Undescended and Maldescended Testes

Normal testicular development in utero begins along the mesodermal ridge of the posterior abdominal wall. By 28 weeks, the right and left testes reach their respective inguinal canals and, by 28-40 weeks, each testis has usually reached the scrotum.

Classification

An undescended testis is a testis that is absent from the scrotum. The term cryptorchidism, from the Greek kryptos (hidden) and orchis (testicle), is also used.

Absence may be due to:
- Testicular agenesis (anorchia) - uncommon.
- Retractile testis.
- The ascending testis syndrome.
- Testicular maldescent.

Retractile testes[1]
- Prepubertal boys can have an exaggerated cremasteric reflex.
- The testis may retract out of the scrotum in the cold, on examination, on excitement or on physical activity.
- It is normal and will descend when relaxed and warm, or it can be manipulated back into the scrotum.
- Retractile testes do not need any treatment but do need close follow-up until puberty, as they can become ascendant.

The ascending testis syndrome
- A previously normal or a retractile testis can become high with a shortened spermatic cord that prevents the testis from staying in the scrotum.
- It is a rare condition which occurs more commonly on the left side.[2]
- It is usually diagnosed in those aged 8-10 years.

Maldescended testes
- These are usually unilateral.
- The scrotum may be underdeveloped.
- Maldescent may be due to an anatomical abnormality or due to hormone lack or hormone resistance. The release of testosterone from the fetal testis, the release of substances from an intact genitofemoral nerve and gonadotrophin hormone have all been cited as having possible involvement in normal testicular descent.
- Most undescended testes migrate into the lower scrotum within the first three months of life, presumably as a consequence of a postnatal testosterone surge, with less than 1% remaining undescended by 1 year of age.[3]
- Descent can be:
  - Arrested - where descent is along the normal path but incomplete. The testis may be located near the pubic tubercle, in the inguinal canal (80%), or, uncommonly, in the abdomen. The testis is often small and abnormal with a short spermatic cord. There may be associated inguinal hemia.
  - Ectopic - where descent deviates from the normal path. The testis is most often found in the superficial inguinal pouch. Perineal, abdominal, pelvic, crural, penile and femoral positions are also all possible. The testis and spermatic cord are usually normal.

Epidemiology[4]
- Undescended testis is the most common birth defect among boys.[3]
- Undescended testes affects 1-6% of males.
- There is a higher incidence in premature babies (up to 30%).
- Unilateral cryptorchidism is four times more likely than bilateral.

Aetiology
- The aetiology of cryptorchidism is multifactorial (genetic, maternal and environmental factors).
- However, it occurs most often as an isolated disorder with no obvious cause.

Diagnosis[5]
- This is by physical examination.
- Around 70% of all undescended testes are palpable.
- It can be difficult to distinguish undescended testes from retractile testis.[3]
Imaging or ultrasound does not add any benefit to differentiating between palpable and non-palpable testes.

Examination should take place while the child is supine and in a cross-legged position. Cover the following steps:

- Perform a visual examination of the scrotum.
- Inhibit the cremasteric reflex with one hand above the symphysis in the groin region before touching the scrotum.
- 'Miking' of the groin region towards the scrotum may help to move the testis into the scrotum. It can also help to differentiate between an inguinal testis and enlarged inguinal lymph nodes.
- A retractile testis can usually be moved into the scrotum and will remain there until it retracts back into the groin again with a cremasteric reflex (eg, touching the inner thigh).
- Look at the femoral, penile and perineal region for ectopic testes.
- Diagnostic laparoscopy is usually the preferred method to confirm or rule out an intra-abdominal, inguinal or absent/vanishing testis (non-palpable testis). However, an examination under anaesthetic is often carried out before laparoscopy, as a previously non-palpable testis may become palpable.
- Abdominal and pelvic ultrasonography may be required if intersexuality is suspected.

Guidelines from the European Association of Urology (EAU) [5]
The EAU's Paediatric Urology guidelines suggest that undescended and maldescended testes should be categorised into palpable and non-palpable testes, as the location and existence of the testis affects clinical management.

Bilateral, non-palpable testes and any suggestion of sexual differentiation problems (eg, hypospadias) require urgent, mandatory endocrinological and genetic evaluation.

Syndromes associated with cryptorchidism

- Prader-Willi syndrome.
- Kallmann's syndrome.
- Laurence-Moon syndrome.
- Intersexuality/congenital adrenal hyperplasia.
- Prune belly syndrome.

Management [5]

- If, by the age of one year, descent has not occurred, spontaneous descent is unlikely. Treatment should be initiated, as there is also potential for histological deterioration and loss of testicular quality (may affect future fertility).
- If there is unilateral undescended testes still present at 3 months then the child should be referred to an appropriate paediatric surgeon, ideally before the age of 6 months. [6]
- The ideal management of cryptorchidism is a highly debated topic within the field of paediatric surgery.
- Treatment should be completed by 12-18 months of age.
- However, despite early diagnosis in many patients with undescended testes, many are still referred and operated after 1 year of age. [4]

Medical treatment

- Testicular descent is hormonally dependent.
- Treatment with human chorionic gonadotrophin (hCG) or gonadotrophin-releasing hormone (GnRH) can be used.
- Success rates are best the lower the undescended testis is located.
- Maximum success rates are 20%.
- Medical treatment may be useful before or after surgery and may have a beneficial effect on later fertility.
- However, hormonal treatment is not usually recommended anymore. [7]
- Side-effects of hCG treatment can include enlargement of the penis, pubic hair growth, increased testicular size and aggressive behaviour during treatment.
- While some authors suggest that hormonal treatment increases the number and maturation of germ cells in cryptorchid testes, others believe the opposite. [8] Studies have shown that hCG treatment may decrease sperm counts together with the future fertility potential. [9]

Surgical treatment

- The ideal management of cryptorchidism is still a highly debated topic within the field of paediatric surgery. Orchiopexy before 10-11 years may protect against the increased risk of testicular cancer associated with cryptorchidism. Orchiopexy should not be performed before 6 months of age, as testes may descend spontaneously during the first few months of life. [10]
- If the testis is palpable: an inguinal approach is usually used. Orchiopexy or orchidofuniculolysis (mobilisation of the testis and cord) can be performed. Success rates are up to 92%. [11] Orchiopexy involves mobilisation of the testis on its essential structures (the vas, the testicular vessels and the spermatic cord) so that the testis can be brought down into the scrotum. The testis may also be fixed within the scrotum. Early surgical intervention in infancy may allow the normal development of stem cells for spermatogenesis. Some experts recommend that orchiopexy be performed between 6 and 12 months of age to maximise the future fertility potential and reduce risk of testicular cancer. [12]
- If the testis is non-palpable: examination under anaesthetic may reveal the previously non-palpable testis. The role of laparoscopy in the case of non-palpable cryptorchidism is both diagnostic and therapeutic. Laparoscopic orchiopexy for non-palpable testes is usually the preferred surgical approach among paediatric urologists. [13] Removal, orchidolysis or orchidopexy can then be performed laparoscopically.
If a prepubertal boy is aged 10 years or older and there is a normal contralateral testis, an intra-abdominal testis should be removed. If he is younger than 10 years, or there are bilateral intra-abdominal testes, an attempt at moving the testis/testes into the scrotum should be made using special surgical techniques.\(^5\)

**Complications**

- Increased risk of testicular torsion. This may be associated with the development of a testicular tumour. Torsion of an intra-abdominal testis may present as an acute abdomen.
- Increased risk of testicular trauma.

**Effect on fertility\(^5\)**

- Boys with one undescended testis have a lower fertility rate but the same paternity rate as boys with bilateral descended testes.
- Boys with bilateral undescended testes have lower fertility and paternity rates.\(^{12}\)

**Risk of testicular malignancy**

- There is a three-fold increase in the incidence of testicular cancer in males with undescended testes.\(^{14}\)
- There is a history of cryptorchidism in 5-10% of testicular cancers.\(^5\)
- Prepubertal orchidopexy for cryptorchidism may be associated with a lower risk of testicular cancer.
- Orchidopexy facilitates testicular self-examination.

**Cosmetic appearance**

- Surgical transfer of the testis into the scrotum produces a better cosmetic appearance.
- Prostheses may be used if the testis is removed. Prostheses should be implanted during adolescence.

**Prevention of complications**

The National Screening Committee Policy - 'cryptorchidism screening' - states that screening for undescended and maldescended testes should take place in the routine physical examination of boys within 72 hours of birth and at the six- to eight-week check.\(^5\)

**Further reading & references**

5. Guidelines on Paediatric Urology; European Association of Urology (2015)
6. Undescended testes; NICE CKS, August 2014 (UK access only)
15. Cryptorchidism: The UK NSC policy on cryptorchidism screening in newborn boys; UK National Screening Committee Policy Database, 2012

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Dr Louise Newson

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