KNOW BEFORE YOU KNOCK

RARE GENETIC DISORDERS

Every child and family has unique aspects of their situation, experience, and diagnosis. This page is intended to provide a general overview, but is not attempting to represent every situation or child you encounter. Every family deserves your expertise to assess and adapt your prior knowledge. This resource is best viewed electronically.

DEFINITION

"Genetic disorders are health problems that happen because of some type of abnormality in a person's genetic material. There are several types of genetic disorders. Some disorders are caused by a genetic change (mutation) in a single gene; some are caused by an abnormality in one of the chromosomes; and some are complex, involving numerous genes and influences from environmental factors."(1)

PREVALENCE

1 in 10 people (2)

LANGUAGE

TO USE/AVOID

- It's ok to treat the parent as the expert on their child's diagnosis. No provider can know everything about every rarre genetic disorder.
- Still research and learn what you can!

CONSIDERATIONS

*THAT **MAY** IMPACT SERVICES*

- If the gene variation was passed down from one of the parents, there may be feelings of guilt
- Many diagnoses have online communities that may be beneficial for families
- If a child presents with certain signs and symptoms but does not yet have a diagnosis, it may be appropriate to request a referral to genetics. (5)(6)

NATIONAL/STATE ORGS

- The Genetic and Rare Diseases (GARD) Information Center "...our mission is to support people living with rare diseases and their families by providing free access to reliable, easy-to-understand information in English and Spanish."
- National Organization for Rare Disorders "(NORD) advances practical, meaningful, and enduring change so people with rare diseases can live their fullest and best lives. Every day, we elevate care, advance research, and drive policy in a purposeful and holistic manner to lift up the rare disease community."

TOP RESOURCES

- Genetics Referral Guide
- RareChromo.org
- Amish, Mennonite, and Hutterite Genetic Disorder Database
- Children with a rare congenital genetic disorder: a systematic review of parent experiences
- Check for a foundation for the specific diagnosis, for example: Prader Willi, Dandy Walker, Williams Syndrome.

DIFFERENCES

*THAT **MAY** BE PRESENT*

- Presentation and prognosis varies greatly not only depending on the diagnosis, but for some, within each diagnosis.
- 89.6% of pediatric rare disorders compromise the nervous system (3)
- Cognitive deficits
- Challenges with speech or social skills.
- Eating and digestive issues, such as difficulty swallowing or an inability to process nutrients.
- Limb or facial anomalies, which include missing fingers or a cleft lip and palate.
- Movement disorders due to muscle stiffness or weakness.
- Neurological issues such as seizures or stroke.
- Poor growth or short stature.
- Vision or hearing loss. (4)

REFERENCE LIST

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WISCONSIN RESource RESPONSIVE EQUITABLE SUPPORT A CESA 5 Subsidiary Professional Development Grounded in Research