

Opis choroby *

Definicja

Shwachman-Diamond syndrome (SDS) is a rare multisystemic syndrome characterized by chronic and usually mild neutropenia, pancreatic exocrine insufficiency associated with steatorrhea and growth failure, skeletal dysplasia with short stature, and an increased risk of bone marrow aplasia or leukemic transformation.

Dane

Klasyfikacja

Choroba

Synonimy

Pancreatic insufficiency and bone marrow dysfunction

Niewydolność trzustki i dysfunkcja szpiku kostnego

SDS

Zespół Shwachmana

Zespół Shwachmana, Bodiana i Diamonda

SDS

Shwachman syndrome

Shwachman-Bodian-Diamond syndrome

Kod ORPHA

811

Kod OMIM

617941

Kod ICD10

D61.0

Kod ICD11

3A70.0

*Źródło

orphanet