Fabry's Disease — A Case Report

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Fabry's disease is a rare inherited sex linked disorder of glycolipid metabolism with defect of ceramide trihexosidase activity. As compared to skin, kidney, cornea and nervous system intestinal manifestations are uncommon. Impaired motility is the prominent disorder. There are only very few reports of the ultrastructural changes of rectal mucosa in Fabry's disease. In this report, electronmicroscopy of rectal mucosal biopsy from a patient with Fabry's disease who was investigated for obscure gastrointestinal bleed is presented.

Case report: A 50 year old male, known hypertensive for 2 years, presented with a history of 2 episodes of malena in the last 3 years.

Physical examination revealed multiple slightly elevated, hyperkeratotic purplish papules since childhood on forehead and scrotum suggestive of angiokeratomas. Similar lesions were seen in his younger brother. Investigations revealed anaemia and stool occult blood was positive. Small bowel enema study, gastroscopy, colonoscopy, Meckel's Scan and Ultrasound scan of abdomen were normal. Ophthalmic examination did not reveal any abnormalities.

Pathology: Duodenal, ileal and rectal mucosal biopsies showed no histological abnormalities by light microscopy. Rectal mucosal biopsies were processed for electron microscopy by standard methods. Ultrastructural examination revealed amorphous osmophilic deposits within smooth muscle cells of muscularis mucosae and lamellated deposits within endothelial cells lining arterioles, venules and capillaries (Fig 1) but not in enterocytes.

Discussion

Widespread accumulation of glycolipid in the endothelium, smooth muscle and supporting cells of blood vessels in kidney, skin, gastrointestinal tract, central nervous system, heart and reticuloendothelial tissue, is believed to contribute to the protean manifestations of Fabry's disease.

In rectal mucosas characteristic inclusions have been shown in neuronal bodies and axons of Meissner’s plexus, in endothelial cells of the microvasculature and in smooth muscle cells but not in enterocytes.

Fig. Part of an endothelial cell with lamellated deposits (x 75000)

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The pathophysiologic basis for gastrointestinal bleeding in our case, could be glycolipid deposition in small vessels which can induce severe vasculitides, though no evidence of vasculitis was detected in the tissue sampled.

References


Chronic recurrent Parotitis Complicated by Salmonellosis in a Renal Transplant Patient

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Renal transplant recipients are susceptible to a wide range of unusual infections, this being the single most important cause of morbidity and mortality. A high index of suspicion, adequate antibiotic therapy and surgical intervention wherever necessary are essential for successful treatment. Gram negative bacilli have been increasingly implicated in the pathogenesis of parotid infections, which is consistent with the increasing prevalence of Gram negative bacillary infections in the last two decades. Parotid infections due to salmonellae are an exceedingly rare entity; only one case has been reported previously in medical literature. We report a case of parotid infection caused by Salmonella typhimurium in a renal transplant recipient.

Case Report: A 60 year old man presented at the surgery outpatient department with a 1 week history of left sided facial pain and swelling. The patient had received a live related donor renal transplantation 20 years ago due to end-stage renal disease as a result of chronic glomerulonephritis. He was on maintenance therapy with azathioprine and prednisolone and his graft was functioning well. He later developed extra-pulmonary tuberculosis involving the tarsal bones of his left foot. He was treated with an 18-month course of antituberculosis therapy, following which he has been receiving isoniazid prophylaxis. Physical examination revealed a healthy man, afebrile and well nourished.

There was a large, warm, tender and erythematosus swelling, 3x3 cms in lobe region of the left parotid gland. He was treated with broad spectrum antibiotics following which the infection resolved. A