RODGER C. HAGGITT GASTROINTESTINAL PATHOLOGY SOCIETY

CASE 6

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Distal Intestinal Obstruction Syndrome (Meconium Ileus Equivalent)

Cystic Fibrosis

Cystic fibrosis (CF) is an autosomal recessive disorder of cyclic AMP regulated chloride transport. It results from a defect in the CF transmembrane regulator (CFTR). This regulates chloride secretion and consequently water flow in the apical membranes of ductular epithelial cells. It sites of aqueous gland secretion (such as sweat glands) this defect causes few clinical problems. However, in glands where the secretion is protein rich (mucin or pancreatic enzymes) deficient water and high electrolyte concentration cause elevated protein levels that slow flow within the lumen. Ultimately, this leads to precipitation and plug formation. The mucin that is produced is semi-solid rather than being watery.

There are a large number of possible mutations to the CF gene and many patients with this condition are compound heterozygotes. Different mutations may disrupt CFTR function in at least four different ways. Not surprisingly, therefore, these different mutations or combinations of mutations produce different phenotypic patterns of disease. Some CF patients may have predominant pulmonary disease and some have major manifestations at other sites. The major non-gastrointestinal manifestations of CF include: sweating defect (volume depletion), lung disease (infections leading to bronchiectasis), rhinosinusitis, azospermia and female hypofertility (hyperviscous endocervical mucus). The gastrointestinal complications include: pancreatitis (steatorrhea

and diabetes), DIOS, silent appendicitis and appendiceal intussusception, rectal prolapse, biliary cirrhosis and micro gallbladder.

The term “meconium ileus equivalent” is now old fashioned and has been replaced by the less descriptive term “distal intestinal obstruction syndrome” (DIOS). Patients may present with acute or chronic disease. Acute DIOS produces cramping lower abdominal pain, a palpable mass in the right lower quadrant and decreased stool frequency. If these symptoms are unrelieved patients may develop abdominal distension and bilious vomiting. Chronic DIOS presents with cramping abdominal pain that often follows meals. Anorexia is a method of avoiding these symptoms which may then remit for weeks or months at a time.

The presented case is typical of DIOS. By naked eye inspection the colon and distal small bowel are distended and contain brownish putty-like material that is difficult to dislodge from the underlying mucosa. Typically the bowel wall is thickened but the mucosa shows no obvious distortion or ulceration. Histologically, the crypts are dilated and filled with mucus that may have a laminated appearance. The crypt mucus is continuous with the luminal mucus. The colonic goblet cells are enlarged and distended. Inflammatory changes are rarely prominent although there may be a non-specific increase in lamina propria lymphocytes and eosinophils. Myocytes in the muscularis may contain increase amounts of lipofuschin. This finding is not confined to DIOS patients and in seen in individuals with malabsorption or maldigestion of any type. It is thought to result from a deficiency of vitamin E.

Hypermucinous mucosa may also be seen in other conditions such as chronic inflammatory bowel disease. The mucosal contour may be villous or flat. In ulcerative colitis hypermucinous mucosa may contain K-ras mutations and is regarded as pre-malignant.

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