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Methods

Participants were identified by patient advocates and patient advocacy groups and were screened and consented by Health Outcomes Solutions (HOS). Children/adolescents between 8 and 17 years of age with confirmed molecular diagnosis were eligible to participate. Parents completed a background questionnaire containing demographic and clinical questions.

The percentages of medical challenges (ACH-related symptoms/complications) and impacts of these challenges to HRQoL were calculated separately for children and parents.

Saturation tables were developed to categorize each challenge/impact mentioned by each participant. Saturation, the point at which no new concepts are mentioned by each participant, was reached by the 9th interview.

Concept elicitation findings:

Medical/physical challenges reported by children/adolescents and parents (Figure 1)

- The challenges most commonly reported by children/adolescents were feeling hot/sweaty (88%), pain (83%), balance issues (73%), fatigue (64%), muscle fatigue/loose joints (63%), and speech issues (63%). With the exception of speech apnea, saturation was reached by the 9th interview.
- The challenges most commonly reported by parents were pain (53%), air infections (71%), feeling hot/sweaty (71%), and sleeping (71%). Saturation was reached by the 7th interview.
- Some differences were observed between parents and children/adolescents in the impact of ACH. Difficulties with toileting, dressing, and walking were more commonly reported by parents, whereas children/adolescents more often reported difficult breathing and sleeping.

Concept elicitation findings: Impacts reported by children/adolescents and parents (Figure 2)

- The impacts most commonly reported by children/adolescents were difficulty reaching things (88%), running (86%), and sleeping (71%). Saturation was reached by the 7th interview.
- The impacts most commonly reported by parents were difficulty reaching things (76%), toileting (58%), bathing (56%), running (56%), and sleeping (55%) (Figure 2). Saturation was reached by the 10th interview.
- Some differences were observed between parents and children/adolescents in the impact of ACH. Difficulties with toileting, dressing, and walking were more commonly reported by parents, whereas children/adolescents more often reported difficult breathing and sleeping.

Results

Demographic and clinical characteristics

- Eight children/adolescents were interviewed. Most were female, Caucasian, married, and had a college degree or higher (Table 2).

Table 1. Demographic and clinical characteristics at enrollment: Children/adolescents

Table 2. Demographic characteristics at enrollment: Parents

Table 3. Medical challenges and impacts on health-related quality of life in this population.

Figure 1. Most commonly reported medical challenges

Figure 2. Most commonly reported impacts

Table 4. Medical challenges and impacts on health-related quality of life in this population.

Concept elicitation findings: Medical/physical challenges reported by children/adolescents and parents (Figure 1)

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Concept elicitation findings: Impacts reported by children/adolescents and parents (Figure 2)

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References