



# An Exploration of Health Concerns and Needs for Care in Women with Trisomy X (47, XXX)



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## Purpose

- To explore the personal stories of women with Trisomy X to understand their health issues and how they would like healthcare providers to address their concerns.
- To identify gaps in understanding and needs for care in this population.

## Background

- Sex chromosome aneuploidies are random genetic variations of X and Y chromosomes.
- Women with Trisomy X have one extra X chromosome.
- They often have physical, neurocognitive, and psychosocial health issues and are at a greater risk for chronic health problems.
- The reported incidence is 1:1000 female births, but the condition is often diagnosed late or missed altogether
- Late or absent diagnoses increases risk for adverse health and psychosocial outcomes
- Lack of research and awareness contribute to uncertainty and confusion as they navigate complex healthcare pathways.

Participant	Age in Years	Age at diagnosis	Region
1	59	55	Central US
2	29	15	NE-US
3	21	21	SW-US
4	44	42	NW-US
5	22	17	Canada
6	36	21	China

Figure 1: Demographic Table

## Methods

- Design:** pilot qualitative study
- Sampling method:** purposive maximum variation
- Inclusion criteria:** Trisomy X diagnosis, English-speaking, 18 years of age or older
- Sample:** 6 women with trisomy X between 21 and 59 years of age ranging from mild to severe functional impairment
- Data collection:**
  - One-hour long online interviews
  - Semi-structured interview guide informed by the literature
  - Interviews (conducted from September to December 2017) - audio recorded and transcribed
- Data analysis:**
  - Interviews were analyzed independently by 2 members of the research team. A list of start codes were created and compared, leading to the development of themes that were extracted for meaning.
  - Interpretive description (Thorne)

## Results

### Independence vs. dependence

- "There are many times... that I'd have ambition to do something, but I've always needed a friend to stand next to me..." (Participant 3)
- "I'm at the age where I really want to support myself and it's a struggle... being so emotional and dealing with stress... it kind of takes a really big toll on my body." (Participant 5)

### Trust vs. betrayal

- "My friends don't stay. They turn on me and I don't know if it's something I'm doing or what." (Participant 4)
- "People use you and the world's terrible place." (Participant 5)

### Giving help vs. needing help

- "I'm very thankful that I had supportive parents. I don't know how someone with this disorder would be able to go through life without that support." (Participant 4)
- "If I accept who I am I think life can be much easier, and actually I want to inspire more people to see positive things in their life rather than only see the negative things in their life." (Participant 6)

## Results (cont.)

### Belonging vs. isolation

- "I don't want everybody staring at me like I'm some kind of freak." (Participant 4)
- "I'm different, yes, but I see it in a positive way. I'm kind of special. Unique." (Participant 6)
- "I know what holds me back. I'm afraid of getting rejected." (Participant 6)

## Summary and Conclusions

- This study describes unique personal experiences of women with Trisomy X.
- Trisomy X is an understudied and challenging condition to have and to manage.
- Dichotomous themes highlight dynamic challenges the women face as they search for stability in their lives.
- Mental health concerns and calls for support and awareness predominated in this sample
- Searching for central places of caring at home, school and within health care delivery is the common overarching theme in this sample.

## Implications

- The healthcare community needs to become knowledgeable about Trisomy X and how complexities of the condition affect individuals
- Healthcare providers need to recognize the complexity of healthcare needs women with Trisomy X.
- Mental health screenings and discussion of mental health issues are unmet needs in this population.
- Future research is needed to develop interventions in which families, schools and healthcare providers can improve their understanding of women with Trisomy X and provide meaningful aspects of caring.

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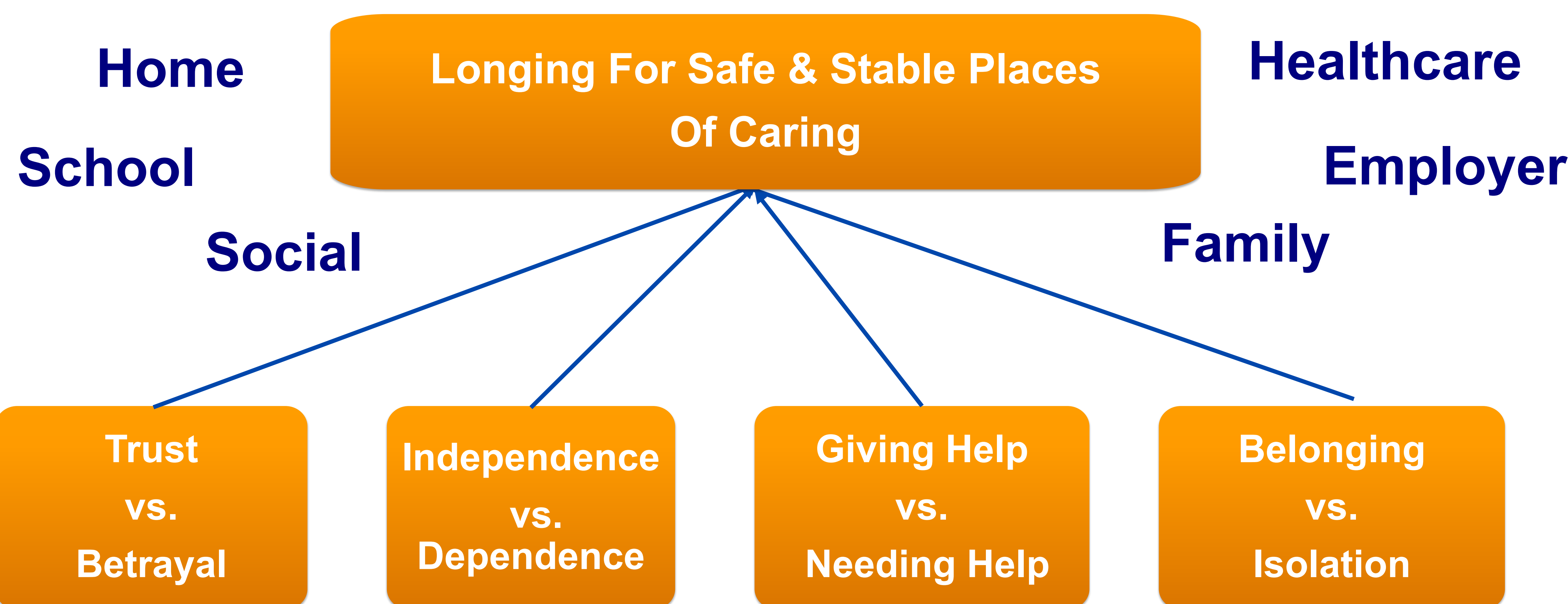


Figure 2: Qualitative Themes, Needs