Genitourinary Issues in CdLS

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Genitourinary (GU) manifestations are commonly found in patients with Cornelia de Lange Syndrome (CdLS). These can encompass anatomic and functional anomalies of the kidney, urinary tract, reproductive organs, and external genitalia. Reflux of urine into the kidneys, improper drainage of the kidneys, and abnormal development are the most common abnormalities of the urinary tract, and can lead to a decline in renal function and recurrent infections. For males, undescended testis is the most common anomaly of their external genitalia, which can affect fertility and increase their risk of developing testicular cancer. Hypoplastic (underdeveloped) genitalia and hypospadias, where the urethral opening is located on the underside of the penis rather than at the tip, can also be present. Similarly, females with CdLS can have small external genitalia, abnormal uterus, and ovarian atrophy (decreased size/function).

The first step in management is a detailed family, medical, and surgical history. This can aid in identifying genetic or pre-existing risks factors for development of a concomitant GU disease. A review of systems should focus on a detailed analysis of urinary and bowel habits, nutritional habits, and abdominal or groin pain. The physical exam should assess the abdomen for fullness or masses, curvature and size of the penis, position of urethral opening, circumcision status, asymmetry or discoloration of the scrotum/labia, location and ability to examine the testicles, location and patency of the vagina, patency of the anus, and lower back anomalies. As an infant, ultrasound of the bladder and kidneys should be completed to help detect any GU anomalies which may prompt further imaging. Ultrasonography is an ideal initial study as it is non-invasive, cost effective, and does not expose the child to any radiation.

A number of these exams can be accomplished at home during routine care for the patient. If hypospadias is a concern, observing the patient while voiding can help locate the urethral opening. A testicular exam is usually best accomplished in a warm bath if presence of testes within the scrotum is questionable. Any concerns should be brought to the attention of your primary care physician.

If an undescended testis is diagnosed at birth, it is observed for positional changes during the first 6 months of life to allow for spontaneous testicular descent. After six months of age, surgical treatment by a pediatric specialist is encouraged. Since hypospadias does not

routinely affect the patient's ability to empty their bladder, surgical repair is not recommended until at least six months of age and may require multiple procedures depending on its severity. If renal or bladder anomalies are detected, the patient may be placed on antibiotics to prevent infections or require future interventions.

By increasing awareness of the GU manifestations that can present in CdLS, early detection and appropriate management can be better achieved. There are many other, more rare diagnoses related to the GU system that the patient may develop. Therefore, it is imperative to be vigilant to any change in the patient's exam or symptoms which may warrant a formal evaluation with a board certified pediatric urologist.

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