

# Exploring the Relationship Between Anxiety and Quality of Life in Women With Trisomy X



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association for x and y  
chromosome variations

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# Anxiety and Quality of Life in Trisomy X

## Key Terms:

- ★ **Quality of Life (QOL)**—an individual's satisfaction with their life (Mendlowicz & Stein, 2000)
- ★ **Generalized Anxiety Disorder (GAD)**—persistent and excessive worry about common situations that causes serious distress (Hurtado et al., 2020)

- 47,XXX females have an increased risk of anxiety (Bender et al., 1995)
- Symptoms of GAD interfere with daily life (Hurtado et al., 2020)

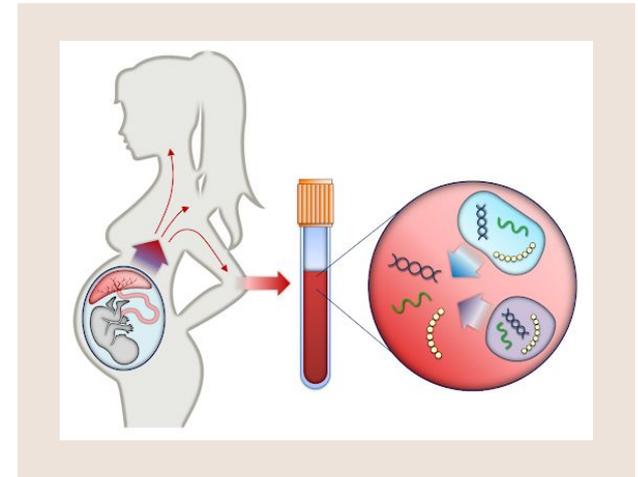


# Rationale

- Advancements in noninvasive prenatal testing are expected to increase diagnostic rates for trisomy X in the future (Wigby et al., 2020)
- Mental health problems within the 47,XXX population remain unaddressed by medical providers (Wei & Close, 2019)

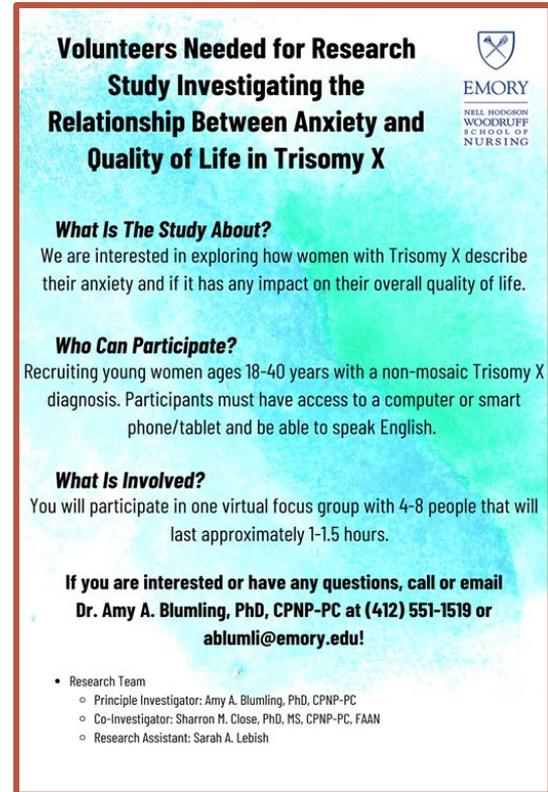
## Key Term:

- ★ **Sex Chromosome Aneuploidy (SCA)**—a genetic disorder that involves the addition of an extra X or Y chromosome in males and the addition or deletion of an X chromosome in females (Harmon et al., 1998)



# Data Collection

- Research flyer was posted on the AXYS website to recruit study participants
- Volunteers were screened over the phone and oral consent was obtained
- Moderator's guide questions were created and adapted from the domains used to measure QOL in **credible surveys**
- Focus group discussion was conducted over Zoom



**Volunteers Needed for Research**  
**Study Investigating the**  
**Relationship Between Anxiety and**  
**Quality of Life in Trisomy X**



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**What Is The Study About?**  
We are interested in exploring how women with Trisomy X describe their anxiety and if it has any impact on their overall quality of life.

**Who Can Participate?**  
Recruiting young women ages 18-40 years with a non-mosaic Trisomy X diagnosis. Participants must have access to a computer or smart phone/tablet and be able to speak English.

**What Is Involved?**  
You will participate in one virtual focus group with 4-8 people that will last approximately 1-1.5 hours.

**If you are interested or have any questions, call or email**  
**Dr. Amy A. Blumling, PhD, CPNP-PC at (412) 551-1519 or**  
**ablumli@emory.edu!**

- Research Team
  - Principle Investigator: Amy A. Blumling, PhD, CPNP-PC
  - Co-Investigator: Sharron M. Close, PhD, MS, CPNP-PC, FAAN
  - Research Assistant: Sarah A. Lebish

# Focus Group Moderator's Guide

Tell me about any experiences you have had or currently have with anxiety in your life.

How would you describe your overall QOL?

Tell me about your social life.

Do you think anxiety impacts your social life?

# Demographics

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Participant	Age	State of Residence	Country of Residence
1	32	Massachusetts	United States
2	18	Texas	United States
3	27		New Zealand
4	40	Colorado	United States
5	34		United Kingdom
6	29	New York	United States

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# Theme 1: Comparison to Others

- Constantly compared aspects of their lives (friendships, memory, etc.) to those of partners and peers
  - Used adjectives like “common” and “typical” to describe men and women without SCAs
- Anxiety stemmed from fear of judgement and other people’s perceptions
  - “What if I say the wrong thing?”
  - “Am I gonna look weird?”

# Themes 2 & 3

## Lack of Knowledge

- Healthcare providers are uneducated
  - “It seems like every time I’d go to the doctor, they’d be like I’ve never really heard of that...”
- Public stigma and ignorance
  - “I find my close friends... don’t really understand exactly what I have and... they kind of poke fun at it...”

## Moderate Quality of Life

- Described either improved or high QOL
  - Developed tools to cope with anxiety and maintain QOL

# Conclusions

- **Hypothesis #1:** Women with trisomy X will state that their anxiety relates to psychosocial interactions.

- **Supported**

- Meeting new people and attending social gatherings are stressful

- **Hypothesis #2:** Participants will express that their psychological symptoms decrease their self-reported QOL.

- **Not Supported**

- Participants have learned to accept their differences and limitations

Any Questions?