
Urinary system birth defects

Summary

- Common birth defects of the urinary system include hypospadias, obstructive defects of the renal pelvis and renal agenesis.
 - Hypospadias is characterised by the location of the urethral opening on the underside of the penis.
 - Obstructive defects of the renal pelvis prevent urine from entering the bladder.
 - Renal agenesis means one or both kidneys are missing.
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The urinary system consists of the kidneys, ureters, bladder and urethra. The human body has two kidneys (one on either side of the middle back) that filter wastes from the blood and produce urine. Each kidney has a tube called a ureter. Urine leaves the kidneys via the ureters and enters the bladder for temporary storage. The urethra is a tube that connects the bladder to the outside of the body, allowing urine to exit. Common birth defects of the urinary system include hypospadias, obstructive defects of the renal pelvis and renal agenesis.

Hypospadias

Hypospadias is a birth defect of the penis that commonly has four characteristics:

- The urethral opening is located on the underside of the penis, instead of the tip, and may exit the penis anywhere along its shaft as high as the scrotum.
- The urethral opening is unusually narrow.
- The entire foreskin may be bunched on the topside of the penis.
- The penis itself may be curved to one side.

Hypospadias is one of the most common birth defects in Victoria, occurring in around one in 300 births. Most often, hypospadias is noticed at birth; however, if the abnormalities are particularly mild, diagnosis may come later in life.

Causes of hypospadias

The causes of hypospadias are unknown. There seems to be a genetic association, since a baby boy with a family history of hypospadias is slightly more likely to be born with the condition.

Treatment for hypospadias

Hypospadias is treated with surgery, usually when the child is between six and 18 months old. The aims of surgery include repositioning the urethral opening at the tip of the penis, removing the abnormal foreskin (which gives a circumcised appearance), and correcting the bend in the penis (if it is present) to allow sexual function. Repositioning the urethral opening is relatively straightforward if the tube exits through the head of the penis. If the urethra exits higher on the shaft, a skin graft using the foreskin may be needed. It is important not to have your son circumcised before the hypospadias repair, in case the foreskin is needed.

Obstructive defects of the renal pelvis

An obstructive defect of the renal pelvis means that urine can't drain properly from the kidneys into the bladder. This is caused by a blockage in the ureters. One or both ureters may be affected. The urine backs up the ureters into the kidneys and, without treatment, can lead to persistent urinary tract infections and kidney failure. This common birth defect occurs in around one in 350 Victorian babies. Symptoms include recurrent urinary tract infections and may also include impaired growth. Other names for this condition include uteropelvic junction obstruction and pelvo-uterero junction obstruction.

Causes of obstructive defects of the renal pelvis

The exact causes of this defect are unknown, but genetic factors are thought to contribute. The structural abnormalities that block the passage of urine can include:

- Unusual twists or bends in the ureter
- A blood vessel pressing on the ureter
- Malformations of the surrounding muscle tissue.

Treatment for obstructive defects of the renal pelvis

Obstructive defects of the renal pelvis can be detected during pregnancy ultrasounds. A subsequent ultrasound of the baby is usually needed, once it is born, to confirm the diagnosis. It is important to correct this problem as soon as possible to maximise normal kidney function. The blockage is surgically corrected, usually with open surgery. Possible complications of surgery include infection and trauma to structures of the urinary system.

Renal agenesis

Renal agenesis means that one or both kidneys are missing. Renal agenesis and dysgenesis (malformation of the kidney) occurs in around one in 1,500 births in Victoria. If only one kidney is missing, the chances of survival are excellent, since the remaining kidney is capable of handling the workload of its absent partner. The situation is different for babies with both kidneys absent. Around four in 10 babies with both kidneys absent (bilateral renal agenesis) are stillborn, and the remainder die within a few hours of birth.

Other abnormal characteristics that may be associated with bilateral renal agenesis include:

- Absent bladder.
- Underdeveloped lungs (hypoplastic lung syndrome).
- Absent sex organ structures, such as the vas deferens and seminal vesicles in males, and the uterus in females.
- Absent rectum and anus (anorectal atresia).
- Gap in the oesophagus (oesophageal atresia).
- Malformations of the legs.

Causes of renal agenesis

During fetal development, the kidneys grow in three distinct stages. The initial structures called 'pronephros' are replaced by the 'mesonephros' structures. By the end of the first trimester, the 'metanephros' (fully functioning kidneys) are formed and producing urine. Renal agenesis is thought to occur during the last stage of kidney development, when the 'metanephros' buds fail to mature. The exact causes are unknown, but genetic and environmental factors seem to contribute. For example, a woman with untreated diabetes is at increased risk of having a baby with renal agenesis, among other birth defects.

Treatment for renal agenesis

If both kidneys are missing, the baby will not survive. A baby can manage with one functioning kidney, since the organ enlarges to cope with the extra workload.

Where to get help

- Your doctor
- Paediatrician.

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