

# A Brief overview of endocrine issues in Klinefelter syndrome

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## Hypogonadism in KS

- ▶ Primary testicular failure
  - Central hormones responsible for onset of puberty are normal
  - The testis itself is abnormal and unable to respond appropriately to LH and FSH
- ▶ Once "pituitary puberty" starts
  - Pituitary attempts to drive response from the testis
    - Sperm cell production abnormal – FSH rises
    - Testosterone production abnormal – LH rises
- ▶ Hypergonadotropic hypogonadism
- ▶ LH and FSH won't be elevated prior to puberty

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## Timing of testosterone therapy

- **Early or late childhood?**
  - Small penis
  - Behavioral benefits?
  - Physical benefits of low doses?
    - Theoretical based on low doses present in typical boys
- **Age driven**
  - Normal time of puberty in boys – 12 to 12.5 years
- **LH driven**
  - Start testosterone when LH begins to rise
  - Indicates that the pituitary is trying and failing
- **Gynecomastia – breast development in males**

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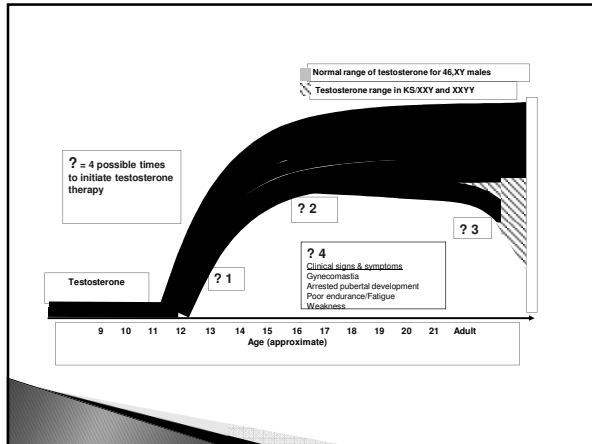
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### Consequences of Testosterone Deficiency

- ▶ reduced body hair
- ▶ decreased muscle mass and strength
- ▶ increased fat mass & altered fat distribution
- ▶ unfavourable cholesterol changes (increased LDL and reduced HDL)
- ▶ decreased hemoglobin
- ▶ decreased libido, erectile dysfunction
- ▶ osteoporosis
- ▶ depressed mood

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### Benefits of testosterone

- Virilization
  - Outward signs of pubertal development
  - Promotion of normal sexual function and development
  - Maintain similarly with peers
  - Self-esteem
- Muscle development/fat distribution
- Body proportions – reduction in excess limb length
  - Testosterone promotes bone maturation
  - Testosterone promotes spine growth
- Bone maturation and mineral accumulation
- Prevention/treatment of gynecomastia
- Behavior?

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## Disadvantages of testosterone

- ▶ Acne
- ▶ Increased sexual activity
- ▶ Increased strength
- ▶ Short stature if started too early or titrated too fast

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## Options for testosterone therapy

- Oral
- Injected
- Transdermal
- Buccal
- Axillary
- Implant

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## Other hormonal abnormalities in KS

- ▶ Hypothyroidism
- ▶ Diabetes
  - Generally related to excess body fat
- ▶ Low bone density
  - Vitamin D deficiency
  - Hypotonia, low muscle mass
  - Delayed testosterone replacement or testosterone deficiency
  - Overt osteoporosis (fracturing) is uncommon

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# *eXtraordinary Kids Clinic*

Children's Hospital Colorado  
University of Colorado School of Medicine  
Aurora, Colorado

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## *eXtraordinary Kids Clinic*

### **Goals**

- ▶ Comprehensive Interdisciplinary Clinical Care
- ▶ Research
- ▶ Education and Support

for children, adolescents, and young adults with X&Y chromosome variations

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## *eXtraordinary Kids Clinic*

### **TEAM:**



**Nicole Tartaglia, MD**

- Clinic Director
- Developmental-Behavioral Pediatrics
- Medical Evaluations
- Medication Management of Behavioral / Emotional Disorders / ADHD, etc.



**Susan Howell, MS, CGC**

- Genetic Counselor
- Clinic and Research Coordinator

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*eXtraordinary Kids Clinic*

**TEAM:**

Child Psychologists:

- ▶ Rebecca Wilson, PsyD
- ▶ Jamie Blume, PsyD
  - Social-Emotional Problems
  - Behavioral Problems
  - Developmental Assessments



Neuropsychologists:

- ▶ Richard Boada, PhD
- ▶ Jennifer Janusz, PsyD
- ▶ Christa Hutaff-Lee, PhD
  - Learning Disabilities
  - Executive Functioning
  - ADHD Assessment



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**TEAM:**



- Speech/Language Therapy
  - Jackie Frazier, MS, CCC-SLP
  - Shelley Pfeiffer, MS, CCC-SLP
  - Speech/Language Evaluations



- ▶ Occupational Therapy
  - Syd Martin, MS, OTR
  - Motor Skills Assessment
  - Sensory Processing

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*eXtraordinary Kids Clinic*

**TEAM:**



- ▶ Pediatric Endocrinology
  - Philip Zeitler, MD, PhD
  - Testosterone treatment
  - Obesity
  - Type 2 Diabetes
  - Growth
  - Thyroid disorders

Also:

- NURSING – Karen Regan, RN
- SOCIAL WORK – Sarah McSwegin, LCSW
- CLINIC / RESEARCH ASSISTANTS –
  - Lisa Cordeiro, Tanea Tanda, Vinita Patel

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*eXtraordinary Kids Clinic*

- ▶ Evaluations
  - All X&Y chromosome variations
  - Infants, Children and Adolescents
  - Some young adults depending on what needs to be evaluated
  - Evaluation team members are individualized to what the patient needs
  
- ▶ Private Insurance and Medicaid for most states accepted
  - From out of state, we are usually "out of network"
  - Medical evaluations usually covered
  - Different plans have different coverage for psychological assessments

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*eXtraordinary Kids Clinic*

- ▶ Call Susan Howell for intake: **720-777-8361**
  - Susan talks with families on phone to collect background information and sends intake packet
  - Packet and medical/therapy/school records completed by parents and sent back to Clinic
  - Once packet is received, we work with family to determine team members for evaluation
  - Family will be called to schedule appointment(s) and discuss plans for checking insurance coverage
  - Waiting list depends on if full team evaluation is needed
    - currently 6-12+ months for new patients
  - Some patients have evaluation that lasts all week, with feedback session at the end
  - Others just see one or a couple providers in 1-2 days

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