

Opis choroby *

Definicja

A rare 46,XY disorder of gonadal development characterized by congenital complete absence of testicular tissue in an individual with an otherwise normal male phenotype and normal karyotype. In addition, a small penis is a frequent finding in anorchid patients. Typical hormonal characteristics are elevated basal levels of gonadotropins (especially FSH), low concentration of testosterone, and lack of increase of plasma testosterone in response to hCG administration. The GnRH stimulation test induces a prolonged increase in FSH and LH levels.

Dane

Klasyfikacja

Wada morfologiczna

Synonimy

Bilateral anorchia

Anorchia obustronna

Kod ORPHA

325124

Kod OMIM

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Kod ICD10

Q55.0

Kod ICD11

LD2A.2

*Źródło

orphanet