Putting A Spotlight On...

Kleefstra Syndrome

What is Kleefstra Syndrome?

- Kleefstra syndrome is a rare genetic condition that affects development and involves many body systems.

What are the signs/symptoms?

- Microcephaly
- Seizures
- Hearing loss
- Wide, short skull (brachycephaly)
- Limited or absent speech
- Autism spectrum disorders
- Heart defects
- Sleep disorders
- Kidney defects
- Gastrointestinal problems such as gastroesophageal reflux (GERD)
- Protruding jaw
- Single eyebrow
- Widely spaced eyes
- High birth weight and childhood obesity
- And more...

What is Kleefstra Syndrome a result of?

- Kleefstra syndrome is a genetic condition that is caused by the loss of function of one copy of the EHMT1 gene. In about 50 percent of cases, a variation or “mutation” in one copy of the EHMT1 causes the loss of function. The remaining cases are caused by the deletion of one copy of a segment of chromosome 9, which includes the gene EHMT1.

What are the complications of Kleefstra Syndrome?

- Structural brain abnormalities, congenital heart defects, genitourinary abnormalities, seizures, and a tendency to develop severe respiratory infections

References:

- https://tinyurl.com/yck4wnjb
- https://tinyurl.com/4dh3un76