

Patient Concept Elicitation Interviews: Insights Into Spinocerebellar Ataxia Patient Experiences

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BACKGROUND

- Spinocerebellar ataxias (SCAs) are a group of rare cerebellar degenerative disorders¹⁻³ that are clinically characterised by progressive lack of motor coordination, gait impairment, loss of balance and associated falls, visual disturbances, and speech and swallowing difficulties^{1,4}
- Additionally, SCA can substantially impact patient mental health,⁵ activities of daily living (ADLs)⁶ and health-related quality of life⁶
- It is important to understand the full spectrum of SCA symptoms and impacts from the patients' perspective to promote the development of SCA clinical outcome assessments (COAs) that are sensitive to detecting meaningful change over time
- The recently developed Patient-Reported Outcome Measure (PROM)-Ataxia is a COA comprised of 70 questions across 3 domains (physical, ADLs and mental health) for assessment of patients with ataxia⁷
- Independent assessment of the content validity of the PROM-Ataxia has not yet been conducted

OBJECTIVE

- Conduct qualitative interviews with individuals with SCA to understand patients' lived experiences of the disease and assess the content validity of the PROM-Ataxia

METHODS

- Individuals with SCA were recruited via clinician or self-referral from a patient advocacy organisation (National Ataxia Foundation) or via Engage Health, Inc.
- Two sets of semi-structured qualitative interviews were conducted in October to December 2022 and April to May 2024 via video call with United States-based individuals with SCA to explore the signs, symptoms and impacts of SCA
- Interviews consisted of 2 phases:
 - Collection of background information
 - Concept elicitation to understand the most relevant and bothersome symptoms experienced by individuals with SCA
- Relevance and comprehensiveness of the PROM-Ataxia was assessed by mapping concepts discussed by individuals with SCA against the 3 PROM-Ataxia domains
- Interviews were audio recorded, transcribed, coded and analysed by ATLAS.ti v23 software, following established methods⁸

RESULTS

- Interviews were conducted with 31 individuals with SCA
- Demographics and clinical characteristics are presented in **Table 1**
 - Most (n=16; 51.6%) individuals with SCA were female
 - SCA3 was the most common genotype among individuals with SCA (n=16; 51.6%)
 - Most (n=13; 41.9%) individuals reported that their SCA severity was mild

Table 1. Demographics and clinical characteristics for individuals with SCA

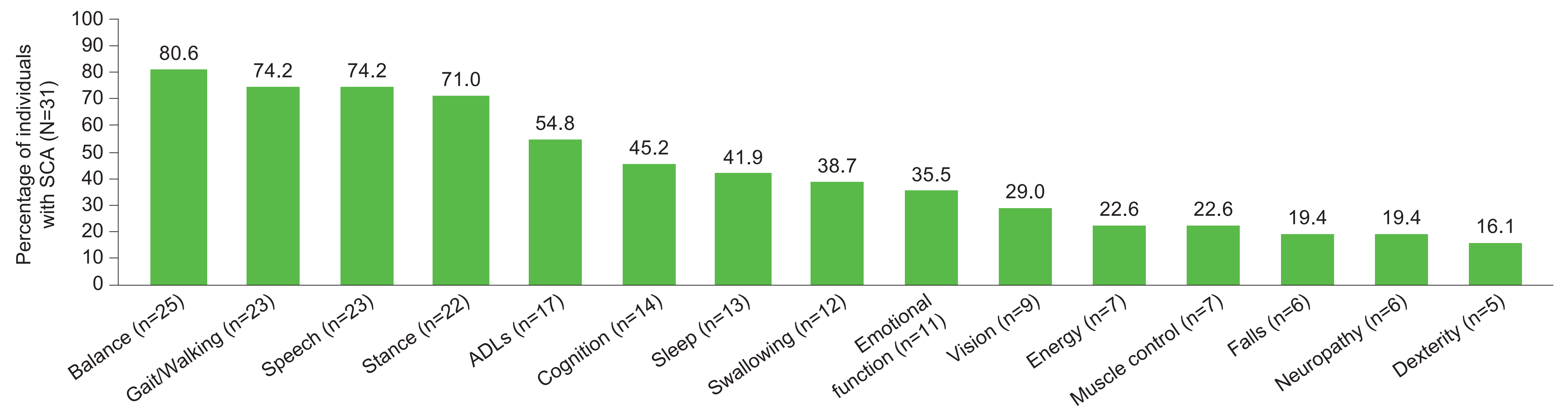
Individuals with SCA demographics and clinical characteristics	N=31
Sex, n (%)	
Male	15 (48.4)
Female	16 (51.6)
Age in years, mean (range)	52.5 (34–75)
Age in years at diagnosis, mean (range)	44.2 (22–72)
SCA genotype, n (%)	
SCA1	7 (22.6)
SCA2	6 (19.4)
SCA3	16 (51.6)
SCA6	2 (6.5)
Self-reported SCA severity, n (%)	
Mild	13 (41.9)
Moderate	9 (29.0)
Severe	9 (29.0)

SCA, spinocerebellar ataxia.

Concept elicitation: signs, symptoms and impacts of SCA

- A total of 87 concepts reflecting the signs, symptoms and impacts of SCA were reported by >1 individual with SCA
- The most frequently reported overarching concepts are shown in **Figure 1**
 - Overall, balance (n=25; 80.6%), gait/walking (n=23; 74.2%) and speech (n=23; 74.2%) were the most frequently reported concepts by individuals with SCA
 - Within the ADL concept, difficulties with relationships/socialising (n=15; 48.4%), driving (n=12; 38.7%) and working (n=10; 32.3%) were most common
 - Individuals with SCA also reported feelings of depression, anxiety, self-consciousness, fear, embarrassment and frustration related to SCA symptoms and impacts
- Illustrative quotations regarding the impact of SCA concepts reported by individuals with SCA are shown in **Table 2**

Figure 1. Overarching concepts frequently reported by >10% individuals with SCA



ADLs, activities of daily living; SCA, spinocerebellar ataxia.

Table 2. Illustrative quotations regarding SCA concepts from individuals with SCA

Concept	Quotations from individuals with SCA
Gait/Walking	"Gait and walking around is essential to seeing the world and being part of the world around you. And if you can't move and you can't walk, you can't be out in the world."
Balance	"Difficulties with balance, I mean, from getting drinks to the table to being able to walk my pee jug to the bathroom, not being able to balance that is a pain. It's a daily struggle."
Energy	"Fatigue, tiredness, lack of energy, I just hate not being able to want to do stuff. You just get too tired and you're willing to accept less productivity than you used to."
Speech	"People currently understand what I'm saying, but I fear for the day when they don't."

SCA, spinocerebellar ataxia.

Most bothersome signs, symptoms and impacts of SCA

- Of the concepts reported by individuals with SCA, the most bothersome were issues with stance/balance (n=16; 51.6%), difficulties with gait/walking (n=14; 45.2%) and difficulties with speech (n=11; 35.5%) (**Table 3**)
- Illustrative quotations regarding SCA concepts considered most bothersome by individuals with SCA are shown in **Table 4**

Table 3. SCA concepts considered most bothersome by individuals with SCA

Symptom	Most bothersome (N=31), n (%)
Stance/Balance	16 (51.6)
Gait/Walking	14 (45.2)
Speech	11 (35.5)
Energy	4 (12.9)
Swallowing	3 (9.7)
Neuropathy	2 (6.5)
ADLs	1 (3.2)
Sleep	1 (3.2)
Emotional function	1 (3.2)

ADLs, activities of daily living; SCA, spinocerebellar ataxia.

Table 4. Illustrative quotations regarding SCA concepts considered most bothersome from individuals with SCA

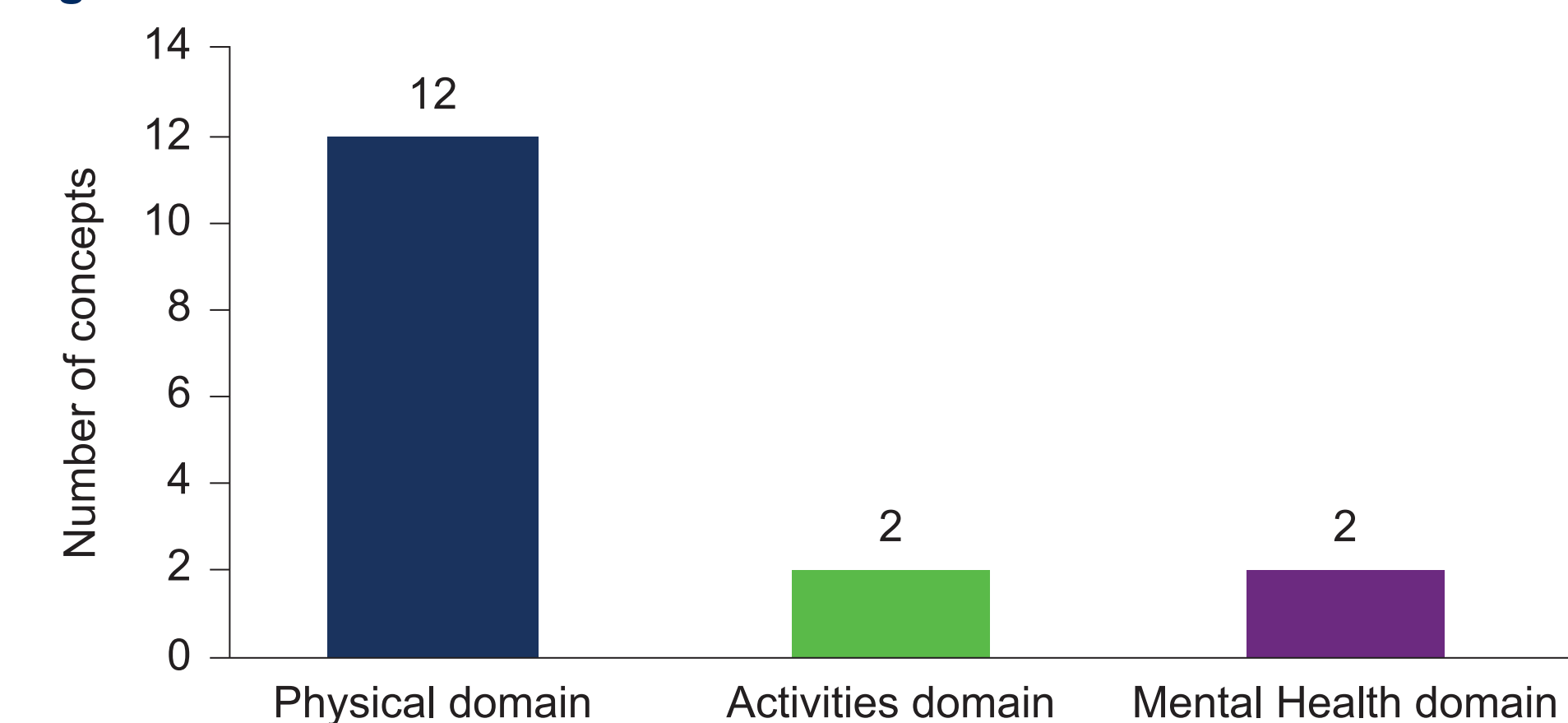
Symptom	Quotations from individuals with SCA
Balance	"The balance disorder is definitely the most bothersome, and the reason is because it just inhibits my ability to move."
Speech	"...the speech. That's what bothers me the most. Because I'm a single mom and my job is so important to me that not being able to speak, I'm so scared that I'll lose my job because of that."
Neuropathy	"The neuropathy, because it causes the most pain."
ADLs	"There's no way I could work again. I used to have a speaking job where I would address crowds, small crowds, and I can't do that anymore. For two reasons, one is I can't stand or do anything like that, and second, my slurred speech would really get in the way of trying to be convincing."

ADLs, activities of daily living; SCA, spinocerebellar ataxia.

Mapping concepts reported by individuals with SCA onto PROM-Ataxia domains

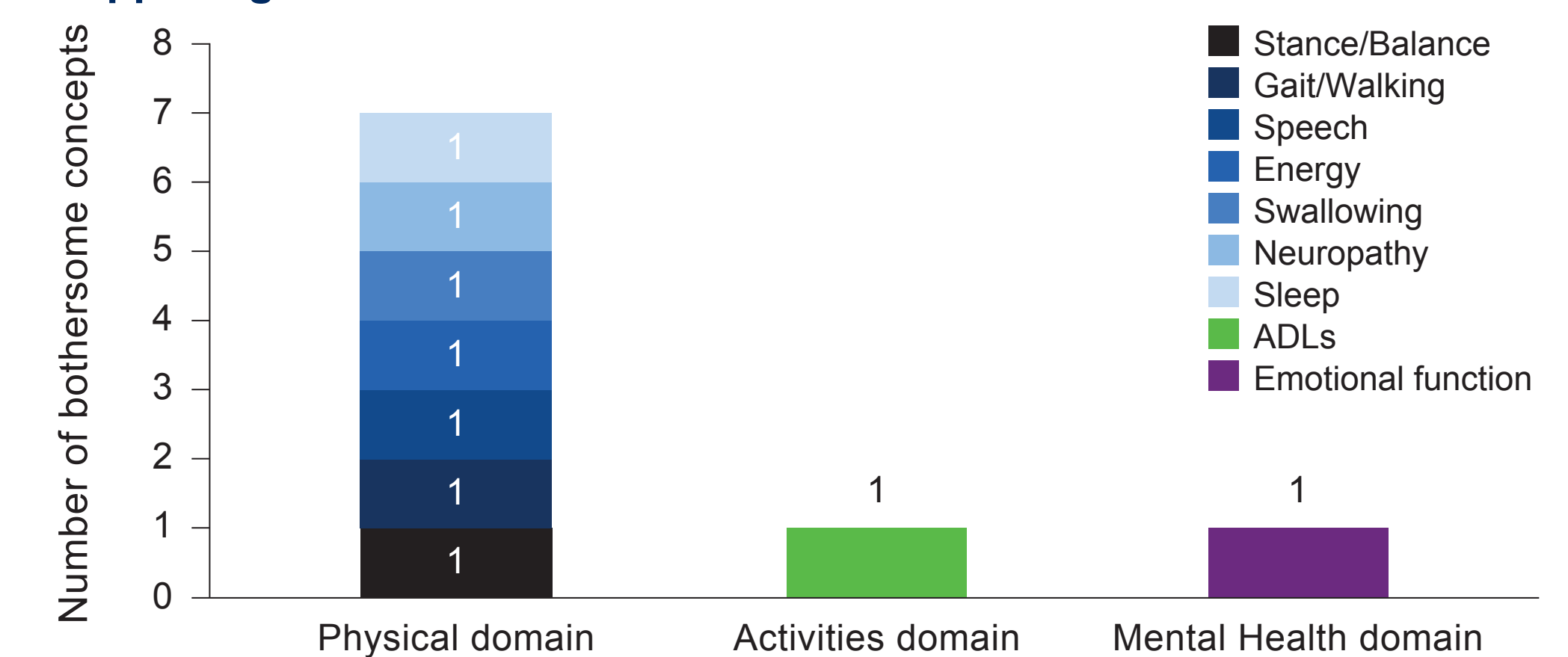
- The overarching concepts reported by >10% individuals with SCA were mapped against the PROM-Ataxia domains and sub-domains
 - All concepts reported by individuals with SCA were captured by the PROM-Ataxia across ≥1 domain (**Figure 2**)
 - The PROM-Ataxia Physical domain also includes the sub-domains of 'Dizziness' and 'Sexual Ability', which were not commonly reported among individuals with SCA during concept elicitation

Figure 2. Concepts frequently reported by individuals with SCA mapped against PROM-Ataxia domains



PROM-Ataxia; Patient-Reported Outcome Measure-Ataxia; SCA, spinocerebellar ataxia.

Figure 3. Most bothersome concepts reported by individuals with SCA mapped against PROM-Ataxia domains



ADLs, activities of daily living; PROM-Ataxia; Patient-Reported Outcome Measure-Ataxia; SCA, spinocerebellar ataxia.

PROM-Ataxia: Most bothersome concepts of SCA

- All concepts reported as most bothersome were captured by the PROM-Ataxia
 - The most bothersome concepts spanned all 3 PROM-Ataxia domains (**Figure 3**)
 - Seven of 10 sub-domains in the PROM-Ataxia Physical domain were reported as most bothersome by individuals with SCA
 - One of 2 sub-domains in both the Activities and Mental Health domains were reported as most bothersome by individuals with SCA

CONCLUSIONS

- The signs, symptoms and impacts of SCA are heterogeneous and have a profound effect on daily functioning
- Findings support the content validity of the PROM-Ataxia and other measures, such as the modified functional Scale for the Assessment and Rating of Ataxia (f-SARA), to evaluate disease progression in individuals with SCA
- The PROM-Ataxia is a comprehensive outcome measure that captures concepts considered relevant to individuals with SCA

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