

# Klinefelter Syndrome

## Table of Contents

|   |   |
|---|---|
| At a Glance .....   | 2 |
| Medical and Dietary Considerations.....   | 3 |
| Education Supports.....   | 3 |
| Behavioral and Sensory Support.....   | 5 |
| Physical Activity, Trips, Events .....  | 6 |
| School Absences & Fatigue .....   | 6 |
| Emergency Planning.....   | 6 |
| Resources .....   | 7 |
| Info for School Nurse and Primary Care Staff .....                              | 8 |
| Meet a Young Man with Klinefelter Syndrome: <i>Graham, Mystery Writer</i> ..... | 9 |

## Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

## Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

# Klinefelter Syndrome

## At a Glance

Klinefelter Syndrome (KS) is caused by an extra “X” chromosome. It occurs only in males. Boys and men with KS are genetically male and have male characteristics. They usually have average intelligence. Very often individuals will have minimal symptoms and not realize they have this extra chromosome. However, occasionally it can cause problems that may require treatment.



Meet Graham on page 9.

## Common Features of Klinefelter Syndrome:

### In babies and toddlers

- weak muscles and very flexible (hypermobile) joints
- learning to sit up, crawl, walk and talk later than usual
- being quieter and more passive than usual
- having undescended testicles, only one testicle, or a smaller penis

### In childhood

- shyness and low self-confidence
- problems with reading, writing, spelling and paying attention
- mild dyslexia or coordination problems
- low energy levels
- difficulty socializing or expressing feelings

### In teenagers

- growing taller than expected for the family (with long arms and legs)
- broad hips
- poor muscle tone and slower than usual muscle growth
- reduced facial and body hair that starts growing later than usual
- small testicles and possibly enlarged breasts
- may continue to have problems with reading, writing, spelling and paying attention
- may continue to have difficulty socializing or expressing feelings
- may continue to have difficulty socializing or expressing feelings

## In Depth

### Medical and Dietary Considerations

#### What you need to know

It is important to ask the parents about the unique medical issues of their child and what medical management challenges they may face at school. Klinefelter Syndrome occurs in males with an extra X chromosome (XXY rather than XY.) Low testosterone is the reason for many issues in KS. Many boys with Klinefelter Syndrome are followed by an endocrinologist when they reach the age of puberty. They may be placed on testosterone treatments. No special diet is required for KS although a well-balanced diet is important. Here are some characteristics that might be present:

- Hypogonadism (reduced or absent hormone secretion from the testes)
- Absent, delayed or incomplete puberty
- Swelling of breast tissue during puberty
- Weak bones
- Scoliosis
- Dental problems
- Cognitive, reading, and language difficulties
- Depression and/or social differences
- Increased risk of diabetes and hypothyroidism
- Increased risk of breast cancer and lupus as adults

#### What you can do

- Encourage a yearly check-up in the child's Medical Home and request updated recommendations.
- Be aware of any academic or behavioral changes. Contact parents when any differences are noticed.
- Provide and seek appropriate supports as needed, whether school based or more medically based, e.g.:
  - Communication supports
  - OT or PT for supporting motor development, coordination, tone and sensory needs
  - Supports for emotional or behavioral issues that might arise, as needed

### Education Supports

It is important to have high learning expectations for children who have Klinefelter Syndrome utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

## What you need to know

Many individuals with KS grow up to be successful in academics, careers, and personal lives. They do not typically have an intellectual disability. Most have average to low average intelligence. However, there are differences related to the presence of an extra “X” chromosome. Boys with KS may have developmental delays, issues with sensory integration, or ADHD. Individuals with KS often have auditory processing and language-based learning disabilities. If learning difficulties aren’t addressed it can lead to lower academic achievement, reduced self-esteem, and behavior problems. Specific difficulties may include:

- Receptive language issues - difficulty processing what they hear
- Difficulty with reading speed and/or reading comprehension
- Problems with spelling and math
- Lower skills in auditory, memory, confrontation naming (word retrieval), and verbal fluency
- May have difficulty putting thoughts, ideas, emotions into words
- May find it difficult to concentrate in a noisy class.

## What you can do

### Academic Supports for Reading Comprehension, Math, Language

- Help with organization
- Provide routine, structure, and consistency
- Present information in concrete manner
- Use manipulative materials to demonstrate concepts
- Simplify verbal information and explain concepts clearly
- Provide visual cues and instructions
- Repeat information and use positive reinforcement
- Provide quiet learning environment; background noise may be distracting
- Help prioritize work and activities
- Allow a concentration “break”
- Teach good study skills
- Use technology when appropriate

### Communication Supports

- Appropriate and timely intervention by a speech and language pathologist can help keep speech and language skills on track.
- Promote language understanding by using simple short sentences, visual prompts, and pictures.
- Use a child’s experiences and interests to engage child in learning.
- Allow extra time, repeat directions, provide lesson summaries, and record lessons so the child can listen again.
- Have child repeat directions.

- Create situations in which children can practice their skills in natural settings.
  - Allow opportunities to use descriptive expression throughout the day.

### Promote Language Development

- Provide ample time for responding
- Increase the child's self-confidence by calling on them when they know answers
- Encourage a child to repeat the questions before responding
- Allow a child time to rehearse and respond

### OT or PT – if needed

- Support motor development, coordination, tone and sensory needs

### Interventions for Auditory Processing

- Repeat instructions
- Provide non-distracting environment

## Behavioral and Sensory Support

### What you need to know

Many social and behavioral symptoms in KS may be related to language and learning difficulties. Children with KS may have trouble using language to express their thoughts and needs. This may inhibit social interactions.

#### Children with KS may be:

- Quieter/shy
- Less assertive or self-confident, or immature
- Sensitive
- Delayed in social skills
- Anxious or depressed
- Restless
- Less physically active
- More helpful and eager to please

#### Teenage years

- Teenagers may notice differences between themselves and other typical teenagers.
- They are at risk for depression, substance abuse, behavior problems.
- They may feel withdrawn and/or sad.
- Note - They are no more likely to have serious psychiatric disorders or get in trouble with the law than their typical peers.

### What you can do

#### Support healthy emotional and behavioral development

- Teach self-talk to help the child develop self-control. Use specific, short phrases such

as “stop and think.”

- Assist with social skill development
  - Teach basic rules of social behavior
  - Model, rehearse, and practice and provide feedback
- Support development of self-esteem
  - Encourage the child to participate in sports, clubs, or other structured activities that are meaningful to them.
- Make sure language supports are in place to help with social development
- Monitor emotional and behavioral health and refer as necessary.
  - Learn how to tell when the child is getting frustrated and utilize early intervention
  - If the child has anxiety and/or depression, medication or counseling may be helpful

## Physical Activity, Trips, Events

### **What you need to know**

- Exercise is important and should be encouraged,
- Coordination may be a problem and make individuals a target for teasing.
- Lack of stamina and energy levels can be problem

### **What you can do**

- Encourage exercise, especially those exercises that build muscle mass and motor skills.
- Encourage practice to help build coordination.
- Teach children who have KS how to pace themselves and learn when to rest.

## School Absences & Fatigue

Children with Klinefelter Syndrome should not have excess absences or fatigue.

## Emergency Planning

Develop an emergency plan if necessary, depending on the needs of individual children.

## Resources

### **Medline Plus**

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.

<https://medlineplus.gov/genetics/condition/klinefelter-syndrome/>

### **Living with XXY**

Living with XXY is a positive, supportive organization with a comprehensive website. Check out the great info under the “Resources” and “Early Intervention” tabs. <https://livingwithxxy.org/>

### **American Association for Klinefelter Syndrome Information and Support**

AAKSIS is a national volunteer association with the mission of education, support, research, and understanding of 47,XXY and its variants. <https://www.aaksis.org/>

### **Klinefelter Syndrome Association UK**

The Klinefelter Syndrome Association offers support and information to all affected by, or having an interest in Klinefelter Syndrome. They have information available which can assist employers, teachers, social workers, and medical professionals. <http://www.ksa-uk.net/>

## Info for School Nurse and Primary Care Staff

Klinefelter Syndrome is a genetic condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. The American Academy of Family Physicians published a series of reviews on genetic conditions to provide a overview for primary care providers. This article on Klinefelter Syndrome from *The American Family Physician* provides a brief description of presentation, diagnosis, and treatment. <https://www.aafp.org/pubs/afp/issues/2005/1201/p2259.html>.

### HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, ensure that the parents have signed a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

### Example letter for physician to provide to school

"My patient \_\_\_\_\_ has been diagnosed with Klinefelter Syndrome. This is a rare genetic condition in boys that can impact physical and social development. These boys often have difficulties with reading, math and language. Communication support is often necessary. This condition requires an Individualized Education Program (IEP) or 504. Because of this, \_\_\_\_\_ needs the following accommodations:"



## Meet a Young Man with Klinefelter Syndrome: *Graham, Mystery Writer*

At age 21, Graham loves to spend time on his favorite pastime of writing mysteries and screen plays. The topics vary but they often involve science-fiction. The stories have intrigue- like explorers going into a cave and finding gold but then someone is kidnapped and, and, and....they always have a cliff hanger to leave the reader wondering! He also recently started painting and he is exploring this interest and hopes to continue it as he transitions into the developmental disabilities services in his state.

Graham is outgoing, approachable and not shy about asking questions! He is athletic and likes to play basketball. He has a girlfriend and Lori describes the two as “fantastic friends.” In addition to his artistic endeavors, he loves to play Xbox games and loves social events like parties.



Graham is the 4th out of 5 children and lives in Massachusetts. As a young child he was involved in Early Intervention following his diagnosis at age 2 of Klinefelter (47 XXY). His mother advocated for genetic testing when she noticed that he was small for his age, his speech was difficult to understand, and he had some behaviors that she thought were unusual.

Throughout school he has received related services such as Speech and PT. Although his mother wanted him to be included in regular classes, the school system was in favor of him being in special education classes. After much advocating, this year Graham began attending a residential school during the week and lives at home on the weekends. His mother explains that this situation imitates college life in a way.

Lori explains that, like a person on the autism spectrum, Klinefelter Syndrome encompasses a wide variety of abilities and challenges. Some of his challenges include difficulty with executive functions and at times making decisions that are not safe or wise. He has difficulty generalizing concepts. His natural curiosity may sometimes cause him to, for example, run off to explore. Medically, he has had teeth removed and has some joint pain and low tone. Weight control has been an issue as has some difficulty moving his ankle joints through a full range. A decision that might occur in families is whether or not to have testosterone shots.

“He is pretty independent and laid back,” explains Lori. Graham is close to everyone in his family and loves them very much and as a young adult transitioning, he will have lots of support in making choices about the next steps in his life.

**Lori's Advice to Parents:**

- Because of the awareness level around autism, it might be helpful to advocate for your child describing this condition as “similar to autism” if it truly is.
- Don't put limits on your child because of a diagnosis. There is no cap! Look for their strengths and build on those- like Graham's artistry and writing skills.
- See them as unique and advocate for them.
- Educate yourself. Use the AXYS website located at [www.genetic.org](http://www.genetic.org).
- Watch for bullying. Because your child may not notice teasing or recognize bullying and be more gullible, make sure to be aware of potential issues.

**Lori's Advice to Teachers:**

- Many of the ideas above also apply to school personnel.
- They may have cognitive, language and some behaviors that are similar to students who have autism. You may need to advocate for supports and services that are used for children on the autism spectrum.

*Thank you so much to Graham and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.*