ANZSPED

AUSTRALIA AND NEW ZEALAND SOCIETY FOR PAEDIATRIC ENDOCRINOLOGY AND DIABETES

Want to know more about

Congenital Adrenal Hyperplasia (CAH)

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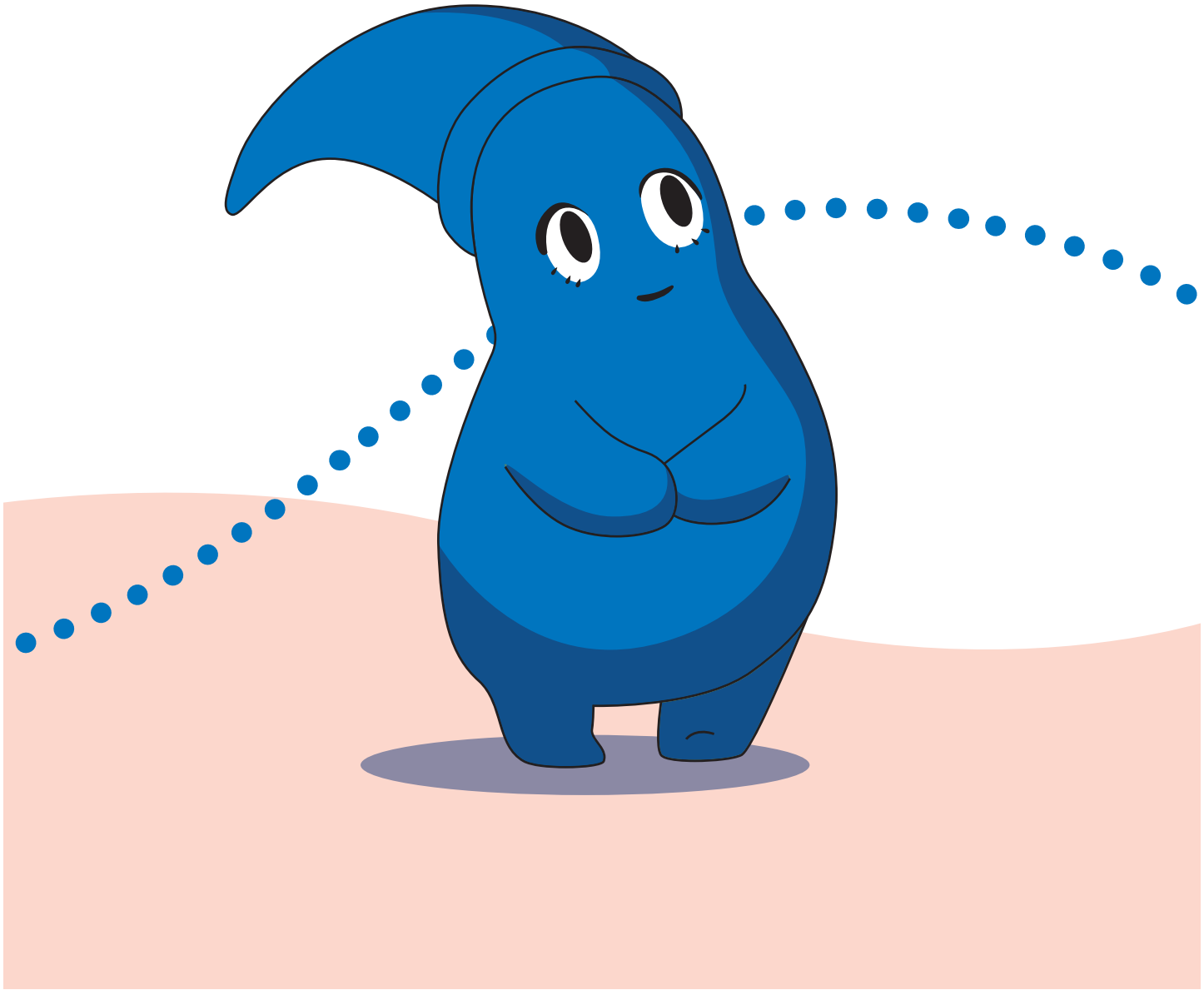
What is Congenital Adrenal Hyperplasia (CAH)?

CAH is a genetic condition present from birth (congenital) and associated with enlargement of the adrenal glands (hyperplasia). The most common type of CAH is caused by the lack of an enzyme called 21-hydroxylase.

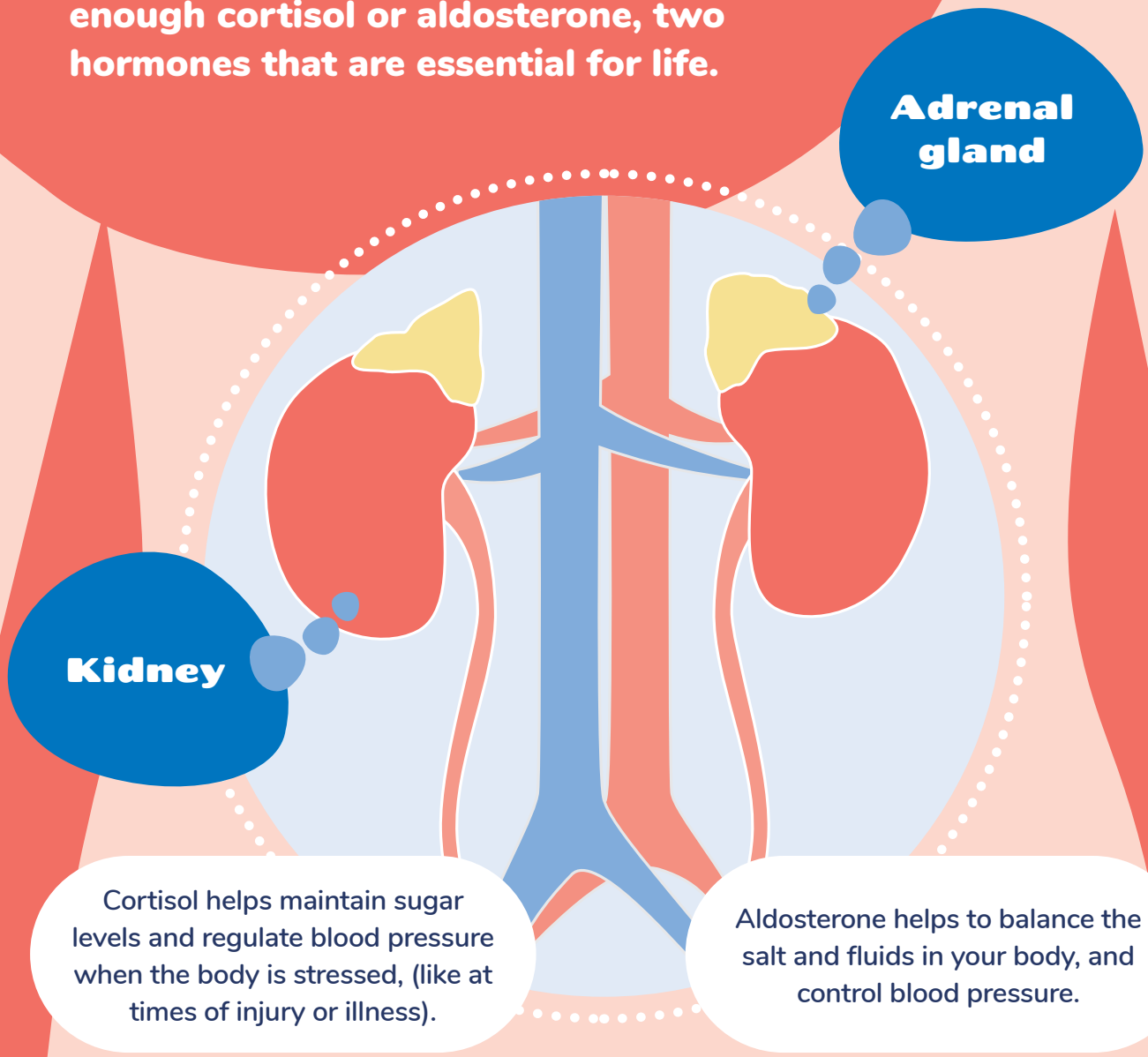
Your doctor will be able to explain this in more detail.



What are adrenal glands and how are they affected in CAH?

Adrenal glands are two small glands that sit on top of the kidneys.

In CAH, the adrenal gland cannot produce enough cortisol or aldosterone, two hormones that are essential for life.



Cortisol helps maintain sugar levels and regulate blood pressure when the body is stressed, (like at times of injury or illness).

Aldosterone helps to balance the salt and fluids in your body, and control blood pressure.

In both boys and girls, the adrenal gland also produces male hormones called androgens. In CAH, excessive amounts of **androgens** are produced.

What are the symptoms of CAH?

CAH symptoms vary by person and depend on the type of CAH a person has.

Salt wasting CAH (sometimes referred to as classical CAH)

- Newborn babies may present as pale, lethargic, with poor feeding, vomiting and diarrhea due to a lack of cortisol and aldosterone. They may also have low blood sugar and low blood pressure.
- This condition can be life-threatening.

Simple virilising CAH

- The adrenal glands make enough aldosterone but not enough cortisol and too much androgen. Female infants may present early with changes that make their genitalia appear more male-like (viralised). Both boys and girls may present with early puberty changes such as pubic hair and underarm hair.

Non-classical CAH

- Symptoms may include early puberty, acne, and rapid growth. Girls may have increased facial hair, irregular menstrual periods, and voice deepening. Some people with this form of CAH may not have any symptoms over their lifetime.

How is it diagnosed?

CAH is usually diagnosed after birth, based on symptoms and signs or the results of newborn screening. Some tests that may be carried out to confirm the diagnosis in infants suspected of having CAH include:

- Blood tests to check adrenal hormone levels and salt levels.
- Imaging tests, such as ultrasound.
- Genetic testing.

What treatment is required?

The hormones affected can be replaced by medications. These should be taken for life, with regular follow-up by specialist health professionals. Hydrocortisone replaces cortisol, fludrocortisone replaces aldosterone and additional salt supplements may be needed.

Increased doses of hydrocortisone are needed when your child is unwell e.g. at times of illness or broken bones, as our bodies require increased cortisol at times of stress.

Does the treatment have side effects?

The medications are very well tolerated. Sometimes a child's medication dose may be too high or too low due to their body's natural changes and cortisol needs.

What monitoring is required throughout life?

Treatment will need to be monitored with regular medical review. The goal of treatment is to keep adrenal hormone levels in range and ensure normal growth and development. It is valuable for families to have access to psychosocial support such as counsellors, psychologists, and peer support. There may be other health professionals in a person's health team such as a urologist, gynaecologist, geneticist, and others.

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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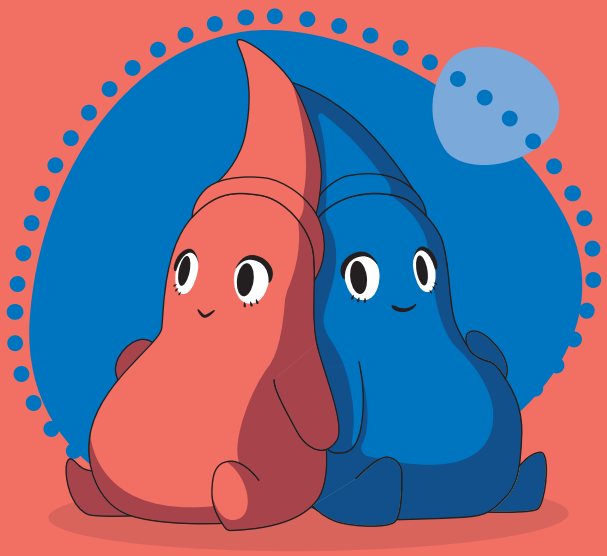

Remember

It is very important to increase doses of hydrocortisone during times of illness or injury and seek medical review early if unwell. Close review with your medical team is essential.

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