The Association of motor skills with Adaptive functioning in Children with 47,XXY/Klinefelter and 48,XXYY syndrome

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Introduction

- Klinefelter syndrome (47,XXY) and 48,XXYY syndrome are two genetic disorders in which males have additional sex chromosome compared to the typical male karyotype of 46,XY.
- The conditions have been previously associated with cognitive, motor and adaptive (AF) delays.
- The aim of this study was to evaluate motor skills and their association with adaptive functioning in these groups.
- In this project we aimed to further describe and compare the visual motor integration (VMI) and motor skills in children and adolescents with 47,XXY/XXS and 48,XXYY, and to further analyze factors associated with motor deficits such as age, cognitive abilities and adaptive functioning skills.

Klinefelter Syndrome

- Occurs in 1:500-1:1,000 births
- Tall stature
- Clinodactyly (curved 5th finger)
- Low muscle bulk and tone
- Joint hyperextensibility, tremor
- Low testosterone production, infertility
- Language-based learning difficulties
- Delays in early motor development
- 5% or less have intellectual disability
- Visual perceptual skills are a strength
- Attention difficulties

48,XXYY syndrome

- Occurs in 1:18,000 births/shared medical presentation including:
  - Cubitus varus (narrowed carrying angle at elbow), Radioulnar synostosis
  - Clubfoot, seizures, scoliosis
  - 90% speech and language delays
  - 75% motor skill delays
  - Learning and educational difficulties are present
  - 30% with intellectual disability
  - Anxiety, attention difficulties, impulsivity, social problems and autism

Methods

- 64 male participants with 47,XXY/XXS and 46 participants with 48,XXYY (mean age KS= 12.16) (mean age XXY= 13.27, years old, age range 4-21 years).
- Participants were recruited for a study on neurodevelopmental and psychological features of sex chromosome aneuploidy through national advocacy groups for individuals with sex chromosome aneuploidy.
- The parents of each participant signed consent, IRB approval was granted.
- Instruments included: BOT-2, VMI, Vineland Adaptive Behavior Scales, WASI and WiSC

Results

- This is the first study to analyze and compare visual motor and motor skills in SCA.
- Both 47,XXY/XXS and 48,XXYY showed motor coordination deficits on the VMI, and deficits in the manual dexterity tasks of the BOT-2.
- Nonverbal cognitive skills (performance IQ) and visual perceptual skills were an area of relative strength for both groups.
- Low visual motor abilities were associated with lower cognitive scores in both 47,XXY/XXS and 48,XXYY groups.
- Low adaptive scores in the daily living skills in 47,XXY and low adaptive composite score in 48,XXYY.

Conclusions

- This study was part of an ongoing study at the University of California Davis, MIND Institute and The Children’s Hospital in Denver, Colorado through the eXtordinary Kids Clinic.
- Special Thanks to Nicole Tartaglia, M.D.
- Acknowledgement to the Occupational Therapy Department at Children’s Hospital Colorado.