



DIESC

Development of Individuals with an Extra Sex Chromosome

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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.

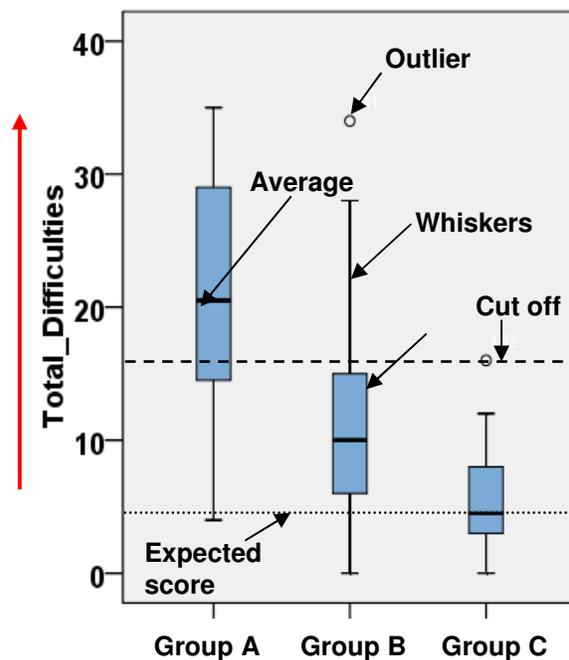
- *Adaptive Behaviour* – Behaviours which indicate how well an individual might be able to cope in everyday life, in terms of listening, speech, hygiene, play, interacting, and understanding of money, time and danger.
- *Maladaptive Behaviour* – A behaviour which might be viewed as challenging or an 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.

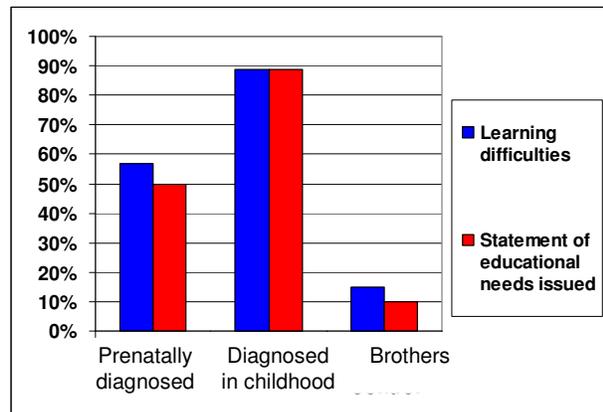
Boys with XYY

Overview

- Boys were described as having a great sense of humour, and being *very kind and caring*.
- They sometimes struggled with frustration, as they were reported to find school more difficult than their brothers and found making friends hard, despite being outgoing and often enjoying the company of others.
- Although it affects only a minority of these boys, *autism spectrum disorder is more common than in the general population, something that parents had alerted us to at the Unique XYY day*.
- They were often described as artists with good attention to detail, and *having a great understanding of technology*.

School Performance

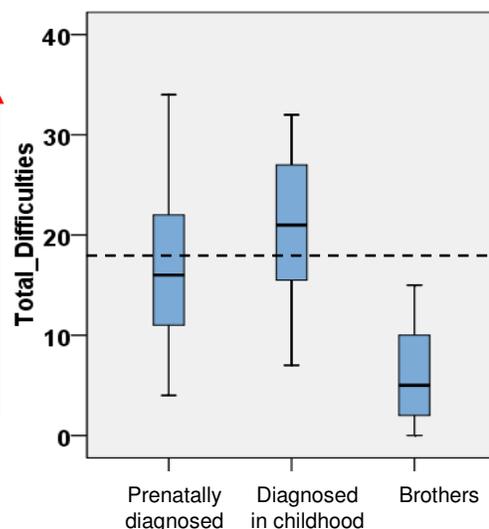
Boys were reported to be *good at Maths*, although many found English a little more challenging. Almost 60% of boys with XYY had some learning difficulties, with 50% of prenatally diagnosed boys having a full statement of educational needs. Despite this, the majority of these boys coped well with classroom support in mainstream schooling. Around a third attended a special unit or school, usually being referred at ages 11-13 because of lack of resources for learning support in large high schools. For boys diagnosed in childhood, a greater proportion needed assistance in school, with almost 90%



having a statement of educational needs, and around half attending specialised schools or units. Statements were typically given around age 4-6, with difficulties with language the key concerns. Parents reported that getting help at school made a big difference to their son's happiness, and they noticed an improvement in progress when help was in place.

Strengths and weaknesses

Boys with XYY showed a wide range of outcomes, with just over half of prenatally diagnosed boys having few difficulties and only around 30% showing elevated levels of difficulty (above the dotted line). However, despite this the majority of boys show higher levels of difficulty than their brothers. Difficulties were most marked in the areas of emotional understanding, hyperactivity levels, conduct problems and peer problems. Pre- and postnatally diagnosed groups show similar patterns of difficulties with social relationships and understanding emotions, something that parents have reported as a concern as the boys mature. In addition often these boys are energetic, and many of them are trying to make friends, even if at times they find it challenging.



For an explanation of graph see pg 2

Boys with XYY

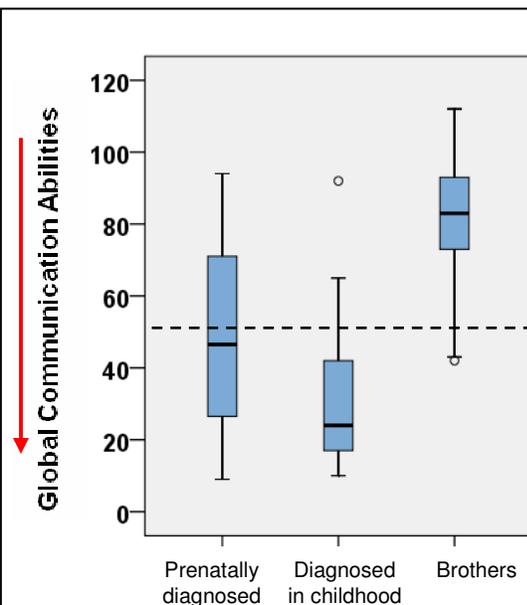
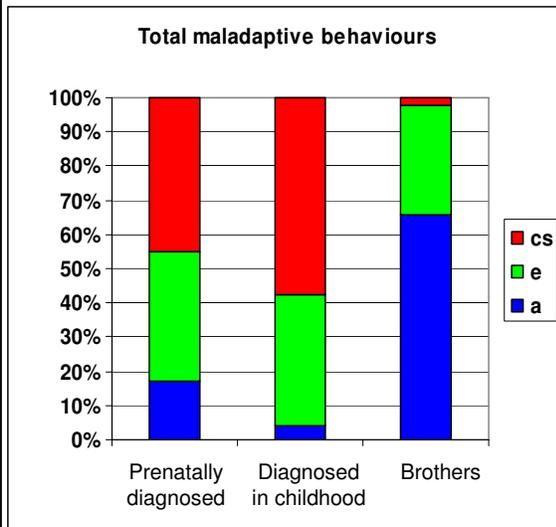
Behaviour

Adaptive Behaviours

Boys with XYY were observed to be helpful around the home and good at cooking, although often they were given less responsibility than their brothers. Cleanliness was a concern for some of the parents, who reported their sons having poor understanding of personal hygiene and tidiness; while this may be common for boys in general, it did seem unusually so for boys with XYY. The majority of boys had a relatively good understanding of money and time, and enjoyed playing a wide range of games, with many parents reporting particular strengths in understanding complex computer games. However some of the boys were observed to find interpersonal relationships challenging, and at times found it difficult to relate to others and understand their point of view. This may be linked to difficulties with both communication and listening, making it more difficult to understand others. Despite this, families found that the boys were relatively good at saying sorry, and were usually polite.

Maladaptive behaviours

Around 20% of prenatally diagnosed boys showed low levels of *maladaptive behaviours*, with 40% showing elevated levels, and the remainder showing concerning (indicated by “cs”) levels of behaviours. This is in contrast to 35% of brothers showing elevated or concerning levels of behaviour. *These behaviours primarily included temper tantrums, difficulties reading others' emotions and being easily led.* Parents reported that usually temper tantrums were caused by frustration, often as a result of a build up of relatively minor incidents. Some parents reported being able to prevent outbursts by calming the situation before frustration builds up. Boys diagnosed in childhood showed a similar pattern, but with a higher rate of concerning behaviours. It is likely that their problems led parents to seek a diagnosis.



For an explanation of graph see pg 2

Communication

Parents reported a relatively good understanding of tone of voice, gesture and body language in these boys, but communication was often a difficulty. A relatively large proportion of the boys scored below the expected level for communication, reinforcing observations by parents. There were some difficulties observed in all 10 subscales, but fewer problems were observed in *interests, non verbal communication, stereotyped language* and *inappropriate initiation* compared to other scales. This suggests that whilst boys may find it challenging to express themselves clearly, they have a range of interests and can rephrase sentences others might not understand. However the boys diagnosed in childhood tended to find all aspects of communication challenging, especially understanding the meanings of words and how they might change with context.

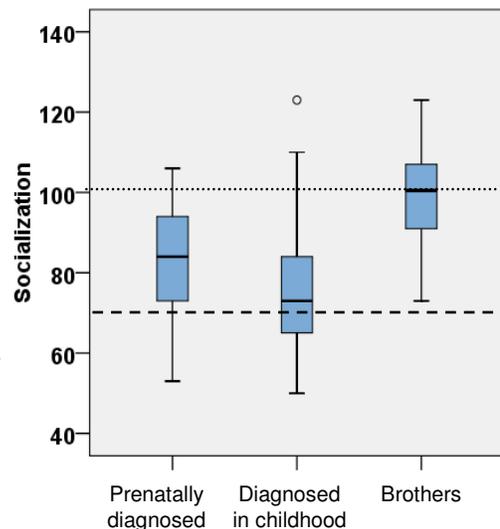
Boys with XYY

Social Skills

When many different aspects of socialisation are considered, over a range of different situations, only a fifth of prenatally diagnosed males showed difficulties, despite many families expressing concern about social skills. This was assessed over how well they play with others, share, and cope with the disappointment of losing, their understanding of their own emotions, and recognising emotions in others, leading on to their understanding that others have different interests and thoughts, and finally how well they behave when introduced to others, their manners, and change of routine. In particular many families commented that their son found it challenging to relate to peers, despite being keen to make friends and be part of a group. Some families were concerned that this had led to their son behaving badly at school to try and fit in with or

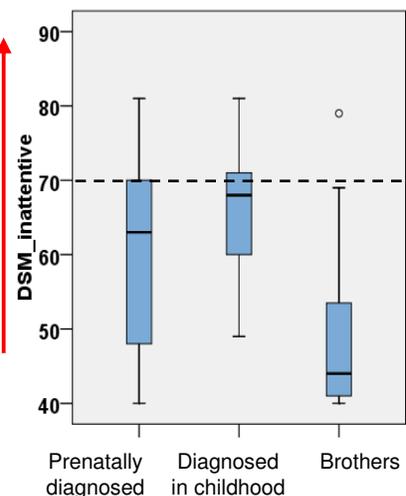
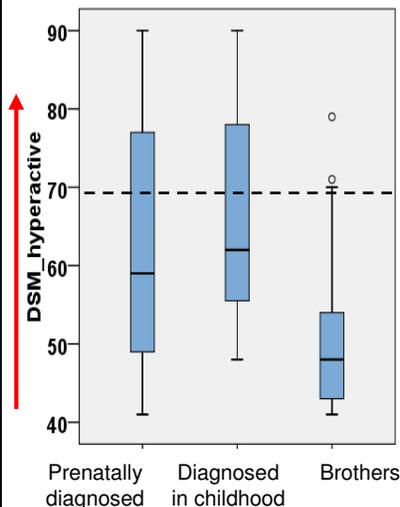
impress other boys. When these aspects are considered in more detail it becomes apparent that these boys are relatively good at understanding play, sharing, and coping with losing, but find it more challenging to understand others' thoughts, feelings and emotions. This pattern is also apparent with regard to postnatally diagnosed boys who find social skills particularly challenging, however, like prenatally diagnosed boys, they are keen to try to make friends.

For an explanation of graph see pg 2



Hyperactivity and Inattention

Boys with XYY show fewer difficulties with *anxiety and shyness* compared to other problem behaviours, as well as low levels of worry *psychosomatic* difficulties. Despite this, they show significantly higher hyperactivity and inattention than brothers, with very similar patterns of behaviour regardless of time of diagnosis. Over 30% of boys in both groups show concerning levels of



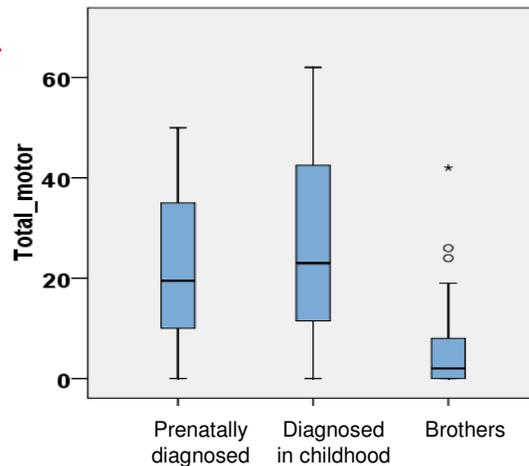
hyperactivity, with only around a fifth of brothers showing concerning levels of hyperactivity. When considered in more detail prenatally diagnosed boys tend to be described as restless, hyperactive or inattentive, and are often busy and energetic. In addition they show similar levels of *perfectionism* to their brothers, with some boys showing fewer perfectionist behaviours than brothers. Despite pre and postnatally diagnosed boys being similar on the majority of scales, postnatally diagnosed boys are inclined to show increased levels of anxiety and are described as real worriers. *Overall they were described as energetic boys* who may have difficulty concentrating but were great fun!

Boys with XYY

Motor Control

Boys with XYY were often described as arty and showing great attention to detail. Despite this, they had some difficulties with both *fine and gross motor skills* relative to their brothers. Fine motor skills such as writing and using a knife appropriately were particularly challenging for these boys, but they were reported to find drawing easier and many improved with age and practice. The boys also enjoy sports with many favouring individual sports such as biking and running.

For an explanation of graph see pg 2



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 be aware 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 boys with XYY with large groups of typically developing children of the same age) change with age. There are clear indications that as prenatally diagnosed males with XYY mature they have increasing levels of difficulties with communication and socialisation skills compared to their peers. These are larger changes than those found in brothers of a similar age. Coupled with this is an increase in concerning or challenging behaviours, especially around ages 10-12, reported by parents to be linked with frustration. However as the boys mature further there is some evidence that their behaviour begins to improve, something that has been reinforced by many parents. This may in time be linked to improvements in communication and socialisation skills, and would support the suggestion from many parents that their son is maturing but more slowly than peers. This is reinforced by a change in the types of behaviour shown from conduct to emotional difficulties. In contrast, postnatally diagnosed males show a stable pattern of strengths and difficulties as they mature.

Conclusions

Overall boys with XYY tended to be kind and caring with a good sense of humour. Boys were described as creative and arty with good attention to detail and a practical approach. However many needed assistance in school and found communication challenging, making it more difficult to relate to peers. They also displayed increased levels of hyperactivity, and many were reported to have difficulty controlling their temper and monitoring their behaviour. Many parents reported that temper was often a result of frustration built up after many seemingly small incidents. Boys with XYY were described as outgoing and keen to make friends, with many showing flair for computing or mechanics.

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:

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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:

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