Family Management Style and Diagnosis Disclosure in Klinefelter Syndrome: Preliminary Findings

Looking to the Future
For X and Y Chromosome Variations
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Learning Objectives

• Explore how KS affects the child and the family system
• Describe the vulnerabilities and needs of parents of boys with KS and how they manage the health of their sons
• Discuss how to initiate conversation with health care providers concerning how we can optimize clinical support for KS families.

It’s a Family Affair

Phenotype
Developmental Trajectory
Psychosocial Maturation
Learning Potential
Health Risks: Metabolic, Bone and Mental Health

S e v e r i t y
Families of Children with Chronic Conditions

- Parental attitude shapes the way families manage the health of the affected child. (Gallo et al. 2005)
- How family life is affected by childhood chronic conditions:
  - Systems
  - Developmental Life course
  - Symbolic Interaction
  - Stress and Coping
  - Resilience
  - Family Management Style (Knafl, VanRiper & Knafl, 2012)

Family Management

- Families have different responses to chronic illness based upon real and perceived capabilities. (Summers et al. 2005)
- Management styles may vary from
  - Accommodating
  - Enduring
  - Struggling
  - Floundering (Knafl et al. 1996)

Theoretical Framework: Family Management Style

Knafl, Deatrick & Havill, 2012
Previous Study

"An Exploratory Study of Physical Phenotype, Biomarkers and Psychosocial Health in Boys with Klinefelter Syndrome"
Close et al. (2011)
• Recruitment Challenges:
  – Some parents had not disclosed the diagnosis
  – Difficult to reach target population with traditional recruitment means
• Collection of demographic information and medical history of boys:
  – Parents wanted to talk about their stories
  – Expressed frustration about lack of information
  – Parents complained that they did not know how to care for their sons

Current Study: Family Management and Diagnosis Disclosure in Klinefelter Syndrome

• Challenges from the previous study led to new research question:
  "What is it like to have a son with KS?"
  – How do parents manage information?
  – How do parents care for their sons?
  – How do parents decide to disclose the diagnosis to their sons and others?

Purpose: Aim 1

• To explore the needs of families who have sons with KS
• How they feel about raising a son with KS
• How they manage the health of their son
• How they make the decision to disclose the diagnosis to their sons
Purpose: Aim 2

• To describe family management style, family quality of life and perceived stress in parents

• To examine the relationships among parental disclosure of diagnosis and these factors

Methods

• Design:
  Concurrent Triangulated Mixed Method
• Qualitative interviews
• Quantitative Surveys
• Sample: 40 families of boys with KS using purposive sampling for maximum variation

Data Collection

Qualitative Interviews
• Interviews conducted using the semi-structured interview guide
• 45 minute interviews conducted via SKYPE or telephone
• All interviews were recorded and transcribed.
• Analysis by Interpretive Description Method (Thorne, 2008) using Atlas.ti software

Quantitative
• Survey’s were self-administered via Qualtrics® Survey software.
• Participants were invited by email link.
• Responses were recorded and downloaded to an Excel database.
Quantitative Measures: 3 Self-Report Surveys

- **Family Quality of Life Survey** (Hoffman et al. 2006)
  - 25 Item Survey
  - Measures family quality of life.
  - 5 subscales: family interaction, parenting, emotional well-being, physical/material well-being and disability-related support.

- **Family Management Measure** (Knafl et al. 2011)
  - 45 Item Survey
  - Measures how families manage caring for a child with a chronic condition and how they incorporate condition management into everyday lives.

- **Stress** (Cohen et al., 1983)
  - 10 Item survey
  - The degree to which situations in one’s life is appraised as stressful.

Qualitative Approach

Semi-Structured Interview

- What is it like to have a son with KS?
- How do you find information about KS?
- How do you manage your son’s health care?
- How did you disclose the diagnosis to your son?
- What do you worry about?
- What do families of sons with KS need?

Timeline to date

- IRB Approval: August 2012
- Recruitment: September 2012 - February 2013
- Data Collection: Completed February 2013
- Preliminary Results: Ongoing Analysis with Preliminary Results
Results: Characteristics of the Sample

N=40

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency</th>
<th>Percent</th>
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<tbody>
<tr>
<td>Parent Education</td>
<td></td>
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<tr>
<td>HS Diploma or less</td>
<td>7</td>
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<tr>
<td>College Degree</td>
<td>23</td>
<td>57.5%</td>
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<tr>
<td>Graduate Degree</td>
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<td>25%</td>
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<tr>
<td>Son’s Karyotype</td>
<td></td>
<td></td>
</tr>
<tr>
<td>47, XXY</td>
<td>37</td>
<td>92.5%</td>
</tr>
<tr>
<td>47, XXY/46XY</td>
<td>3</td>
<td>7.5%</td>
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<tr>
<td>Diagnosis of KS</td>
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<td></td>
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<tr>
<td>Pre-natal</td>
<td>21</td>
<td>52.5%</td>
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<tr>
<td>Post-natal</td>
<td>19</td>
<td>47.5%</td>
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<tr>
<td>Disclosure of Diagnosis to Son</td>
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<td></td>
</tr>
<tr>
<td>Yes</td>
<td>25</td>
<td>62.5%</td>
</tr>
<tr>
<td>No</td>
<td>15</td>
<td>37.5%</td>
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</table>
Preliminary Results N = 40:
Perceived Stress
Higher Scores Are Associated with Higher Perceived Stress

Stress Total by Age Group

Preliminary Results N = 40:
Family Quality of Life
Higher Scores Are Associated with Better Quality of Life
FQOL Family Interaction
The degree to which families interact in a positive family-supportive way

Mean Subscale Scores

Son's Age Group in Years

FQOL Parenting
Use of skills by parents and other adults in the household to help the affected family member

Mean Subscale Scores

Son's Age Group in Years

FQOL Emotional Well-Being
Measure of support and self-renewal within the family

Mean Subscale Scores

Son's Age Group in Years
FQOL Physical/Material Well-Being
Availability of physical and material resources for the family to obtain necessary medical care and to feel secure at home.

Mean Subscale Scores
Son's Age Group in Years

FQOL Disability Support
Support for the affected family member to receive home, school and social support as well as good relationships with service providers.

Mean Subscale Scores
Son's Age Group in Years

Preliminary Results N = 40:
Family Management Style

Parent Perception of Child's Life
Condition Management
Condition Management Effort
Family Life Difficulty
Parent Mutuality
View of Condition Impact

Parents of Sons with KS
FaMM Child Life
Parents Perception of Child’s Life in Spite of the Condition

Mean Subscale Score vs. Son’s Age Group in Years

FaMM Management Ability
The parents’ perceptions of overall manageability including knowing what needs to be done to take care of the condition and their ability to competently handle it Higher is Better

Mean Subscale Score vs. Son’s Age Group in Years

FaMM Management Effort
Addresses the time and work needed to manage the condition Lower is Better

Mean Subscale Score vs. Son’s Age Group in Years
FaMM Family Life Difficulty
Parent perceptions of the extent to which having a child with a chronic condition makes life difficult for the family
Lower is Better

Mean Subscale Score
Son's Age Group in Years

FaMM Parent Mutuality
Perception of support, shared views and satisfaction with how partners work together to manage the child's condition
Higher is Better

Mean Subscale Score
Son's Age Group in Years

FaMM View of Condition Impact
Parent perceptions of the seriousness of the condition and its implications for their child's and their family's future
Lower is Better

Mean Subscale Score
Son's Age Group in Years
Preliminary Results N = 40
Qualitative Themes

- Double Edge Sword
- In the Dark
- Internalized Worry
- Diagnosis Disclosure Dilemma
- Need for a Roadmap

What Is It Like to Have a Son with KS?

“It’s a Double-Edge Sword”  Mother of a 16 year old

Wonderful Precious Son

Serious and Chronic

Qualified Normal

“Soothing”
“Caring”
“Helper”
“Desire to Belong”

Speech and Language Problems
Learning Difficulties
Social Interaction Struggles
Increased Health Risks
Low self-esteem

Fear and Worry
Uncertain & Unpredictable

Frustration
Confusing

Lack of Networking
Lack of Support
Lack of Research
Lack of Information
Lack of Public Awareness
Lack of Training and Information for Health Care Providers
Lack of Training and Information for Educators

“We’re in the dark…..in the dark.
The dark, dark, dark……. ”

Mother of 14 year old son
What Parents Worry About

- Labeling
- Stigma
- Education
- Independence
- Social Relationships, including marriage
- Infertility

What Parents Think About Disclosing the Diagnosis of KS to Their Sons

"And so we were battling over this, but each time we talked about telling him, you know, you don't want to tell him in middle school because the kids are all mean and nasty, you know? Then you're in high school and they're dealing with all these other issues, so you don't want to disrupt that. But what ended up finally pushing us over the edge to tell him was an article that I read about them finding sperm in teenage boys, and that you could extract it and bank it for the future in the event that you could get sterile as an adult, and that the earlier you did that, the less likely the extraction procedure would be invasive. And so we finally decided to tell him so that we could take him and have him bank his sperm in case that he wanted to have children later. And that's why we told him."

Father of a 16 year old son

Tipping Point: Diagnosis Disclosure

"We debated numerous times as to whether to tell him when he was little, or tell him when he was 12, or when he was 15, or maybe not tell him at all and let him discover it when he was married and was going to be sterile."

Father of a 16 year old

"He was studying about Biology and he said he thought he had small testicles compared to other kids. He learned about KS in Biology Class."

Mother of a 14 year old

"He'd actually asked me. He was like, 'Mom, why do I have to keep going to all these doctor's appointments?' That's kind of how it started."

Mother of a 16 year old

"So we told him in bits and pieces, I would say, as much that seemed like enough."

Mother of a 15 year old
Observing the HOW to Disclose the KS Diagnosis

"I said to him, son, you know how you’ve always wondered why things don’t come easy to you when you’re learning and you say you can’t remember things as easily as other kids? Well, the good news is that you are not crazy and it’s real and it’s called Klinefelter Syndrome….and now we’re going to put our game faces on and we’re going to figure it out with you."

Mom of a 14 year old

What Parents and Families Need: A Roadmap

"When do we start Testosterone?" Mother of a 6 year old

"What part of this is KS and what part is my son?" Mother of an 8 year old

"How can we give our son his best shot at life?" Father of a 3 year old

“Who is supposed to be in charge of my son’s care?”

Preliminary Impressions of This Study

Explore how KS affects the child and the family system

- Parents are distressed and frustrated about how to help their sons throughout the life span
- Confusion leads to uncertainty and unpredictability for how to help their sons.
- The seriousness and chronic nature of KS symptoms contributes to the families’ inability to define their new “normal”.
- Parents feel lost (in the dark) about the ability to manage their son’s health.
Preliminary Impressions

Describe the vulnerabilities and needs of parents of boys with KS and how they manage the health of their sons

- Stress is higher: parents of younger & older children
- Family quality of life is decreased for emotional well-being and disability support: scores are strong for family interaction and parenting
- Family Management Measure shows interesting patterns and we are in the process of linking those scales with the qualitative findings.

Potential Implications for Practice

- Awareness and training of HCPs for higher index of suspicion.
- Need for more research on decision support for how to treat KS.
- Need to develop interventions to help with individual & family management of KS through the life course
- Parents need help and support for how to disclose all aspects of KS to their sons and to others

Significance

- Since one in 450-500 male births results in having a son with KS, there is a need to understand how parents care for the physical and psychosocial needs of their sons.
- This study adds to the understanding:
  - How families manage KS
  - How families choose to disclose the KS diagnosis
- Specific problem identification leads to the development of an intervention to help families in the future
Limitations

• Results are preliminary, so conclusions are not final
• Potential for biased sampling
• Self-report questionnaires
• Full qualitative rigor has not been assessed yet
  – Dependability, Credibility, Trustworthiness, Triangulation

Acknowledgments

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Much Appreciation To Parents who participated in this study

Questions Are Welcome

“All I can say is that we need to find ways to help more families because KS occurs in one in 500 boys. That's a lot of kids.”

Mother of a 16 year old son