

Living with XYY: Voices of Patients and Caregivers

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Introduction & Background

- 48 XYY is a fairly rare genetic condition with an estimated prevalence of 1 in 18,000 to 1 in 50,000 male births.
- Formerly described as a “variant” of the more common sex chromosome variation known as Klinefelter Syndrome (KS) 47 XXY
- 48, XYY has its own unique pattern of physical, neurocognitive and psychosocial characteristics compared to other SCAs.
- While some adults with XYY are employed and live independently.
- A majority are not able to launch to independence and self-support requiring part-time or full-time caregiving in group living, institutional living or remaining with family members throughout the lifespan.
- Unrelenting and often intense caregiving can have negative effects on the physical and psychosocial health of caregivers.
- Caregivers rarely receive respite from constant need for surveillance and care

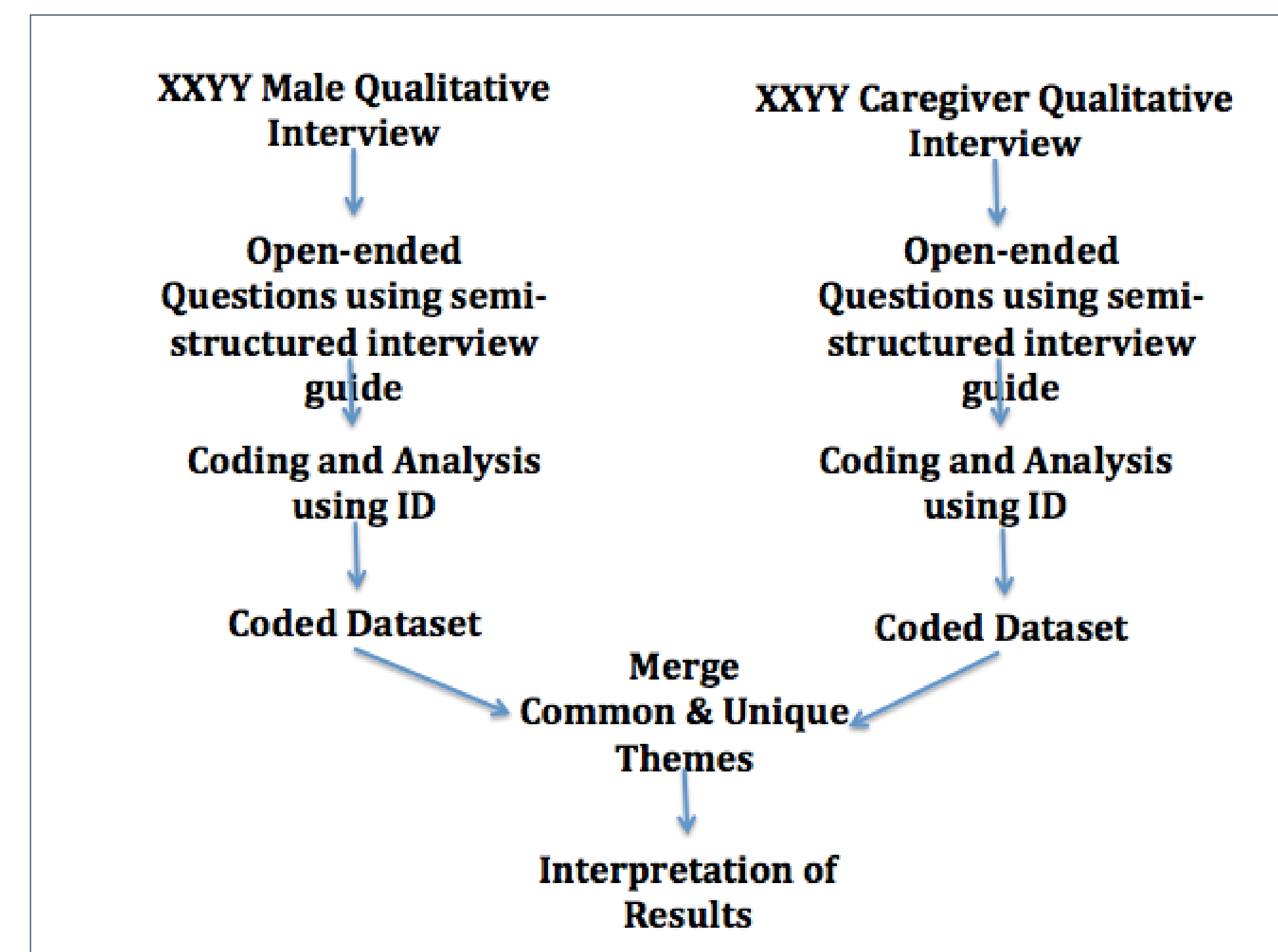
Significance & Purpose

- 48, XYY may be considered an “orphan” genetic condition since it is rare and very under-studied.
- The genetic variation itself presents challenges and consequences for not only the affected person but also for those caring for them
- The purpose of this study was to **explore the life experiences** of patients and caregivers affected by XYY and to **identify problems** that interfere with daily living and quality of life.
- By **deeply exploring narrative stories**, we will identify problems that will inform future intervention development for the benefit of patients and caregivers.

Methods

- This study was approved by the Emory University Institutional Review Board
- We used a qualitative methods approach known as Interpretive Description
- Participants were recruited through AXYS, the XYY Project and various social media sites.
- We used a maximum variation purposive sampling approach to recruit adults and caregivers affected by XYY.
- Men with XYY and an adult caregivers were interviewed separately for an hour each.
- Interviews were audio-recorded and transcribed verbatim by a HIPAA-certified transcription agency.
- Privacy was maintained by removing all identifying information from the transcript and destroying the audio files of the interviews.
- Analysis was conducted according to the method of Interpretive Description.
- 2-coders independently coded transcripts and collaboratively extracted categories and themes from the data.

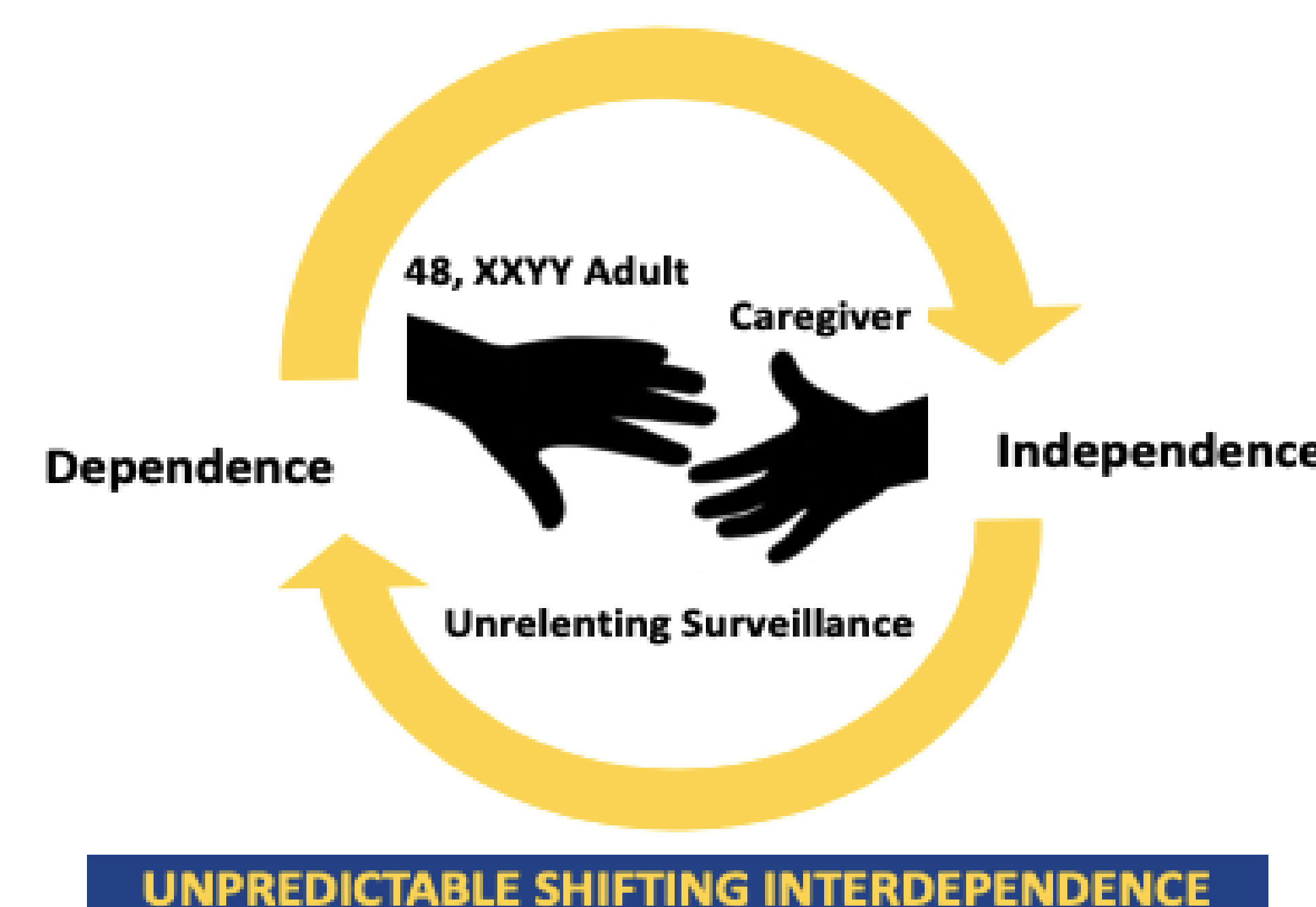
Qualitative Methods Approach



Participant Characteristics N = 22

Adult Son Age	Caregiver Age	Region
24	54	US
45	66	US
33	56	US
22	56	US
23	48	CAN
31	63	CAN
32	63	US
20	53	Germany
20	43	US
38	66	US
21	52	US

Overarching Theme



Exemplar Quotes

CAREGIVERS

- “So we do leave him home and we feel uncomfortable doing that, but at the same time I feel like it’s a good thing for him because it’s forcing him to take care of the house”.
- “So I would say we’re actually in some ways at one of the hardest points, which is how are we going to transition my son to a more independent life and what is that going to look like?”
- “I mean my life it could have been much fuller. There were things that I would like to have done or gotten involved with but it’s almost like my son is at the forefront of everything.”

SONS

- “I still do, I still feel like I kind of got the short end of the straw in this family. My mom takes care of me a lot, I think a little too much. So there’s a lot of things I don’t get to experience in my life. I’ll probably never get to experience what I want”
- “So I still need to get -- make more money and that way I can have a chance in -- so it may not be physical now, but I am getting closer and closer to my ability to live on my own, but for now I’m just enjoying my job and just doing what I can to have a happy life.”
- “What bothers me about having XYY is that I can’t get married because I can’t live on my own. I need my mom. If I need to go to the doctor, she takes me there. I feel like I want to live on my own, though,”

Discussion

Narratives depicted stories of mutual love, dedication and attachment with prominent dichotomous themes describing unpredictable shifts in interdependence:

- Wish for Independence vs Need for Dependence
- Desire for Privacy vs Need for Surveillance
- Immature Decision-making vs Mature Decision-making
- Media-based socialization vs In-person socialization
- Risky entrustment vs Trustworthiness

Caregivers reported fewer negative physical complaints than expected. Caregivers frequently reported problems with worry and anxiety related to unpredictable situations with their sons and their persistent struggles and dissatisfaction with education, employment, living environments, disability support and medical care.

Self-described characteristics of 48, XYY participants showed social isolation, limited personal engagement with others and difficulty establishing intimate relationships. Social needs were satisfied during videogaming, texting and other forms of social media interaction.

Conclusions & Implications

Further research is recommended to more fully characterize the unique phenotype and quality of life for those with 48, XYY and their caregivers so that targeted interventions can be developed. Also of great importance is raising awareness in medical, educational and disability support communities.