Diagnosis of Kleefstra Syndrome

Signs from birth

The first signs of Kleefstra syndrome vary from person to person. In a study of 14 babies, 5 appeared developmentally typical at birth and were only seen as a cause for concern a few months later. One baby seemed to progress as expected until experiencing seizures at 3 months old. In the other 9 babies there were immediate signs from birth. Signs and symptoms varied from a weak cry to feeding difficulties and unusual facial features. Additional information on the signs and symptoms can be found in the health section of this website.

Diagnosis

Kleefstra syndrome can be caused by a deletion in the chromosome region labelled 9q34.3. Deletion means that a small part of the chromosome is lost during DNA replication. Alternatively, Kleefstra syndrome can be caused by a mutation in the EHMT1 gene. This mutation in the gene leads to the following characteristics:

- Intellectual disability
- Childhood hypotonia (low muscle tone)
- Characterised facial appearance such as arched eyebrows, a short nose and open mouth with a protruding tongue
- Some individuals with Kleefstra syndrome may also have congenital (from birth) heart and kidney problems. Research has found that approximately half of individuals with Kleefstra syndrome experience these congenital defects. For more information from Dr Kleefstra about this click here.

Detection of gene deletion or mutation can take place by:

- Conventional cytogenetic analysis – this analysis technique looks at the chromosome in such detailed resolution that even tiny alterations can be detected.
- Chromosomal microarray (CMA) – This technique finds extra or missing (deleted) segments of the chromosome.

Prenatal screening

Prenatal screening is also offered to women with high risk pregnancies. High risk pregnancies include women who are of a higher maternal age, have a family history of chromosomal abnormality or have had foetal abnormalities detected in a prenatal ultrasound.

The genetic testing registry provides more information about the types of genetic tests available for Kleefstra syndrome.

The information on this website was developed in collaboration with www.kleefstrasyndrome.org. This is the parental support group for Kleefstra, a well-developed community with an active facebook group and parent support conference. Please visit their website for further information and to get in touch with families of individuals with Kleefstra syndrome.