Columbia University & Yale University
Research Update

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

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Research Program & Trajectory

2006-2010
Pre-Doctoral Study of KS Development of Biopsychosocial Framework in KS
Funded

2009
"An Exploratory Study of Phenotype, Biomarkers and Psychosocial Health in Boys with KS"
Funded

2009-2011
Development of Biopsychosocial Framework in KS
Funded

2011-2013
"Family Management and Disclosure of KS"
Funded

Future Research

Testing of an OPIS Application Development of KSphIMeasure Testing of the KSphIMeasure Family Management in other SCA Development of a registry for contact, biomarkers and developmental data
Grant Development Underway

Biopsychosocial Framework in KS

2009

An assessment of the biopsychosocial determinants of the quality of life of persons of a family members of a pediatric and adult patients with Klinefelter syndrome in the United States.
Adapted BPS for KS

Awareness and Index of Suspicion 2010

Klinefelter Syndrome Clinical Observation Guide
Bone Mineral Density in Boys with KS
2010

Cardiometabolic Risk Factors
2011

Psychosocial Health in Boys with KS
2012
Development of a Phenotype Index for KS (The KSphi)

<table>
<thead>
<tr>
<th>Physical Trait</th>
<th>Measurement Limits</th>
<th>Frequency (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hip Dorsiflexion</td>
<td>&gt;90° presence</td>
<td>48 (24.2%)</td>
</tr>
<tr>
<td>Upper Body Fatigue Perceived</td>
<td>&gt;60</td>
<td>46 (23.2%)</td>
</tr>
<tr>
<td>Distance to Height Ratio</td>
<td>&gt;3 in of standing height</td>
<td>49 (25.9%)</td>
</tr>
<tr>
<td>Facial Dysmorphism</td>
<td>&gt;90° presence</td>
<td>33 (16.9%)</td>
</tr>
<tr>
<td>BMI</td>
<td>&gt;90° presence</td>
<td>30 (15.6%)</td>
</tr>
<tr>
<td>Dysmorphia</td>
<td>Present or absent</td>
<td>5 (2.6%)</td>
</tr>
<tr>
<td>Facial Fatigue</td>
<td>&gt;3 in of standing</td>
<td>42 (21.7%)</td>
</tr>
<tr>
<td>Deletion/Restriction</td>
<td>Present or absent</td>
<td>1 (0.6%)</td>
</tr>
<tr>
<td>High School Failure</td>
<td>Present or absent</td>
<td>29 (15.2%)</td>
</tr>
<tr>
<td>Divorce</td>
<td>Present or absent</td>
<td>33 (16.9%)</td>
</tr>
<tr>
<td>extremes</td>
<td>Present or absent</td>
<td>20 (10.4%)</td>
</tr>
<tr>
<td>Depression</td>
<td>Present or absent</td>
<td>0 (0.0%)</td>
</tr>
</tbody>
</table>

An Exploratory Study of Boys with KS 2012

Theoretical Framework: Family Management Style 2013

Knaff, Deatrick & Havill, 2012
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

Research Dissemination

Publications

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

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)
Questions?