



How to Best Deliver a Diagnosis of an X/Y Variation to Parents AXYS 2021 Virtual Family Conference

Who should deliver the diagnosis?

The diagnosis should be delivered by a provider with knowledge in genetics and the X/Y variation. Providers should:

- Be prepared to provide detailed information about the condition.
- Refer parents to genetic counseling/genetics and other specialties as needed.
- Be aware that receiving an X/Y variation diagnosis is a formative event. The most upsetting provider encounters were ones where the provider seemed unaware of the condition, had little information to share with parents, and were perceived as uninterested in helping parents process the diagnosis.

What information should be provided at the time of diagnosis?

Parents should be provided with comprehensive information about the condition, including discussion of:

- The wide phenotypic spectrum of most X/Y variations.
- Information about their child's developmental trajectory.
- Information about medical, learning, behavioral, etc. impact at each life stage.
- What "real life" might look like with a child with an X/Y variation.
- Potential challenges and needs. These should be discussed and not minimized, but should not be the sole focus of the conversation.
- The distinction between X/Y variations and other aneuploidy conditions, such as Down syndrome.

Parents should be provided with written or online materials about the condition. These materials should:

- Be easy to understand and not contain technical jargon.
- Be up to date and reflective of the latest scholarship, including recent findings from children diagnosed as infants or prenatally.
- Not include findings from studies from the 1960's and 1970's conducted in prisons or other institutionalized populations.
- Contain non-medicalized pictures of children to show that X/Y variations do not typically cause noticeable facial or physical features.
- Be able to be explored on parents' own time after they have had time to process the diagnosis.

Other Parent Needs at the time of diagnosis:

- Connect parents to a fellow parent of a child, or an adult with the same condition.
- Connect parents to a support organization, such as AXYS.
- Many parents find the diagnosis to be overwhelming and may need to be referred to a counselor or support group for help with emotional processing.
- Parents may be concerned and need guidance about the implications of disclosing the diagnosis to family, friends, and community members.

Special Considerations of an X/Y Variation Diagnosed Prenatally:

- Recognize that parents may be anxious and upset at the return of any positive genetic result.
- Parents should be counseled *before* non-invasive prenatal screening (i.e., NIPT or NIPS) that sex determination may return an X/Y variation diagnosis.
- NIPT is not diagnostic. Results must be confirmed prenatally through chorionic villus sampling or amniocentesis or determined after delivery.
- Pregnancy continuation decisions should not be made based on NIPT results alone.
- Results and discussion of the condition should be presented *nondirectively* (i.e., neutrally) so as to not imply provider recommendation or undue pressure towards pregnancy termination.
- Parents with a prenatal result or diagnosis should be provided with the same materials and supports as those with a postnatal diagnosis.

Special Considerations of an X/Y Variation Diagnosed in Childhood:

- Diagnoses typically follow a lengthy diagnostic journey. Parents may be relieved to have a diagnosis, yet frustrated that their child was not diagnosed earlier.
- Parents need to be reassured that the diagnosis does not change who their child is as a person or their existing relationship with their child.
- Parents should be provided with concrete next steps regarding specialty referrals or a treatment plan. Preferably, their child's care should be coordinated to reduce parental burden.
- Parents may need assistance in accessing education supports, such as an IEP or 504 plan.
- Elementary and young adolescent children should not be present when the diagnosis is delivered. Parents prefer the diagnosis is delivered when they feel their child is developmentally ready and/or once they have processed the diagnosis themselves.

Special Considerations of an X/Y Variation Diagnosed in Adolescence/Young Adulthood:

- At this stage, individuals begin to develop greater autonomy over their medical care. Respect for this autonomy must be balanced with their ability to understand and process medical information.
- Confidential concerns, including sexual development and activity, may be discussed without parental presence. It may be in this context that an X/Y variation is suspected.
- Teens and young adults should be asked if they want a parent or other support person present when the diagnosis is delivered.
- The diagnosis should be delivered with sensitivity towards their self-image and self-esteem, especially as it relates to discussions about infertility and sexual development.
- Receiving the diagnosis may be an identity forming event. This may bring clarity to prior learning or social struggles, but may also reinforce feelings of difference from peers.
- Offhand comments about infertility, genital size, etc. should not be made. These comments are distressing and remembered years, even decades, post-diagnosis.
- The diagnosis may have direct implications on their romantic relationships and future education and employment plans. Teens and young adults may need referral for counseling to emotionally process the diagnosis and its resulting impact on their life and plans.