Pre-natal diagnosis: personal stories

Lucy's story

I had to arrange my 13 weeks scan privately and Sydney UltraSound for Women recommended the NIPT test. I willingly had this due to the greater accuracy for Downs Syndrome… this was my third pregnancy and I was over 40, plus it now only cost a little more than the standard Nuchal Translucency Scan.

I knew something was "wrong" when they phoned the day after the test (a Friday) – multiple times… and started calling me and then my partner again on the Monday. We were on holiday, so slow to answer.

I called back… already feeling sick at the thought that there was something seriously wrong with the third child I so badly wanted. I had never heard of XXY or Klinefelters, and to be honest, only heard part of what the nurse was telling me.

- What I heard…..infertility; my little boy would never be able to have children of his own
- What I asked….life expectancy; I learnt that there wasn't any difference in life expectancy of an XXY boy (and I was in too much shock to ask much else…..and my other two kids were just outside the bedroom door)
- What I remembered… her insistence that I book in a CVS and/or an amnio to confirm the diagnosis before deciding on a termination… and her telling me not to Google
- What I did on putting the phone down… Googled (and cried)

I spent the next two days Googling every chance I got (whilst trying to act normal for my eldest's fifth birthday!) It was pretty bleak, particularly some of the images of men with breasts. It was also frustrating; I just wanted the facts, but there seemed to be little that was up to date.

I found a Facebook group….this was also scary as many of the people on there had older children and there seemed to be so many issues …..fortunately someone in that group directed me to an XXY babies Facebook group.

Finally I had found the other side of the coin, the happy stories, the beautiful boys and the (rightly) proud parents. The more I read, the more I realized that this was something I could cope with… and not because I'm an amazingly strong person; I'm totally not… but because XXY is a spectrum and early intervention could make a huge difference.

It took me a little while to get over knowing the gender of my third child; the first two were surprises and this would have been to, had it not been for the NIPT test throwing up the genetic flag. I worried about XXY, but chose not to confirm the diagnosis until after birth as I wanted to hold onto the small chance it was an incorrect reading… and I don't really like needles… and didn't want to increase the chance of miscarriage. I also reasoned that if I terminated and tried again, if I was lucky enough to get pregnant, it could be even worse (I'm old!)

This was my last pregnancy and I tried to enjoy every moment of it… it went by too quickly (running after the other two!). I had a few more scans than "normal" as the first obstetrician I saw at the public hospital flagged genetic abnormalities could cause growth to slow/stop. I welcomed these as it was a chance to confirm my baby was OK (I was much more scared the third time… older and more aware of the dangers). I also chose to talk to the Genetics team. They were reassuring and learning more about the prevalence of XXY all the time.

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I'm lucky in that I have easy pregnancies… the only difference in this one was the time I spent reading about XXY – mainly via other people's stories on Facebook.

When my son finally made his appearance he was beautiful and everyone said how much he looked like the other two. I loved him fiercely and within moments, couldn't imagine life without him

We had the blood test… results took over a week (Easter got in the way). I kept hoping we would be "lucky" and it would be negative. Of course it wasn't… then I had another week hoping it would just be mosaic (in one cell). Of course it wasn't (it's in every cell).

At 6 months old, it's too early to tell where he is on the spectrum… but he is doing everything around the same time as my first son. We've had the T-shots, I'm continuing to read the latest research and I'll intervene if I need to.

XXY can't be "cured", but on the other hand, it isn't something that needs to be managed every minute or even every day. It isn't life-threatening (unlike some common allergies). It isn't obvious to others. We just need to watch that milestones are met and that difficulties are addressed as soon as possible

Overall, I'm so incredibly grateful that I know about the extra X so I can make sure I give my little boy every chance to shine.

Our XXY Journey – Lisa's story

I still remember that phone call only a couple of weeks out from Christmas where my husband and I were chasing the results of our NIPT test. We really just wanted to find out the sex of our second baby (as we were too impatient to wait until the 20 week scan) and naively I didn't think for one minute our results would come back with anything more than whether our baby was going to be a boy or a girl. I had been chasing my results for over a week and when I finally received "that call" with the results we were advised we needed to come in ASAP to discuss the results of the NIPT with my Obstetrician. I still remember the sick feeling in the pit of my stomach.

Sitting in my Obstetrician's office I was so happy to have my hubby by my side as all I heard from the discussion was extra chromosome, infertility, learning difficulties and termination was put on the table as an option. We walked out of there with our heads spinning and jumped onto google not even knowing how to spell Klinefelters as we had never heard of it. I didn't want to have a CVS done but felt like I should so we could understand the full picture and confirm our baby boy was XXY.

Two conversations greatly impacted me in those early days – one with the Genetic Counsellor prior to the CVS and the other a mother of an XXY boy for which I am so grateful. The three key things I took away from these discussions were:

- My unborn sons diagnosis of XXY will not define him
- It is a spectrum, be careful not to label or confuse XXY traits as a lot of these traits are consistent with those of regular boys
- You didn't sign up for this for your son, however with your other children there will be things you never signed up for either

After having the CVS done a week out from Christmas the hardest part was waiting for the results and also the possibility I might miscarry from this procedure. I was actually relieved when the results came in that it was just XXY. I decided from that moment I was going to enjoy the pregnancy and realistically it won't be for at least another couple of years where the symptoms of XXY may show.

All this dark and negative thinking wasn't good for my mindset and having the ability to be conscious of this and think positively really made a difference for me.

After my gorgeous son was born there have been some key moments that have stuck with me over the past 2.5 years:

- Our Paediatrician at birth and then again at the two year check-up saying how lucky we were to have a prenatal diagnosis and he is not displaying any classic signs
- An old work friend at a lunch cuddling my baby saying he is perfect and wants a baby just like him
- Walking at 12 months – I was in disbelief our other son didn't walk until 16 months
- Participating in a Klinefelters speech research study at the Royal Children's Hospital with my son's speech results falling in 'normal' range
- Watching the strong bond grow between my XXY son and his older brother – they are so close and just do regular boy stuff like run and wrestle

My gorgeous boy is only 2.5 years old and is early days I know. Our journey (post those early dark days) has been wonderful and he has brought so much joy to our family. I am conscious that kindergarten is only 18 months away, and challenges may appear. I feel so lucky to have had a prenatal diagnosis as we are aware and informed and I wouldn't change him for the world, he truly is an exXtraordinary boy.

Sam's story:

We had done IVF for a few years and at the age of 41 and 42 my husband and I came to the decision that we would stop and accept that it was very unlikely we were ever going to be parents. I took four months of long service leave from a busy job and we went on a holiday and came back pregnant from that trip!

We had the usual screening and the relatively new NIPT. I was expecting results back before the long weekend in June, hoping that we could catch up with a lot of family in the country who were all heading home that weekend and announce our pregnancy. I called Thursday or Friday before the weekend but was put off until after the weekend which was so annoying as it seemed like more than enough time had elapsed between my NIPT and results time. I later found out they had results but the radiologist wasn't there to communicate results to us. An appointment was made for Tuesday after the long weekend, I remember the wait before the appointment and feeling like I was getting a lot of sympathetic looks from the reception area which did nothing to calm our nerves!

Toby's diagnosis was delivered by the radiologist who had done the 12 week scan. I had never heard of XXY or Klinefelters syndrome. Her first sentence mentioned high incidence of prison and high chance of autism. I was reeling and remember burying my head in my hands whilst I tried to end the appointment as quickly as possible so I could consult google! She also went on to mention that her son had autism and had she known she might not have continued the pregnancy! While this sounds horrific to her credit she got on the phone to the genetic counsellors and we were able to go straight in to see someone in the middle of a busy clinic. This appointment was much more balanced and talked spectrum etc. As an aside our obstetrician also told us most people abort this condition.

I off course googled which wasn't the best move and did as much reading as I could which I had to stop during my pregnancy as I found it quite distressing and a bit depressing. I wanted to embrace my pregnancy and enjoy my pregnancy so towards about 2/3 through stopped researching etc. I was so lucky to receive a call from our support network and so grateful that this person offered for us to

meet her 10yr old XXY boy. This was a great comfort to us and really helped us see that XXY is a spectrum condition and that we would deal with challenges if and when they arose, it was lovely to meet this family and see the warmth and love in place.

Our gorgeous boy is now almost 3 and I'm sooo thankful for him, he is a great joy, he does have some speech delay which we are working on but has given us plenty of fun and action! He has great receptive language and he is saying new words every day now.

Louise's story

Our journey began after 6 years of IVF . I had decided not to go one more round and my husband Pete decided we would (yay for Pete) we received our early positive pregnancy result. It wasn't uncommon for us to return positive test with and without assistance (IVF) as we returned several positives that didn't progress. In the December we had a CVS test done as I was 43 and my obstetrician recommended it. I approached this with naivety and one positive to come from that nauseating painful experience was the imaging they used was amazing. We got to see a brilliant picture of our gorgeous baby. It left me feeling very uncomfortable, although I had been experiencing some morning sickness at this point. Days passed and we were anxiously awaiting the results.

I decided to call our Obstetrician/IVF doctor to see if and why I hadn't had any results. We had a great relationship with him as we had been trying to conceive with his assistance for several years at this point. When my call was transferred to him he was surprised we hadn't been contacted as this is what the plan was as far as he knew from the imaging place in East Melbourne. He then delivered the Klinefelter syndrome result to me with no preparation for delivering that news. This is how I recall the phone conversation Klinefelter syndrome, tall, you are having a boy, infertility and that's about it. After the call of course I frantically started to google & became incredible scared. A meeting with a genetics councillor was arranged which had us drive 2.5 hours from home to maybe the hospital where we had a 10 minute appointment with a professor in Genetics. We left with not much more information. Just that his IQ would probably be 10 points lower than other biological siblings. Unfortunately he didn't even have fact sheet or booklet for us to take away with us.

Our Obstetrician throughout our pregnancy was wonderful with snippets of encouraging information regarding the KS, even though the way he delivered this diagnosis to me wasn't great initially. He reinforced that so many guys lead very normal healthy lives and don't know until they start to try and have families. I am very grateful for the pre-natal diagnosis even though I think we could have been more proactive in the early years (up to 4) as he was just a regular baby / toddler. I did take him to OT and speech therapists but he was never majorly behind so we just kept up the wait and see approach. However from the end of prep we started to see speech therapist, OT and informed both of his diagnosis and they still support him to this day - speech weekly, OT fortnightly. It is more about us choosing to have this support rather than it being a must… In summary a pre-natal diagnosis in our opinion is the best tool you can have for supporting a child with KS.