

## 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

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#### \*Źródło

orphanet