Clinical Guideline

CONGENITAL PENILE PATHOLOGY

SETTING	Primary and Secondary Healthcare Institutions in the Western Cape which refer to Red Cross War Memorial Children's Hospital (RCWMCH)
FOR STAFF	Primary and Secondary Doctors and Nurses who refer patients to RCWMCH Doctors in nurseries and NICUs who refer patients to RCWMH
PATIENTS	Patients under the age of 13 years old with congenital penile pathology

Introduction

Significant congenital problems of the penis will most often be identified at birth when the patient undergoes a full neonate head-to-toe examination. In some cases, conditions can become more obvious as the genitals grow leading to presentation at a later age.

It is often a very emotional experience for parents to find that their child has an anomaly of the penis. For this reason, sensitivity and clear counselling on what to expect is valuable.

It is the intension that this guideline will assist doctors and nurses to correctly identify anomalies of the penis, counsel parents on what they can expect next and then make the appropriate referral to the urology services.

Background

There is a time in early foetal development when the foetal genitalia have not yet started to differentiate into the male or female phenotype. There are multiple factors that influence this process of differentiation, including anatomical considerations, hormonal control and local messaging systems.

In some patients, sexual differentiation is not complete and this results in an atypical appearance of the genitalia. The implication of this may be extreme, as in the case of ambiguous genitalia – where there is no clear male or female pattern to the appearance of the external genitalia. In other cases, the gender of the patient might be clear but the appearance of the phallus is abnormal.

Important details on history

When was the anomalies identified?
Is there a family history of similar abnormalities?
Has the patient been passing urine freely?
Did the mother have any unusual exposures during pregnancy?
Does the patient have any other medical conditions?

Important details on examination

Confirm the patient is otherwise well with no other features of dysmorphology.

Inspect the perineum:

- Confirm whether you are able to see where the urethral meatus is.
- Are there palpable gonads and where are they positioned?
- Assess if there is a urogenital sinus present.

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- Confirm the anus is patent

Making the diagnosis

Diagnosis	Clinical features
Buried penis or congenital Megaprepuce.	Normally developed penis that appears to be hidden by foreskin tissue.
Hypospadias	1. Abnormal opening of the urethral meatus on the ventral side of the of the penis, the scrotum or perineum 2. Abnormal foreskin called a dorsal hood 3. Varying degrees of abnormal angulation of the penis called chordee
Epispadias	Abnormal opening of the urethral meatus on the dorsal side of the penis
Ambiguous genitalia	An external manifestation of a possible disorder of sexual development (DSD)

Buried Penis or Congenital Megaprepuce

This is a normally developed penis that appears to be hidden by pre-preputal tissue. Parents will usually describe the following concerns:
- Ballooning of the preputial tissue upon urination which can be as large as a tennis ball
- Constant dribbling of urine
- A need to express urine from the preputial space to relieve build up
- These patients are at risk for recurrent infections & dysuria

The main causes seem to be inelasticity of the dartos fascia or a very large congenital Megaprepuce which causes a valve effect at the level of the foreskin.

Some conditions which can be mistaken for this are a tight ring secondary to neonatal circumcision or a large suprapubic fat pad in a chubby baby that hides the penile shaft.

Referral guidance: Non urgent urology OPD review.

Parental counselling:

There are various surgical techniques for correction with varying cosmetic outcomes. The most important thing that all the operative techniques have in common is that the foreskin is opened to allow urine to pass freely through the tip of the foreskin and no longer pool behind the foreskin.

Hypospadias

Three Components
1. Abnormal opening of the urethral meatus on the ventral side of the of the penis, the scrotum or perineum
2. Abnormal foreskin called a dorsal hood
3. Varying degrees of abnormal angulation of the penis called chordee

The scrotum and testis should be examined, looking for:

- A bifid scrotum
- Penoscrotal transposition
- Undescended testis

Referral guidance: Non urgent urology OPD review.

Parental counselling:

- Parents should be reassured that this is not a life threatening condition and that there are surgical options to correct the defect.
- The treatment is not urgent and repair usually takes place between 6 and 18 months of age
- The patient should NOT be circumcised

Epispadias

This is the abnormal opening of the urethral meatus on the dorsal side of the penis. It is a rare defect. It may occur in isolation but can be seen as part of the exstrophy- epispadias complex.

In the case of exstrophy-epispadias complex, the anterior bladder wall has not fused and the patient presents with an open bladder plate on the abdominal wall. This is considered a urological emergency and the bladder closure is usually attempted within 48 hours of birth when possible.

Referral guidance:

Isolated epispadias: Non urgent urology OPD review
Newborns with exstrophy-epispadias complex: Urgent referral to the urologist on call for possible early closure.

Parental counselling:

- In epispadias: Parents should be reassured that this is not a life-threatening condition and that there are surgical options to correct the defect.
- The treatment is not urgent and repair usually takes place between 6 & 12 months of age
- The patient should NOT be circumcised

- In exstrophy- epispadias complex there is urgency to close the bladder where possible to allow it to grow more normally and to prevent damage to the inside surface of this structure.

Ambiguous Genitalia

Ambiguous genitalia are an external manifestation of a possible disorder of sexual development (DSD). In some patients a phenotypic gender may not be possible to identify while some may appear phenotypically male or female.

A phenotypic male may appear under virilized, with severe peno-scrotal or perineal hypospadias, a bifid scrotum and undescended testes.

A phenotypic female may appear virilized with features of a large clitorophallus, rugae and pigmentation of labia majora and urogenital sinus (single opening for the urethra and genital tract) and palpable gonads.

Congenital Adrenal Hyperplasia (CAH) may present with ambiguous genitalia as females with this condition appear over-virilised. CAH is a true medical emergency because these patients have a salt-wasting syndrome that must be urgently addressed. They can develop dehydration, hyponatraemia and hyperkalaemia which may be life-threatening.

Management guidance

Patients with ambiguous genitalia should at the very least have electrolytes taken to evaluate for possible CAH. More extensive bloods may be requested by the endocrinology team who will be taking responsibility for the patient.

Referral guidance

Patients with possible CAH should be referred to neonatal specialists and paediatric endocrinology emergently.

Patients with ambiguous genitalia should be referred to a paediatric endocrinology team with experience in the management of DSD. This team will perform the necessary work-up and refer to a paediatric surgical or urological service as necessary thereafter.

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