Shwachman-Diamond syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Shwachman-Diamond syndrome. ORPHA:811

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.