

DISEASE:
Shwachman-Diamond syndrome

NAME:	Shwachman-Diamond syndrome
DESCRIPTION:	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.
ORPHACODE:	811
SYNONYMS:	Pancreatic insufficiency and bone marrow dysfunction SDS Shwachman syndrome Shwachman-Bodian-Diamond syndrome
XREF(S):	Orphanet MedDRA ICD-10 OMIM OMIM

ANALYTE(S):	<u>SBDS</u> <u>EFL1</u> <u>SRP54</u> <u>DNAJC21</u>
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