



Dear Parents:

I am writing this letter to parents who have received a prenatal diagnosis of *sex chromosome aneuploidy*, one or more extra X and/or Y chromosomes. First, I want to tell you that I have been in your shoes, having learned at 18 weeks pregnancy that I was carrying a boy with an extra X chromosome, a condition known as Klinefelter syndrome, or 47,XXY. It is one of a group of genetic conditions that also includes 47,XYY (an extra Y chromosome), and 47,XXX or Trisomy X. Sometimes these conditions may involve 48 or 49 chromosomes.



I also know that this diagnosis is an indescribable shock, and that it causes great apprehension for your child's future. You may even have been told that you need to make a decision, quickly, about whether or not to continue with this pregnancy.

Let me tell you that my son is now 25. He lives independently in an apartment, he drives, he can work, and he has also had girlfriends and hopes to get married in the future. I can tell you, however, that he had substantial delay in developing speech as a young child, along with low muscle tone and poor coordination, which is fairly typical of many children with X and Y chromosome variations. He responded well to early intervention, and largely caught up with his peers. Although he has an IQ in the normal range, he has significant learning disabilities and received special education services during school. He did, however, play violin and received a number of musical awards. He is kind, and compassionate, and has a great sense of humor. He plays golf, and badminton, and although he dropped out of college in his second year because of his learning difficulties, he has determined that he now has the maturity to return, obtain the extra help that he needs, and complete a degree.

If I were to characterize John's functioning on the "spectrum" of sex chromosome aneuploidy (SCA), I would say that he is squarely in the middle of the bell curve. There is enormous variation in how these children and adults present. In perhaps one-third of kids, there are no learning disabilities and few other complications or signs of the syndrome. In the other two thirds, there can be mild to moderate learning disabilities, but intellectual disability (mental retardation) is rare. Most adults with SCA establish themselves in careers and live independently. Many have satisfying relationships and become parents. Most adults will tell you that while it took them a bit longer to become independent than their peers, perhaps earning a college degree and settling into a career, they have had good lives, even though they may have had some struggles in childhood and adolescence.

I can't tell you specifically what to expect. I can tell you that parenthood is always both a joy and a challenge and that the mild or moderate disabilities that may be present in children with X and Y chromosome variations are manageable. These conditions are not devastating, either to the child or to his or her family. When you know about the condition before birth, you have a great advantage. Without prenatal diagnosis, these conditions often cause such subtle symptoms that diagnosis and the therapies that can minimize developmental difficulties are delayed,



sometimes by years. Children with SCA often are remarkably responsive to early intervention therapies, and to special education programs that target their difficulties with reading and with verbal and written expression. When you are aware of the specific deficits that SCA may cause, you can more quickly qualify your child to receive the services that he or she needs. When parents are committed to learning about SCA and addressing developmental and learning difficulties promptly with effective therapies, potential problems can be minimized.

AXYS is committed to assisting families in helping their children live successfully with X and Y chromosome variations. The organization also provides substantial support and education for adults with SCA. I am a member of the Board of Directors of this organization. We have created a collection of materials to help expectant parents to understand SCA, and the particular trisomy condition that affects their future baby. They can prepare for the baby's birth in a knowledgeable manner, and can consult with their pediatrician about any questions that they may have. The organization can put you into contact with one or more parents of children who have been diagnosed with your baby's genetic condition.

AXYS's website features a number of resources that summarize some of the childhood manifestations of SCA, and provide you with a listing of resources available to you through AXYS to learn more about what you may expect in childhood with children with X and Y chromosome variations. I want to emphasize that these conditions are surprisingly common, affecting 1 in 500 children. You do not need to feel that you are coping with this news alone; our community is there for you to provide support and education that will help you promote your child's development and well-being.

Yours sincerely,

Virginia Isaacs, MSW