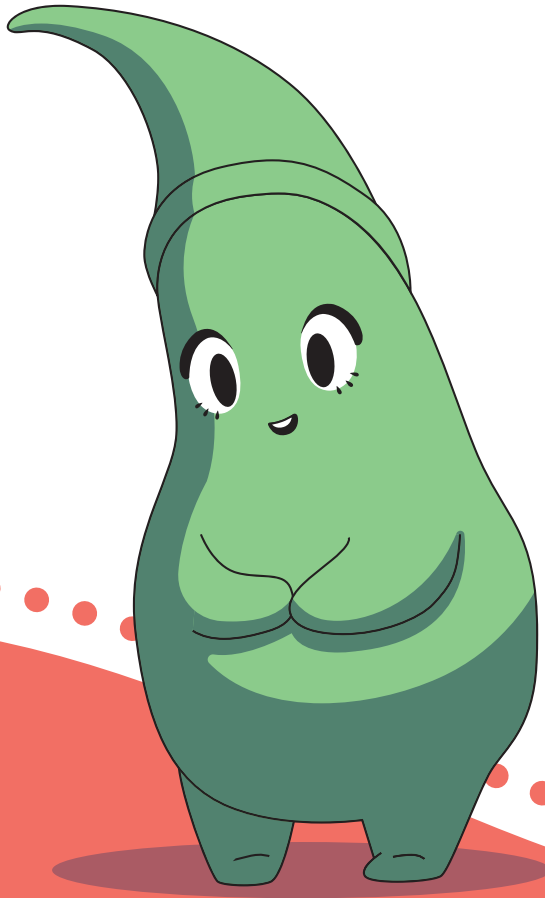
ANZSPED
AUSTRALIA AND NEW ZEALAND SOCIETY FOR PAEDIATRIC ENDOCRINOLOGY AND DIABETES

Want to know more about

Klinefelter syndrome



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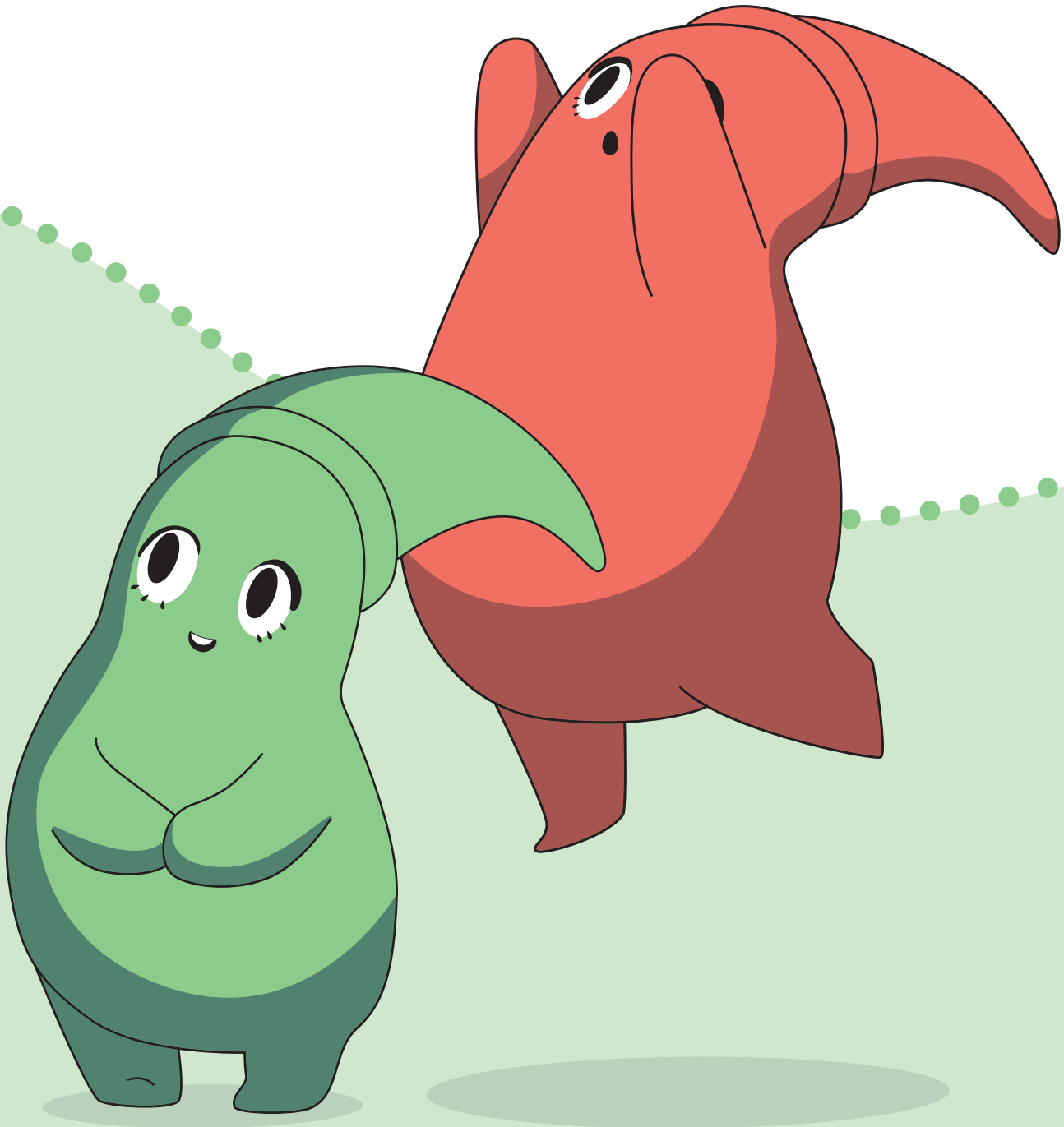
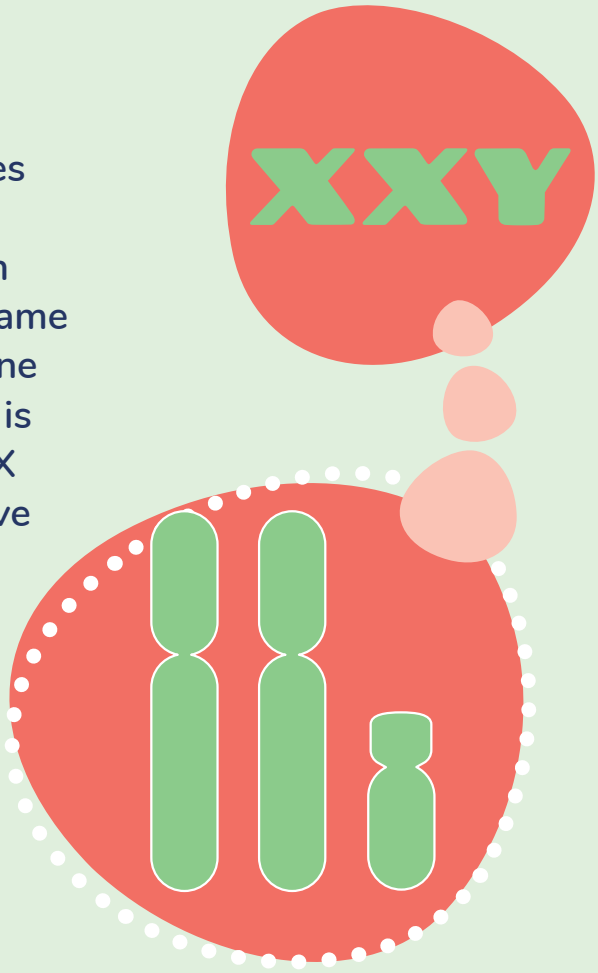
What is Klinefelter syndrome?

Klinefelter syndrome (KS) is a condition that occurs in males due to the presence of one or more extra X chromosomes e.g. 47XXY.



Chromosomes

Most people have 46 chromosomes per cell. These occur in pairs with one of each pair coming from each parent. 22 of these pairs are the same between males and females but one pair, known as sex chromosomes, is different. Most females have two X chromosomes and most males have one X and one Y chromosome.



How common is Klinefelter syndrome?

It is estimated Klinefelter syndrome occurs in approximately 1 in every 500 to 1000 males. However, it is thought that only 25-35% of cases are ever diagnosed and only 10% are diagnosed during childhood or adolescence.

How does Klinefelter syndrome affect boys?

The features of Klinefelter syndrome vary between people, with some showing many signs and others few or none. Boys usually have a typical male appearance but may experience delays in puberty and most will develop testosterone deficiency and have fertility challenges.

These boys are often taller than average and may have developmental delay, behavioural and learning issues, or mood issues. They may benefit from extra help at school. Adolescents and men will typically have small testes and may also have reduced body hair and breast enlargement (gynaecomastia).

How is it diagnosed?

Klinefelter syndrome is diagnosed by genetic testing on a blood test. The most common time of diagnosis is when adult men are being investigated for infertility. Sometimes it is identified by genetic screening in pregnancy.

Are there other medical conditions associated with Klinefelter syndrome?

Boys with Klinefelter syndrome have a slightly higher risk of developing some medical problems such as osteoporosis, type 2 diabetes and autoimmune thyroid disease. They also have a slightly higher risk of developing some cancers (very rare). Therefore, it is important to have regular medical check-ups.

What treatment is needed?

There is no treatment required in childhood for Klinefelter syndrome but different signs and symptoms can be addressed if required. For example, behaviour and learning issues may be managed by a paediatrician, therapists and/or psychologist. Delayed puberty or testosterone deficiency is usually managed by an endocrinologist.

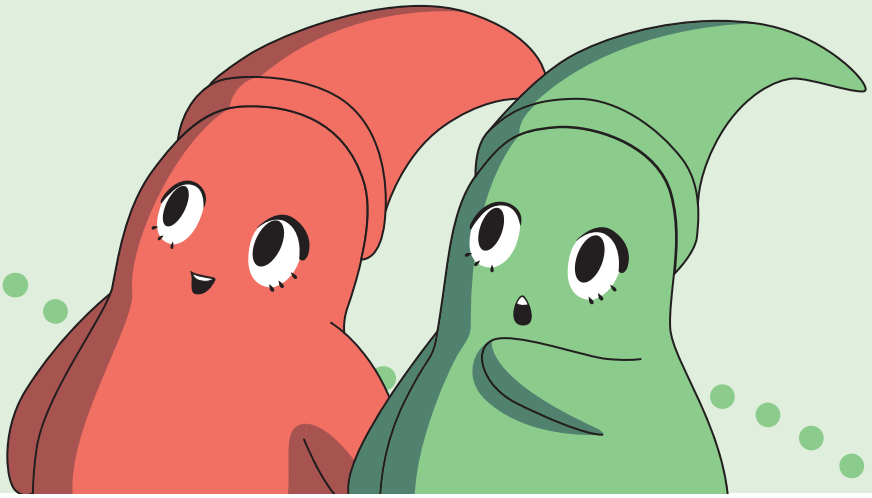
Testosterone supplementation is usually required in adulthood and may be started in adolescence. There are different ways to give testosterone including injections and gels/creams. The best type of testosterone treatment for you should be discussed with your doctor.

Are there fertility treatments for Klinefelter syndrome?

Fertility can be challenging for people with Klinefelter syndrome. If you would like to find out more about fertility treatments, this is best discussed with your doctor. You may be referred to a fertility doctor who specialises in this area.

Disclaimer

This leaflet has been written by members of ANZSPED. It is designed to give you some general information about your child's condition and treatment. If you have any questions about your child's condition and treatment, it is best to speak to your child's doctor or specialist nurse.



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Remember

Regular review with your child's medical team is essential to guide treatment.

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