In our series of 24 cases, endoscopic treatment was performed in 10 cases, surgery as the first treatment in nine cases, biopsies in three cases, chemotherapy in one case, and monitoring in one case due to the patient's age and associated diseases.

The prognosis depends on the size and whether or not they have metastases and develop carcinoid syndrome (less than 10%). In our cases the figure was 4% (one case of d-NET with liver metastases).

Gastroduodenal NET were the most common in our series, probably due to search bias; they were resolved by endoscopic treatment (mainly AP or band EMR). EUS contributed to a higher complete resection rate, with a complication rate similar to that described in the literature.[5]

In conclusion, the overall incidence of NET may be increasing. The proportion of d-NET has also increased compared to historical series, probably due to the widespread use of endoscopy. In our experience, most d-NET smaller than 1 cm can be treated endoscopically with snare resection after submucosal injection or band-assisted mucosectomy, with very good results and a low complication rate.

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2444-3824/ © 2024 Published by Elsevier España, S.L.U.

Challenging the disease: Esophageal achalasia type II in a patient with myotonic dystrophy type I

Retando a la propia enfermedad: acalasia esofágica de tipo II en paciente con distrofia miotónica de tipo I

Myotonic dystrophy type I or Steinert's disease is caused by the abnormal expansion of the CTG trinucleotide of the *DMPK* gene, and its severity appears to increase proportionally to the number of repeats.[1,2] It is a myotonic phenomenon accompanied by progressive weakness of the muscle fibres which, given its multisystemic nature and genetic variability, affects different areas in addition to the motor sphere (gastrointestinal, respiratory, cardiological and endocrine).[1]

Gastrointestinal symptoms sometimes manifest first, with dysphagia being the most important of these for the patient, as the progressive loss of oesophageal motility and swallowing reflex lead to problems such as an increased risk of bronchoaspiration.[1,3] This type of dysphagia cannot usually be detected in endoscopic or radiological studies, so manometry is the test of choice for diagnosis.[2]

As Steinert's disease involves progressive degeneration of the striated muscle and the upper oesophageal sphincter (UOS) is composed of fibres of the cricopharyngeus muscle, the manometry finding most expected in these patients is decreased sphincter tone.[2,4] Although more controversially, hypotonia of the lower oesophageal sphincter (LOS) is also frequently observed in most of these manometry tracings when compared to healthy controls,[4] suggesting some smooth muscle involvement in this disease (and explaining the high rate of gastro-oesophageal reflux observed in these patients).[2] However, the absence of LOS relaxation with swallowing is a highly unusual finding in the literature.[3,5,6]

We describe the case of a 56-year-old male with type II diabetes mellitus, primary hypogonadism and obstructive sleep apnoea, on treatment with metformin. At the age of 54 he began to experience progressive loss of strength in both hands and, after neurological investigations, was finally diagnosed with muscular dystrophy type I. Two years later, he was admitted for dysphagia of liquids and solids with significant associated weight loss. Oesophagogastroduodenoscopy was reported as normal, with no hiatus hernia or other relevant findings. On discharge, oesophageal manometry was performed where, in addition to decreased UOS tone, there was also elevated LOS tone, increased integrated pressure relaxation and pan-oesophageal pressurisation with all swallows without peristaltic wave generation, these findings being compatible with type II achalasia (Fig. 1). At the patient's clinic follow-up, in view of the impact on his quality of life, it was decided to perform endoscopic pneumatic dilation (taking into account the patient's opinion and the experience of the centre). This was done with great success, as three months later the patient remains clinically asymptomatic.

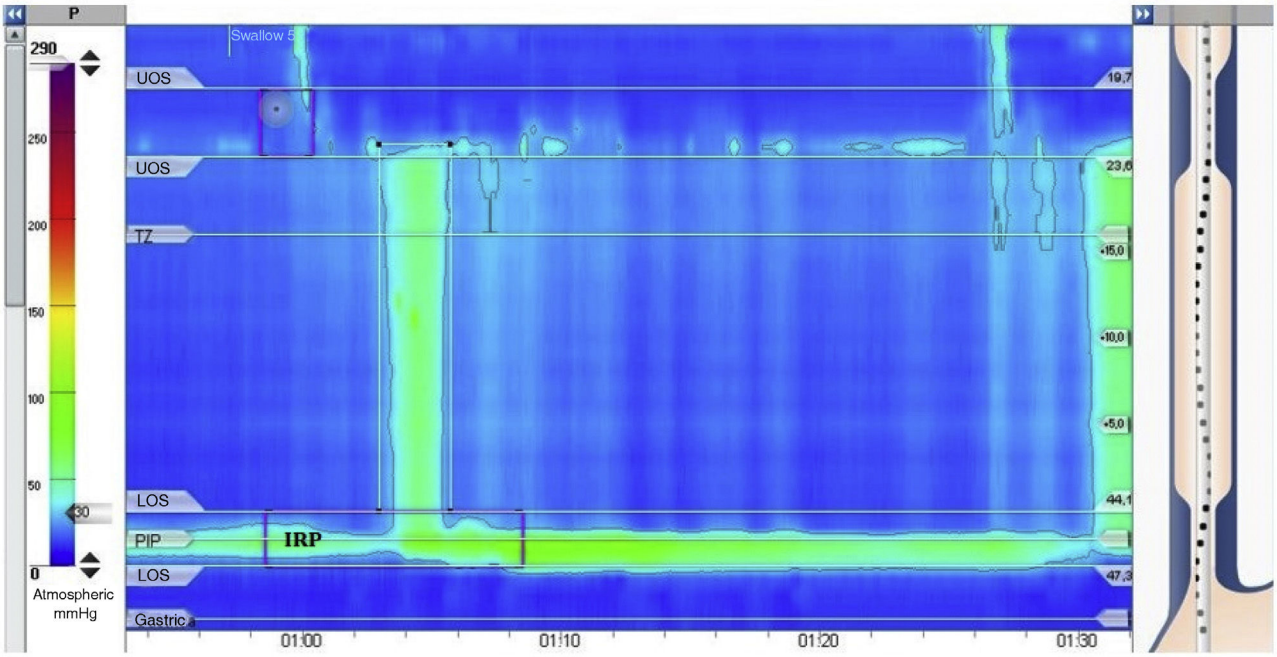

Figure 1 Manometry recording of the patient. In addition to increased LOS tone (48 mmHg), we found increased integrated relaxation pressure (25 and 23 mmHg/4 s lying down and upright respectively) and pan-oesophageal pressurisation without peristaltic wave generation, manometry findings compatible with type II oesophageal achalasia. PIP: pressure inversion point; TZ: transition zone.

Steinert's disease has a tendency towards hypotonia of the LOS, without generation of resistance to the passage of the bolus.[1,2] Although somewhat open to debate, the neural theory could explain this frequent involvement of smooth muscle at this level, suggesting a mixed myopathic and neurological aetiology[2]; while the myopathic component seems to explain the weakness of the pharyngeal contraction, the reduced UOS tone and the reduced amplitude of the peristaltic wave (with normal coordination), the neurogenic component explains both the lack of coordination of peristalsis (with normal amplitude) and the weakness of the tone of the predominantly smooth LOS.[1,2] This neurological dysfunction appears to be due to inhibition of nitric oxide synthase, which causes alterations in smooth muscle hyperpolarisation and relaxation, which in turn could explain its functional compromise in the absence of significant histological abnormalities in the smooth muscle.[3]

However, the manometry diagnosis of oesophageal achalasia in patients with Steinert's disease is unusual. We found a total of three more cases published in the literature. Our patient appears to be the second case of type II oesophageal achalasia described after Doukas et al., who report a directly proportional relationship between the number of CTG triplet repeats (in their patient, >200) and the expression of the disease.[1,4] Ghazaleh et al. report on a patient with type I achalasia and concomitant cricopharyngeal achalasia (permanently increased basal tone of the UOS with no relaxation during swallowing).[3] In the case of Sato et al., their patient's manometry changed dramatically over three years (in parallel with the progression of the disease) from type I achalasia to exaggerated relaxation of the LOS on bolus passage. These authors point out that such a development could be explained as part of the natural process of the disease (when in the early stages myotonic phenomena predominate over muscle weakness).[5] More reports are needed to support such a theory and, whether or not it is true, in our opinion, it should not delay the patient's symptom relief if their dysphagia is significantly impairing their quality of life.

The development of achalasia in patients with Steinert's disease therefore continues to represent a theoretical challenge to the natural progress of the disease which we believe it is necessary to further investigate, in order to improve the management of this condition and help our patients have the best possible quality of life.

Funding

No funding was received for this study.

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