INTRODUCTION

Proctalgia fugax is an uncommon condition and is characterized by intermittent but severe attacks of anal pain. The origin of the pain is still a matter for debate but may result from transient spasms of the pelvic floor muscles [1].

Internal anal sphincter myopathy is a rare condition that has characteristic histological and radiological features. It has been described as an unusual cause of proctalgia [2]. In this report, we present a case of a 64 year-old woman who presented with chronic constipation and proctalgia, and the diagnosis of internal anal sphincter myopathy was eventually confirmed by muscle biopsy.

CASE REPORT

A 64 year-old woman was referred to our colorectal clinic, from another hospital, with a provisional diagnosis of a possible anal malignancy. She complained of intermittent but severe anal pain. She also suffered from chronic constipation, which was only partially relieved by laxatives. She could not recall whether her deceased parents had any difficulty with constipation or anal pain, but she distinctly remembered that her sister, who had died a few years earlier, suffered from similar anorectal symptoms.

Examination under anaesthesia revealed anal stenosis, which had been initially attributed to her previous haemorrhoidectomy. The anal sphincter was thickened circumferentially, but the surrounding and overlying mucosa were normal. A colonoscopy was performed and no other abnormality was seen in the rest of the large bowel.

Anal manometry was carried out and this registered a persistently elevated resting anal tone with prominent periodic ultraslow wave activity. Endoanal ultrasound was then performed using a dedicated endoanal ultrasound machine (B and K Medical, type 1846) and a 10 MHz transducer (endosonic probe type 1850). This revealed a uniformly thickened, but well-defined, internal anal sphincter measuring 9 mm thick (Fig. 1). The puborectalis and external sphincter were of normal thickness and appearance. Magnetic resonance imaging (MRI) (Philip Gyroscan ACS/NT Powertrak 3000, 1.5 T) of the pelvis was unremarkable apart from confirming the presence of a grossly thickened internal anal sphincter (Fig. 2). The three distinct parts (deep, superficial and subcutaneous) of the external anal sphincter were also well demonstrated especially on the coronal section and no hypertrophy was noted.

All the clinical and radiological features pointed to the diagnosis of internal anal sphincter myopathy. The anal sphincter muscle biopsy specimen was sent for specialist histological examination and this showed smooth muscle with no evidence of inflammation or malignancy on conventional staining. On the periodic acid–Schiff (PAS) staining, positive ovoid inclusion bodies up to 160 μm in length within the smooth muscle were evident and the diagnosis of polyglucosan myopathy of the internal anal sphincter was made [3,4].

Guarantor and correspondent: F. W. Poon, Department of Radiology, Glasgow Royal Infirmary, 16 Alexandra Parade, Glasgow G31 2ER, UK. Tel: +44-141-2115521; Fax: +44-141-211-4783; E-mail: fw.poon@northglagow.scot.nhs.uk

Fig. 1 – Endoanal ultrasound showing a markedly thickened internal anal sphincter. This internal anal sphincter (+) measures 9 mm thick.
As the anal discomfort was probably due to spasms of the abnormal anal sphincter, the patient was asked to undergo a trial of topical glyceryl trinitrate cream to the perianal region. This succeeded in providing some relief and suggested that partial internal anal sphincterotomy might give some symptomatic relief. In order to avoid the risk of causing incontinence, the patient agreed to have a staged procedure. Unfortunately, her symptoms were only partly relieved even after two partial internal anal sphincterotomies. Anal manometry continued to show a very high resting pressure of over 100 mmHg. The ultrasound and MR were repeated after surgery, and apart from the presence of the surgical scar, there had been no significant change in the appearance (Fig. 3).

DISCUSSION

Isolated cases of internal anal sphincter myopathy have been reported in the literature. A family with hereditary internal anal sphincter myopathy causing proctalgia fugax and constipation was first described in 1991 [2]. Since then, several similar cases have been reported [3–6]. It has been noted that not all family members are affected by this unusual condition, and the nature of inheritance is probably autosomal dominant with incomplete penetrance [2,6]. We are unaware of any family links of our case to previously reported patients.

The clinical, histological, ultrasonographic and manometric findings are very similar in all the described subjects. Symptoms begin in the fourth decade with chronic constipation, excessive straining and hard, pellet stools. The anal pain is spasmodic but severe, occurs mainly at night, and can be precipitated by a change in posture. As in our case, manometry in all cases reveals high resting pressure with massive ultraslow waves, and the anal pain is noted to coincide with these ultraslow wave peaks [5]. Histology of the resected internal anal sphincter tissue may show degrees of disorganization and a moderate increase in endomysial connective tissue. All reported cases show typical inclusion bodies positively reactive with PAS stain before and after diastase digestion, which may be associated with vacuolation [3,4]. This characteristic finding was also demonstrated in the present case. In previously described cases, ultra-structural studies of the inclusion bodies revealed a granulofibrillar structure consistent with polyglycosan bodies. These polyglycosan bodies are not seen in any other smooth muscles, but can be found in rare sporadic
limb-girdle myopathy [2]. These changes therefore suggest the presence of a localized internal anal sphincter myopathy.

On ultrasound, the normal external anal sphincter is hyperechoic with an average thickness of 4 to 6 mm and by comparison, the internal anal sphincter is relatively hypoechoic with an average thickness of 2 mm. As with all cases of internal sphincter myopathy, the present patient had a grossly thickened internal anal sphincter (measuring 9 mm across). The external anal sphincter and puborectalis are normal. MR imaging of the anal sphincter showed similar appearance with a thickened internal anal sphincter and a relatively normal external anal sphincter muscle. Post-operative MR imaging also showed the partial sphincterotomy scar accurately.

Myopathy of a single muscle is a recognized feature in a variety of rare neurological disorders, but the classically described entities affect skeletal muscles. Selected smooth muscle myopathy is unusual and can affect the internal anal sphincter giving rise to anal pain and chronic constipation.

REFERENCES

5 Orkins BA. Internal anal sphincter myopathy and severe constipation. Gastroenterology, 1992;102:A496.