Down syndrome and Urology

Introduction

- Trisomy 21, which appears in approximately 1/1000 live births.
- 50% survival at 50 years of age.
- Extraurological abnormalities: heart problems in up to 50% of cases, eye diseases in 60%, hearing impairment in 75%, sleep apnea in 75%, gastrointestinal atresia in 12%, and thyroid disease in 15%.

Kidney abnormalities

- Renal hypoplasia: in 20% of cases there is a significant reduction in nephron mass.
- Glomerular microcysts: in over 20%. Their clinical impact is unknown.
- Obstructive uropathy: in 7%.
- Simple renal cysts: in 6%, with cystic dysplasia in 3%.
- Glomerulonephritis, especially of the mesangiocapillary type.
- Decreased creatinine and uric acid clearance.

Ureteral abnormalities

- Obstructive megaureter.
- Ureteral atresia.
- Vesicoureteral reflux, usually high grade and with a poor prognosis.

Vesicourethral abnormalities

- Emptying voiding dysfunction.
- Posterior urethral valves: with a greater incidence than that of the general population.
- Hypospadias: increased incidence.

Genital abnormalities

- Cryptorchidism in at least 25% of patients.
- Hypogonadism: with increased FSH and LH, although testosterone and estradiol levels are usually normal in men and women, respectively.
- Testicular cancer: occurs more frequently than in the general population, linked to higher incidences of cryptorchidism and hypogonadism. The most common histological type is seminoma.
- Secondary sex characteristics tend to be fairly normal, although with a slight decrease in the size of the penis and testicles.
- Changes in spermatogenesis and oligospermia: although there have been no reported cases of paternity, cases of women with Down’s syndrome giving birth have been described.

Urological recommendations for patients with Down’s syndrome

- Prophylactic circumcision in patients with heart valve disease to avoid complications from bacteriemia linked to urinary tract infections.
- Monitoring of cryptorchidism to prevent the occurrence of testicular cancer.
- Clinical evaluation of ureteral-renal abnormalities and voiding dysfunction.
- For patients undergoing surgery: an echocardiographic evaluation is advisable. Anesthetic intubation presents 3 problems: the high incidence of infraglottic stenosis, the invariable presence of macroglossia, and the fact that up to 17% of patients with Down’s syndrome exhibit vertebral atlanto-axial instability, requiring extra care when manipulating the neck.