

## Columbia University & Yale University Research Update

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Looking to the Future  
For X and Y Chromosome Variations  
July 2013 Conference  
Denver, Colorado

Sharron Close PhD, MS, CPNP-PC Ilene Fennoy MD, MPH Arlene Smaldone PhD, CPNP-PC, CDE Nancy Reame PhD, MS, FAAN	Sharron Close PhD, MS, CPNP-PC Lois Sadler PhD, PNP-BC, FAAN Margaret Grey, Dr PH, RN, FAAN
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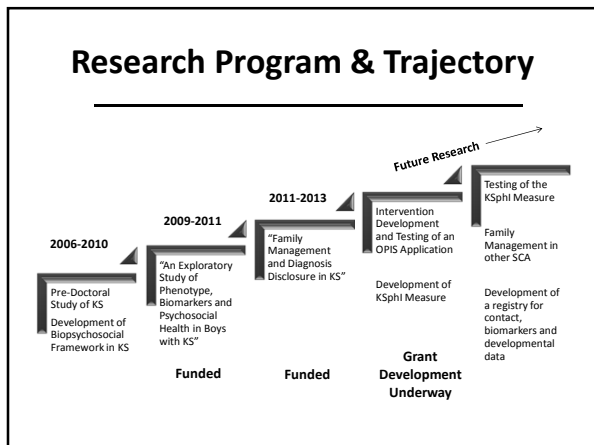
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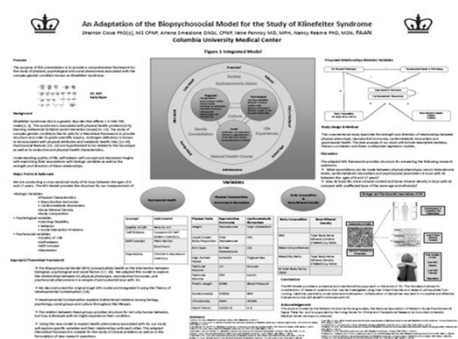
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## Biopsychosocial Framework in KS 2009



**An Adaptation of the Biopsychosocial Model for the Study of Klinefelter Syndrome**  
Sharron Close PhD, MS, CPNP-PC, Ilene Fennoy MD, MPH, Arlene Smaldone PhD, CPNP-PC, CDE, Nancy Reame PhD, MS, FAAN  
Columbia University Medical Center

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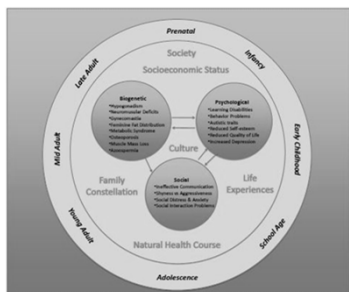
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## Adapted BPS for KS




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## Awareness and Index of Suspicion 2010

Phenotype, Biomarkers & Psychosocial Health Parameters in Boys with Klinefelter Syndrome: A Clinical Guide

Simon Stone PhD, MS, CRNP-PC, Arlene Smalton DNP, CRNP-PC, Jane Fenno MD, MPP, Nancy Krasner PhD, MS

Developmental Period	Characteristics
Infancy: Birth to 1 year	<ul style="list-style-type: none"> <li>Generally normal appearing</li> <li>Occasional genital anomalies such as hypospadias, undescended testes, shielded scrotum</li> </ul>
Early Childhood: 2-5 years	<ul style="list-style-type: none"> <li>Physically normal appearing</li> <li>Speech and language delay</li> <li>Clumsy &amp; uncoordinated movement</li> <li>Behaviorally passive and shy or aggressive</li> <li>Physical traits such as cleft-lip or high arched palate</li> </ul>
School age: 6-11 years	<ul style="list-style-type: none"> <li>Height velocity accelerations</li> <li>Leg longer than upper body segment</li> <li>Uncoordinated movement</li> <li>Speech and language delay</li> <li>Language based learning problems</li> <li>Slow auditory processing</li> <li>Social interaction problems, difficulty making friends</li> <li>Behaviorally shy or aggressive</li> </ul>
Adolescent: 12-18 years	<ul style="list-style-type: none"> <li>Enters puberty normally, phallus grows but testes remain small</li> <li>Reduced body hair compared to unaffected boys</li> <li>Reduced upper body strength</li> <li>Feminine pattern of fat distribution</li> <li>Osteopenia in later adolescence</li> <li>Lipomas or gynecomastia</li> <li>Autism spectrum</li> <li>Uncoordinated movement</li> <li>Hand tremor</li> <li>Social difficulties</li> <li>Behavioral issues associated with passivity, aggression and depression</li> <li>Other comorbid psychosocial conditions such as low self-esteem and low quality of life may be present</li> </ul>

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## Klinefelter Syndrome Clinical Observation Guide

Developmental Period	Characteristics
Infancy: Birth to 1 year	<ul style="list-style-type: none"> <li>Generally normal appearing</li> <li>Occasional genital anomalies such as hypospadias, undescended testes, shielded scrotum</li> </ul>
Early Childhood: 2-5 years	<ul style="list-style-type: none"> <li>Physically normal appearing</li> <li>Speech and language delay</li> <li>Clumsy &amp; uncoordinated movement</li> <li>Behaviorally passive and shy or aggressive</li> <li>Physical traits such as cleft-lip or high arched palate</li> </ul>
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# Bone Mineral Density in Boys with KS 2010

Observation of Early Bone Mineral Density in a Cohort of Peri-pubertal Boys with Klinefelter Syndrome: Preliminary Findings  
Sharon Cook PhD (S), MEd, CHMP, Liane Fennell MD, MPH, Anne Straburaj DMD, CRNP & Nancy Reame PhD, PAW

Columbia University Medical Center

**Introduction & Background**

Bone mineral density (BMD) is a key indicator of bone health. It is a measure of the amount of bone mineral in a given volume of bone. BMD is a predictor of fracture risk. In children, BMD is a predictor of peak bone mass, which is a predictor of osteoporosis risk in old age. The purpose of this study is to examine the relationship between BMD and clinical characteristics in a cohort of boys with KS.

**Methods**

A retrospective analysis of BMD data from 100 boys with KS. The data were analyzed for correlations between BMD and clinical characteristics such as height, weight, and testosterone levels.

**Results**

Figure 1 shows the mean BMD values for the cohort. Figure 2 shows the relationship between BMD and height. Figure 3 shows the relationship between BMD and weight. Figure 4 shows the relationship between BMD and testosterone levels.

**Conclusions**

BMD is a predictor of clinical characteristics in boys with KS. The relationship between BMD and height, weight, and testosterone levels is significant.

# Cardiometabolic Risk Factors 2011

Cardiometabolic Risk Factors in Boys with Klinefelter Syndrome  
Sharon Cook PhD (S), Anne Straburaj DMD, CRNP, Liane Fennell MD, MPH, Nancy Reame PhD, PAW

**Introduction**

Cardiometabolic risk factors are a major cause of morbidity and mortality. The purpose of this study is to examine the relationship between cardiometabolic risk factors and clinical characteristics in a cohort of boys with KS.

**Methods**

A retrospective analysis of cardiometabolic risk factor data from 100 boys with KS. The data were analyzed for correlations between risk factors and clinical characteristics such as height, weight, and testosterone levels.

**Results**

Figure 1 shows the mean cardiometabolic risk factor values for the cohort. Figure 2 shows the relationship between risk factors and height. Figure 3 shows the relationship between risk factors and weight. Figure 4 shows the relationship between risk factors and testosterone levels.

**Conclusions**

Cardiometabolic risk factors are a predictor of clinical characteristics in boys with KS. The relationship between risk factors and height, weight, and testosterone levels is significant.

# Psychosocial Health in Boys with KS 2012

Psychosocial Health Parameters in Boys with Klinefelter Syndrome  
Sharon Cook PhD (S), CHMP, MEd, Anne Straburaj DMD, CRNP, MEd, Liane Fennell MD, MPH, Nancy Reame PhD, PAW

**Introduction**

Psychosocial health parameters are a major cause of morbidity and mortality. The purpose of this study is to examine the relationship between psychosocial health parameters and clinical characteristics in a cohort of boys with KS.

**Methods**

A retrospective analysis of psychosocial health parameter data from 100 boys with KS. The data were analyzed for correlations between psychosocial health parameters and clinical characteristics such as height, weight, and testosterone levels.

**Results**

Figure 1 shows the mean psychosocial health parameter values for the cohort. Figure 2 shows the relationship between psychosocial health parameters and height. Figure 3 shows the relationship between psychosocial health parameters and weight. Figure 4 shows the relationship between psychosocial health parameters and testosterone levels.

**Conclusions**

Psychosocial health parameters are a predictor of clinical characteristics in boys with KS. The relationship between psychosocial health parameters and height, weight, and testosterone levels is significant.



### Family Management & Diagnosis Disclosure in KS

- Concurrent Triangulated Mixed Method Study
- 40 Parents of sons with KS
- Qualitative Interviews
- Quantitative Measures
- Combined analysis to describe how families manage the health of their sons and how they handle disclosure of diagnosis

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### Research Dissemination

**Publications**

- Close S, Fennoy I, Reame N, & Smaldone, (2010) "Klinefelter Syndrome: Overview and Index of Suspicion" Journal of Pediatric Nursing 25 (6) 592-4.
- Close, S. Smaldone, A. Fennoy, I. Reame, N. Grey, M. (2013) "Using of information technology and social networking strategies for recruitment of research participants: Experience from an exploratory study of pediatric Klinefelter Syndrome. Journal of Medical Internet Research doi:10.2196/jmir.2286

**Current Writing Projects:**

- Close, Smaldone, Fennoy & Reame: An Exploratory Study of Physical Phenotype, Biomarkers Psychosocial Health in Boys with KS
- Close, Smaldone & Fennoy: Pubertal Comparison of Bone Mineral Density in Boys with KS
- Close, Sadler & Grey: Family Management in KS
- Close, Sadler & Grey: Diagnosis Disclosure
- Close, Sadler & Grey: How to care for families who have sons with KS

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### Funders & Support

- Alpha Zeta Chapter of Sigma Theta Tau
- National Association of Pediatric Nurse Practitioners
- Pediatric Endocrinology Nursing Society
- Irving Institute for National Center for Advancing Translational Sciences at Columbia University Medical Center (NIH Grant UL1 RR024156)
- Yale School of Nursing T32 Post-Doctoral Fellowship Training Grant (NIH 5T32NR008346-08)

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**Questions?**



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