Chronic Pancreatic Insufficiency-Think of Shwachmann Diamond Syndrome

Exocrine pancreatic insufficiency which is not due to cystic fibrosis is a diagnostic dilemma until attention is paid to the other systemic findings. We present such a child where focusing on other systems helped us reach a diagnosis. A one year one month old male child was brought to us with onset of complaints since the first few months of life. Parents complained of failure to thrive since birth, repeated anemia needing transfusions to maintain his hemoglobin, extensive seborrheic dermatitis of the scalp, radiologically confirmed pneumonia at four months needing hospitalization and intravenous antibiotics, BCG lymphadenitis at four months needing two drug AKT isoniazid and rifampin, acute otitis media at 6 months, and frequent oily stools since birth. Examination revealed a child with a weight of 6 kg, height of 68 cm and a head circumference of 43.5 cm. Systemic examination revealed seborrheic dermatitis on the scalp and florid rickets. Investigations showed anemia, thrombocytopenia, and intermittent neutropenia. The immunoglobulin pattern and T cell and B cell distribution on flow cytometry was normal.

Bone marrow aspiration revealed low grade myelodysplasia. Stool pancreatic elastase I was less than 15mcg/g suggestive of severe pancreatic insufficiency. The karyotype was normal and when the sweat chloride too came normal [10 meq/L], we were at a dead end until a CT of the abdomen showed a classical appearance of extensive fatty infiltration of the entire pancreas diagnostic of Shwachmann Diamond Syndrome. He was put on pancreatic enzyme replacement [Creon] with every meal along with lansoprazole. His height and weight percentiles now approach 25, he been able to maintain a hemoglobin of 11 without transfusions and his stool output has considerably reduced.

Schwachmann Diamond Syndrome is an autosomal recessive syndrome characterized by exocrine pancreatic insufficiency, neutropenia which may be cyclical, metaphyseal dysostosis [the most variable feature] and failure to thrive with short stature.

These children typically present in infancy with poor growth and greasy foul smelling stools as did our child. The pancreatic insufficiency may be transient and steatorrhea may spontaneously improve with age. Our child may have either responded to enzyme replacement or may be following the natural course of spontaneous improvement with time. Recurrent pyogenic infections are common [otitis media, pneumonia, osteomylitis, dermatitis, sepsis] and a frequent cause of death. Such infections could be related to the cyclical neutropenia or to the neutrophil hemotactic defects that some of these children possess. Thrombocytopenia occurs in 70%; anemia occurs in 50% while neutropenia occurs in virtually all patients. Severe anemia needing repeated transfusions, as experienced by our patient, is unusual. Development of a myelodysplastic syndrome or transformation to acute myeloid leukemia has been reported in upto 33% and 24% patients respectively, especially associated with monosomy 7 [1]. While our child's bone marrow did reveal mild MDS, subsequent follow up did not demonstrate any deterioration. Shwachmann Diamond Syndrome is the second commonest cause of exocrine pancreatic insufficiency after cystic fibrosis from which it is readily differentiated by normal sweat chloride levels, lack of cystic fibrosis gene, metaphyseal lesions and the characteristic hypodense appearance on CT or MRI. Though a large number of invasive and non-invasive tests are available for pancreatic insufficiency [2], fecal elastase is a standard screening test with a sensitivity and specificity [3] of over 90%. A detailed study of the SBDS gene is planned for this family since they have requested for genetic counseling prior to the subsequent pregnancy. Only occasional case reports appear from India, some of which highlight the importance of the study of the SBDS gene located on chromosome 7q11 and its pseudogene [SBDSP] located in the vicinity of the SBDS gene [4]. Such a molecular diagnosis is critical before therapy such as stem cell transplant is attempted as also for genetic counseling for subsequent pregnancies.

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Infected Urachal Cyst - An Uncommon Cause for Incessant Cry in Newborns

A 33 day old baby presented with episodes of incessant cry of 13 days duration, along with drawing up of knees to chest while micturating; intermittent episodes of vomiting, decreased feeding and mild abdominal distension, but no constipation. Two days before presenting to us the mother noticed a swelling in the umbilicus, with increasing periumbilical redness, which ruptured draining pus and blood via the umbilicus, following which the incessant cry subsided. On examination, there was periumbilical redness, umbilical discharge of pus and blood, local rise of temperature and periumbilical induration per abdomen, but no other signs. Ultrasound of abdomen revealed infected urachal cyst. We managed the child conservatively with intravenous antibiotics. On the 3rd post admission day, he developed incessant cry again, along with bilious vomiting, constipation, abdominal distension and decreased bowel sounds (but no fever/ guarding/ rigidity). *X*-ray showed multiple fluid levels consistent with paralytic ileus/ subacute intestinal obstruction. Oral feeds were stopped and IV fluids were administered along with gastric decompression. Laparotomy was planned in the event of non-improvement. However, the child improved in 24 hours, feeds were restarted after 48 hours and the baby was discharged after 1 week, to undergo complete excision at a later date.

In neonates, patent urachus presenting as umbilical discharge is usually seen rather than infected urachal cysts, which have a higher age of presentation [1]. Complications like intestinal obstruction are even rarer [2]. An urachal cyst usually presents when infected as lower abdominal pain, fever, voiding symptoms, a palpable mass and evidence of urinary infection. If left untreated it may drain into the bladder or through umbilicus. The urachus lies in an extraperitoneal fascial plane, hence an urachal remnant is unlikely to cause an intra-abdominal pathology, particularly intestinal obstruction. However this is seen in neglected infections [2,3]. Ultrasound can be diagnostic in 80 to as much as >90% of cases, where diagnosis is doubtful, a CT scan/ MRI is diagnostic [1,4]. Management is controversial with one group advocating a 2 stage procedure - incision and drainage followed by delayed resection [4] and another group arguing that the former was developed in the pre-antibiotic era; and that the use of appropriate antibiotics followed by complete excision as a primary procedure is both possible and safe [1, 5].

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