

XXY Update – Fall, 2017

In case you have not had a chance to visit the AXYS website lately, several new research papers have been added in the library that you may want to review and also share with your health care providers and others. Two of these articles involve research by Dr. Claus Gravholt and his team at the Department of Endocrinology and Internal Medicine at Aarhus University Hospital in Denmark. This multidisciplinary clinic specializes in treating XXY individuals over the age of 15 and is one of the few clinics in the world that has that distinction. They see more than 400 adult patients with KS and have published a number of excellent research papers in the past based on their experiences.

Their latest publications focus on cognitive performance and quality of life issues associated with KS adults and they bring out many important points, including:

1. Adult issues related to neurocognitive challenges are complex, difficult and do not receive adequate research attention or effective care.
2. There are very few places in the world that provide expert multidisciplinary treatment for XXY adults
3. Most KS adults have difficulty finding skilled, effective health care from health professionals that are aware of current research in KS.

We know it can be difficult when reading these recent research papers by Dr. Gravholt and his associates because they can appear to paint a discouraging picture about KS adults. However, it is important to remember several key points when reviewing their results:

- The findings are real and provide indicators of the kinds of challenges KS individuals can face as they grow older. We do not want to pretend these challenges do not exist and we need to have adequate and effective treatment options to deal with them.
- Knowing what issues may be prevalent for KS adults can be extremely helpful in designing effective early intervention programs that can help decrease the severity of potential problems as people get older.
- There are no simple answers, i.e., testosterone is not a magic solution for all problems. It can be very important as part of an overall treatment plan, but it will not resolve executive function or cognitive issues or challenges with social engagement. Those things require different interventions.
- Saying someone has KS always implies a constellation of challenges ranging from significant to mild. Using the term KS to describe someone does not imply anything other than the genetic signature of that individual. Every person is unique and may benefit from more or less care, which again is why it is so important to have specialized centers and physicians that can create best practice standards that can be used by other health professionals. When someone needs help, they should be able to get the most effective help possible wherever they may live.

These are all very real issues that present significant challenges for the adult KS population. They have been documented and written about many times in the past but despite this awareness, there are still very few specialty clinics available for adults. There are many reasons for this including lack of understanding, few models of excellence, organizational inertia, cost considerations, funding and the scope of services needed. While recognizing these multiple challenges, AXYS remains determined in finding solutions.

Thanks to early pioneers like Dr. Nicole Tartaglia and her team at the Denver Children's Hospital, we have seen significant progress in the development of specialty clinics for kids with SCA conditions across the U.S. Now we need to recognize the critical need for adult clinics where comprehensive evaluations can be completed and multi-disciplinary treatment plans designed for adults struggling with different medical, neuropsychological and independent living problems. It is imperative that we find ways to effectively address these issues to provide adequate health care and support for KS adults. Specialty adult clinics that can serve as centers of excellence are the best way to begin.

The plain truth is that a majority of health professionals do not understand KS. They are not keeping up with research and we do not have best practices defined or widely published, especially for adults. The encouraging news is that we have pretty good ideas on how to improve this, but it will take time, money, support and involvement from many people. We need all of you to help us with this and there are many ways you can provide support: read and learn more.....share information with all people you come into contact with, especially doctors and teachers.....provide financial support when you can.....look at ways to volunteer your time with AXYS.....confront marginal care or misunderstanding whenever you see it, and as always, let us know how we can help.

It is through AXYS, and the dedicated professionals and families and individuals we work with, that positive change happens. We have made great progress in the past 5 years and I firmly believe we can achieve even more. Many thanks to all of you for your past, present and future support and interest. Together we will continue to make things better.



Gary Glissman
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