

What is the name of the autosomal disorder associated with exocrine pancreatic insufficiency, cyclic neutropenia and skeletal defects?

ANSWER: Shwachman-Diamond syndrome

Shwachman-Diamond syndrome is an autosomal disorder characterized by exocrine pancreatic insufficiency, hematologic abnormalities including cyclic neutropenia, skeletal defects, short stature and normal sweat electrolyte values. Johanson-Blizzard syndrome, also autosomal recessive, features nasal alar hypoplasia, hypothyroidism, pancreatic achylia, and congenital deafness. Pearson's Marrow-Pancreas syndrome is an autosomal dominant mitochondrial DNA breakage syndrome, characterized by sideroblastic anemia and exocrine pancreatic dysfunction. Iverson's syndrome features asplenia with cystic liver, kidney and pancreas. Pancreatic agenesis syndrome results in intra-uterine growth retardation, insulin-dependent diabetes and pancreatic exocrine insufficiency.