Disclosing the Diagnosis

AXYS 2017 Families Conference

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Quick Review of Conditions

- Extra X and/or Y chromosomes
- Many Names
- Most common chromosomal aneuploidy conditions ~1/400 (in total), although under-recognized
- Males with extra X’s (XXY, XXXY, XXXY, etc.) also need testosterone and have fertility problems
- Every child and family is unique
Perspectives on Disclosure

• Parents often worry when facing disclosure
  – Common concerns
  – When, how, what to say/share
  – Family and environmental dynamics
  – Individual’s perspective
  – Do they really need to know?

• Professionals respect and support for patient’s autonomy
  – Self-advocacy
  – Transition to adulthood
  – Ongoing medical and psychological needs
Common Concerns

• Impact on child’s emotional health and fear that the knowledge will have a negative impact
  – Change in self-perception
    • If I tell him, he will know he’s different
    • If I tell her, she will think there is something wrong with her
    • If I tell her, she’ll have an excuse not to try hard
    • He’s already struggling, I don’t want to give him an excuse to give up

• Privacy
  – Discrimination and/or stigma
    • If I tell him, he may tell other people
    • If I tell her, she will tell everyone she knows
Potential Benefits to Disclosure

• Helping children adjust to the information
  – Before they need to make choices
• Shaping the conversation
  – Addressing concerns upfront
• Enhancing communication and trust within family
• Avoiding disclosure from a non-desirable source
• Respecting child’s autonomy
Decisions?

- Do you tell them?
- When to tell them?
- What to tell them?
- How to tell them?
- Who else to tell?

Recognize that every child is different with regard to his or her needs, comprehension, communication, and coping skills. You know your child best.
Research on Disclosure
Past 10 years
Issues Identified in Research

• Multiple influencers about sharing information
  – Family rules and boundaries
  – History and culture
  – Personal beliefs, attitudes & values
  – Child’s level of cognitive development and psychosocial maturity
  – Assessment of children’s readiness and interest

• Spectrum of strategies to sharing information
  – Openly, selectively, used clinic conversations to share, did not share
"How should I tell my child?" Disclosing the Diagnosis of Sex Chromosome Aneuploidies

Study aimed to explore the experiences of parents disclosing the diagnosis to their affected child as well as individuals with a diagnosis learning about their diagnosis

• 139 parents and 67 affected individuals answered survey questions regarding topics discussed, parent preparedness, resources accessed for preparation, parental concerns, and recommendations for disclosure
### Dennis study: Diagnosis Disclosure Recommendations

<table>
<thead>
<tr>
<th>Parents</th>
<th>Individuals</th>
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</thead>
<tbody>
<tr>
<td><strong>How to tell them</strong></td>
<td><strong>How to tell them</strong></td>
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<tr>
<td>- Be honest</td>
<td>- Be honest</td>
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<tr>
<td>- Gradually, over time</td>
<td>- Gradually, over time</td>
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<tr>
<td>- Inform yourself first</td>
<td>- Do not lie, omit, or mislead</td>
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<tr>
<td>- Be positive</td>
<td>- Be supportive &amp; open</td>
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<tr>
<td><strong>When to tell them</strong></td>
<td><strong>When to tell them</strong></td>
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<tr>
<td>- Early</td>
<td>- Early</td>
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<tr>
<td>- Before puberty</td>
<td>- Before puberty</td>
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<tr>
<td>- Child asks questions</td>
<td>- Child asks questions</td>
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<tr>
<td>- Based upon child’s maturity</td>
<td>- When treatment/HRT is needed</td>
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<tr>
<td><strong>What to tell them</strong></td>
<td><strong>What to tell them</strong></td>
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<tr>
<td>- Everyone has challenges</td>
<td>- It is not a disability, disease, or weird</td>
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<td>- You will help your child</td>
<td>- Encourage questions &amp; feelings</td>
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<tr>
<td>- Identify child’s strengths</td>
<td>- Treatments (HRT)</td>
</tr>
<tr>
<td>- Privacy issues</td>
<td>- Advancements/future possibilities</td>
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When to tell him/her?

• No perfect time
• Both parents and individuals surveyed support:
  – Early
  – Before puberty
  – When the child asks questions
• Based on child’s ability to understand and maturity
• Possible times to consider sharing information
  – At the time of diagnosis, or if prenatal, when going to therapy or doctor
  – When/If they start to struggle or before
  – When they start asking questions
Information sharing is a process that will continue throughout a child’s lifetime

Opportunities to continue information sharing

- Appointments with the doctor
- IEP meetings
- Meeting other kids with condition
- Learning about it together
- Talk about it when it’s on their mind
What to tell them?

Ways to prepare

• Become informed
  – Healthcare professionals, genetic counselors, support groups, other families, reading materials

• Thoughtful and consistent terminology

• Age and developmentally appropriate

• Focus on your child and how it relates to him/her
  – Wide spectrum of physical, cognitive, medical and psychological features

• Identify what “take aways” you want them to remember whenever it’s discussed
How to tell them?

**Keys to Success**

- **Get comfortable**
  - Parent emotions, okay to say “I don’t know”, awkward can be misinterpreted
- **Maintain a positive attitude**
  - Do not dwell on the potential negative
- **Truthful in the context of the child’s age and level of understanding**
- **Use simple and direct language**
- **Express support and reassure**
- **Encourage your child to ask questions**
- **Information sharing as a continuum**
  - Learning process that evolves through childhood and adolescence
Things to Try to Avoid….

- Everything at once
- Focus on genetics and chromosomes
  - “girls have two Xs and boys have an X & Y”…..XXY
  - “sex” “abnormality”
- Fertility terminology
  - Infertility = no sperm in semen, but normal sexual function otherwise
- Changing or terminology may get confusing
Resources

Diagnosis Disclosure Handouts

“Talking with your child about his/(her) diagnosis” handouts created from Dennis research for XXY, XXX, XXXY, and XYY available at conference and through the eXtraordinarY Kids Clinic

Talking with your child about his/(her) diagnosis of XXY (Klinefelter syndrome)

Talking with your child about his/(her) diagnosis of Triple X syndrome

Talking with your child about his/(her) diagnosis of XYY syndrome

Talking with your child about his/(her) diagnosis of XXYY syndrome

Why is talking about the diagnosis important?

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Resources: Diagnosis Disclosure Books

• Series of children’s books created by Arlie Colvin (genetic counseling graduation project at the University of Colorado) and available at conference or on Amazon.com or createspace.com (publisher)

![Book Covers](image)

• AXYS ([www.genetic.org](http://www.genetic.org))
  – booklets about XXY, XYY or XXX, written specifically for children
  – The book for parents, "Living with Klinefelter Syndrome, Trisomy X or 47,XYY"
Telling Others?

• School Disclosure:
  – Advocacy for resources
    • IEP qualification
  – Misperception as lazy
    • Self-esteem

• Family / Friends Disclosure:
  – Child’s right to privacy
  – Child will grow up and others will know already
  – Misinformed by internet / perception by others leading to possible stigma

• Doctor disclosure:
  – Providing medical care appropriately
  – Absence of parents

• Community and cultural considerations
Example: Age 4-7(ish)

• Do you know why you have to go to speech therapy?
• Your body is made a little differently than other kids, and your brain is made a little differently too
• It’s called…Triple X, XXY, Klinefelter’s
• When you have Triple X, you need some extra therapy to help with your speech
• It doesn’t mean you can’t learn, but sometimes you’ll have to work a little/lot harder than other kids because of triple X
• There are some good things about Triple X too
• If you ever have questions, you can always ask me about Triple X. If I don’t know the answer, we will find out together from the doctor.
Example: Age 7-12 (ish)

• Do you know why you have to go to tutoring?
• When you were little, we found out there was something different about you. That your genes and chromosomes are different from other kids.
• The genes and chromosomes are like the map or code of how your body is made. Genes tell your body what color to make your hair and your eyes, and how tall or short you will grow.
• Your genes and chromosomes are a little different from other kids, because you have something called XXY, Klinefelter’s, XYY, Triple X … That means you have an extra X chromosome or extra genes.
• When you have XXY, your brain is made a little differently and guys with XXY sometimes need extra tutoring.
• It doesn’t mean you can’t learn, but sometimes you’ll have to work a little/lot harder than other kids because of XXY.
• There are some good things about XXY too.
• If you ever have questions, you can always ask me about XXY. If I don’t know the answer, we will find out together.
Example: Age 12(ish)+

(Older than 12 or 13, really depends on the child)

- We want to talk to you about you
- When you were little, we found out there was something different about you.
- The doctors did a blood test and found out that your genes and chromosomes are different
- You have something called XYY, Triple X, XXY, Klinefelter’s, … That means you have an extra Y chromosome or extra genes.
- XYY is a very common condition, in fact 1/1000 people have it
- When you have XYY, there can be some differences in the way your brain develops, and that might explain some of the problems you’ve been having
- If you ever have questions, you can always ask me about XYY. If I don’t know the answer, we will find out together.
Questions for the Guys?

1) Do you remember when you learned about your diagnosis? If yes, what do you remember about when you learned?
2) Do you think the information you were told about the diagnosis was adequate and accurate?
3) What advice would you give to parents who are considering telling their child the diagnosis? When to tell them? How to tell them? What to tell them?
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- eXtraordinarY Kids and Families
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QUESTIONS?

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