Opis choroby *

Definicja

A rare endocrine disorder characterized by primary hypogonadism and partial alopecia. Females present with Mûllerian hypoplasia, absent or streak ovaries, hypoplastic internal genitalia, primary amenorrhea, and sparse or absent axillary and pubic hair. Some patients also presented sparse eyebrows, microcephaly, flat occiput, dorsal kyphosis or mild intellectual disability. The only described male presents with germinal cell aplasia. Affected individual all present partical scalp alopecia.

Dane

Klasyfikacja
Choroba

Synonimy
Al Awadi-Farag-Teebi syndrome
Zespół Al Awadi, Farag i Teebi

Kod ORPHA
2232

Kod OMIM
241090

Kod ICD10
E29.1

Kod ICD11
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*Źródło

orphanet