

Opis choroby *

Definicja

A rare congenital hypogonadotropic hypogonadism characterized by hypogonadism due to selective deficiency of follicle stimulating hormone (FSH). Clinical manifestations are primary amenorrhea, absent or incomplete breast development, and infertility in women, and small testes, azoospermia, and infertility in men. Luteinizing hormone is elevated in the gonadotropin-releasing hormone stimulation test, while the FSH response is impaired.

Dane

Klasyfikacja

Choroba

Synonimy

Isolated FSH deficiency

Izolowany Niedobór FSH

Kod ORPHA

52901

Kod OMIM

229070

Kod ICD10

E23.6

Kod ICD11

5A61.0

*Źródło

orphanet