LETTER TO THE EDITOR

Cornelia de Lange syndrome in association with ulcerative colitis: A case report

Dear Sir,

Cornelia de Lange syndrome (CdLS) is a rare multi-system genetic disorder with an incidence rate between 1:10000 and 1:50000. It is characterized by growth and developmental delay, distinctive facial dysmorphism, limb malformations and multiple organ defects. Patients with CdLS usually also have behavioral problems. CdLS has been found associated with mutations in the NIPBL, SMCTA, and SMC3 genes. A variety of gastrointestinal abnormalities have been described, including malrotation, colonic duplication and non-fixation of the colon. A high mortality rate caused by cecal volvulus in CdLS patients has also been reported. There are several reports of association of ulcerative colitis with rare genetic syndromes. However, no case of coexistence of CdLS with ulcerative colitis has been reported so far.

We report a 20-year-old male with CdLS that was diagnosed with ulcerative colitis. He was admitted to the hospital with a two month history of bloody diarrhea (about 10 bowel movements per day), abdominal pain and weight loss of 6 kg. He was diagnosed since childhood with CdLS mainly based on the clinical findings. He had a positive family history (mother) with ulcerative colitis. Physical examination revealed facial dysmorphism (Fig. 1) as well as significant growth and mental retardation. The laboratory tests revealed mild anemia (hemoglobin 12.5 g/dL, hematocrit 37.3%) and elevated ESR (80 mm/h) and CRP (12.5 mg/dL) with normal liver and kidney functions. The patient underwent colonoscopy that showed all the parts of the colon having multiple ulcers, marked erythema and friability (endoscopic partial Mayo score 3). Histology showed loss of goblet cells, crypt abscesses and chronic inflammation in the lamina propria findings consisted with the diagnosis of ulcerative colitis. Initial treatment of the patient was mesalazine 3 gr/d per os and enema 4 gr/d as well as prednisolone 30 mg per os but without any response mainly due to the behavioral problems and the poor compliance of the patient to the treatment. Even if his parents were giving him the medications he was trying to avoid them by voluntary emesis. Afterwards infliximab 5 mg/kg (loading dose at 0, 2 and 6 weeks) was administered and the patient showed prompt clinical response. Currently, the patient is in clinical remission under treatment with infliximab 5 mg/kg every 8 weeks.

The presented case represents a rare coexistence of CdLS with ulcerative colitis that has not been previously reported in the literature. An interesting point also is the problems with the treatment in this situation which were overcome by the IV administration of infliximab. The pathophysiology of the association between CdLS and ulcerative colitis remains unknown but the positive family history of this patient could play also a prominent role.

Conflict of interest statement

All authors do not have any conflict of interest regarding this report.

References

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