CASE REPORT

Orofacial granulomatosis as a presenting feature of Crohn's disease

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SUMMARY

An 11-year-old girl was referred to oral medicine with persistent facial swelling. She was diagnosed with orofacial granulomatosis (OFG) and was treated conservatively for 7 years, with no evidence of systemic illness. Aged 17 she re-presented with a flare up of her OFG, watery diarrhoea and fluctuating febrile episodes. Inflammatory markers were raised and an MR enterogram revealed terminal ileal Crohn's disease. This case highlights that OFG may precede the onset of intestinal Crohn's disease.

BACKGROUND

Orofacial granulomatosis (OFG) is a rare disorder describing non-caseating granulomatous inflammation of soft tissue in the orofacial region. It can present as persistent soft tissue enlargement of the lips, the cheeks and the periorbital region, and as ulceration of the mouth. It can also cause erosions, erythema and hypertrophy of the oral mucosa.^{1 2} OFG can be an extra-intestinal presenting symptom of Crohn's disease. OFG diagnosed in children can be related to systemic disease, which may not manifest itself for years after initial presentation.³ The aetiology of OFG is not clear hence treatment options can be limited and their effectiveness unsatisfactory. This case highlights the fact that OFG may precede the development of symptomatic intestinal Crohn's disease. This is important to consider at presentation and may prompt further investigation to exclude underlying systemic disease.

CASE PRESENTATION

An 11-year-old girl was presented with the lower lip swelling. Her parents described episodes of persistent swelling in the lower lip, which eventually became permanent. She was referred to the oral medicine clinic for further investigation.

The patient had no significant medical history. At the time of presentation there were no intestinal symptoms to suggest Crohn's disease. Extra-oral examination revealed a full lower lip, symmetrical in appearance with a red blush to the right cheek. There was no cervical lymphadenopathy. Intra-oral examination revealed erythematous and swollen buccal mucosa.

A clinical diagnosis of OFG was made; the patient did not undergo a biopsy to confirm the diagnosis histologically. The diagnosing doctor was confident of his diagnosis. Further investigation of her gastrointestinal (GI) tract was not performed. Conservative treatment was initiated, involving

dietary modification, reduced consumption of sodium benzoate and elimination of 'trigger foods', including sweets, chocolate, tomatoes, fizzy drinks and biscuits; she was also asked to monitor her symptoms. The lip swelling and intra-oral swelling and erythema improved. Over time she reported only mild intermittent lip swelling and continued her childhood in a normal manner.

She re-presented to the oral medicine clinic aged 17 with a flare up of her OFG; examination revealed a slight blushing of the right upper lip and below the nose. Intra-oral examination revealed healthy non-ulcerated morphologically normal mucosa with no lesions present. The flare was suspected to be secondary to the reintroduction of chocolate into her diet. She also developed mild intestinal symptoms of diarrhoea occurring once per day; she denied abdominal pain and urgency. She was prescribed soluble prednisolone (mouthwash three times daily for 5 days) and Orabase protective paste, which she applied topically as required for a short period. Additionally, she was referred to a gastroenterologist to investigate for possible Crohn's disease. While waiting for outpatient investigations she deteriorated.

She was referred to the acute medical in take with a 2-week history of fluctuating temperature spikes, worsening watery diarrhoea with no blood, decreased appetite and a low body mass index of 12.9 kg/m². Inflammatory markers were raised: C reactive protein 58 mg/L and white cell count 10.9/L.

INVESTIGATIONS

Abdominal radiograph displayed dilated bowel loops with faecal impaction in the rectum. Ultrasound of the abdomen found only a small amount of free fluid. An MR enterogram revealed terminal ileal Crohn's disease with some involvement of the right-sided small bowel.

DIFFERENTIAL DIAGNOSIS

- Crohn's disease
- Sarcoidosis
- Angio-oedema
- Amyloidosis
- Foreign body
- Infections—leprosy, tuberculosis and fungal infections
- Anaphylaxis

TREATMENT

The patient was started on budesonide 3 mg three times a day.

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OUTCOME AND FOLLOW-UP

The patient is making good progress.

She now opens her bowels once a day describing normal stool. She is at college and has resumed normal activities. Her weight has increased by 7 kg.

DISCUSSION

OFG is an uncommon disease and the clinical presentation is variable and unpredictable. OFG includes the previously recognised clinical entities of Melkersson-Rosenthal syndrome and cheilitis granulomatosa (Miescher's cheilitis). The precise cause of OFG is not known,^{4 5} although there does appear to be immunological and histological comparability between OFG and Crohn's disease. The current literature suggests that 15% of OFG cases have an underlying systemic disease. In those cases where OFG precedes the diagnosis of Crohn's disease or is diagnosed concurrently it is often difficult to know if these are separate diseases.⁶ The oral changes noted in OFG could be oral Crohn's disease.

Cutaneous and oral lesions are often the presenting features of asymptomatic inflammatory bowel disease (IBD),⁷ especially in children and adolescents.^{8 9}

Careful examination of the mouth should reveal the diagnosis. The patient with oral Crohn's disease involvement suffer from linear and aphthous ulcers, the lip swelling and characteristically have a cobblestone oral mucosa. A linear ulceration in the buccal vestibule surrounded by hyperplastic mucous is highly suggestive of Crohn's disease.^{1 10 11} Patients with OFG present with variable symptoms. The most frequent presentation is non-tender persistent swelling involving the lips.¹² As the clinical features of OFG can be produced by a variety of underlying pathologies, patients should be investigated for these.

All patients should have a biopsy for histological diagnosis. Further investigations should be directed by presenting symptoms. Chest radiograph can exclude sarcoid and tuberculosis, in conjunction with ACE levels and acid-fast bacilli staining of samples. IgE levels and patch testing are important if hypersensitivity is suspected. If there are GI symptoms, colonoscopy and colonic biopsy are justified. Our patient should have had a biopsy to determine her diagnosis—this may have led to an earlier diagnosis of her Crohn's disease.

Treatment for OFG is often aimed at symptom resolution and clinical appearance. National guidelines are absent and treatment is varied.¹³ Treatment options include a cinnamon-free and benzoate-free diet, intralesional or topical corticosteroids, hydroxychloroquine,¹⁴ dapsone, danazol, thalidomide and antimicrobials (eg, minocycline¹⁵). The use of long-term systemic corticosteroid treatment is limited due to the relapsing chronic nature of the disease and the significant side effects associated with long-term steroid use.

There is concern that the radiation associated with diagnosing and monitoring disease activity in inflammatory bowel disease adds to the already increased risk of lymphoma. New guidelines from the ECCO-ESGAR¹⁶ suggest that MR enterography/enteroclysis has a similar diagnostic accuracy to CT, with the enormous advantage of not imparting ionising radiation. This is of particular significance in young patients who have a lifetime of follow-up. MRI as an imaging modality for inflammatory bowel disease is likely to become more frequent. A survey of current

UK practice¹⁷ showed that clinicians regularly consider ionising radiation dose when organising scans. However, modalities using ionising radiation such as barium follow through and CT are still used frequently, probably as a consequence of the limited availability of MRI in some hospitals.

Learning points

- ▶ Orofacial granulomatosis (OFG) may precede the development of intestinal inflammatory bowel disease (IBD), especially in children and adolescents.
- ▶ Careful examination of the mouth should reveal the diagnosis; all patients should have a biopsy of their lesion to confirm the diagnosis.
- ▶ MRI of the bowel has comparable diagnostic accuracy to CT in IBD with the advantage of reducing the level of ionising radiation exposure to patients.
- ▶ Clinicians should have a low threshold of referral to exclude IBD in patients diagnosed with OFG.

Competing interests None.

Patient consent Obtained.

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