



DIESC

Development of Individuals with an Extra Sex Chromosome

Investigators: Victoria Leggett, Dr Gaia Scerif, Prof Dorothy Bishop, Prof Kate Nation, Prof Patricia Jacobs, Dr Angela Barnicoat, Dr Patricia Boyd, Dr Alan Fryer, Dr Katherine Lachlan, Dr Kay Metcalfe, Prisca Middlemiss, Dr Debbie Shears, Dr Sarah Smithson and Dr Diana Wellesley.

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The Study

This study aims to provide more complete information for parents whose children are given a pre-natal diagnosis of a sex chromosome trisomy (SCT). We focused mainly on pre-natally diagnosed cases because information is particularly limited for them. Our study also included a small number of cases who were diagnosed in childhood, but in some of these cases chromosomes are investigated only because the child is experiencing difficulties. We therefore keep their results separate, as we might otherwise exaggerate the rate of problems associated with a SCT. Our study included information about 20 boys with XXY, 57 boys with XYY, 55 girls with XXX and their brothers and sisters without a SCT (40 boys and 26 girls respectively). We used both standardised questionnaires and semi-structured interviews with parents to consider school performance, attention, social skills and behaviours. We focused both on both strengths and weaknesses, taking into account variation in development from child to child, as well as the average result. We hope that this information will be useful for both parents and professionals such as GPs and teachers who may have little experience of children with SCTs.

Thank you very much to all parents who have taken part in this study, and been so generous with their time. It has been fantastic to visit you and find out about your children, and I hope that our findings are of interest to you.

Victoria

Results – What did we do with what you told us?

We have shown the results in graphs that indicate the range of scores as well as the average for each group, to give an idea of variability from child to child. It is important that we draw attention to areas where children may need help, but we want to stress that not all children with a SCT are the same. In some cases, we have shown how scores relate to established cutoffs used by clinicians, so we can tell how many children have problems that are likely to give clinical concern. A quick user guide for the presentation methods we used is provided on page 2.

Terms

When these terms appear in the newsletter, they are highlighted in italics to allow you to refer back to their meaning in this table if you want to.

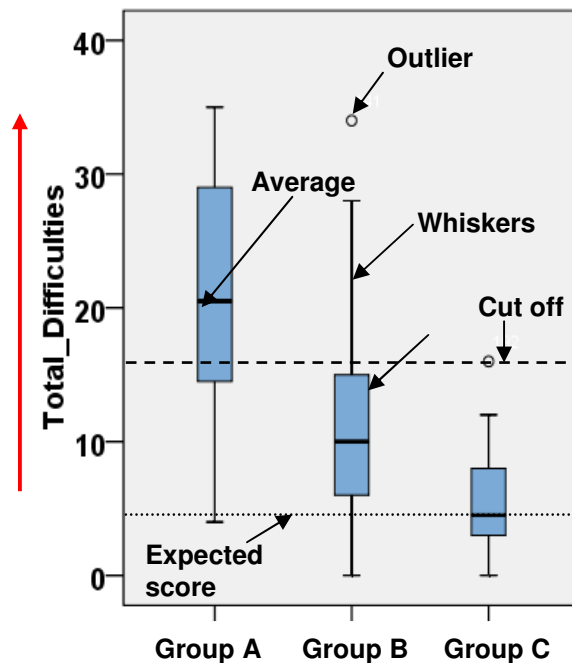
- *Adaptive Behaviour* – Behaviours which indicate how well an individual might be able to cope in everyday life, in terms of listening, speech, hygiene, play, interacting, and understanding of money, time and danger.
- *Maladaptive Behaviour* – A behaviour which might be viewed as challenging or an individual may need help addressing.
- *Prosocial Behaviour* – Trying to make friends and caring for younger children.
- *Communication* – This has been broken down into: speech (clarity), syntax (sentence structure), semantic (understanding of words), coherence (putting sentences together), inappropriate initiation (talking too much), stereotyped language (reusing phrases or structures repetitively), use of context (to understand different word meanings), non verbal (gestures and body language), social interaction (getting along with others) and interests (what they like to talk about).
- *Hyperactivity and Inattention* – These are measured by compiling a range of subscales; oppositional (difficult behaviours), inattentive (difficulty concentrating), hyperactive (overly energetic), anxious/shy (worries), perfectionism (will redo something many times to get it just right), social problems, psychosomatic problems (illnesses which might be linked to worry), restlessness (inability to sit still), emotional lability (seeming to have unstable moods, and swinging from happy to sad).
- *Gross Motor Skills* – This considers attributes like jumping, skipping, hopping, throwing and catching.
- *Fine Motor Skills* – This considers skills such as writing, art work, using a knife and fork and using scissors.

Graphs

We have presented the findings as a series of graphs, primarily in the form of what are called “box and whiskers plots”. In all graphs, the direction of the red arrow denotes increasing difficulties. Half the data for each group fall in the range shown by the blue box, with the line across the box representing the average. The remainder of the data is represented by the whiskers. Any data points which do not fit easily within the plot (known as “outliers”) are plotted separately by a dot as shown. The dashed line across the graph denotes a cut off (for example, one used by clinicians), with any values beyond this point as raising concerns. The dotted line denotes the expected average score for children of this age.

Data regarding difficult behaviours are presented as percentage of children falling in one of three categories: average levels (indicated by “a”), elevated levels (“e”), and levels of clinical significance (“cs”). Individuals who have behavioural difficulties of clinical significance may require extra help in managing their behaviour.

For all groups of children and adolescents, the graphs are organised to first present school performance, behavioural difficulties and adaptive behaviours, followed by overall areas of difficulty and strength, and then a more detailed look at communication, social skills and motor skills. Finally we present data concerning hyperactivity and inattention, before considering differences across younger and older children and overall conclusions.



Thoughts on your feedback during the study visits and on study days

Telling your child

A concern for many parents was how and when to tell their child about their extra sex chromosome. There was a wide range of different approaches and these varied greatly from child to child. Some parents had found that the diagnosis was unintentionally mentioned by their GP when they visited, so they suggested informing the GP if their child was unaware of their extra chromosome.

In many cases, if their child was having difficulties, families referred to differences from a relatively young age as 'their special thing'. As the child grew up they gradually gave them more detail. Parents whose children were having fewer difficulties favoured telling the child around 10, when they were considered mature enough to start to understand, but before they had reached teenage years. One family described telling their daughter 'we wished so hard for a little girl that you have an extra girl chromosome!'.

All families said it was important to stress the positives, such as beautiful long legs for girls with XXX, and great memory or manual skills, before explaining that it might be why they found school a little bit harder than others. However all parents said it was important to stress to their children that a diagnosis did not mean they could not do anything, just that it might be harder, as a general concern was that they might give up trying. For families with a son with XXY, the majority of families told their son when it became necessary to check whether they needed testosterone treatment, and in the majority of cases their GP referred them back to a genetic counsellor who was able to give their son more detail on XXY.

A word of warning

It is important to bear in mind that an understanding of the kinds of difficulties children might face has increased dramatically over the last decades, especially with regard to communication and socialisation difficulties. This understanding has led to increased provision for individuals who need assistance and earlier recognition of any difficulties. These factors may lead to some clear differences between the youngest and oldest individuals in our sample, which may purely reflect different attitudes of professionals towards the sex chromosome trisomies. Despite lower awareness of specific difficulties for individuals with SCTs, in the past it may have been easier to obtain assistance through a statement of educational needs, but obtaining this has become very difficult more recently. By taking part in this study you have also made a positive contribution to future children as better understanding can lead to better provision where difficulties arise.

Feedback from Unique XXX and XYY study days

When analysing the results from this study, in addition to our study hypotheses, we have considered the concerns or strengths that parents identified in the show of hands on the study days organised by Unique. These were attributes noticed by many parents, who were questioning whether they might be a result of having an extra sex chromosome, or simply a characteristic of their own child. When these characteristics also appear in our data, *we have highlighted them in blue and in italics*. We hope that this information might begin to bridge the gap between the scientific and medical findings and the reality of having a child with an extra sex chromosome for you, the families.

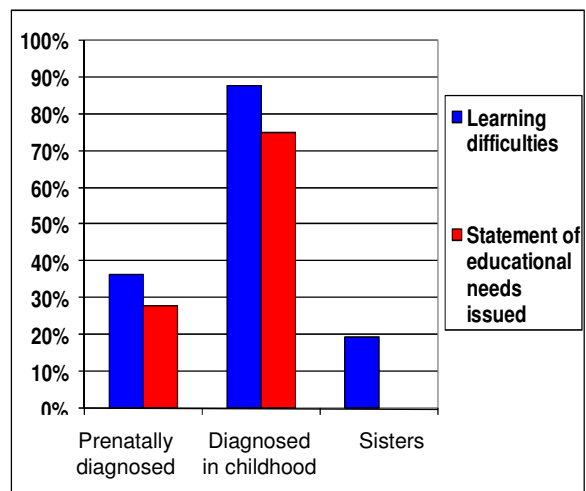
Girls with XXX

Overview

- *Girls were described as very caring, especially with animals and younger children.* Many were described as artistic, with a great sense of fun.
- A quarter of the girls were described as good at sports, whilst some others found *coordination challenging*.
- Biggest concerns for families were difficulties with learning, in addition to difficulties making or keeping friends and some degree of emotional immaturity. However there is no evidence of additional diagnoses in these girls.
- Overall the girls were described as very loving and kind.

School Progress

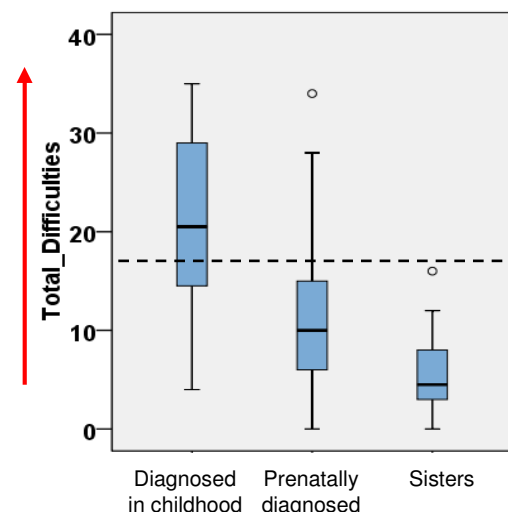
Almost 70% of girls with XXX seem to do fine at school, with just over 30% of prenatally diagnosed girls reported to have learning difficulties. 28% of prenatally diagnosed girls had a statement of special needs, allowing them access to additional help at school, but very few girls were in specialist units or schools. This suggests that whilst some of these girls do show difficulties with learning these are relatively mild. The majority of girls were given a couple of hours specialist tuition as part of the school week, with literacy being the most common focus. Almost 90% of postnatally diagnosed girls show some difficulties with learning, with 75% having a statement of educational needs. The majority of girls were reported to enjoy school and many particularly enjoyed maths and spelling.



Strengths and weaknesses

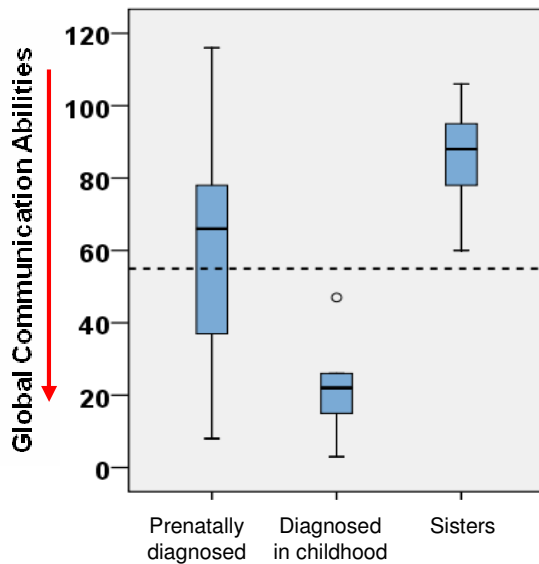
Prenatally diagnosed girls with XXX show relatively few overall difficulties, with no significant differences relative to their sisters in emotional understanding, peer problems, and prosocial behaviours. They do show significantly more difficulties with conduct and hyperactivity, but only 15% score in concerning levels. In contrast the majority of postnatally diagnosed girls with XXX show difficulties in emotional understanding, peer relationships, conduct and hyperactivity, *with relatively intact prosocial behaviours*.

We will now consider possible outcomes in more detail.



For an explanation of graph see pg 2

Girls with XXX



For an explanation of graph see pg 2

Communication

Prenatally diagnosed girls with XXX show communication abilities within the expected range for their age, but these are *poorer than their sisters with regard to speech, syntax, semantics and use of context*. Despite this they show similar levels of *inappropriate initiation, stereotyped, non verbal communication, and interests* relative to their sisters. This was reinforced by parents who reported that their daughters were chatty, but had a slightly more limited vocabulary than their sisters. In contrast the postnatally diagnosed girls have greater difficulties with communication, perhaps leading to diagnosis.

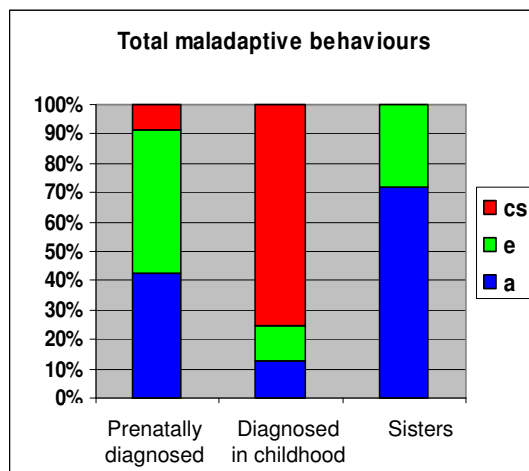
Behaviours

Adaptive Behaviours

Prenatally diagnosed girls with XXX find many everyday behaviours harder than their sisters, but the majority are performing within the range expected for their age. Particular strengths are in understanding of money, time, and play and leisure skills. This suggests that they are happy to play a wide range of games and understand sharing, taking turns and scoring games. In addition written communication is a particular strength, with girls showing relatively good reading and writing skills. Postnatally diagnosed girls with XXX find many *adaptive behaviours* more challenging than their sisters, with personal hygiene, listening, cleanliness and cooking abilities of particular concern. Many parents commented on these areas, which were of particular importance for older girls, but reported that these girls show a relatively good understanding of other everyday skills such as time and money.

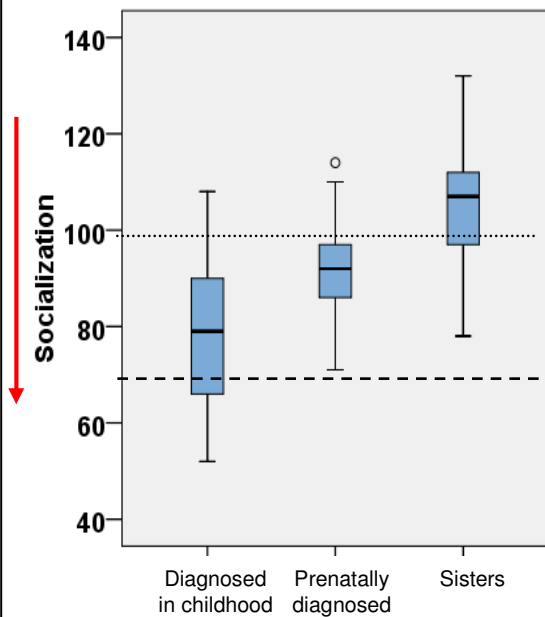
Maladaptive Behaviours

90% of prenatally diagnosed girls with XXX showed normal (indicated by “a”) or slightly elevated (“e”) levels of difficult behaviour, with only 10% displaying concerning (“cs”) levels of difficulties behaviour. The girls showed similar levels of internalising behaviours to their sisters, ranging from anxiety, to crying easily, and avoiding others. However girls with XXX showed an increased number of problematic behaviours relative to their sisters, including temper tantrums, stubbornness, and *being easily led*. This finding was reinforced by parental reports of increased numbers of temper tantrums, often due to frustration. The pattern is very similar for girls diagnosed in childhood, although almost 75% of these girls showed concerning levels of behaviour.



Girls with XXX

Socialisation

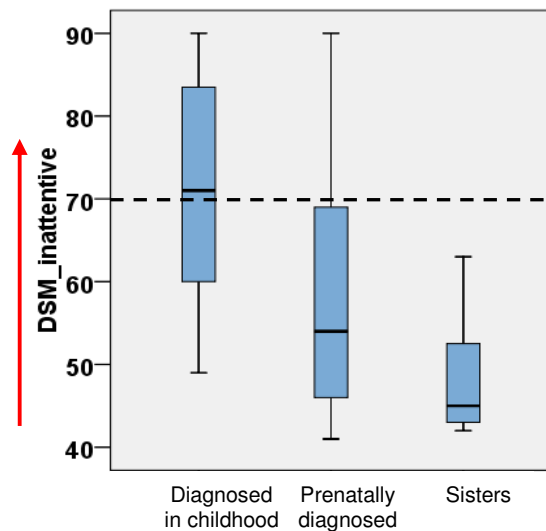


For an explanation of graph see pg 2

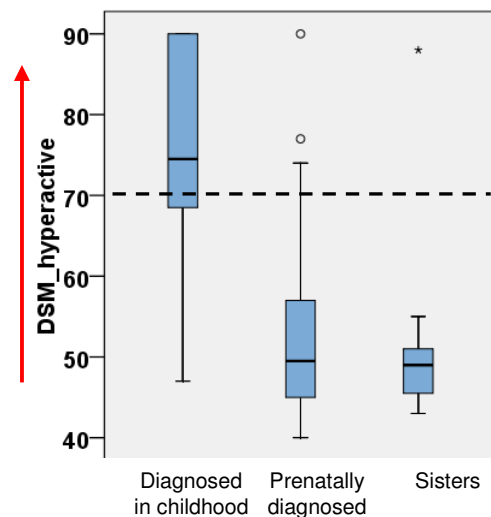
All of the prenatally diagnosed girls have social skills within the range usually expected for their age. However, they have been observed to find social skills more challenging than their sisters. The girls show the expected levels of play and leisure behaviour, and only a slight decrease in their coping skills, e.g., awareness of others, manners, coping with change and awareness of danger. In addition they show relatively good interpersonal skills, understanding of emotions and knowledge of the dynamics of friendship; however some parents commented that their daughter may tend to smother friends, not understanding that there may be times when the friend does not want to interact. A small proportion of the postnatally diagnosed girls score below the normal range, displaying greater difficulties with play and interpersonal skills. The majority of girls in both groups are reported to have close friends.

Hyperactivity and Inattention

Prenatally diagnosed girls with XXX show similar levels of hyperactivity to their sisters, with no evidence for increased levels of fidgeting, restlessness, or impulsivity. However these girls do show increased levels of inattention, although it is not a cause for concern in the majority of individuals. This suggests that these girls are inclined to have difficulties concentrating but are unlikely to be excessively energetic.

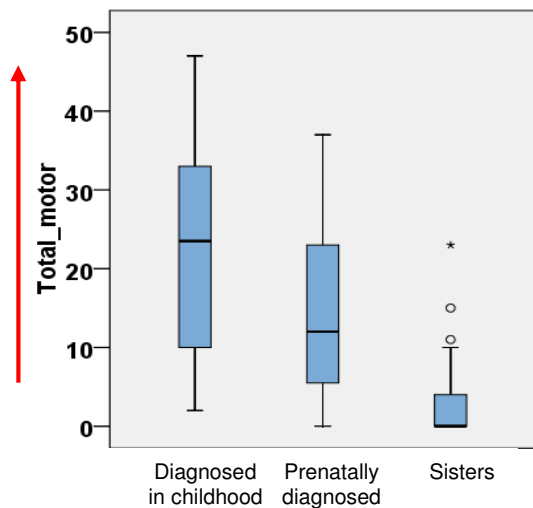


For an explanation of graphs see pg 2



When considered in further detail, the girls display slightly increased *emotional lability*, such as crying easily, but showed *no evidence of anxiety, perfectionism or oppositional behaviour*. This is in contrast to postnatally diagnosed girls who displayed increased levels of inattention, hyperactivity and anxiety, alongside normal levels of perfectionism. Girls are generally described as calm and relaxed.

Girls with XXX



For an explanation of graph see pg 2

Motor Skills

Both groups of girls with XXX find *fine and gross motor control* more difficult than their sisters, with both groups finding fine motor control particularly challenging. This leads to slightly messy handwriting, and some girls appearing uncoordinated when using cutlery. However, despite this *a large proportion of the girls were good at art* with parents describing the girls to have great imaginations. Furthermore whilst many of the girls found gross motor skills challenging parents reported that if they persevered they soon built up muscle strength with girls enjoying swimming, dancing and horse riding.

Age trends : Are there differences between younger and older children?

Many of you asked about the possible future for your child and whether you should expect different areas of strengths and weaknesses over time. Please note that this is extremely difficult to establish without following individual children over time: when we compare younger and older children who have only been assessed once, any differences may depend on the two groups being different by chance or reasons other than age. Keeping this in mind, one way in which we can look at whether our data suggest age differences is by considering how our standardised measures (i.e., the comparison of data from individual girls with XXX with large groups of typically developing children of the same age) change with age. There is some evidence of emotional immaturity, which becomes more apparent as the girls grow up. This is particularly evident in their socialisation skills, which develop more slowly than expected, with girls finding socialisation particularly difficult around the age of 11. As girls with XXX mature, there is evidence that their socialisation skills begin to improve until they reach the level which would be expected for their age. These difficulties with socialisation are often coupled with an increase in challenging behaviours. However, there is some evidence that after the age of 13 these behaviours begin to reduce and have reached a similar level to those of their sisters by 16. By the age of 16 there was no evidence of clinically significant behaviours, but some girls continued to show elevated levels of *maladaptive behaviours*. There is also some evidence that communication and socialisation skills may follow a similar pattern, developing well when the girls are young, then plateauing aged 10 to 13, before beginning to improve further aged 13 to 16. These data were reinforced by observations from parents that their daughter was becoming happier at school or college, making more friends and showing fewer challenging or concerning behaviours.

Conclusions

Overall these girls tend to be happy, relaxed, and caring, displaying few behavioural problems. Many were doing just as well as their sisters in everyday behaviours such as play, domestic skills, and understanding time and money. However, some had difficulty concentrating at school, and many found social skills increasingly difficult as they matured. Parents reported age 11 as a particularly challenging age as their daughter seemed emotionally immature for her age, and was still happy to undertake make believe play which their peers might find childish. However the majority of girls appear to catch up with peers, finding it easier to make friends again by the time they were 14 or 15.

What Next?

Helpful Publications

Available through UNIQUE:

- XYY Syndrome
- XXX Syndrome

The UNIQUE documents will be available within the next few months.

We hope to publish as many of our study findings as possible in journals read by clinicians, educational psychologists, researchers and other professionals to increase awareness about sex chromosome trisomies. As articles are published, we will upload them on the study website (address below), but unfortunately academic publications may take as long as a couple of years to be published, so please be patient. Alternatively, let us know and we will endeavour to send you the articles in which you are particularly interested.

Future Directions

We are interested in following this study up with an additional investigation, during which we would hope to meet the children themselves. This would most likely involve computerised tasks and a series of puzzle games which would allow us to assess the children in a little more detail, focusing on the aspects identified by this study.

We are currently applying for funding to undergo a further study, and will be in contact with information in due course. Once you have considered the study information, it is up to you whether or not you would like to take part.

Finally...

Thank you again for all your help.

If you have any questions about the study please contact:

Dr Gaia Scerif: 01895 271403

gaia.scerif@psy.ox.ac.uk

For further information about the study and links to publications:

<http://psyweb.psy.ox.ac.uk/abcd/Research.html>

For more information about the parent support group UNIQUE please contact:

info@rarechromo.org 01883 330766