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

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*Źródło

orphanet