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

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
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

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?
Disadvantages of testosterone

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

Options for testosterone therapy

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

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
**eXtraordinary Kids Clinic**

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

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**Goals**
- Comprehensive Interdisciplinary Clinical Care
- Research
- Education and Support

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

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**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
**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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**eXtraordinarY Kids Clinic**

**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 Cordero, Tanea Tanda, Vinta Patel
**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