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Case report

Idiopathic hypertrophy of the oesophagus in children

A case report and review of the literature

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Summary. Idiopathic hypertrophy of the oesophagus is a rare entity. Of approximately 50 cases reported in the literature, only 5 are in children. The case of an 8-year-old girl is presented and compared with those previously reported.

Key words: Oesophageal tumours – Mediastinal tumours

Introduction

Idiopathic hypertrophy of the oesophagus is a rare condition mostly occurring in adults, males seeming to be more often affected than females. In only five cases has the condition been reported in children: Pritchard and Hillier (1920), Guthrie (1945), Spencer and Hudson (1961), Blank and Micheal (1963), and Uhrich (1965). The morphological features and the clinical symptoms of this lesion are quite different from those in megaoesophagus, achalasia and to some extent in diffuse spasm of the oesophagus, although these entities are sometimes confused. We report the condition in an 8-year-old girl who had suffered from respiratory disorders from 3 years of age.

Case report

Clinical history. The girl was born in September 1980, the first child of healthy parents. Pregnancy and birth were uneventful, birthweight 3.4 kg, length 50 cm. The child was healthy during infancy and early childhood. At the age of 42 months she had a sudden episode of cyanosis, interpreted as a breath-holding spell. One month later she developed an upper urinary tract infection which was treated with antibiotics. A urological workup revealed a vesico-ureteric reflux grade III and a distal stenosis of the urethra for which a submucous urethrotomy was performed. The infection and the reflux resolved completely but microhaematuria persisted. A routine chest X-ray showed unexpected widening of the mediastinum which was found to be due to a slightly thickened oesophagus. The diagnosis of achalasia was made, although the girl had no symptoms of dysphagia.

At the age of 75 months, the girl was hospitalized because of attacks of dyspnoea, on one occasion losing consciousness. The chest X-ray revealed an extremely thickened oesophagus (Fig. 1). There was a history of dysphagia with solids and recurrent respiratory infections. There was no history of chest pain. CT scan of the chest (Fig. 2) revealed compression of the trachea by the oesophagus, with a wall 2 cm thick and increased peristalsis throughout the oesophagus. The patient was transferred to a paediatric surgical department where Heller's myotomy and a Nissen plication were performed. The child's condition did not improve markedly. The difficulties in eating solids improved somewhat but the respiratory problems persisted. She had several respiratory infections and these difficulties increased to such an extent that removal of the oesophagus became imperative.

This was carried out in February 1988. She was fed post-operatively through a gastric fistula. Her condition improved rapidly and some months later the stomach was divided into halves and the left half was interposed between the cervical oesophageal remnant and the cardia. She remains healthy at the time of this report.

Macroscopic findings. Examination of the resected material revealed a 22-cm-long portion of the lower two-thirds of the oesophagus (Fig. 3). The wall was greyish-white and thickened uniformly to a depth of 25 mm, due to hypertrophy of the muscle layers (Fig. 4). The mucosa was smooth and white with longitudinal folds. The lumen of the oesophagus was slightly dilated. The inner-muscle layer showed prominent oblique striations. According to the operating surgeon's description, the oesophagus proximal to the upper-thoracic inlet was normal and distally the thickening of the oesophageal wall ceased abruptly at the cardia.

Histology. Histological examination reveals normal mucosa but the muscularis mucosa is hypertrophied. The submucosa is thickened and shows a scanty lymphocytic infiltration. The thickening

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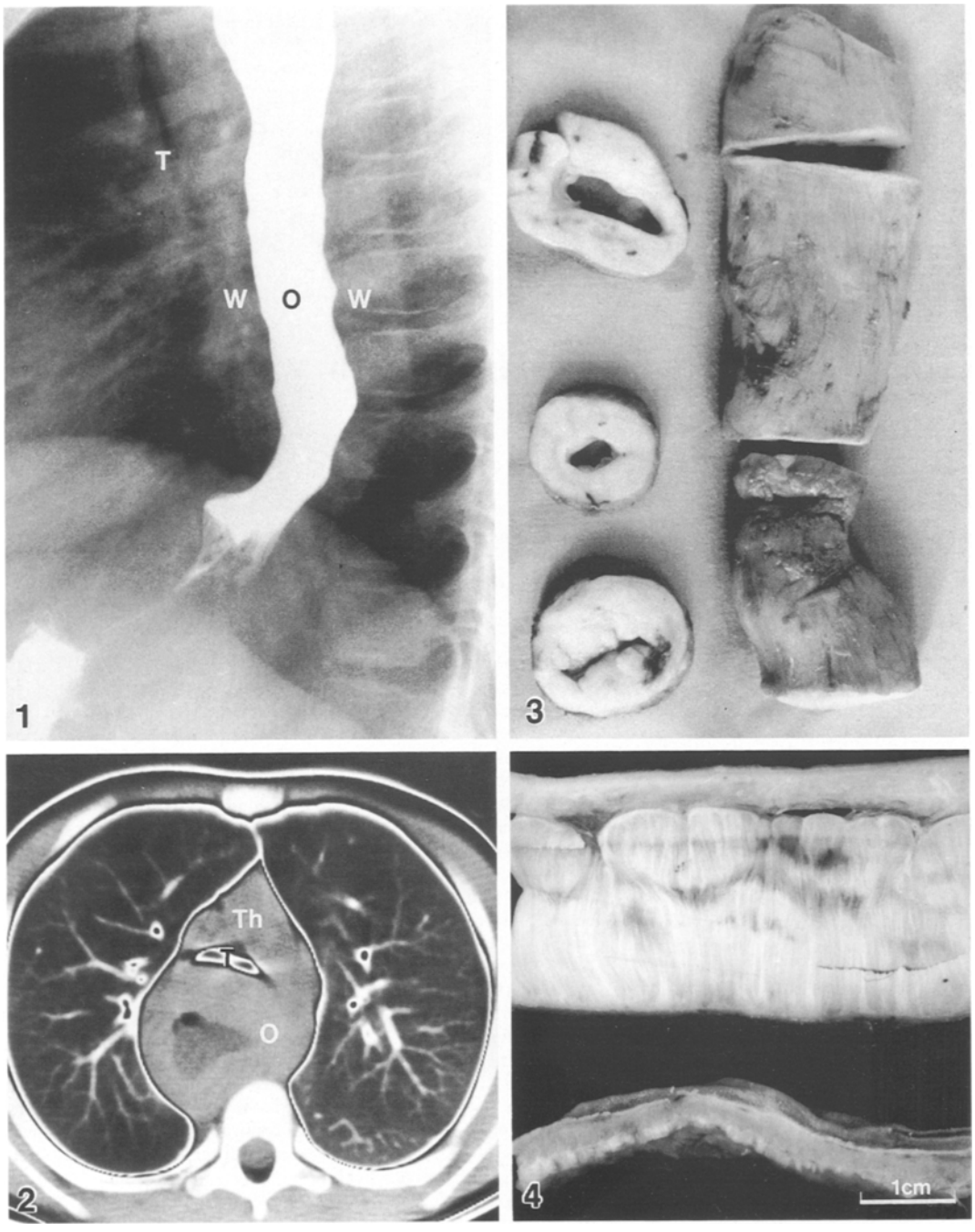

Fig. 1. Lateral view of the barium swallow: increased thickness of the wall (*W*) of the oesophagus (*O*); anteriorly the highly compressed trachea is seen (*T*)

Fig. 2. CT scan showing the hypertrophic oesophagus (*O*) and compression of the trachea (*T*) shortly below the bifurcation; thymus (*Th*)

Fig. 3. Macroscopically: the resected oesophagus

Fig. 4. Longitudinal dissection of the oesophagus; note the marked transverse striations; below, a normal oesophagus

of the wall is due to giant hypertrophy of the inner-circular muscle layer by hypertrophy and increase in number of smooth muscle cells. The macroscopically described ablique striations are in fact hypertrophic muscular bundles (Fig. 5). Also small myomas and areas with myolysis can be seen which are thought to be due to trophic disorders. The outer-longitudinal muscle layer shows only slight hypertrophy. The number of ganglion cells is moderately reduced.

Discussion

Excessive diffuse hypertrophy of the oesophagus, normal motility, normal oesophagoscopy and, in comparison with the extent of the pathology, minor problems in swallowing led us to the diagnosis of idiopathic hypertrophy of the oesophagus.

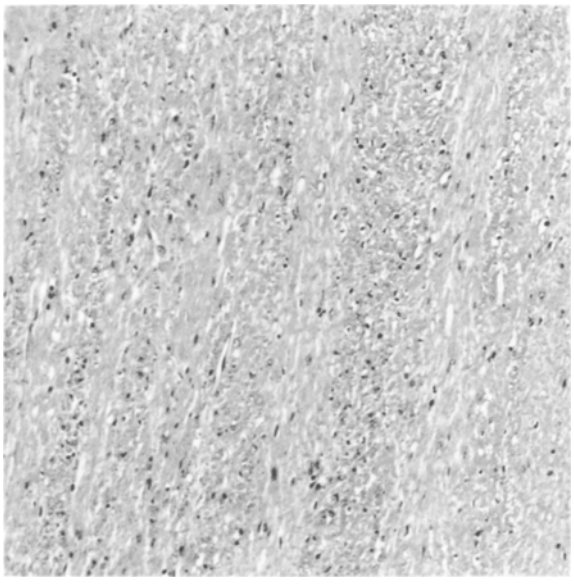

Fig. 5. Histological aspect: the transverse striations are seen to be hypertrophic muscular bundles. ×100

This rare entity is rather poorly defined as a diffuse hypertrophy of the muscle layers, mainly the inner-circular layer, of the lower oesophagus, beginning in the mid-portion and extending to a maximal thickness of 15 mm at the cardia. The intramural nerve plexuses appear normal; sometimes the number of ganglion cells is slightly reduced. There is only moderate dilatation of the lumen of the oesophagus and in many cases the finding was incidental. We want to emphasise the fact that the clinical symptoms of idiopathic hypertrophy of the oesophagus largely depend on the extent of the hypertrophy. If the hypertrophy is slight the disease can be asymptomatic and the lesions are found incidentally at autopsy. In cases with marked hypertrophy and involvement of a large portion of the organ, respiratory disorders and dysphagia are leading symptoms. In achalasia and in its congenital form, the megaoesophagus, the major finding is of increased tonus of the cardial sphincter with the inability to relax. This leads to dilatation of the oesophagus and subsequent loss of ganglion cells in the intramural nerve plexuses of the dilated portion. Hypertrophy of the smooth muscle is only slight. The major clinical symptom is dysphagia.

Another differential diagnosis is diffuse spasm of the oesophagus. In this entity the basic condition is a characteristic distortion of oesophageal motility. On swallowing, the primary peristaltic wave may be obliterated or replaced in the involved areas of the oesophagus by non-progressive prolonged contractions. These accentuated tertiary-type peristaltic waves produce a distinctive radiological appearance of the barium-filled oesophagus, known as curling. They are thought to be responsible for the oesophageal pain the major clinical symptom which accompanies swallowing in patients with diffuse spasm (Katz et al. 1974). Hypertrophy of the smooth muscle is much less than in idiopathic hypertrophy of the oesophagus. In the latter disease, pain accompanying swallowing is very uncommon.

The first description in the literature was given by Albers (1839). He quoted Baillie (1799), who described as submucous tumour which in our opinion was probably a neurinoma. Sloper (1953) found 32 cases in the literature, only 25 of them with microscopic descriptions. Since then only a few new cases have been added: Spencer and Hudson (1961), Blank and Micheal (1963), Uhrich (1965), Ferguson et al. (1969), Katz et al. (1974). Semantic confusion makes it very difficult to isolate the cases of true idiopathic hypertrophy of the oesophagus.

There are only five paediatric cases reported in the literature. Pritchard and Hillier (1920) reported the case of a 3-month-old boy with hypertrophy of the oesophagus, cardiac, pyloric and ileocaecal sphincters; Guthrie (1945) described a case of a 11-year-old girl, who suffered from diffuse hypertrophy of the oesophagus, duodenum, jejunum, and pylorus. The other sphincters, including the cardiac sphincter were normal. The histological examination of the kidneys revealed acute glomerulonephritis. Spencer and Hudson (1961) reported the case of a 3-year-old pseudohermaphrodite male with idiopathic muscular hypertrophy of the oesophagus, stomach, pylorus, duodenum jejunum, and ileum. Vomiting was the principal symptom. Blank and Micheal (1963) reported the case of a girl, who, 6 months after birth, developed progressive respiratory disorders, caused by a hypertrophic oesophagus, which led to tracheostomy at the age of 26 months. Several weeks later, she suddenly began to gasp, suffered a cardiac arrest, and died despite temporary improvement following open cardiac massage. Finally Uhrich (1965) reported the case of massive hypertrophy of the oesophagus, stomach, and small bowel in a 10-day-old male infant. The hypertrophy of the pylorus of the stomach apparently was present at time of birth; the involvement of the small bowel occurred approximately 20 days after birth.

Our case is the sixth reported paediatric case of idiopathic diffuse hypertrophy of the oesophagus. In contrast to the cases of Pritchard and Hillier (1920), of Guthrie (1945), of Spencer and Hudson (1961) and of Uhrich (1965), all sphincters are normal in our case. The clinical history of the case of Blank and Micheal (1963) seems to be very similar to our own case and respiratory disorders are also a major feature. We did not find a single description with similar thickening of the oesophagus which reached 2·5 cm thick in our case whereas in the literature the maximum was 1·5 cm.

Several theories of the aetiology of diffuse muscle hypertrophy of the oesophagus have been discussed. An inflammatory aetiology was suggested by Foerster (1863) based on cellular infiltration. This is not a very constant finding in other reports and was only scanty in the submucosa and absent in the muscular layers in our case. The neoplastic theory suggested by Hall (1916) appears improbable because of the diffuse and symmetrical extension of the lesion; small additional myomas are sometimes seen and may cause radiological filling defects. A congenital aetiology was proposed by Guthrie (1945), who saw an 11-year-old girl with a combination of jejunal, duodenal, pyloric and oesophageal hypertrophy. The combination of oesophageal hypertrophy with

pyloric hypertrophy was also described by Ehler (1907), Helmke (1939), Sloper (1953), Spencer and Hudson (1961) and Uhrich (1965). In fact, most of these patients were elderly and less than 10% of all reported cases are children. Helmke (1939) found an association with myocardial hypertrophy and postulated reflex vagal overactivity, responsible for the cardiac, oesophageal and pyloric hypertrophy. Myocardial hypertrophy was absent in most cases, including ours.

Some authors have reported an association with extra-oesophageal lesions, such as carcinomas of bronchus and large intestine, gastrointestinal ulcers and gastritis. Some patients suffered from renal disorders such as glomerulonephritis and pyelonephritis. All of these lesions seem to be coincidental.

As none of these hypotheses or associations seems convincing, we think it is proper to call the entity "idiopathic".

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