Cloacal malformation

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of cloacal malformation and where to get help.

What is cloacal malformation?

Cloacal malformation is a congenital (present at birth) problem that only affects girls. Very early in pregnancy, the rectum, urethra and vagina fail to separate into separate tubes. This means that urine and faeces drain into a common channel opening in the perineum (the area where the anus and vagina are normally located). It occurs in 1 in 50,000 births and can be associated with other congenital malformations.

What causes cloacal malformation?

Cloacal malformation occurs very early in pregnancy, around five to six weeks after conception. We do not know what stops the rectal, urethral and vaginal tubes separating but it is unlikely to be caused by anything you did or did not do before conception or during early pregnancy.

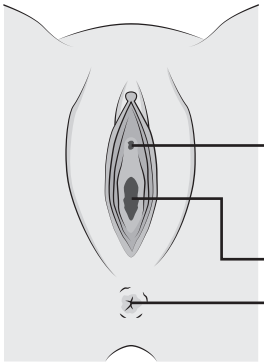
What are the signs and symptoms of cloacal malformation?

In cloacal malformation, there is a single drainage channel in the perineum instead of a separate rectum, urethra and vagina. Initially, an anorectal malformation (absent anus) may be suspected. The external genitalia may be small and the clitoris oversized, which may lead to an initial diagnosis of a disorder of sex development. On closer examination, a single opening in the perineum becomes apparent.

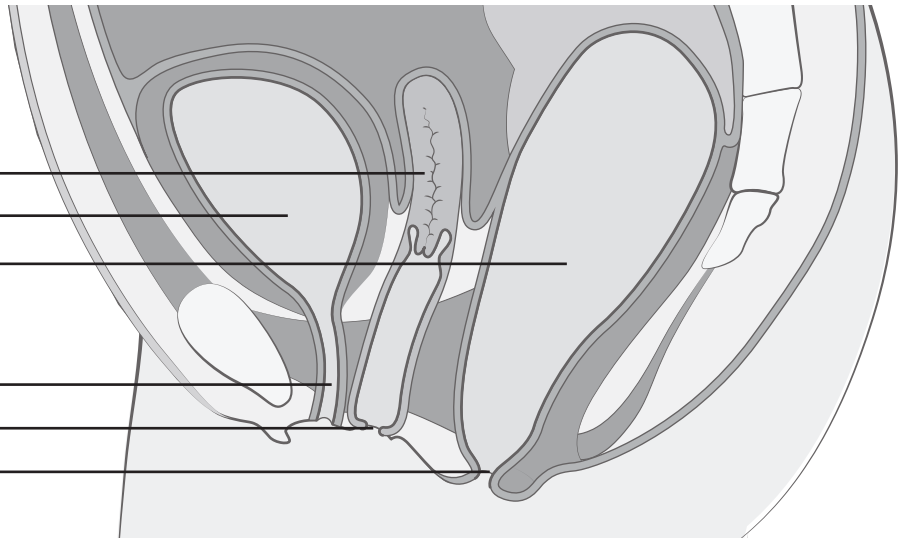
Cloacal malformation is associated with other malformations such as:

- Oesophageal atresia and tracheo-oesophageal fistula (OA/TOF) where the bottom end of the oesophagus is not developed or is joined to the trachea (windpipe).
- Structural heart problems.
- Structural problems with the lower spine.
- Kidney problems such as hydronephrosis, vesico-ureteric reflux, duplex kidney or ectopic ureters.

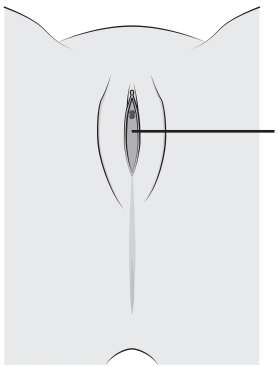
Normal external appearance of female perineum



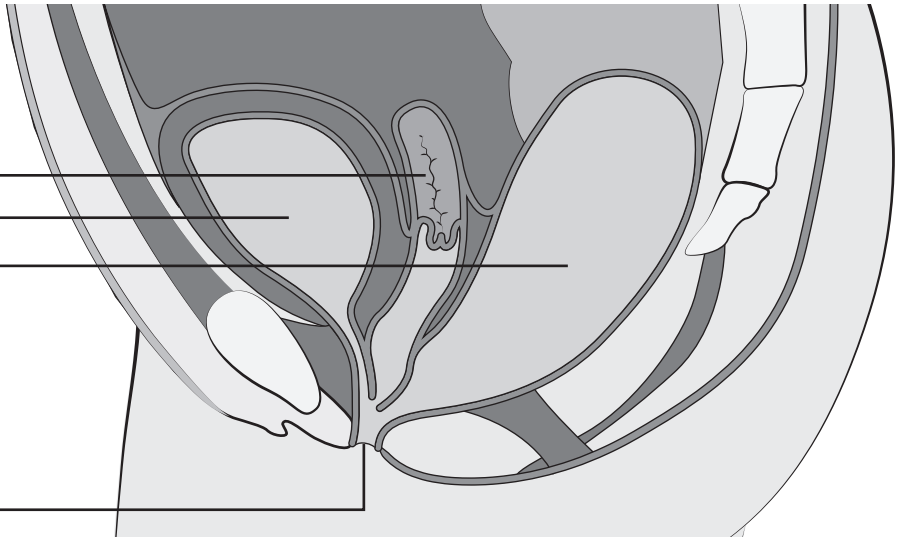
Normal internal structures



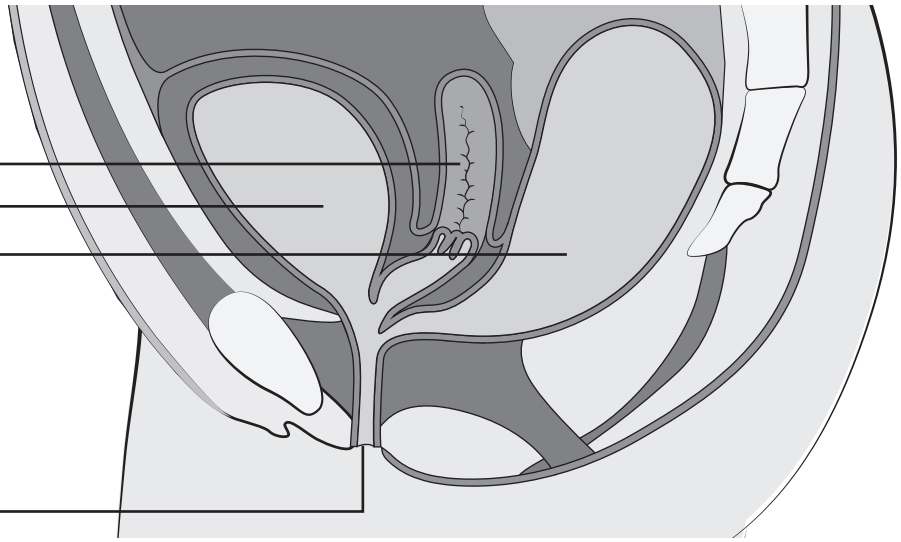
External appearance of female with cloacal malformation (single opening)



Short common channel cloacal malformation



Long common channel cloacal malformation



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How is cloacal malformation diagnosed?

Cloacal malformation can be suspected before birth (prenatally) on routine ultrasound, which may show bilateral (both sides) hydronephrosis – that is, the kidneys are swollen with urine because they cannot drain properly. The bladder may not be very clear on the scan and there may also be a cyst-like swelling in the abdomen. If cloacal malformation is suspected, repeat ultrasound scans will be carried out throughout pregnancy to monitor the problem.

Soon after birth, cloacal malformation may be suspected if the meconium (the dark faeces passed in the first few days of life) is not passed or it comes from a different place. Physical examination will confirm the diagnosis. Imaging scans, such as ultrasound and echocardiogram, will be used to identify any associated problems.

How is cloacal malformation treated?

Initial management

Immediately after birth, babies will need to be stabilised if there are any associated problems that are life-threatening. This may involve a stay in intensive care with breathing support and/or transfer to a specialist centre for treatment. For example, babies with oesophageal atresia will be transferred to GOSH for an operation soon after birth to enable them to feed safely.

They will usually have an intravenous (into a vein or IV) infusion of fluids as they will not be able to feed initially. They will also have antibiotics intravenously. A naso-gastric (NG) tube will be passed down the throat into the stomach to drain off any fluid and air to make the baby more comfortable.

A urethral catheter will be passed into the common channel to drain off fluid to reduce any swelling and potential damage to the kidneys. Rarely, a catheter may need to be inserted through the abdominal wall into the bladder if insertion into the common channel is not successful.

When the baby is stable, surgery to construct a stoma (artificial opening to the bowel) will be carried out so that faeces can be passed safely and feeds by mouth (breast or bottle feeding) can be introduced. The stoma will also allow the bowel to 'deflate' or shrink back to normal size as it will vent off excess air. For further information about the stoma formation operation, please see our information sheet. During the operation to create the stoma, the surgeons may examine the cloacal malformation more closely using a cystoscope – a tube containing a small camera and a light.

Children can usually be discharged home when the stoma has settled – usually after a stay of five to ten days in hospital. Regular check-ups will be needed until the next stage of treatment and all children will continue to have antibiotics to prevent infection affecting the kidneys. Regular blood tests will be continued to confirm kidney function. If urine is not draining freely from the bladder, catheterisation either through the common channel or through the abdominal wall into the bladder will be needed.

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Definitive reconstruction (cloacal repair)

Cloacal malformation is a complex problem, best dealt with at a specialist centre with input from both surgeons and urologists. Other members of the multidisciplinary team looking after children with cloacal malformation will usually include clinical nurse specialists, radiologists, nephrologists, gynaecologists and psychologists.

Two to three months after birth, the surgeons at GOSH will start to plan the next stage of surgery. This will involve further imaging scans, such as a loopogram – this uses contrast liquid, which shows up well on x-rays, inserted into the stoma to view the large intestine. Kidney function scans, such as a DMSA or MAG3 scan, will be suggested. Further tests such as a cystoscopy and cystogram will be required. Further information about these scans is available on our website.

The aim of the definitive reconstruction is to create three separate channels. Most operations will include a procedure to separate the channels and bring them to the surface in the perineum creating a new urethral, vaginal and anal opening. Your child will continue to use the stoma so that the bowel and anus can rest and heal. Children usually stay in hospital for three to seven days while they recover and heal from the operation.

Around six to eight weeks after the operation, the surgeon will close the colostomy by disconnecting the bowel from the stoma to allow your child to pass faeces through the anus. The urologist will also check the urethral and vaginal passages using a cystoscope at the same time.

After the series of operations, regular follow up appointments will be needed throughout childhood until puberty and beyond. These appointments will include imaging, such as ultrasound scans, blood tests, and urodynamics to monitor kidney, bladder and bowel function. Psychology input will be an integral part of their ongoing care.

What happens next?

The aim of surgery is to provide the potential for the child to achieve bowel and bladder control, protect kidney function and the best chance for normal sexual and reproductive function in later life. Most children achieve urinary control (remaining dry), but around half will require further treatment, such as additional surgery or bladder training. Many children achieve faecal or bowel control as well, but again some may need extra help such as bowel washouts or further surgery later in childhood.

Most girls will go through puberty at the expected age however some may not have periods (menses) or have difficulty with menstrual blood flow from the body. Further surgery may be needed in adolescence to enlarge the vaginal opening to facilitate this. Follow up with a gynaecologist (specialist in female reproductive problems) is essential for all girls approaching puberty. Pregnancy and childbirth may be possible, however extra support and monitoring will be needed throughout pregnancy and delivery is usually using a caesarean section.

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Kidney problems are relatively common in children who have had cloacal malformation, so regular kidney function tests and imaging will be needed throughout childhood and beyond. Around 60 per cent of children with cloacal malformation have kidney problems, some of whom may develop kidney failure needing dialysis or transplantation.

Further information and support

There is no support group specifically for those with cloacal malformation, but Contact a Family may be able to help. You can call their helpline on 0808 808 3555 or visit their website at www.cafamily.org.uk.

Notes

Compiled by the Specialist Neonatal and Paediatric Surgery and Urology departments in collaboration with the Child and Family Information Group

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