	0	2 (17)

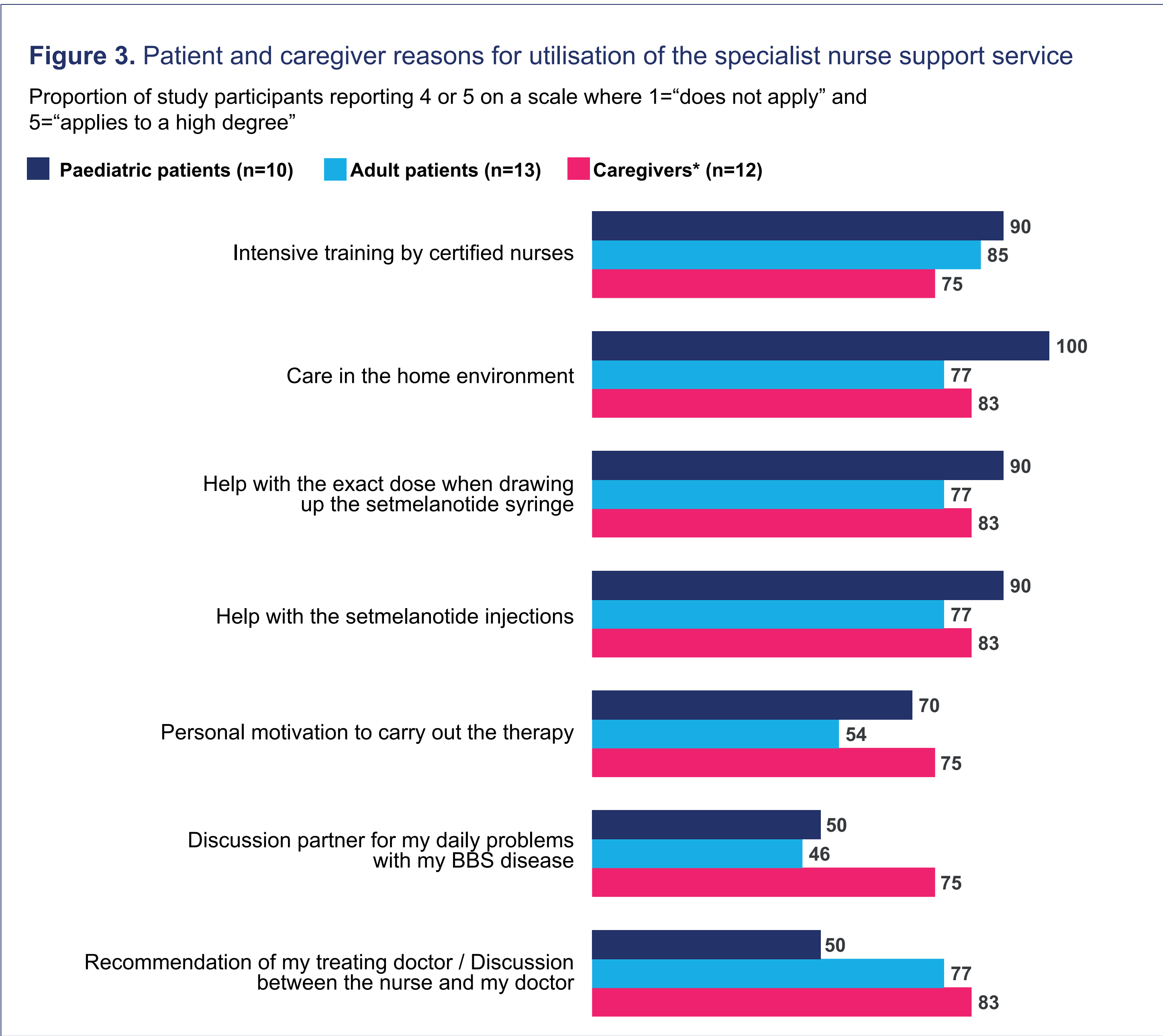
Effects of setmelanotide treatment

- In paediatric patients, the most reported improvements with setmelanotide therapy were improved mobility (90%), reduced hunger (80%), reduced body weight (80%), and better mood (70%) (Figure 1)
- In adults, the most common improvements were reduced hunger (69%), reduced body weight (62%), and better mood (38%) (Figure 1)
- Mean BMI z-score ± SD of all patients at the start of setmelanotide treatment was 3.12 ± 0.89, which was reduced by 0.47 ± 0.37 after 6 months
- Almost all patients (≥92% of respondents across all three survey groups) reported feeling satiated after meals (Figure 2)
- The majority of paediatric patients and caregivers reported a reduction in snacking between meals (60% and 58%) and caregivers reported less aggressive behaviour when food was not available on time (67%) (Figure 2)
- The most commonly reported treatment-emergent events were changes in skin pigmentation (paediatric 40%; adult 15%; caregivers 50%), (initial) vomiting, diarrhoea, and nausea (paediatric 30%; adult 15%; caregivers 8%)



Experience with the support service at the start of setmelanotide treatment

- The most commonly cited reasons for using the support service were home care, help with injections and dosing, access to certified nurses, and the recommendation of the treating physician (Figure 3)
- The median time to being able to independently administer injections was 1 month among adult patients and caregivers of paediatric patients, and 5 months among paediatric patients
- At the start of therapy, specialist nurse support was provided daily to 60%, 85%, and 67% of the paediatric patients, adult patients, and caregivers, respectively
- At the follow-up, between 30–42% of the respondents were no longer visited by the support service, or required visits only as needed by 20%, 23%, and 8% of the paediatric patients, adult patients, and caregivers, respectively
- All participants rated the overall support package provided by the specialised nurse support service as excellent. Most patients (paediatric patients 100%; adult patients 92%) and caregivers (92%) would recommend the support service to others, and many patients (paediatric patients 30%; adult patients 38%) and caregivers (42%) did not express any further need for support



Conclusions

- This real-world survey of patients with BBS and their caregivers demonstrated the meaningful benefits of setmelanotide in improving insatiable hunger and obesity
- The specialised nurse support service contributed to high patient and caregiver satisfaction and enabled most of them to administer the drug independently
- Our results illustrate how a personalised approach to healthcare support can address a high unmet need among patients living with BBS, contributing to high rates of patient satisfaction

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