

Opis choroby *

Definicja

Lung fibrosis-immunodeficiency-46,XX gonadal dysgenesis syndrome is characterised by immune deficiency, gonadal dysgenesis and fatal lung fibrosis. So far, it has been described in two sisters born to consanguineous parents. Both karyotypes were normal female (46,XX). No genetic anomalies could be identified by comparative genome hybridization analysis of their genomes or by analysis of genes known to be associated with these types of anomalies.

Dane

Klasyfikacja

Choroba

Kod ORPHA

137631

Kod OMIM

611926

Kod ICD10

D82.8

Kod ICD11

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*Źródło

orphanet