Scottish Differences of Sex Development (SDSD)
Bilateral Impalpable Testes Pathway

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Document control

Key personnel

Title:	Scottish Differences of Sex Development (SDSD) Pathway for the management of Bilateral Impalpable Testes picked up on routine newborn examination.
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Version history

Version	Date of revision	Summary of changes
0.1		First draft prepared by Martina Rodie
0.2	26 October 2020	Revisions following Steering Group
V1	March 2021	

Distribution

Name	Organisation
Mr Phil McNicol & Ms Alex Brown	NHS National Services Scotland
Dr Martina Rodie	Lead Clinician, NHS Greater Glasgow & Clyde
SDSD Steering Group	Various

Introduction

Bilateral undescended testes usually result from delayed testicular descent in an otherwise healthy male infant, but can (rarely) represent a serious underlying condition (e.g. congenital adrenal hyperplasia or abnormality of the gonads). Full clinical assessment in the early newborn period is essential, in order to identify potential problems, and also to avoid worrying parents unnecessarily.

If a baby presents only with palpable but incompletely descended testes or with an isolated unilateral undescended testis, send a letter to the GP and ask them to re-check at the 6 week check and refer to Paediatric Surgery or Urology if warranted.

Undescended testes are more common in premature infants so do consider the gestational age of the baby, it is still worth highlighting if the testes are impalpable and ensuring they are examined again once closer to term corrected age.

If the testes are not palpable – this should be confirmed by the most experienced clinician available. The testes may be high but palpable, and it may be possible for an experienced person to feel them in the inguinal region. Paediatric surgeons are particularly skilled at this. Ensure the baby is not crying while being examined and consider the use of sucrose to calm the baby and lubricating gel to slide fingers along the inguinal canal.

If both testes are truly impalpable, please notify the Neonatal and Paediatric Endocrine Consultants. Do not discharge the baby until they have been discussed with a Consultant.

The immediate aim is to exclude Congenital Adrenal Hyperplasia (CAH) before an affected infant runs into a salt losing crisis. CAH is very rare but can be life-threatening if missed.

1) Look for, and document the following:

- birth weight centile
- results of antenatal investigations eg amniocentesis for karyotype
- length of phallus (cm) - should be at least 2.5cm
- position of urethral opening
- appearance and position of scrotum, and whether bifid
- any other midline opening

Presence or absence of

- pigmentation
- other congenital abnormalities/dysmorphic features
- parental consanguinity

2) Perform investigations:

a) Request pelvic USS for as soon as possible (but no longer than 7 days): to view gonads and for presence of any internal structures (uterus).

b) Send blood urgently to the genetics laboratory for QF-PCR / FISH / karyotype / microarray. A taxi may need to be organized in order to get the sample there promptly.

c) Send blood urgently to the biochemistry laboratory for UEs, glucose, cortisol, 17-OH progesterone, anti-Mullerian hormone, FSH, LH, ACTH (these tests should be performed >36 hours of age).

d) Send a urine steroid profile to the biochemistry laboratory.

3) Ensure the baby remains clinically well:

If the baby is well with no evidence of electrolyte derangement (high potassium and/or low sodium), stable blood sugars and no vomiting then they can be considered for discharge pending results of the above investigations. Make sure that parents are given robust worsening advice and know who to contact should concerns arise. The Neonatal and Paediatric Endocrine Consultants should be aware if the baby is being discharged.

4) Ensure follow up is in place:

The majority of the results should be available within 48-72 hours – a clinician who will review the results promptly needs to be clearly identified. Take a note of the parents' telephone numbers so that the results can be communicated via telephone and the family can be asked to re-attend immediately if required.

If the baby truly has undescended testes, refer to Paediatric surgeons, as there is an increased risk that the testes may not descend spontaneously.

If any of the results are concerning or indicate CAH, discuss with the on call Paediatric Endocrine Consultant immediately even if out with normal working hours.

What to say to parents if genitalia are otherwise typically male, but neither testes palpable:

1. It is likely that the testes will come down on their own over time, but your baby will be reviewed by the paediatric surgical team to monitor this.

2. In the mean time, we need to exclude a rare (and treatable) problem of the adrenal gland that can give this appearance and cause salt loss - this will involve a blood and urine test today (if your baby is over 36 hours old). The blood test measures salt levels, and hormones to assess how the adrenal gland is working.

3. We will also arrange for an ultrasound scan to look for the testes that we cannot feel, and for the presence of internal sex organs.

4. For the testes to make male hormones, male sex chromosomes are required, and we would like to send blood today to look at those chromosomes. Chromosomes are the genetic packages in every cell of the body. Usually sex chromosomes show a typical male pattern (XY), but occasionally the chromosome pattern can be more complex.

We will arrange a plan for contacting you with these results, and this plan should be made clear to you at the time the sample is taken.

It is also important that you know who to contact/where to go if you have any questions, or if you are concerned about your baby at any time.