23/09/19

I am writing with reference to 48XXYY

I have been contacted by many families and asked to write about how the medical condition 48XXYY affects daily living.

I am the founder of the 48XXYY Family Support Group UK and have organised annual conferences with medical and educational professionals and family events. I also have a son of 13 years old with this medical condition.

I am not a medical professional and can only draw upon my own personal knowledge of 48XXYY and those that I have met with this medical condition.

48XXYY does impact greatly upon those that have it and their family. It does have a spectrum and some boys are affected more than others. It presents with developmental delays and milestones tend to be reached much later than by neuro-typical children. Often mental age is far lower than chronological age and development is often static for some time then suddenly makes a big leap. All the boys / men that I have met, clearly have some level of learning disability and the degree of this differs massively from one to another. They tend to be visual learners and think differently, they often get confused and have trouble making sense of the world around them. They dislike busy places and often go into meltdown if overwhelmed by too much e.g choice, noise etc They tend to flourish in quieter surroundings and when given only one thing at once to deal with. They do not read facial expression, body language and social cues well.

Most have traits of the following: Sensory Processing, Autism, Anxiety, Speech disorder, Slow Processing, Easily Fatigued, OCD, PDA, Mental Health Issues, Dyspraxia, Dyslexia, Verbal and Oral Dyspraxia.

Other features are infertility, tall stature, bone anomalies (particularly elbows, feet and fingers).

All of the above have a massive global impact upon our boys. They get frustrated easily which leads to behavioural issues, temper outbursts and complete meltdown. School tends to be very challenging for them and most end up with an EHC Plan and in specialist schooling due to their additional needs and their slow academic progress. They are bright enough to be aware of their differences and this can lead to problems fitting in with their peers leading to bullying, frustration, anger and behaviour issues. They are generally very sensory seeking (a lot of them still suck their thumbs even as adults). Most are very particular about clothing, socks, layers, labels and how it feels against their skin. They struggle to understand and manage money, also being able to understand time is a big issue. They are vulnerable as children and adults because of their differences, their levels of understanding, tendencies to trust others, misreading of situations and social cues. They need a lot of support, reassurance and understanding as children and this continues throughout adulthood.

If you have any further questions please feel free to contact me: info@xxyy.co.uk

If you would like further advice about the condition I would suggest you contact: our groups endorsing medical professionals: Dr. Gary Butler gary.butler1@.nhs.net Endocrinologist GOSH, Dr. Nicole Tartaglia Nicole.Tartaglia@childrenscolorado.org (Denver Childrens Hospital USA) or the Social Communications Disorders Clinic GOSH

Yours Sincerely

Jocelyn Eldridge