

The Relationship of Physical Function and Psychosocial Health on Quality of Life in Individuals with 48,XXYY: Preliminary Results

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Introduction & Background

- The SCA 48,XXYY has a prevalence of 1 in 18,000-40,000 male births.
- Individuals with 48,XXYY present with tall stature, hypogonadism, but with more moderate to severe developmental, cognitive, and psychosocial impairment than the more common 47,XXY SCA.
- Other symptoms include autistic-like characteristics, poor impulse control, mood disorders, sleep difficulty, tremors/seizures, and respiratory disorders.
- The increased severity and wide range of symptoms associated with 48,XXYY places affected individuals at an increased risk for adverse physical and psychosocial health outcomes.
- Psychological comorbidities such as mood disorders, impulsivity, and depression place individuals at increased risk for poor social relationships and the need for ongoing support and/or supervision.

Significance & Purpose

- Interventional research addressing symptom abatement in SCAs is lacking.
- In-depth descriptions of patient-centered problems are also lacking
- Current literature in 48,XXYY focuses on research or clinician descriptions of the disorder, with limited direct input from parents, caregivers, or the individuals affected by 48,XXYY.
- Studies have yet to address the specific needs of this population through first-hand accounts or self-reported measures from the affected men.
- Despite the availability of evidence describing physical, neurocognitive, and physiologic symptoms of 48,XXYY, most available literature lacks any use of qualitative methodology to express how men with 48,XXYY men describe their quality of life or needs from the research and medical community.

Methods

- Participants were recruited from AXYS and the XXYY Project website.
- Data were collected in real-time with the principal investigator via an online meeting platform that allowed participants to see and hear the questions.
- The first phase of this project involved collecting quantitative data on demographics, physical and psychological functioning, and quality of life.
- The following quantitative surveys were administered: the 36-Item Short-Form Survey (SF-36), the Beck Anxiety Inventory (BAI), and the Beck Depression Inventory (BDI).
- The SF-36 is a 36-item self-report questionnaire with 8 subscales addressing the following issues: vitality, physical functioning, bodily pain, general health perception, physical role functioning, emotional role functioning, social role functioning, and mental health
- The BAI is a 21-item self-report questionnaire measuring anxiety levels.
- The BDI is a 21-item self-report questionnaire measuring attitudes and symptoms of depression.
- The second phase of this project involved collecting qualitative data on overall quality of life, physical functioning, and psychosocial health.
- After completion of the quantitative survey data collection, approximately half of the previous participants were selected, using maximum variation sampling, to participate in one-hour long qualitative interviews

Results

Table 1. Participant Characteristics

Variable	Frequency (%)	Variable	Frequency (%)
Age		Employment	
Mean (Minimum – Maximum)	28 (18-42)	Never Employed	5 (22.7)
Race		Previously Employed	8 (36.4)
Caucasian	22 (100)	Currently Employed	9 (40.9)
Ethnicity		Education	
Not Hispanic/Latino	21 (95.5)	Did Not Finish HS	2 (9.1)
Hispanic/Latino	1 (4.5)	Finished HS	6 (27.3)
Country		Started Technical/ Certificate, Did Not Finish	2 (9.1)
US	13 (59.1)	Finished Technical/ Certificate	6 (27.3)
CAN	4 (18.2)	Started College but Did Not Finish	6 (27.3)
UK	1 (4.5)	History of Depression	
EU	1 (4.5)	No/I don't know	6 (27.3)
AUS	2 (9.1)	Yes	16 (72.7)
BR	1 (4.5)	History of Anxiety	
Living Arrangements		No/I don't know	14 (63.6)
With Parents	15 (68.2)	Yes	8 (36.4)
With Roommates	2 (9.1)		
Alone	4 (18.2)		
Group Facility	1 (4.5)		

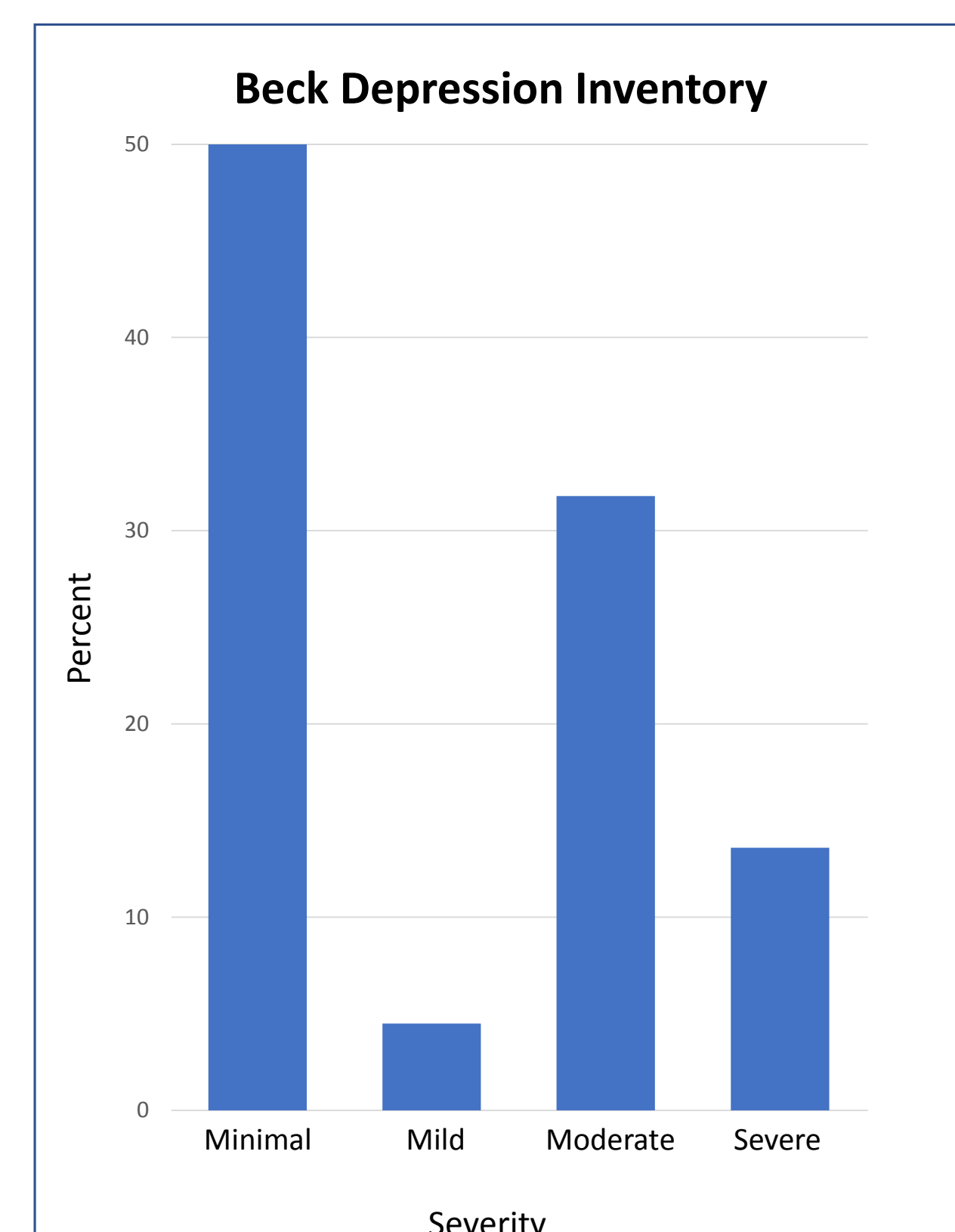


Figure 1. Beck Depression Inventory Results

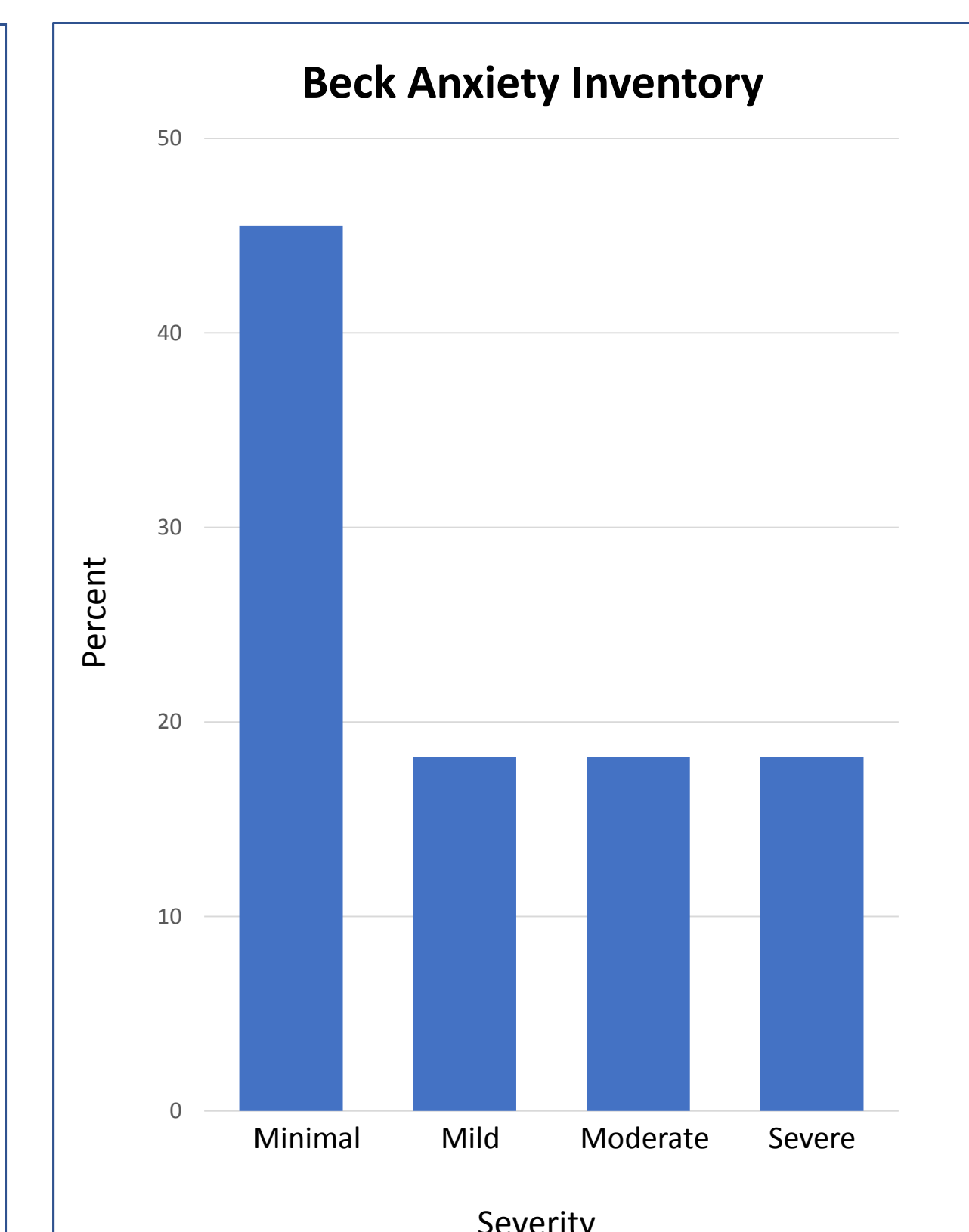


Figure 2. Beck Anxiety Inventory Results

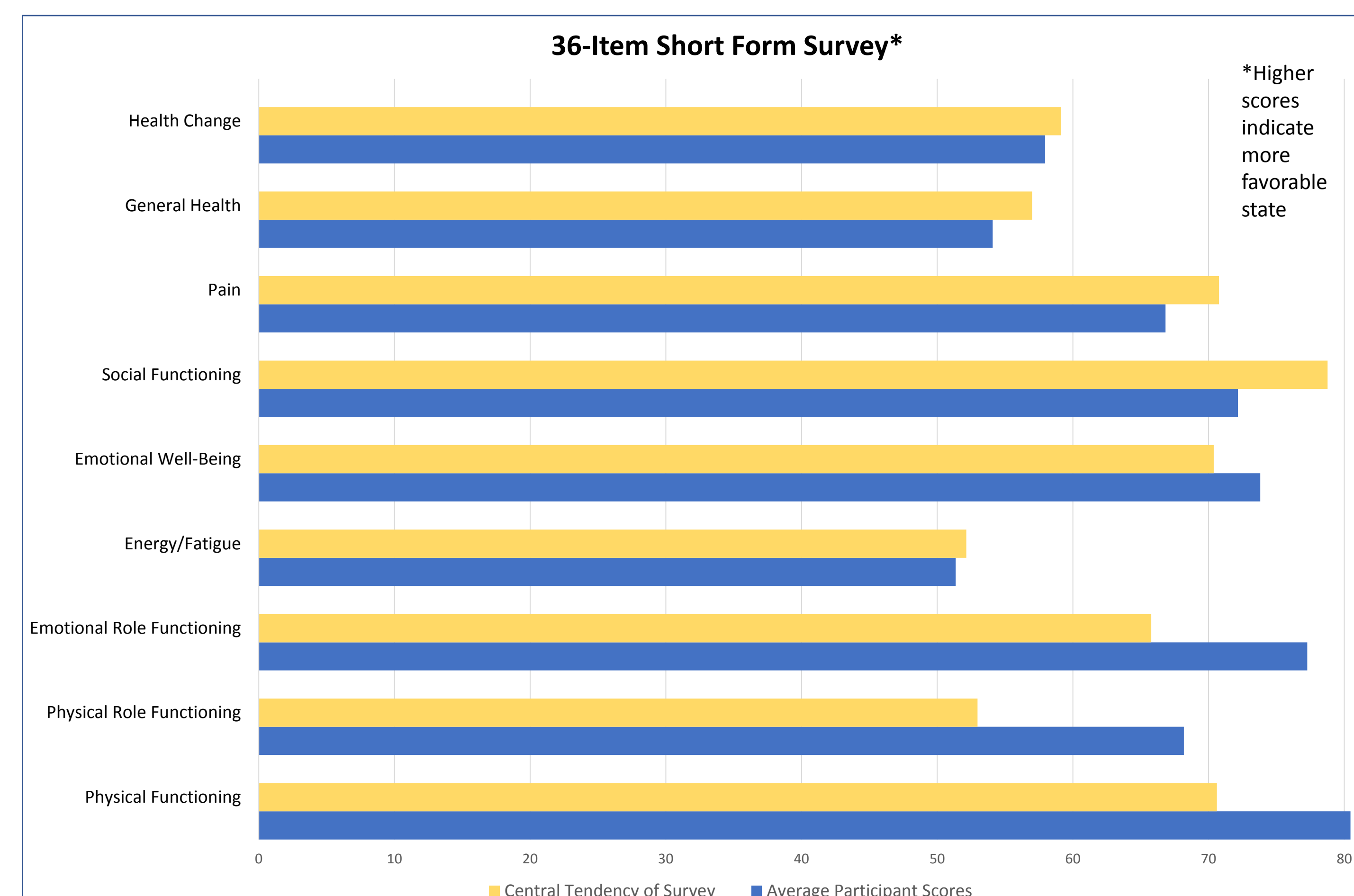


Figure 3. 36-Item Short Form Survey

Exemplary Quotes

"I can't read what people are saying. I think they're being mean with me, but maybe they're just trying to explain it to me, but I don't understand...I get frustrated trying to understand what they're trying to tell me."

"I really don't know what to tell [you]. I don't think there's anything good about my syndrome. That's a hard question."

"I think I've got it pretty good. I think considering the average person's quality of life is down in the dumps. A lot of people feel like...they don't belong anywhere. [I feel] like I'm part of something greater."

"I find that I can do a reasonable amount of work...I'm really into doing a lot of physical sort of sports and stuff. But...I do really like playing video games. I'm sort of someone that really does like to sit around a lot."

"I don't do a lot of thinking about my life, about me...I just take day by day...When it comes to my video games...my consoles, my phone, my girlfriend, and my work, I think. But when it comes to my mental state of mind...I don't really think too much of it because...I will get anxiety...If I have to think too much of something that happened back in the day, like...depression, anxiety, paranoia...[it] causes me to break down."

Discussion

- The study sample was a heterogeneous population in terms of quality of life, reporting with high individual variation.
- Participants had low depression and anxiety scores and had higher emotional well-being and emotional functioning than survey norms.
- Participants scored lower than average on general health, pain, and social functioning scores.
- In interviews, most individuals described their physical functioning as relatively positive and felt that they were able to do normal activities.
- The ability to establish and maintain social relationships was very important to most participants and had a large impact on their overall quality of life.
- Many participants reported satisfaction with their quality of life, but others implied that they wished they had better mental health, more relationships, more work, and more independence.

Preliminary Impressions

- Individuals with 48,XXYY appear to have a wide range of physical and psychosocial functioning, leading to varying levels of satisfaction with overall quality of life.
- When utilizing surveys with participants, standard symptom inventory responses may not align with detailed interview responses.
- Social relationships appear to be a key contributing factor to satisfaction with quality of life.
- Future research should continue to focus on what individual's needs are with 48,XXYY, what aspects of their life are important to them, and how to intervene early when addressing physical or psychosocial healthcare concerns.