

POINTS TO REMEMBER FOR CLINICAL PRACTICE

Endocrine
manifestations

Genital Functions

- There is primary hypogonadism in male subjects with DS from early childhood.
- Congenital or acquired, palpable or non-palpable undescended testis, is common and should be treated surgically before the age of 12 months, and by 18 months at the latest.
- A micropenis is common and requires an endocrinology assessment.
- The age of onset of puberty does not differ from that of the general population, but it is more rapidly established.
- Clinical and/or ultrasound monitoring of the testes is essential due to the high risk of testicular germ cell cancer.
- Fertility in men with DS is variable and the risk of DS transmission is very low; fertility in women with DS is normal and the risk of DS transmission is high (1/3).
- Menopause is early, correlated with the onset of Alzheimer's disease and the need for HRT.