Chapter 20

Genitourinary Complications

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Introduction

Several genitourinary complications have been reported in patients with classic
dyskeratosis congenita (DC), but there are limited data on their incidence or the
extent to which they affect other related telomere biology disorders (TBDs). A
review of the United Kingdom based DC registry found that 5% of males with DC
had urethral stricture and/or phimosis [1]. In a recent analysis of males with DC at
the National Cancer Institute, 10.5% had a history of urethral strictures [2]. While
there are a few anecdotal literature reports on kidney abnormalities in DC patients,
on review of DC cases registered with the NCI no clear DC associated renal
disease was evident [2, 3, 4].
The management of genitourinary complications in individuals with DC should start with a detailed clinical evaluation based on the patient’s symptoms and consultation with subspecialists as needed.

**Males**

**Urethral Strictures**

Urethral strictures occur most commonly in males. This narrowing of the urethra can occur at any location along the urethra. Men and boys with symptomatic urethral strictures typically present with obstructive voiding symptoms including straining with urination, incomplete bladder emptying, and a narrow or weak urine stream [5]. Patients may have a history of hematuria, frequent urinary tract infections, prostatitis, epididymitis, or bladder stones [6, 7]. The diagnosis of urethral stricture should be made in consultation with a urologist [5, 8]. Imaging studies, such as retrograde urethrography, voiding cystourethrography, or cystoscopy, may be used to determine the location and extent of the stricture.

The pathogenesis of urethral stricture in males with DC/TBDs is not known, but it is hypothesized to be due in part to the limited replicative capacity of the cells in these individuals. In individuals without DC who develop urethral strictures, the normal pseudostratified columnar epithelium of the urethra is replaced by squamous metaplasia. The same mechanism is likely in DC/TBDs, but this has not been studied.

Treatment is determined by the degree of symptoms and location of the narrowing [5, 9, 10] and should be managed by a urologist with experience treating complex urethral strictures. Urethral dilation can be performed for relatively short strictures.
Urethroplasty is used for strictures of the anterior urethra, while meatotomy is the preferred treatment for meatal stenosis.

Phimosis

Phimosis is a relatively common condition in which the foreskin cannot be retracted over the glans penis. The foreskin is not normally retractile in infants but gradually becomes so in childhood [11]. Scarring of the foreskin can result in phimosis, which in turn can cause difficulty in urination, balanitis, or urinary tract infections. Topical corticosteroids may be used for local irritation. Antibiotics may be required for infections.

It is not known if phimosis occurs more often in individuals with DC/TBDs than in the general population. It is thought that it will likely respond to the same management as for individuals without DC, but this has not been systematically investigated.

Hypogonadism

Hypogonadism, manifesting as reduced testosterone production, has been reported in a small number of patients with DC/TBDs. This is further reviewed in Chapter 22, Endocrine and Skeletal Complications.

Females

There are anecdotal reports of labial adhesions, and hymenal and urethral strictures in females with DC. Incidence of these complications in DC/TBDs is not known. However, these should be considered in girls and women with DC/TBDs who have frequent urinary tract infections, difficulty urinating, or abnormal menstrual bleeding. Labial leukoplakia has also been noted in some women with DC/TBDs, but its contribution to adhesions or strictures is not understood. Women and girls with DC suspected of having these complications should be referred to a gynecologist and/or urologist with Telomere Biology Disorders Diagnosis and Management Guidelines, 2nd Edition, available at teamtelomere.org
experience in treating these conditions. Additional information can be found in Chapter 21, Gynecological and Obstetric Considerations.

References


