Size Matters: How Missing or Extra Pieces of DNA Can Affect Children

Speakers:

Chaya Murali, MD, Assistant Professor, Dept. Molecular and Human Genetics, Baylor College of Medicine

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

This webinar focuses on 22q11 deletion syndrome, Williams syndrome and 16p11.2 duplication and deletion syndromes. 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.

- Genes are made up of DNA and are like recipes arranged into a cookbook.   We get a set of 23 cookbooks or chromosomes from Mom and a set of 23 from Dad.
- A missing page or extra page in our cookbook or chromosomes is called a copy number variant. Some copy number variants cause a medical problem, many do not.
- Examples of medical conditions due to a copy number variant are 22q11 deletion syndrome, Williams syndrome, 16p11.2 duplication or deletion syndrome.
- A geneticist is a doctor who can diagnose, give advice on what to expect, coordinate ongoing care and help test other family members.

To learn more or find support:

www.medlineplus.gov

www.rarediseases.info.nih.gov

www.rarechromo.org; www.chromodisorder.org

www.22q.org; www.Williams-syndrome.org; www.chromodisorder.org

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"