Klinefelter Syndrome
Prenatal Diagnosis
Fact Sheet

Have you just received a Prenatal Diagnosis?

Australian X & Y Spectrum Support (AXYS) is a not-for-profit organisation that is here to support you.
This fact sheet has been written by parents who have received a prenatal Klinefelter Syndrome diagnosis.
We understand this time can be an emotional and challenging one and can at times be overwhelmingly with a mix of emotions.
This fact sheet has been written to provide you quality and accurate information on Klinefelter Syndrome and what you need to know.

What is Klinefelter Syndrome?

- Klinefelter Syndrome, also known as 47,XXY and XXY syndrome is a common congenital chromosomal disorder, affecting 1 in 450 males in Australia.
- In a male the normal chromosomal arrangement is 46XY, but for males with Klinefelter Syndrome one or more additional X chromosome are present resulting in 47XXY or other variations.
- Not all cells may contain the extra X chromosome, this is known as mosaic Klinefelter syndrome or mosaic XXY syndrome.
- This is not a hereditary syndrome and it is not known whether the extra X is passed on from the mother or father at conception, nor does it appear to be related to either age of the mother or father at conception.
- Klinefelter syndrome presents as a spectrum with everyone being affected in different ways and in different severity.
- The most common features being; hypogonadism (no sperm count and low levels of testosterone hormone production), language and learning difficulties, hypotonia and the individual may be taller than average for their family.
- Whilst there is no cure for this syndrome, early intervention is the key. Treatment options and early intervention strategies help boys and men with Klinefelter syndrome reach their full potential.
- In some instances, a medical diagnosis is not made until childhood, adolescents or when adults are having difficulties in conceiving, and in some cases, males can go through life without ever being diagnosed.
- It's estimated that up to 75% of cases are undiagnosed or misdiagnosed.

Prenatal Diagnosis

With advanced prenatal testing available, this common chromosomal condition is now being identified prior to birth. The incident of pre-natal diagnosis is rapidly increasing.

Non-Invasive Prenatal Testing (NIPT), is currently the most common prenatal test available. This is an analysis of the mother’s blood and is a test that can get an accurate estimate of the chance that their baby potentially has a common chromosome condition such as trisomy 21 (Down Syndrome), trisomy 18, trisomy 13 and also identifies the addition or deletion of the sex chromosomes X & Y (Klinefelter syndrome).

Chorionic villus sampling (CVS) and amniocentesis are invasive diagnostic procedures during pregnancy that can identify for certain if the baby has a chromosomal condition like Klinefelter Syndrome. These tests involve the passing of a fine needle into the abdomen to obtain placenta tissue or amniotic fluid samples for testing.

Both CVS and amniocentesis carry a small risk of miscarriage, generally within 2 weeks of the procedure. It is recommended to talk to a Genetic Counsellor or Specialist before undergoing these invasive procedures. Expectant parents also have the option to wait until their baby is born and confirm a diagnosis through the baby’s blood sample.
What are the main impacts?

- It is important to remember that Klinefelter Syndrome is a spectrum and will affect each individual in different ways and in different severity. Individuals very rarely have all the signs and symptoms listed.
- The most common impact is male infertility also known as hypogonadism which is a result of not producing enough testosterone (male hormone) needed for the body, effecting the ability to produce sperm.
- With advanced reproductive technology, some impacted males have experienced success in having their own biological children.
- Overall health and different stages of physical, language and social development can be impacted.
- Research indicates very few or no signs are present at birth and unless the infant was tested prenatally, it’s unlikely this condition would be suspected. Some common impacts seen in early childhood may include:
  - Speech delay/expressive language disorder
  - Hypotonia impacting gross and fine motor
  - Shy/timid
  - Small testes
  - Delayed motor development e.g. sitting, crawling, walking later
  - Auditory processing difficulties
  - Mental wellbeing concerns including depression and anxiety
  - Learning difficulties e.g. reading and writing

Early intervention is the key and greatly impacts positively on a child’s life. Prenatal diagnosis is helping to equip parents with knowledge and understand what early intervention methods are available post the birth of their child to help them to reach their full potential. In some cases, funding may be available for early intervention. If unsure whether your child needs some early intervention, it can’t hurt to have them assessed.

What Prenatal and Postnatal Care should I access?

Prenatal Care

- A Klinefelter’s pregnancy is like any other pregnancy, however for many couples receiving a prenatal diagnosis can come as a shock and it’s important to have a strong network around you with a focus on self-care.
- This diagnosis is not a result of anything that you have done and having a prenatal diagnosis can help equip with the knowledge and information to assist your son with early intervention strategies depending on impact and severity.

This network includes:

- Genetic Counsellor – A very important resource to discuss the prenatal diagnosis of Klinefelter Syndrome, to ask any questions or identify any concerns you have;
- Australian X & Y Spectrum Support (AXYS) - A peak Australian support network in Australia, who offer access to parents and individuals who have experienced a prenatal diagnosis which can be accessed online, via phone/email or face to face; and casual meet and greets. AXYS can provide information on key health professionals in your area to ensure that you are well supported on this journey.
- Trusted family and friends – It is a very personal choice who you share this diagnosis with, if the right person they can provide enormous support and reassurance.
- Please refer to AXYS website for further information, support and networks within your area

Postnatal Care

After delivery, key specialists to access and work collaboratively with:

- Family GP
- Paediatrician (ideally with Klinefelter’s Syndrome/XY syndrome knowledge and experience)
- Paediatric Endocrinologist
- Early intervention team e.g. Physiotherapist if any delays are identified

Where to from here? AXYS are here to help

AXYS Australia is a volunteer network and not-for-profit organisation that was created in 2013 to support Australian parents who have children diagnosed with an X & Y sex chromosome variation. More broadly it supports men, woman, partners of diagnosed adults to share personal experiences and access quality information.

AXYS offer a variety of peer support and resources which include a confidential Facebook community group, informal get togethers, up to date literature and research, connection to Global networks, email and phone support and annual information seminars. Please contact the following:

Help and enquiry line: 0412 038 142  
Email: contact@axys.org.au  
Website: www.axys.org.au