

Short impression XY Workshop Leiden, The Netherlands, September 2022

From 12-14 September 2022, the third Workshop on sex chromosome trisomies took place. It was all about Klinefelter Syndrome, Trisomy X, and XYY. Sometimes Turner Syndrome (45,X) was mentioned. This was the first time also people with these diagnoses or parents whose child has one of these, took part. I attended the Workshop because of my connection with the Dutch Contact group for Triple X Syndrome.

The topic on Monday morning was how the changes in chromosomes lead to changes in the body: the brain and in XXY also the reproductive organs. The stories went in dept on DNA, genes and hormones. What struck me is the overlap in symptoms between the three trisomies. Not only in this morning but also in other presentations.

In the afternoon ageing with XXY was discussed. A Dutch endocrinologist told us that there is much we do not know yet. However, clinicians see many physical complaints and also lots of early retirement. One of the participants told us, at my question, it is also true for XYY and XXX. The Dutch endocrinologist would like to do more research on ageing and I told him that is what we really need.

On Tuesday morning a mother and an adult told us how important patient advocacy groups are. I made a short video on the importance of Facebook groups and that was also presented. Then Sophie van Rijn, a Dutch scientist, presented the research of her group. She stated it is important to look beyond behaviour. Also the processes in the brain and the thinking is important. About 15-30% of the children with an extra X or Y also have an autism spectrum disorder (ASD) and 30-35% has ADD or ADHD (attention deficit disorder with or without hyperactivity). Even 70-80% has a problem related to developmental language disorder, mainly in the pragmatic aspects. When language is a problem, it is logical that behaviour is too! And they grow into deficit when they get older.

On Wednesday morning a new guideline for XXY was presented. This guideline could be a good example for the development of a guideline for XXX. For Turner Syndrome guidelines already exist but not for XXX or XYY. We all know that finding help when you have symptoms can be difficult. A guideline would really help us.

During the Workshop, fundamental research was an important topic. How DNA, cells and hormones work exactly. What I missed is research focussing on our daily life and how to make our quality of life better. For children there is more information and several studies are done at the moment. I have asked to pay special attention to adults with XXX. The psychological problems many of us have, how many of us face them and what is the best way to help us? Many adults have pain, abdominal but also musculoskeletal. We know that people with Turner Syndrome have issues that are usually seen in older people. It might be true for XXX too.

There is so much we do not know yet and that is not easily solved. The scientists and doctors appreciated our attendance. In 3 years, the next meeting will be in Italy and I hope to travel there and hear the new information.

September 2022, Jessica Langenhoff

