

Letters to the Editors

SCHWACHMAN DIAMOND SYNDROME: CT APPEARANCES

Sir,

A seven yrs old female child presented with recurrent diarrhea since three years. She was short in height for the age (2' 8") and gave history of recurrent infections of the respiratory tract, paranasal sinuses and mastoids. She was underweight, weighing 20 Kgs. On general examination, she was emaciated and pale with minimally protruding abdomen. Skin was dry with a few petechia over the back. Minimally reduced tone in all four limbs was noted. On laboratory investigations, hemoglobin was 8.9 gm% with neutropenia (WBC count of 1500 cells/mm³), thrombocytopenia (platelet count of 70,000 cells/mm³) and hypochromic anemia (RBC counts of 3.2 mill./mm³), signifying pancytopenia. Fetal hemoglobin, SGPT and SGOT levels were minimally elevated. S. albumin levels were minimally reduced. 72-hour fecal fat measurements revealed an increase in fecal lipids and fatty acids. CD3+/CD4+ cell counts were suppressed. Skeletal survey revealed minimal changes of bronchitis on chest radiograph and minimally reduced bone age (medial epicondyle of lower end of humerus not yet ossified, which ossifies between 5-7 years). Minimal osteopenia was observed in almost all the bones. CT scan of the abdomen (CECT - Figure. 1) revealed complete fatty replacement of the pancreas. However size and contour of pancreas was normal. These imaging features with clinical correlation are diagnostic of Schwachman Diamond Syndrome.



Figure. 1: Axial post contrast CT scan showing generalized fatty replacement of the pancreas with CT values of -23 to -56 HU.

Shwachman-Diamond syndrome (SDS) is a rare congenital disorder characterized by pancreatic insufficiency, bone marrow dysfunction, and short stature. SDS is the second most common cause of inherited pancreatic insufficiency, after cystic fibrosis (CF). Approximately 3% of childhood pancreatic dysfunction is attributed to SDS, which occurs in 1 in 10,000-200,000 births. SDS is reported among all racial and ethnic groups with male-to-female ratio of 1.7:1 (1). Synonyms are Burke's syndrome, Schwachman's disease, Schwachman's syndrome, Schwachman-Bodian syndrome. In individuals with this condition, pancreatic acinar cells do not develop in utero and are replaced by fatty tissue with preserved islet cells. In contrast to CF, the pancreatic ductal architecture is spared and sweat chloride levels are normal. Pancreatic insufficiency can be tested by secretin-cholecystokinin stimulation test and steatorrhea. However, absence of steatorrhea can not rule out SDS, since for reasons as yet unknown, the pancreatic lipase secretion increases slightly with age in patients with SDS, resulting in increased pancreatic function with slowing and decrease in fat excretion. Glucose tolerance tests are normal since the pancreatic deficiency is mostly exocrine and islet cells are usually spared (1). Hematological abnormalities may be due to a stem cell abnormality. Almost one half of patients with SDS have pancytopenia with variable degrees of anemia, neutropenia and thrombocytopenia. Hence, pallor, easy bruising, epistaxis, melena, hematemesis or hematuria are frequent. Fetal hemoglobin may be elevated. Irrespective of the total number of neutrophils, patients with SDS have defective neutrophil chemotaxis, which as been linked to Chromosome 7q abnormality (1,2). Recurrent bacterial infections of the upper respiratory tract, otitis media, sinusitis, pneumonia, osteomyelitis, bacteremia, skin infections, aphthous stomatitis, fungal dermatitis, and paronychia are common because of a neutropenia / neutrophil migration defect. Failure to thrive has been attributed to nutritional deficits (malabsorption, steatorrhea), recurrent infections, and skeletal abnormalities as well as decreased or absent growth hormone levels in individuals with SDS (1).

The exact pathophysiology of skeletal anomalies is unknown. Delayed bone age, pubertal delay, thoracic dysostosis consisting of costo-chondral thickening, metaphyseal widening and cupping, tubulation of long bones, clinodactyly, syndactyly, supernumerary metatarsals, valgus deformity at knee and elbows, dental abnormalities, osteomyelitis, otitis media, myelodysplasia, leukemic transformation etc. have been described (1,3). Serial bone marrow evaluation is warranted in view of increased risk of developing leukemias. Ehler's Danlos syndrome, imperforate anus and Hirschsprung disease have been associated with SDS. These

associations may delay diagnosis of SDS because the presenting symptom is constipation and not diarrhea (1). Fatty replacement of the pancreas can be detected with sonography, CT scan and also MRI. Other causes of fatty replacement of the pancreas are cystic fibrosis, history of steroid therapy, Cushing syndrome, Johansson-Blizzard syndrome, obesity, diabetes mellitus, post pancreatitis etc. (1,4,5) Peri-portal fibrosis, cirrhosis, endocardial fibrosis etc. have been described with SDS. The goals of SDS treatment include pancreatic enzyme supplementation, a low-fat diet, multi-vitamins, and fat-soluble vitamins, prevention or treatment of serious and / or invasive infections with early attention to febrile illnesses, correction of hematological abnormalities when possible, growth hormone supplement and prevention of orthopedic deformities (1).

In conclusion, when generalized fatty infiltration of pancreas is encountered in younger patients with features of pancytopenia, pancreatic insufficiency and retarded growth, possibility of SDS should always be considered.

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