

Opis choroby *

Definicja

A rare, genetic disorder of sex development characterized by primary amenorrhea, short stature, delayed bone age, decreased levels of estradiol, elevated levels of follicle-stimulating hormone and luteinizing hormone, absent or underdeveloped uterus and ovaries, delayed development of pubic and axillary hair, and normal 46,XX karyotype.

Dane

Klasyfikacja

Choroba

Kod ORPHA

444048

Kod OMIM

616185

Kod ICD10

Q96.8

Kod ICD11

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*Źródło

orphanet