

Case Report

Coexistence of Coffin-Siris Syndrome and Stercoral Colitis

İS KARA, U KOC

Abstract

Coffin-Siris syndrome is a rare syndrome diagnosed by clinical features such as mental retardation, growth retardation, absence or hypoplasia of the fifth distal phalanx, different facial features and supported by genetic testing. Stercoral colitis is an inflammatory colitis, which is usually caused by an increase in intraluminal pressure in stool material stuck in colon segments, which is rarely seen in patients with chronic constipation and may result in perforation, shock and death. Our 11-year-old male patient was brought to the emergency department with the complaint of inability to walk and the patient was diagnosed with stercoral colitis with appropriate evaluation. This is the first case in the literature in which coexistence of Coffin-Siris syndrome and Stercoral colitis were seen.

Key words

Coffin-Siris syndrome; Paediatrics; Stercoral colitis

Introduction/Aim

Coffin-Siris syndrome (CSS) is a condition characterised by the aplasia or hypoplasia of the fifth distal phalanx or nail, developmental or cognitive delay, varying facial features, hypotonia, hirsutism/hypertrichosis, and sparse scalp hair.^{1,2} It presents with congenital anomalies including malformations of the heart, gastrointestinal, genitourinary and/or central nervous systems.^{1,2} Nutritional difficulties, slow growth, ophthalmological abnormalities, and impaired hearing can also be seen.^{1,2}

Stercoral colitis (SC) is an inflammatory colitis that occurs due to increased intraluminal pressure caused by faecal material impacted in colon segments.³ The most important complication of SC is colon perforation, and the mortality rate varies between 32 and 57%.³⁻⁶

In most patients with chronic constipation, stool becomes harder due to physical inactivity, making it even more difficult to pass through the intestinal system, and thus creating a vicious cycle. This condition is usually seen in the elderly. SC diagnosis may be delayed in patients with neurological and psychological problems.⁷⁻⁹ Since clinical evidence in SC is not clear, imaging-based diagnosis is required. Late diagnosis of SC can lead to serious complications and even mortality.^{3,4}

SC is infrequently seen in young adults and even rarer among children. In this paper, we present a case of a child having the coexistence of CSS and SC for the first time in the literature.

Department of Pediatrics, Erzincan Binali Yıldırım University, Mengucek Gazi Training and Research Hospital, Erzincan / Turkey

İS KARA

MD, Assistant Prof.

Department of Radiology, Erzincan Binali Yıldırım University, Mengucek Gazi Training and Research Hospital, Erzincan / Turkey

U KOC

MD

Correspondence to: Dr U Koc
Email: dr_uralkoc@hotmail.com

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The Case

An 11-year-old male patient previously diagnosed with CSS in another health center presented to our clinic with the complaint of not being able to push his feet down and walk. In addition to tonsil hypertrophy and hyperemia, he

had coarse facial features characterised by thick eyebrows, marked eyelashes, flat nose bridge, short nose, and thick lips. He also presented with small nails on the fifth fingers and toes, hypertrichosis, low anterior hairline, sparse scalp hair, and low height and weight percentiles (Figure 1). Considering the indications of mental retardation and complaints of frequent constipation, the sweat test was undertaken with the suspicion of cystic fibrosis, but the findings and thyroid hormones were normal. The patient had scarred skin on his feet due to frequent rubbing of hands and feet against each other. It was not possible to perform an abdominal examination since he did not allow anyone to touch his abdomen. Considering that he was not able to push his feet down, the orthopaedic clinic was consulted to evaluate the joint graphy in terms of the

presence of arthritis or a fracture. There was no indication of orthopaedic emergency, but the white blood cell count was high ($20 \times 10^3/\text{mm}^3$). Magnetic resonance imaging of the brain revealed two arachnoid cysts. The patient was hydrated and started on antibiotics but continued to exhibit agitation and agony, and he was unable to stand up. An abdominal ultrasonography was attempted, but the findings were not adequate due to the constant agitation. Due to the dilatation of intestines and faecal impaction at plain abdominal radiograph (Figure 2), an abdominal tomography was performed and revealed thickening of the colonic wall with large volume of feces findings compatible with SC (Figure 3). Once the feces impaction was relieved, the patient's complaints were reduced. He was started on an appropriate diet and was discharged in a few days after full recovery.



Figure 1 An 11-year-old male patient previously diagnosed with Coffin-Siris syndrome had coarse facial features characterised by thick eyebrows, marked eyelashes, flat nose bridge, short nose, thick lips, small nails on the fifth fingers and toes, hypertrichosis, low anterior hairline, sparse scalp hair.

Discussion

CSS is a congenital anomaly characterised by shortness of the fifth distal phalanx (65%) or aplasia or hypoplasia of nail (80%), varying degrees of developmental or cognitive delay, different facial features, hypotonia, hirsutism/hypertrichosis (95%), sparse scalp hair (68%), dental anomalies (96%), intrauterine growth retardation

(67%), craniofacial anomalies, short stature (68%), spinal anomalies (66%), congenital heart disease (45%), coarse facial appearance, bushy eyebrows, and thick lips. Nutritional difficulties, slow growth, ophthalmological abnormalities, and hearing impairment can also be seen in patients with CSS.^{1,2,10} The diagnosis of CSS is most commonly based on clinical features, and genetic assessment has a complementary role.^{11,12}

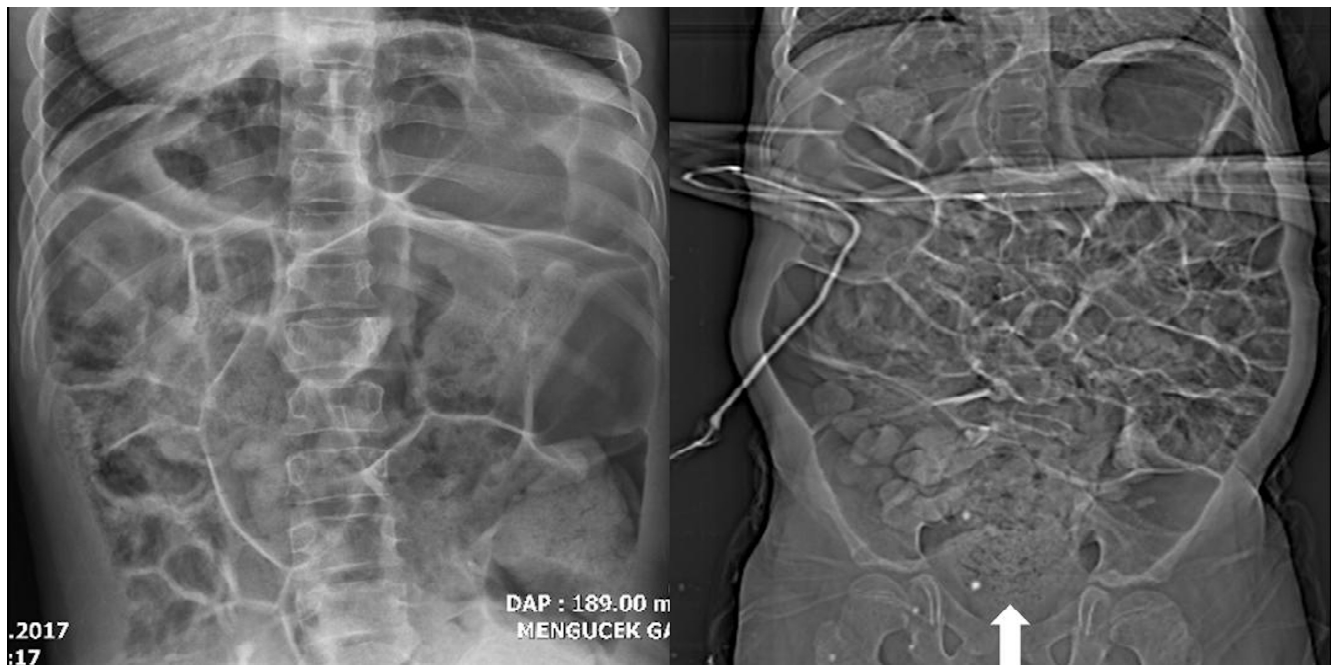


Figure 2 A plain X-ray studies of the abdomen revealed the dilatation of intestines and faecal impaction (arrow).

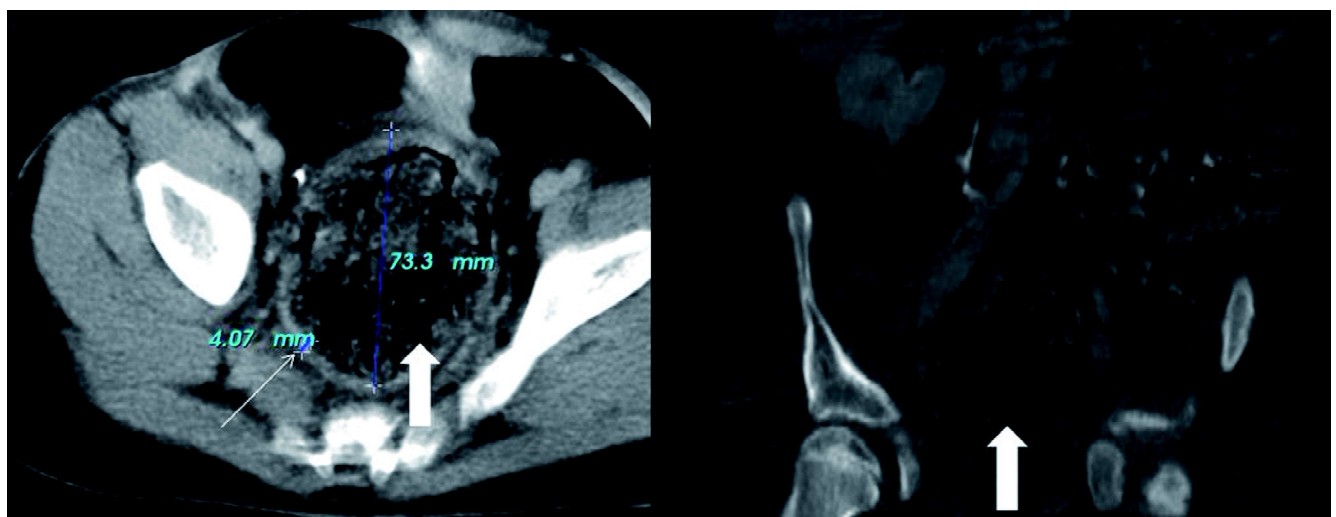


Figure 3 Abdominal tomography revealed thickening of the colonic wall (thin arrow) with large volume of feces (thick arrows) findings compatible with stercoral colitis.

Our paediatric patient presented with coarse facial features, bushy eyebrows and thick lips, short fifth distal fingers, hypoplastic nails, and developmental and cognitive retardation. Hirsutism/hypertrichosis, sparse scalp hair, dental anomalies, and short stature were other clinically significant indications. He did not have a congenital heart disease, but his brain magnetic resonance image revealed two small arachnoid cysts. His clinical findings were consistent with the diagnosis of CSS. His genetic analysis had been performed 10 years earlier and the diagnosis had been confirmed. Since the patient continuously rubbed his feet against each other, there were scarred tissues and new lesions on the contact points.

The development of faecaloma or stercoroma due to the build up and impaction of dry feces causes colonic distention and applies pressure on the wall mucosa, reducing blood flow to the area and eventually leading to ischemia. If the pressure on the intestinal wall continues, necrosis or perforation develops,^{7,8} which, if overlooked or untreated, may result in shock and death.^{9,13,14}

SC has similar prevalence in both sexes and is seen at an average age of 59.⁸ Almost all patients present with conditions and factors implicated in constipation, such as hypothyroidism, diabetes, cognitive disorders, and the use of anticholinergic, narcotic, antacid and tricyclic antidepressants.^{13,14} Eighty-one percent of the SC cases have the complaint of constipation,^{4,12} but diarrhea is rarely associated.^{8,14}

Our patient was mentally retarded and had a previous diagnosis of CSS. He was not taking any medication and he was in constant motion. Since he was not able to explain the reason for his agony, he continuously cried and he was not able to stand on his feet. He also did not allow us to touch his abdomen.

The three most common localisations of stercoral ulceration are the anterior wall of the rectum, the antimesenteric border of the rectosigmoid juncture, and the apex of the sigmoid colon.^{14,15} SC and stercoral ulcers are most frequently seen in the sigmoid colon^{4,14} since it is the narrowest part of the colon with the lowest blood stream, and feces is also the driest in this area.⁴ Our patient was found to have 73.3 mm dilatation of colon involving the rectosigmoid region and his feces were impacted.

Direct abdominal radiography can show faecal loading or calcified faecaloma. Direct graphy of our case revealed markedly dilated intestinal lumen. Abdominal CT facilitates the diagnosis and treatment. The CT findings of SC include build up of feces in the sigmoid colon,

pericolonic stranding, thick mucosa or discontinuous areas of of colonic mucosa (perfusion defect), thickening of the colonic wall of more than 3 mm, and proximal colon expansion.^{7-9,16} The current patient had proximal colon expansion, thickening of the colonic wall of more than 3 mm, and feces accumulation in the colon.

In SC cases, abnormal gas, free fluid, and intense mucosal perfusion suggest perforation and is associated with increased mortality.^{9,14,17} There was no evidence of perforation in our patient. Fifty-two percent of the patients with non-perforated SC are successfully treated with a bowel regimen.⁷ During the follow-up period of the current case, we observed spontaneous faecal discharge and adjusted the patient's diet; therefore, there was no need for surgery.

In the literature, to date, only four paediatric cases of SC have been reported; a two-year-old child due to misuse of ibuprofen,¹⁸ an incidentally diagnosed case presenting with a chronic cough,¹⁹ a 17-year-old girl with an eating disorder,²⁰ and an 11-year-old with stercoral perforation associated with bezoar.²¹ This is the first case report in the literature presenting the coexistence of a very rare CSS and an even rarer SC in a paediatric patient. In clinical practice, we encounter mentally retarded patients with varying complaints and appropriate patient management is of utmost importance in these cases. For the current patient, the careful attention of the radiology specialist concerning the walking difficulty complaint possibly saved the patient's life by preventing the development of a future perforation through the immediate initiation of a laxative diet.

Conclusion

This is the first case of the coexistence of CSS and SC reported in the literature. In patients with mental retardation, chronic constipation, distention, and abdominal sensitivity despite the lack of a voiced abdominal complaint due communication difficulties, the possibility of SC should be considered, and these patients should be managed accordingly.

Informed Consent

Informed consent was obtained from the patient's parents.

Conflict of Interest

No conflict of interest was declared by the authors.

Financial Disclosure

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