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 increase risk for adverse health and psychosocial outcomes.
- Lack of research and awareness contribute to uncertainty and confusion as they navigate complex healthcare pathways.

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 a 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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- Women with Trisomy X who participated in this study

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