

## Opis choroby \*

### Definicja

A rare endocrine disorder characterized by primary hypogonadism and partial alopecia. Females present with Mùllerian 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 partial 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