Case report

Neurofibromatosis with lower G.I.T. presentation

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Summary

Hyperpigmented skin lesions with different benign and malignant neoplasms, which arise primarily from cells of neural-crest origin, these tumors include neurofibromas, neurofibros-arcomas, optic gliomas, and pheochromocytomas is the usual presentation of neurofibromatosis. However, the disease may have an unusual clinical presentation that may need surgical intervention.

A young male patient presented with diarrhea, abdominal pain, fresh blood per rectum and anemia, colonscopy showed large polyps with distinct histopathology. This prompted the reporting of this case.

Case Report

A 25-year-old Iraqi male presented 5 years ago with a history of abdominal pain mainly epigastric region; colicky in nature and aggravated by hunger and associated with nausea. The pain was associated with frequent loose bowel motion; about 8-times per day, small volume, mixed with blood and mucus. The patient was treated as an Amoebic Dysentery and treated with metronidazole for one week with no improvement. The condition of the patient got worse in a form of loss of weight and appetite, easy fatigability and effort intolerance. For this reason, the patient was admitted to Simawa Hospital five years ago and barium meal and follow through and barium enema were arranged, but no specific diagnosis were reached. In 1999 fresh blood per rectum were continued and Haemorrhoidectomy done with no much improvement.

During his illness there were no history of orogenital ulceration, joints pain, eye problems. Regarding family history; his sister has similar multiple hyperpigmented skin lesions. Past, social, and personal history was not remarkable.

Physical examination revealed a young male with signs of anemia and emaciation. There were bilateral clubbing of fingers and toes and diffuse hyperpigmented skin lesions with freckling all over the body; specially the axillae and groins. Caf'e au lait skin hyperpigmentation lesions of variable sizes ranging from 0.5-6 cm and about 12 in number, involving trunk, abdomen and even the buttocks (Fig 1). Also multiple painless slippery masses about 2-7 cm in diameter, soft, smooth, not attached to tissues located in right knee and abdominal wall. Peripheral lymph nodes were not palpable. He was not jaundiced or cyanosed, and his vital signs were normal. Examination of the abdomen showed right umbilical hernia. Liver and spleen were not palpable and there was no ascitis.

Laboratory investigations showed anemia with CBC hematocrit 0.28,WBC 7.4 ×109/L, and

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peripheral blood smear and absolute indices suggestive of microcytic hypochromic anemia. Liver enzymes and other relevant serum chemistry tests were normal.

Upper GI endoscopy showed a flat duodenal folds. Multiple biopsies were taken for histopathological examination. These revealed finger like villi and normal histology. Colonoscopy showed normal vascular pattern up to caecum with three large polyps at the splenic flexure, ranging in size from 2-3 cm in diameter, congested surface. Polypectomy had three large polyps at the splenic flexure, ranging in size from 2-3 cm in diameter, congested surface. Polypectomy had done for histopathological examination (Fig 2). The biopsy revealed hyperplastic mucosa vascular proliferation, bundles of smooth muscles with mixed inflammatory cell infiltration and many eosinophills, spindle cells proliferation; going with hamartomous polyps (Fig 3,4).

Discussion

Neurofibromatosis type 1 is an autosomal

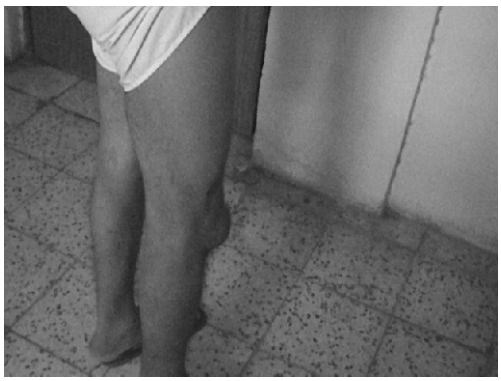

Fig 1c

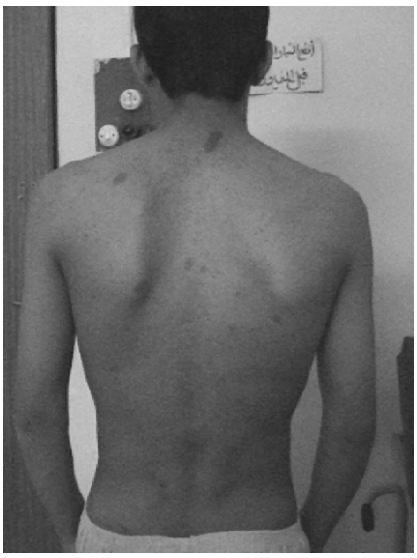

Fig 1b

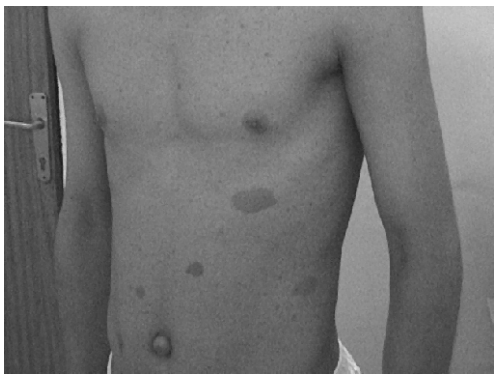

Fig 1a

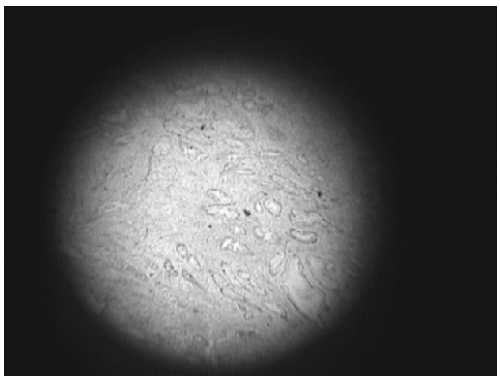

Fig 3a

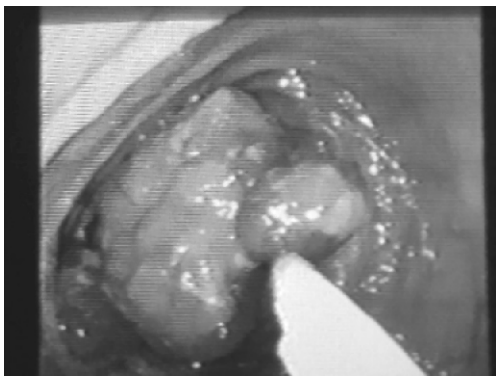

Fig 2

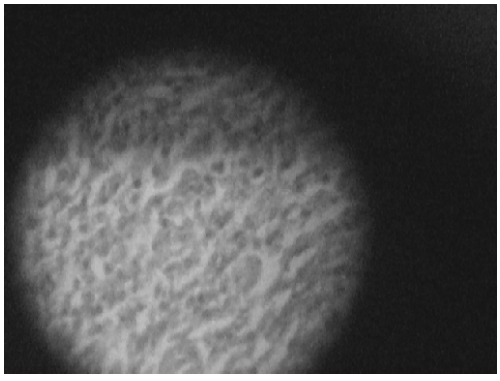

Fig 4

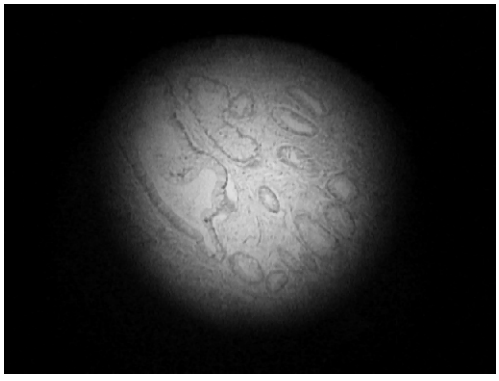

Fig 3b

Discussion

Neurofibromatosis type 1 is an autosomal dominant disorder with an incidence of approximately 1 in 3500 people. Patients with the disease have a predisposition to particular benign and malignant neoplasms, which arise primarily from cells of neural-crest origin and are linked, to a gene on chromosome 17. These tumors include neurofibromas, neurofibrosarcomas, optic gliomas, and pheochromocytomas. In young children with neurofibromatosis type 1, the risk of malignant myeloid disorders, particularly juvenile myelomonocytic leukemia, and the monosomy 7 syndrome, a childhood variant of myelodysplasia, is 200 to 500 times the normal risk (1).

Neurofibromatosis type 2 (NF2) is an autosomal dominant disorder predisposing to multiple neoplastic lesions. The pathognomonic findings are bilateral acoustic neuromas (schwannomas). Astrocytomas, multiple meningi-omas, and occasional ependymomas are also encountered in persons with NF2 (2).

Gastrointestinal involvement of neurofibromatosis occurs in as many as 25 percent of cases. Neurofibroma occurs most frequently in the stomach and jejunum, but the colon may also be involved. Multiple submucosal neurofibromas of the gastrointestinal tract and cafe au lait pigmenta-tion, bony abnormalities, and neurofibromas of both central and peripheral nerves characterize this condition. Gastrointestinal neurofibromas may cause occult bleeding, luminal obstruction, or intu-ssusception. Malignant transformation into neurofibrosarcoma is rare (3,4).

Neurofibromas of the gastrointestinal tract are usually associated with neurofibromatosis type1 (Nfl), or they are exclusive manifestations of the so-called "familial intestinal neurofibroma-tosis". Mesenteric neurofibroma, and watery diarrhoea syndrome due to a VIP-producing adrenal phaeochromocytoma and a case of upper jejunal gastrointestinal stromal tumor (GIST) accompanied with von Recklinghausen's disease had been reported (5,6,7,8).

On reviewing other studies; and in the presence of different gastrointestinal manifestation of neurofibromatosis, there were no associated colonic hamartomous polyps presented as lower G.I.T. blood loss and anemia. Literature reviewing did not mention this association.

References

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