

The Association of motor skills with Adaptive functioning in Children with 47,XXY/Klinefelter and 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/KS and 48,XXYY, and to further analyze factors associated with motor deficits such as age, cognitive abilities and adaptive functioning skills.

Results

Figure 1: Comparison of Age, Cognitive Scores, and Beery VMI results in 47,XXY/KS & 48,XXYY

	47,XXY/KS	48,XXYY	T-test Results
Age	N=64	N=46	
Mean (SD)	12.16 (3.48)	13.27 (4.46)	t(90)=-1.728, p=.087
Range	4.80-21.45	4.43-20.19	
IQ (WASI & WISC-IV)	N=64	N=46	
Verbal- Mean (SD)	92.66 (17.37)	75.93 (14.36)	t(108)=-5.35, p=.000
Verbal- Range	53-119	53-102	
Performance- Mean (SD)	100.64 (15.75)	87.09 (12.99)	t(107)=4.746, p=.000
Performance- Range	61-129	63-108	
Full Scale- Mean (SD)	96.11 (17.74)	79.91 (14.86)	t(107)=5.034, p=.000
Full Scale- Range	46-125	47-121	
Beery Visual Motor Integration	N=62	N=46	
Visual Motor Integration Mean (SD)	92.30 (14.35)	81.48 (11.26)	t(106)=4.239, p=.000
Visual Perception supplementary test	99.63 (15.14)	88.22 (13.58)	t(104)=4.021, p=.000
Motor Coordination supplementary test	87.46 (16.88)	77.73 (15.45)	t(101)=2.999, p=.003*

* p<0.05 for T-test comparing XXY to XXYY

Figure 2: Cognitive and Adaptive scores in 47,XXY/KS and 48,XXYY with below average compared to average VMI scores

	VMI Below Average	VMI Average or Above	T-Test
47,XXY/KS			
N	13	49	t(60)=.464, p=.645
Age	12.5 (2.45)	11.99 (3.71)	
IQ Mean (SD)			
Verbal	78.92 (22.16)	95.98 (14.43)	t(60)=-2.631, p=.010*
Performance	86.62 (14.98)	104.47 (14.12)	t(60)=-3.992, p=.000*
Full Scale	80.62 (20.92)	100.44 (14.9)	t(59)=-3.848, p=.000*
Adaptive Functioning Mean (SD)			
Communication	72.70 (19.60)	87.67 (16.69)	t(53)=-2.482, p=.016*
Daily Living	80.50 (22.82)	92.11 (16.54)	t(53)=-1.861, p=.068
Social	82.70 (20.47)	91.16 (18.27)	t(53)=-1.296, p=.201
Motor	95.00 (23.67)	107.60 (13.22)	t(41)=-1.271, p=.255
Adaptive Composite	78.40 (19.87)	89.24 (16.48)	t(53)=-1.814, p=.075
48,XXYY			
N	22	24	t(44)=2.64, p=.010*
Age	14.99 (4.01)	11.69 (4.33)	
IQ Mean (SD)			
Verbal	74.18 (14.0)	77.54 (14.82)	t(44)=-.789, p=.434
Performance	84.67 (13.9)	89.21 (12.04)	t(43)=-1.175, p=.247
Full Scale	76.36 (13.2)	83.17 (15.78)	t(44)=-1.576, p=.122
Adaptive Functioning Mean (SD)			
Communication	54.65 (16.77)	72.18 (19.31)	t(40)=-3.127, p=.003*
Daily Living	68.85 (10.49)	78.64 (15.55)	t(40)=-2.410, p=.021*
Social	70.80 (11.82)	79.14 (13.51)	t(40)=-2.119, p=.040*
Motor	96.75 (18.50)	96.73 (19.24)	t(21)=.002, p=.998
Adaptive Composite	67.68 (7.27)	75.45 (13.33)	t(39)=-2.357, p=.024*

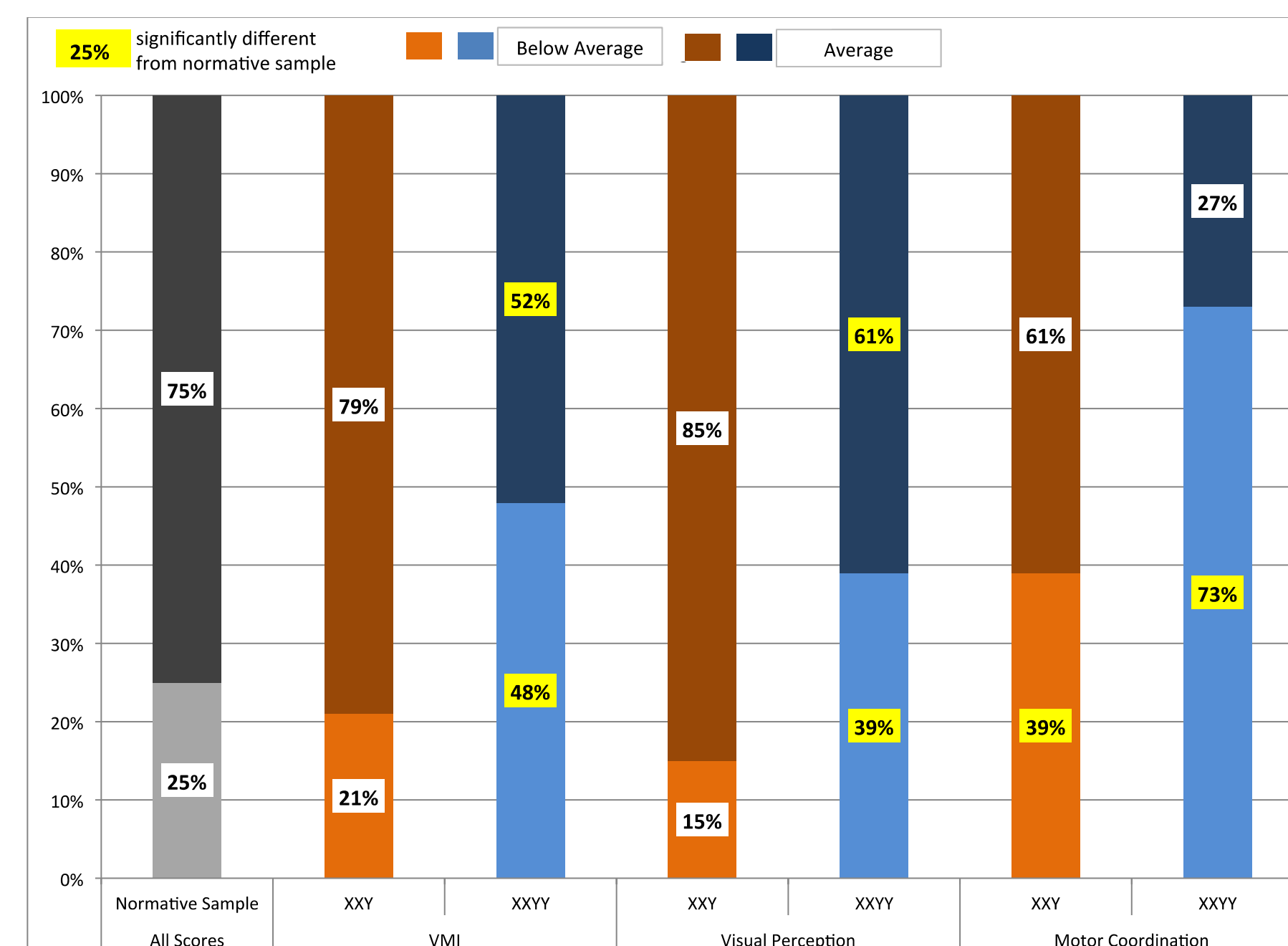
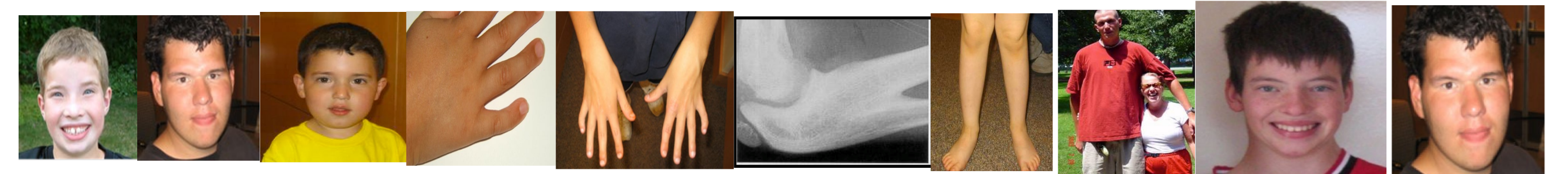


Figure 3: Results of Beery Buktenica VMI, Visual Perception, Motor Coordination in 47,XXY/KS and 48,XXYY

Figure 4: Comparison of Motor Skills on the BOT-2 in 47,XXY/KS and XXYY

BOT-2	XXY	XXYY	T-test and Fisher's Exact Test
N	47	17	
Fine Manual Control			
Mean (SD)	48.5 (12.66)	41.3 (11.68)	t(65)=-2.194, p=.032*
Percent Below Average	52.1% ^{^^}	72.2% ^{^^}	p=.576%
Fine Motor Precision	49.06 (10.33)	42.3 (11.01)	t(65)=-2.404, p=.019*
Percent Below Average	25.5%	60.0% ^{^^}	p=.35.8%
Fine Motor Integration	50.75 (9.69)	46.0 (10.91)	t(66)=-1.699, p=.094
Percent Below Average	14.6%	50.0% ^{^^}	p=.25.0%
Manual Coordination			
Mean (SD)	45.00 (9.37)	34.2 (13.79)	t(63)=-3.686, p=.000*
Percent Below Average	22.2%	62.5% ^{^^}	p=.31.1%
Manual Dexterity	42.35 (8.70)	37.3 (8.62)	t(64)=-2.093, p=.040*
Percent Below Average	15.2%	55.0% ^{^^}	p=.28.8%
Upper Limb Coordination	50.34 (9.42)	43.8 (9.94)	t(64)=-2.552, p=.013*
Percent Below Average	31.1% [^]	47.4% [^]	p=.35.9%
Body coordination			
Mean (SD)	46.37 (9.68)	34.9 (13.55)	t(61)=-3.761, p=.000*
Percent Below Average	29.8% [^]	60.0% ^{^^}	p=.40.2%
Bilateral Coordination	46.48 (9.36)	40.1 (8.36)	t(62)=-2.568, p=.013*
Percent Below Average	13.6%	52.9% ^{^^}	p=.24.6%
Balance	49.11 (10.49)	41.25 (11.81)	t(59)=-2.496, p=.016*
Percent Below Average	22.7%	42.3% [^]	p=.32.3%
Strength & Agility			
Mean (SD)	46.04 (9.18)	38.5 (8.83)	t(59)=-3.105, p=.003*
Percent Below Average	18.2%	64.7% ^{^^}	p=.31.1%
Running Speed & Agility	48.27 (8.76)	38.5 (5.86)	t(59)=-4.198, p=.000*
Percent Below Average	28.9%	70.0% ^{^^}	p=.41.5%
Strength	46.81 (8.16)	38.5 (8.83)	t(60)=-3.205, p=.002*
Percent Below Average	33.3% [^]	77.8% ^{^^}	p=.44.4%
Total Motor Composite			
Mean (SD)	45.68 (9.45)	38.11 (7.63)	t(59)=-2.944, p=.005*
Percent Below Average	27.2%	64.7% ^{^^}	p=.37.7%



Conclusions

- This is the first study to analyze and compare visual motor and motor skills in SCA.
- Both 47,XXY/KS 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/KS and 48,XXYY groups.
- Low adaptive scores in the daily living skills in 47,XXY and low adaptive composite score in 48,XXYY.
- Our study expands upon this finding to further report the supplementary tests of visual perception and motor coordination where motor coordination was found to be a significant area of weakness for both groups.
- Deficits in bilateral coordination were found in SCA than expected for the general population with specific difficulties in sequencing, planning and execution of these motor tasks observed during testing.

- 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 eXtrordinary Kids Clinic.
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Klinefelter Syndrome

- Occurs in 1:500-1:1,000 births
- Tall stature
- Clinodactyly (curved 5th finger)
- Low muscle bulk and tone
- Flat feet with ankle pronation
- Joint hyperextensibility, tremor
- Low testosterone production, infertility
- Language-based learning difficulties
- Delays in early motor development
- 80% of KS receive special education
- 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/KS and 46 participants with 48,xxyy (mean age KS= 12.16) (mean age XXYY= 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

- Results show there is large variability within both conditions.
- Higher proportion of individuals with both 47,XXY/KS and 48,XXYY with motor coordination deficits on the VMI compared to the normative sample.
- 47,XXY/KS and 48,XXYY had significant deficits in the manual dexterity tasks of the BOT-2.
- Boys with 48,XXYY showed more significant overall motor deficits compared to the 47,XXY/KS group.
- Nonverbal cognitive skills and visual perceptual skills were an area of strength for both groups.
- Lower visual motor skills were associated with lower cognitive scores in both groups.
- Lower adaptive functioning scores in activities of daily living with 47,XXY/KS and lower adaptive composite score in 48,XXYY group.