

“Fragile X: A Conversation”

This webinar focuses on the genetics and care of children and families with Fragile X Syndrome. In addition, person-centered practices are reviewed to allow us to see beyond the labels to who your child is and how they are best supported.

Presented by:

Dr. Daryl Scott, Associate Professor, Department of Molecular and Human Genetics, Baylor College of Medicine

Laura Buckner, M.Ed., LPC, Mom, UT Texas Center for Disability Studies

+++++

Fragile X syndrome is the most common genetic cause of intellectual disability in males.

Children with fragile X Syndrome may have delayed development, intellectual disability and autism.

Fragile X can impact the health of multiple family members.

Males and females can have fragile X syndrome, but males are usually more severely affected.

The risk of going from a premutation (55-200 CGG repeats) to a full mutation (> 200 CGG repeats) depends on the mother’s CGG repeat number.

Female premutation carriers are at risk for Fragile X premature ovarian insufficiency (FXPOI).

Fragile X Associated Tremor and Ataxia Syndrome (FXTAS) typically occurs after age 50 and affects males more frequently than females.

Resources:

GeneReviews FMR1 Disorders: <https://www.ncbi.nlm.nih.gov/books/NBK1384/>

FRAXA Research Foundation: www.fraxa.org

National Fragile X Foundation: <https://fragilex.org>

Fragile X Clinics in Texas

- Texas Children’s Hospital Fragile X Clinic: Houston, TX
Website: <https://www.texaschildrens.org/departments/fragile-x-clinic>
Email: dlgodwin@texaschildrens.org Phone: 832-822-3400
- Center for Autism and Developmental Disabilities: Dallas, Texas
Website: <https://www.childrens.com/locations/sc-cadd>
Email: maryann.morris@childrens.com Phone: 214-456-7700

This webinar is sponsored by the Department of Molecular and Human Genetics at Baylor College of Medicine/Texas Children’s Hospital in collaboration with the University of Texas, Texas Center for Disability Studies and the Texas Department of State Health Services.

Person-Centered Practices

Speaker: Ms. Laura Buckner, M.Ed., LPC, Mom, UT Texas Center for Disability Studies

*Person-centered practices allow us to see **beyond** the labels to who your child is and how they are best supported. A Health Care One-Page Profile is one way of supporting someone in a more person-centered way, specific to health care settings and providers.*

- Begin by asking yourself:
 - Where will the Health Care One-Page Profile be used? In what context?
 - What do I want readers of the profile to learn from this?
 - How is support for my child best provided by these readers?
- What do people like and/or admire about my child?
 - Avoid information on the level of child's function
 - Focus on personality gifts, and abilities
- What is important TO this person at medical appointments? What helps the person feel safe, comforted, satisfied, happy in these settings?
- How is support best provided in these settings? Consider what helps ease anxiety, pain, or discomfort. Think about accommodations that might be helpful.
- Free downloadable templates can be found by searching online "one page profile templates"

This webinar is sponsored by the Department of Molecular and Human Genetics at Baylor College of Medicine/Texas Children's Hospital in collaboration with the University of Texas, Texas Center for Disability Studies and the Texas Department of State Health Services.