Disclosing the Diagnosis

AXYS 2017 Families Conference

Susan Howell, MS, CGC, MBA
University of Colorado Denver, Dept. Pediatrics
Children's Hospital Colorado, Child Development Unit
June 2017

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, XXYY, 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

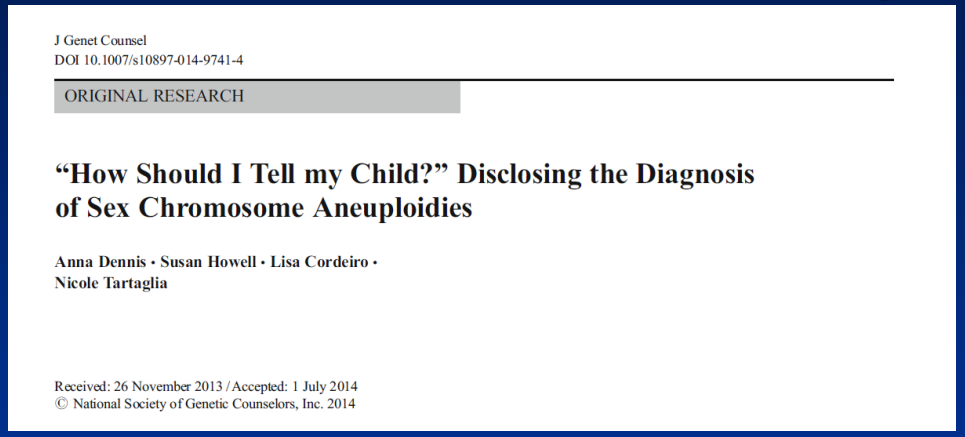
Decisions?

- 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



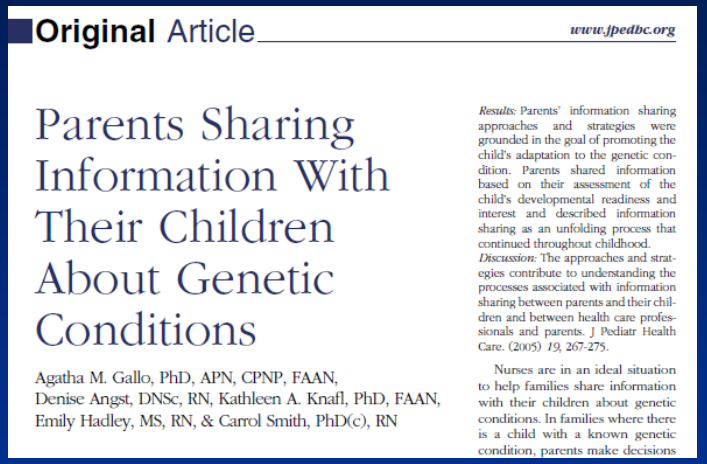
J Genet Counsel
DOI 10.1007/s10897-014-9741-4

ORIGINAL RESEARCH

"How Should I Tell my Child?" Disclosing the Diagnosis of Sex Chromosome Aneuploidies

Anna Dennis · Susan Howell · Lisa Cordeiro · Nicole Tartaglia

Received: 26 November 2013 / Accepted: 1 July 2014
© National Society of Genetic Counselors, Inc. 2014



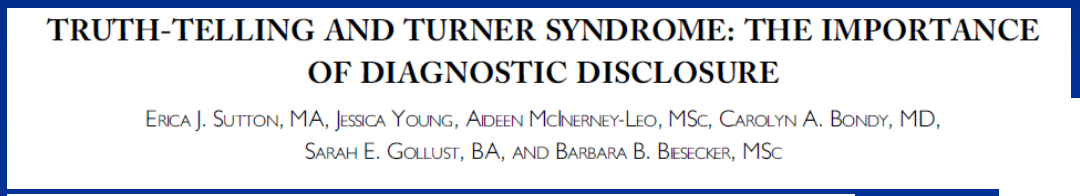
Original Article www.jpedbc.org

Parents Sharing Information With Their Children About Genetic Conditions

Agatha M. Gallo, PhD, APN, CPNP, FAAN, Denise Angst, DNSc, RN, Kathleen A. Knafl, PhD, FAAN, Emily Hadley, MS, RN, & Carrol Smith, PhD(c), RN

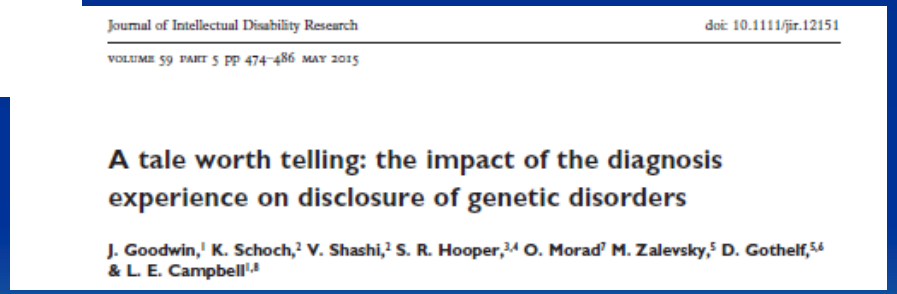
Results: Parents' information sharing approaches and strategies were grounded in the goal of promoting the child's adaptation to the genetic condition. Parents shared information based on their assessment of the child's developmental readiness and interest and described information sharing as an unfolding process that continued throughout childhood.
Discussion: The approaches and strategies contribute to understanding the processes associated with information sharing between parents and their children and between health care professionals and parents. J Pediatr Health Care. (2005) *19*, 267-275.

Nurses are in an ideal situation to help families share information with their children about genetic conditions. In families where there is a child with a known genetic condition, parents make decisions



TRUTH-TELLING AND TURNER SYNDROME: THE IMPORTANCE OF DIAGNOSTIC DISCLOSURE

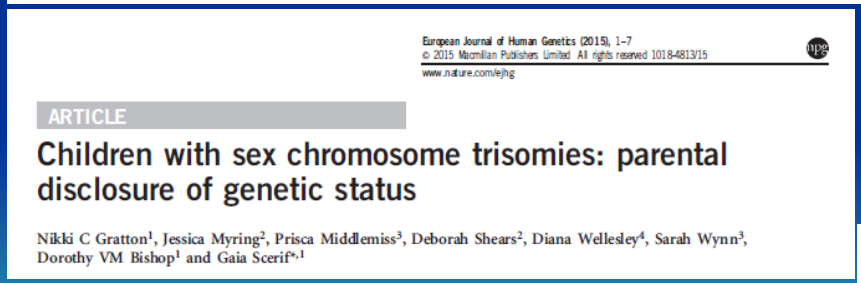
ERICA J. SUTTON, MA, JESSICA YOUNG, AIDEEN MCINERNEY-LEO, MSC, CAROLYN A. BONDY, MD, SARAH E. GOLLUST, BA, AND BARBARA B. BIESECKER, MSC



Journal of Intellectual Disability Research doi: 10.1111/jir.12151
VOLUME 59 PART 5 pp 474–486 MAY 2015

A tale worth telling: the impact of the diagnosis experience on disclosure of genetic disorders

J. Goodwin,[1] K. Schoch,[2] V. Shashi,[2] S. R. Hooper,[3,4] O. Morad[2] M. Zalevsky,[5] D. Gothelf,[5,6] & L. E. Campbell[1,8]

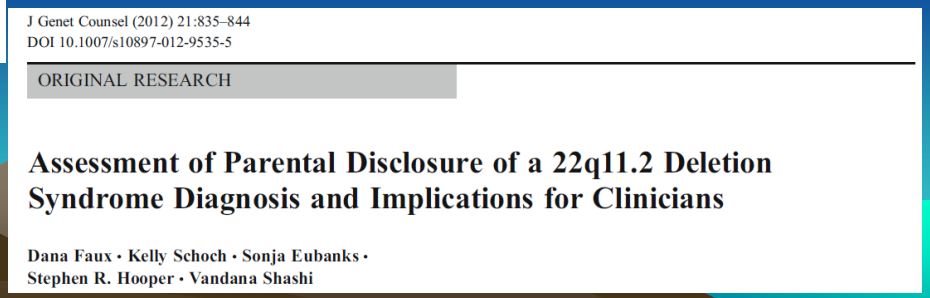


European Journal of Human Genetics (2015), 1–7
© 2015 Macmillan Publishers Limited All rights reserved 1018-4813/15
www.nature.com/ejhg

ARTICLE

Children with sex chromosome trisomies: parental disclosure of genetic status

Nikki C Gratton[1], Jessica Myring[2], Prisca Middlemiss[3], Deborah Shears[2], Diana Wellesley[4], Sarah Wynn[3], Dorothy VM Bishop[1] and Gaia Scerif*[,1]



J Genet Counsel (2012) 21:835–844
DOI 10.1007/s10897-012-9535-5

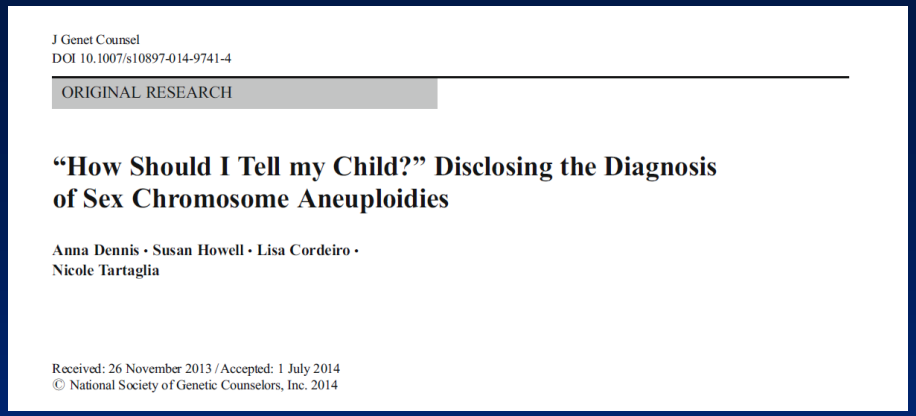
ORIGINAL RESEARCH

Assessment of Parental Disclosure of a 22q11.2 Deletion Syndrome Diagnosis and Implications for Clinicians

Dana Faux · Kelly Schoch · Sonja Eubanks · Stephen R. Hooper · Vandana Shashi

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



J Genet Counsel
DOI 10.1007/s10897-014-9741-4

ORIGINAL RESEARCH

"How Should I Tell my Child?" Disclosing the Diagnosis of Sex Chromosome Aneuploidies

Anna Dennis · Susan Howell · Lisa Cordeiro · Nicole Tartaglia

Received: 26 November 2013 / Accepted: 1 July 2014

"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

Parents

How to tell them

- Be honest
- Gradually, over time
- Inform yourself first
- Be positive

When to tell them

- Early
- Before puberty
- Child asks questions
- Based upon child's maturity

What to tell them

- Everyone has challenges
- You will help your child
- Identify child's strengths
- Privacy issues

Individuals

How to tell them

- Be honest
- Gradually, over time
- Do not lie, omit, or mislead
- Be supportive & open

When to tell them

- Early
- Before puberty
- Child asks questions
- When treatment/HRT is needed

What to tell them

- It is not a disability, disease, or weird
- Encourage questions & feelings
- Treatments (HRT)
- Advancements/future possibilities

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

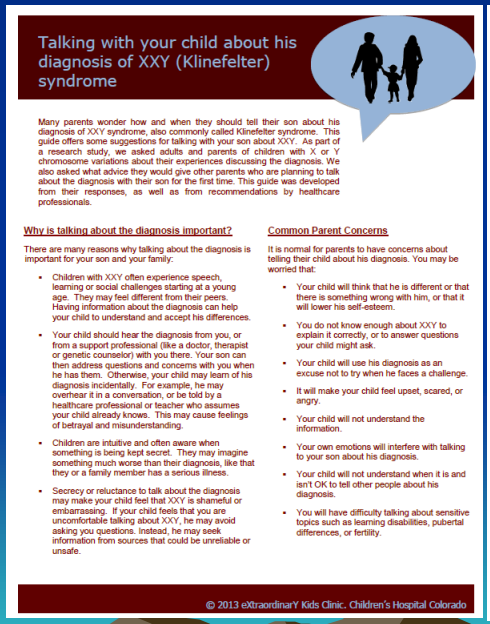
Things to Try to Avoid….

- Everything at once
- Focus on genetics and chromosomes
  - "girls have two Xs and boys have an X & Y"…..XXY ????
- Word choice
  - "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, XXYY, and XYY available at conference and through the *eXtraordinarY Kids Clinic*



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

Many parents wonder how and when they should tell their son about his diagnosis of XXY syndrome, also commonly called Klinefelter syndrome. This guide offers some suggestions for talking with your son about XXY. As part of a research study, we asked adults and parents of children with X or Y chromosome variations about their experiences discussing the diagnosis. We also asked what advice they would give other parents who are planning to talk about the diagnosis with their son for the first time. This guide was developed from their responses, as well as from recommendations by healthcare professionals.

Why is talking about the diagnosis important?

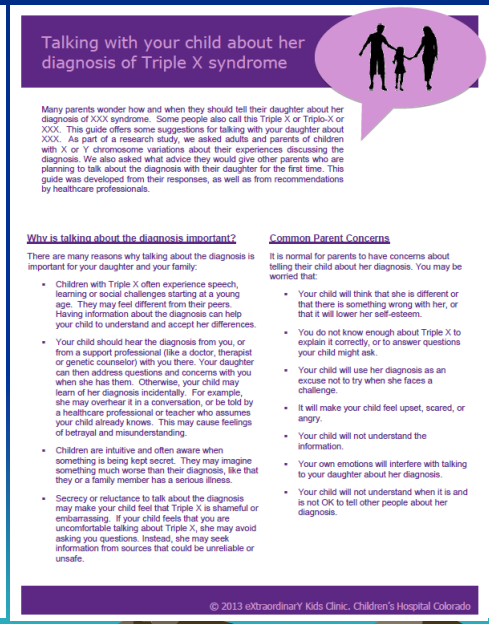
There are many reasons why talking about the diagnosis is important for your son and your family:

- Children with XXY often experience speech, learning or social challenges starting at a young age. They may feel different from their peers. Having information about the diagnosis can help your child to understand and accept his differences.
- Your child should hear the diagnosis from you, or from a support professional (like a doctor, therapist or genetic counselor) with you there. Your son can then address questions and concerns with you when he has them. Otherwise, your child may learn of his diagnosis incidentally. For example, he may overhear it in a conversation, or be told by a healthcare professional or teacher who assumes your child already knows. This may cause feelings of betrayal and misunderstanding.
- Children are intuitive and often aware when something is being kept secret. They may imagine something much worse than their diagnosis, like that they or a family member has a serious illness.
- Secrecy or reluctance to talk about the diagnosis may make your child feel that XXY is shameful or embarrassing. If your child feels that you are uncomfortable talking about XXY, he may avoid asking you questions. Instead, he may seek information from sources that could be unreliable or unsafe.

Common Parent Concerns

It is normal for parents to have concerns about telling their child about his diagnosis. You may be worried that:

- Your child will think that he is different or that there is something wrong with him, or that it will lower his self-esteem.
- You do not know enough about XXY to explain it correctly, or to answer questions your child might ask.
- Your child will use his diagnosis as an excuse not to try when he faces a challenge.
- It will make your child feel upset, scared, or angry.
- Your child will not understand the information.
- Your own emotions will interfere with talking to your son about his diagnosis.
- Your child will not understand when it is and isn't OK to tell other people about his diagnosis.
- You will have difficulty talking about sensitive topics such as learning disabilities, pubertal differences, or fertility.



Talking with your child about her diagnosis of Triple X syndrome

Many parents wonder how and when they should tell their daughter about her diagnosis of XXX syndrome. Some people also call this Triple X or Triple-X or XXX. This guide offers some suggestions for talking with your daughter about XXX. As part of a research study, we asked adults and parents of children with X or Y chromosome variations about their experiences discussing the diagnosis. We also asked what advice they would give other parents who are planning to talk about the diagnosis with their daughter for the first time. This guide was developed from their responses, as well as from recommendations by healthcare professionals.

Why is talking about the diagnosis important?

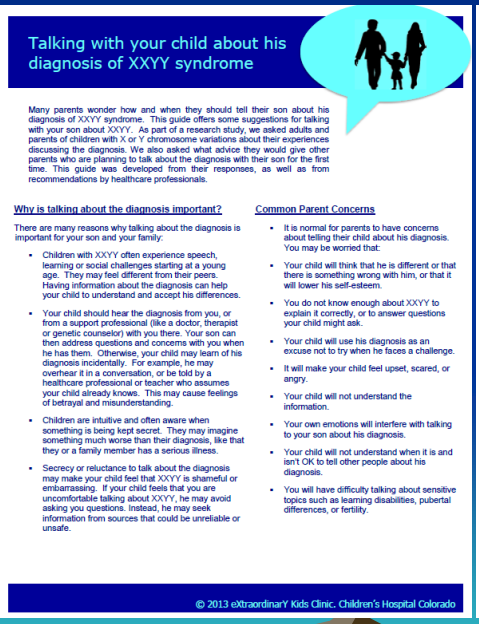
There are many reasons why talking about the diagnosis is important for your daughter and your family:

- Children with Triple X often experience speech, learning or social challenges starting at a young age. They may feel different from their peers. Having information about the diagnosis can help your child to understand and accept his differences.
- Your child should hear the diagnosis from you, or from a support professional (like a doctor, therapist or genetic counselor) with you there. Your daughter can then address questions and concerns with you when she has them. Otherwise, your child may learn of her diagnosis incidentally. For example, she may overhear it in a conversation, or be told by a healthcare professional or teacher who assumes your child already knows. This may cause feelings of betrayal and misunderstanding.
- Children are intuitive and often aware when something is being kept secret. They may imagine something much worse than their diagnosis, like that they or a family member has a serious illness.
- Secrecy or reluctance to talk about the diagnosis may make your child feel that Triple X is shameful or embarrassing. If your child feels that you are uncomfortable talking about Triple X, she may avoid asking you questions. Instead, she may seek information from sources that could be unreliable or unsafe.

Common Parent Concerns

It is normal for parents to have concerns about telling their child about her diagnosis. You may be worried that:

- Your child will think that she is different or that there is something wrong with her, or that it will lower her self-esteem.
- You do not know enough about Triple X to explain it correctly, or to answer questions your child might ask.
- Your child will use her diagnosis as an excuse not to try when she faces a challenge.
- It will make your child feel upset, scared, or angry.
- Your child will not understand the information.
- Your own emotions will interfere with talking to your daughter about her diagnosis.
- Your child will not understand when it is and is not OK to tell other people about her diagnosis.



Talking with your child about his diagnosis of XXYY syndrome

Many parents wonder how and when they should tell their son about his diagnosis of XXYY syndrome. This guide offers some suggestions for talking with your son about XXYY. As part of a research study, we asked adults and parents of children with X or Y chromosome variations about their experiences discussing the diagnosis. We also asked what advice they would give other parents who are planning to talk about the diagnosis with their son for the first time. This guide was developed from their responses, as well as from recommendations by healthcare professionals.

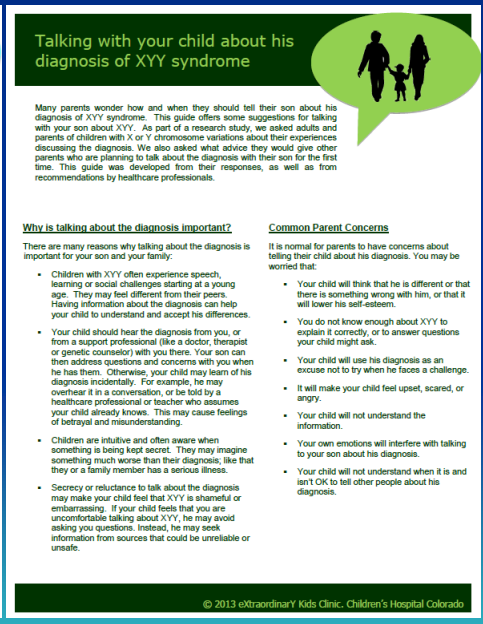
Why is talking about the diagnosis important?

There are many reasons why talking about the diagnosis is important for your son and your family:

- Children with XXYY often experience speech, learning or social challenges starting at a young age. They may feel different from their peers. Having information about the diagnosis can help your child to understand and accept his differences.
- Your child should hear the diagnosis from you, or from a support professional (like a doctor, therapist or genetic counselor) with you there. Your son can then address questions and concerns with you when he has them. Otherwise, your child may learn of his diagnosis incidentally. For example, he may overhear it in a conversation, or be told by a healthcare professional or teacher who assumes your child already knows. This may cause feelings of betrayal and misunderstanding.
- Children are intuitive and often aware when something is being kept secret. They may imagine something much worse than their diagnosis, like that they or a family member has a serious illness.
- Secrecy or reluctance to talk about the diagnosis may make your child feel that XXYY is shameful or embarrassing. If your child feels that you are uncomfortable talking about XXYY, he may avoid asking you questions. Instead, he may seek information from sources that could be unreliable or unsafe.

Common Parent Concerns

- It is normal for parents to have concerns about telling their child about his diagnosis. You may be worried that:
- Your child will think that he is different or that there is something wrong with him, or that it will lower his self-esteem.
- You do not know enough about XXYY to explain it correctly, or to answer questions your child might ask.
- Your child will use his diagnosis as an excuse not to try when he faces a challenge.
- It will make your child feel upset, scared, or angry.
- Your child will not understand the information.
- Your own emotions will interfere with talking to your son about his diagnosis.
- Your child will not understand when it is and isn't OK to tell other people about his diagnosis.
- You will have difficulty talking about sensitive topics such as learning disabilities, pubertal differences, or fertility.



Talking with your child about his diagnosis of XYY syndrome

Many parents wonder how and when they should tell their son about his diagnosis of XYY syndrome. This guide offers some suggestions for talking with your son about XYY. As part of a research study, we asked adults and parents of children with X or Y chromosome variations about their experiences discussing the diagnosis. We also asked what advice they would give other parents who are planning to talk about the diagnosis with their son for the first time. This guide was developed from their responses, as well as from recommendations by healthcare professionals.

Why is talking about the diagnosis important?

There are many reasons why talking about the diagnosis is important for your son and your family:

- Children with XYY often experience speech, learning or social challenges starting at a young age. They may feel different from their peers. Having information about the diagnosis can help your child to understand and accept his differences.
- Your child should hear the diagnosis from you, or from a support professional (like a doctor, therapist or genetic counselor) with you there. Your son can then address questions and concerns with you when he has them. Otherwise, your child may learn of his diagnosis incidentally. For example, he may overhear it in a conversation, or be told by a healthcare professional or teacher who assumes your child already knows. This may cause feelings of betrayal and misunderstanding.
- Children are intuitive and often aware when something is being kept secret. They may imagine something much worse than their diagnosis, like that they or a family member has a serious illness.
- Secrecy or reluctance to talk about the diagnosis may make your child feel that XYY is shameful or embarrassing. If your child feels that you are uncomfortable talking about XYY, he may avoid asking you questions. Instead, he may seek information from sources that could be unreliable or unsafe.

Common Parent Concerns

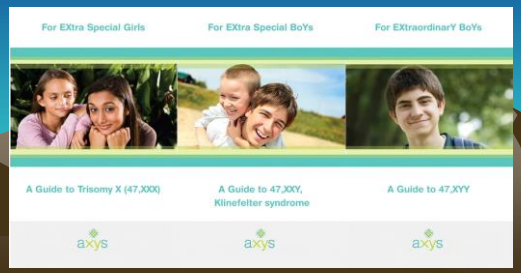
It is normal for parents to have concerns about telling their child about his diagnosis. You may be worried that:

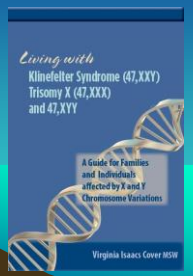
- Your child will think that he is different or that there is something wrong with him, or that it will lower his self-esteem.
- You do not know enough about XYY to explain it correctly, or to answer questions your child might ask.
- Your child will use his diagnosis as an excuse not to try when he faces a challenge.
- It will make your child feel upset, scared, or angry.
- Your child will not understand the information.
- Your own emotions will interfere with talking to your son about his diagnosis.
- Your child will not understand when it is and isn't OK to tell other people about his diagnosis.

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)

- AXYS (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?**

Acknowledgments

- eXtraordinarY Kids and Families
- AXYS
- XXYY Project
- *eXtraordinarY Kids Clinic* Team & Genetic Counseling students

QUESTIONS?

Susan Howell, MS, CGC, MBA

720-777-8361 or susan.howell@childrenscolorado.org