**XXYY Assembly**

1. Good morning my name is . My brother has a rare chromosome anomaly called 48XXYY, it is also referred to as just XXYY. I’d like to tell you a bit more about this condition, how it affects daily living alongside how we support and enhance the lives of those living with this rare anomaly.
2. 48XXYY is a lifelong rare genetic condition which is a chromosome variation. It is not hereditary, it just happens at conception when the cells split and divide too many times…...

 It only affects males.

 A typical male karyotype is 46XY, but these males have an extra X and an extra Y chromosome, making their karyotype 48XXYY

It is a hidden handicap which is difficult to detect and diagnose, as the visible differences are very minimal. A rare diagnosis that many peadiatricians, consultants and geneticists have never seen before. This results in many families being given an incorrect diagnosis and lack of information on how to manage the symptoms of the condition successfully.

The impact upon daily life skills is often misunderstood and support in this area is sadly lacking, often making those who are unaware of it very judgemental.

The condition affects individuals in a variety of ways. This slide has some examples of the more common ones. These are all umbrella titles which manifest slightly differently and to differing degrees within each individual. There a many more alongside these and some are more associated with health, like epilepsy, diabetes and heart defects but I couldn’t fit them all on the slide!...

1. My mum thought there was something wrong with my brother’s foot when he was born, this was disregarded. Due to this and other small indications that his development was different mum continued to pursue his diagnosis journey when he was 12 months old. He had no balance, his speech sounds were unusual, he moved his hands differently and at times his behavior was odd. His first diagnosis of Dyspraxia came when he was 3years old. Although he had many traits of this condition he didn’t quite fit it completely.

 After further investigations and numerous specialist medical appointments with Paediatricians, sign language, speech therapy, chiropody, occupational therapy, physiotherapy and the community dental service. He was finally diagnosed correctly by a Great Ormond Street Hospital geneticist aged 5yrs 11 months.

At just short of 6 years, he was slightly quicker than the average diagnosis. We were told it was highly unlikely that we would ever meet another boy with this condition.

1. So we contacted Unique a rare chromosome support group and we were given a list of 25 UK Families, but not all had contact details. We sent off a few emails and set up 2 dates of open house. Initially 5 families attended and the 2nd meet we had 6 families.

Everyone was amazed at how similar the individuals were and how well these normally socially isolated boys bonded. They were on the same wavelength, and just understood each other completely. We collectively decided we would like to start a UK support group dedicated to just 48XXYY.

1. Whilst 48XXYY is acknowledged as a medical diagnosis, it is not well acknowledged as a disability, particularly within the areas of support. Without data from a UK group it was very difficult to get recognition, prove their needs and navigate the UK educational and health authority systems.

The Support Group was formed in March 2014 and has led to UK statistics being generated. This is improving access to support and is helping making the lives of our XXYY families and individuals less stressful. We have created a UK family network where we all support each other and know that the XXYY boys, their siblings and parents are no longer isolated. We raise awareness, set up conferences, events and family days out. We work alongside the Americans who are the lead in research and management of the condition. We share information with European and American groups giving us all a global connection. Our older XXYY individuals act as Role Models, they share their experiences and make suggestions, helping to inspire our younger and newer members, and enlightening the parents how their younger boys may be feeling. This in turn improves their confidence and self-esteem, aiding them to be more accepting of their differences and making them realise their worth.

1. We run our subsidized conferences over 3 days in Youth Hostel properties in the Midlands making them accessible to the whole of the UK and affordable to all.

We book sole use of the venue which creates a safe and secure environment with lots of space and no outsiders around to judge, ensuring that everyone feels comfortable. We also run an achievement presentation as well as many fun individual and group activities.

We fly the American specialist doctors in from Denver, the only specialised clinic for XXYY in the world. They work tirelessly doing presentations, medical appointments and research as well as networking with our UK doctors, and we all learn from each other.

We run specific sibling workshops, helping them understand and realise how important their role is in their brothers lives. We give them the recognition and support they deserve as this can sometimes be overlooked as the XXYY boys are naturally more demanding of parental time.

This time spent together is so important as it allows for family and interfamily bonding, networking with others and building friendships. Everyone leaves with acceptance and a sense of belonging.

1. Over the 3 days our speakers, from medical professionals, Education specialists and Support services provide us with invaluable information. They cover the broad range of topics our families have been affected by. With all our boys and families in one place we in turn can supply them with a chance to learn from, work with and improve their knowledge and research on the condition.
2. My brother is 17 he has a reading age of 8.9 years and along with this he has learning disability and a developmental delay. This means everything is tricky for him and he has to work twice as hard as us to get the same results. He knows he learns differently and can get very frustrated by this. He struggles to make and keep friends and understand social situations. He is not easy to live with and at times can be exasperating and a bit eccentric… He has no patience, suffers with mood swings and gets angry very quickly, which often leads to complete meltdown. He is impulsive and verbally aggressive when he feels anxious or threatened. He often misreads situations and misinterprets feelings and emotions, this makes him vulnerable in the outside world. He is stubborn, selfish and a real pain at times but he is still my brother and I can’t abandon him…. Even though at times I have felt like I want to! I am one of the few people who understands he sees the world differently and it’s not his fault. I do have to remind myself of this often and have lots of patience with him! We also worry about him when he is out as he struggles to fit in with the friendship groups and has got himself into difficult situations many times. This responsibility for me will grow as he ages as my parents will age too and he will need my help with life skills and control of finances. Its not all bad though, he is still progressing and achieving in small steps. He has just started college studying tree surgery and who knows how far his full potential will take him? Despite our usual teenage sibling rivalry he is a great brother. He has a good sense of humour and is caring, loving and generous. He wants to be sociable, sometimes a bit too much!! and is very family orientated. He is a fabulous uncle to our younger nephews and keeps them amused for hours.

1. My brother sometimes confuses me as he lacks confidence one minute and then the next he is very impulsive and doesn’t believe he is limited by anything. He often has big ideas, but then when it comes to it he tries to back out, but with a bit of gentle encouragement he often manages to succeed. A great example of this was his Silver medal in the National Championships in vaulting. He is always on the go, otherwise he falls asleep, often in funny places! He absolutely loves anything on wheels he rides Motorcross, BMX, mountain bikes, scooters, skate boards and even a unicycle. He may understand and act differently than us but he has tried and excelled at many other things which neuro typical people find hard. He definitely sees the ability within the disability.
2. Our group does not have an income. We run purely through unpaid volunteers and funds raised by our families and supporters in the ways listed on this slide. All our conferences and some of the events we run are subsidised, this stops anyone being excluded due to financial position. 100% of all funds raised are used to support and enhance the lives of our XXYY males and their families and to run our website. Without these funds our group would cease to exist, so I’d like to say a heartfelt thanks to all those that have helped us.
3. Since forming the 48XXYY Family support group UK we have moved forward massively and are creating a lasting legacy:

The condition now has Professor Gary Butler a UK endorsing medical Professional , who is happy to lead and support the group in any way he can.

We run a UK Website full of information and signposting.

We have made national and international contacts.

We have raised awareness in many ways both locally, nationally and internationally.

We have supported and connected families and friendships have flourished.

In the whole of the UK we have grown our membership in 5 years from 25 to 100 families, and have connected with other international groups across Europe and worldwide.

This is all a direct result from our work and public support.

We will be having a day at the end of term to raise funds for the charities our school are supporting this year. I hope you will feel able to help these good causes and that you have enjoyed my presentation. Thank you for listening and thanks again for your support, it means a lot.