

Shwachman syndrome is a rare genetic disorder with multiple and varied manifestations. The disorder is typically characterized by signs of insufficient absorption (malabsorption) of fats and other nutrients due to abnormal development of the pancreas (pancreatic insufficiency) and improper functioning of the bone marrow (bone marrow dysfunction), resulting in low levels of circulating blood cells (hematologic abnormalities). Additional characteristic findings may include short stature; abnormal bone development affecting the rib cage and/or bones in the arms and/or legs (metaphyseal dysostosis); and/or liver abnormalities.