

2018 KLEEFSTRA SYNDROME CONFERENCE – BOSTON, MA

June 29- July 1st, 2018

Doctors and Therapies: A Parent's Perspective

What We'll Cover Today

2

- ❑ The Kleefstra Syndrome team of doctors that are required
- ❑ Therapies to consider
- ❑ Concluding thoughts





Doctors

Genetics

Paediatrics

Cardiology

Nephrology

Urology

Neurology

Psychiatry

Ophthalmology (Eyesight)

Otolaryngology/Audiology (Hearing)

Doctors

6

□ **Genetics**

- Often first contact for KS diagnosis
- Helpful in determining whether KS was genetically passed down (assess risk for future children)
- Also provides sibling counselling
- Usually very limited contact after initial assessment

□ **Paediatrics**

- Likely unfamiliar with KS and rare disorders
- May need to be pressed for referrals or educated on new therapy techniques
- Ultimately need to play quarterback role: coordinator of care

Doctors (cont'd.)

7

□ **Cardiology**

- Up to 50% of KS children are believed to be born with structural or other heart issue
- For great majority of babies, the heart problem is not severe and usually resolves naturally in time
- However, should see a cardiologist and conduct echocardiography/other tests early to determine any issues
- Issues can include atrial septal defect (ASD) and valve defects

□ **Nephrology**

- Many KS kids have kidney issues (renal insufficiency)
- Conditions reported include hydronephrosis (enlarged kidneys), which has many causes including blockages, urine reflux and cystic kidneys
- Children should have their renal and urinary tracts imaged

Doctors (cont'd.)

8

□ **Urology**

- Approx. 50% of KS males are born with immature genital features
- May be harmless, but occasionally requires corrective surgery

□ **Psychiatry**

- Manages behavior changes and mental health development
- Coordinated with behaviour therapists
- Puberty and regression issues

□ **Neurology**

- Assistance managing seizures
- Sleep disorders

Doctors (cont'd.)

9

- **Ophthalmology** (eyesight) and **Otolaryngologist** (hearing) or **Audiologist**
 - ▣ Some KS children have significant challenges with hearing and eyesight

Therapies

Behaviour

Occupational

Speech

Physical

Other

Behaviour Therapy

11

- KS problem behaviors can include invading others' space, chewing objects, biting, hand in mouth, obsessive-compulsive acts, stereotypic movements, outbursts of anger, self-injury and aggression towards others
- Behaviour therapy includes a variety of techniques which look at specific, learned behaviors and how the environment influences those behaviors
- These behaviours may include communication and language, social skills, and adaptive behavior such as eating and toileting
 - ▣ **Applied behaviour analysis (ABA)** is a scientific discipline concerned with applying techniques based upon the principles of learning to change behavior of social significance
 - ▣ ABA can be used with anyone that needs to increase a skill (positive behavior) or decrease a negative behavior

Occupational Therapy

12

- Occupational therapy (OT) covers a number of areas that KS children can benefit from:
 - ▣ **Fine Motor Skills and Handwriting:** Identify specific deficits in the child and suggest activities that will help improve those skills
 - ▣ **Self-Care:** Help a child perform and be independent in self-care skills like eating, dressing, toileting, bathing and grooming
 - ▣ **Play:** Help children participate and interact with others in play
 - ▣ **School:** Help children participate and adapt to schooling. They may suggest aids for writing, and other classroom activities to help them participate
 - ▣ **Environmental Modifications:** Suggest modifications in the home, school, or playground that will help the child participate more easily

Speech Therapy

13

- KS children generally experience speech impairments; severity varies from child-to-child
- Often able to understand more than they can express
 - ▣ Using pictures, signs or tablets can help children develop alternative ways of communicating
- Early intervention from a Speech and Language Therapist is essential to help children with KS develop communication skills
 - ▣ Significant overlap with behaviour therapy (ABA)
- Speech therapy can often help with feeding issues, and vice versa, as mouth muscles impact both speech and feeding – therefore, OT is critical as well

Physical Therapy

14

- Virtually all children with KS have low muscle tone, which generally affects the whole body (including arms and legs)
- Goal of physical therapy services is to enhance function and mobility, given the gross motor delays and slower milestones
- Physical therapists might guide children through a variety of different exercise routines and activities such as:
 - ▣ Developmental activities such as rolling, crawling, walking and climbing stairs
 - ▣ Flexibility exercises to increase range of motion
 - ▣ Balance and coordination activities
 - ▣ Adaptive play
 - ▣ Aquatic (water) therapy
- Some children require supports or insoles steady loose joints and may benefit from practice with a walker

Other Therapies

15

Other forms of therapy include:

- ▣ Hippotherapy (horseback riding)
- ▣ Music Therapy
- ▣ Massage and Relaxation
- ▣ Specialist Sensory Therapy
- ▣ Deep Brain Stimulation

The data set for the effectiveness of these therapies remains small, so broad-based conclusions may be difficult to draw

16

Concluding Thoughts

Concluding Thoughts

17

Doctors

- ❑ KS kids require a multi-disciplinary approach to care
- ❑ Educate your paediatrician and other specialists
- ❑ Proactive management of doctors and related appointments
- ❑ Always consult with your medical team before giving or changing medications – every child is unique

Therapies

- ❑ KS kids require a multi-disciplinary approach to care
- ❑ Early interventions are critical
- ❑ Consider that some therapies have multiple benefits/overlap
- ❑ Consider quality of service providers
- ❑ Leverage government resources where possible
- ❑ Every child is unique