Achalasia

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Pathology and aetiology The term achalasia originated from the Greek word ‘khalasis’, meaning ‘failure to relax’. It is uncommon, with a prevalence of 1.8–12.6 per 100 000 persons per year. The aetiology Leopold Auerbach , 1828–1897, neuropathologist, Breslau, Germany (now Wroc ł aw , Poland). Carlos Chagas , 1879–1934, microbiologist, Rio de Janeiro, Brazil. Jeremy Allgrove , contemporary , paediatric endocrinologist, London, UK. Volker F Eckardt , b. 1942, gastroenterologist, Wiesbaden, Germany . inhibitory ganglion cells in the myenteric (Auerbach’s) plexus, possibly related to a virus-induced autoimmune e ff ect. Histol - ogy of m uscle specimens generally shows a reduction in the number of ganglion cells with a variable degree of chronic inflammation. During a normal swallow , the food bolus will trigger primar y peristalsis in the oesophagus by sequential activation of excitatory lower motor neurones. At the same time, relaxation of the LOS allows oesophageal emptying. The mismatch in excitatory and inhibitory activity results in the failure of LOS relaxation and absent peristalsis. With time, the oesophagus dilates and contractions disappear, so that the oesophagus empties mainly by the hydrostatic pressure of its contents. This is nearly always incomplete, leaving residual food and fluid. The air–fluid level in the stomach evidenced on radiography taken in the erect position in normal individuals is frequently absent, as no bolus with its accompanying air passes through the LOS. The oesophagus becomes progres - sively more tortuous and dilated (megaoesophagus); persistent retention oesophagitis due to fermentation of food residues - may predispose to the increased incidence of carcinoma of the oesophagus. In South America, chronic infection with the parasite Trypanosoma cruzi causes Chagas’ disease and the destruction of the myenteric plexus has marked clinical similarities to achalasia. A rare genetic syndrome (Allgrove syndrome) is associated with familial adrenal insu ffi ciency , alacrimia and achalasia. Clinical features The disease is most commonly diagnosed between 30 and 60 years of age. It typically presents with dysphagia (to both solid and liquid), regurgitation and heartburn (often mistaken for GORD), although chest pain/odynophagia is also common in the early stages. Patients often present late and, having had relatively mild symptoms, remain untreated for many years. Patients may or may not have experienced weight loss. Frequently , patients will adjust their diet according to symptoms and can maintain their body weight after an initial drop. An ‘Eckardt score’ was developed to assess the severity of symptoms and monitor treatment outcome (Table 66.1). - Aspiration-related respiratory symptoms and pneumonia can also occur when there is significant stasis of food residue in the dilated oesophagus. The retained food substance can cause fermentation and therefore halitosis. Patients may report regurgitating food that they have ingested before. Diagnosis A high index of suspicion is needed in the diagnosis of achalasia as symptoms can be mild and chronic and can be easily mis - diagnosed as GORD. Endoscopy typically shows frothy saliva pooling in the oesophagus and the presence of food residue. The oesophagus may be dilated and can be tortuous. T he OGJ appears tight and spastic but can usually allow an endoscope to pass with gentle pressure. A normal endoscopy however does not exclude achalasia, as 30–40% of endoscopies are reported as normal before a final diagnosis of achalasia is made. It is an important

investigation to exclude ‘pseudo-achalasia’, often referring to cancer of the gastric cardia mimicking achalasia (Figure 66.24). Barium contrast study typically shows a hold-up of contrast in the distal oesophagus, abnormal contractions in the oesophageal body and a tapering stricture in the distal oesophagus, described as a ‘bird’s beak’ or ‘rat’s tail’ (Figure 66.25). Progressive dila tation leads to a ‘sigmoidal’ shaped oesophagus. A timed barium oesophagogram is used to quantify the height of the retained contrast at a specific time after ingestion to determine the severity of the disease. All these investigations are suggestive of achalasia but definitive diagnosis relies on HRM. Treatment The treatment goal of achalasia is for symptom palliation since there is no therapy to reverse the neuronal degeneration. Therapies target the LOS, aiming to reduce its contractility by pharmacological means or by destroying the muscle fibres using endoscopic or surgical methods. Medical therapy Pharmacological therapy has a limited role. Calcium channel blockers, nitrates or 5 /uni2032 -phosphodiesterase inhibitors are used to - reduce the LOS pressure. Most have limited e ffi cacy in symp - tom improvement. There are also significant side e ff ects, such as headache, oedema and hypotension, after repeated doses. Medical therapy should be reserved for selected patients who are poor candidates for endoscopic or surgical tr eatments.

Disorders of OGJ out /f_l ow Yes 100% failed peristalsis Achalasia I without POP 100% absent peristalsis Yes Yes 100% failed peristalsis All swallows are either Achalasia II with POP in ≥ 20% swallows failed or premature ≥ 20% swallows with Yes premature contractions. Achalasia III Failed peristalsis ± POP may be present Step 2: (if not done) Wet swallows in secondary position + MRS/RDC Elevated LOS IRP persists in No varying positions + elevated IBP/POP Yes No Yes Abnormal TBO or FLIP OGJOO Figure 66.23 Hierarchy diagnostic algorithm of oesophageal motility disorders according to the Chicago classi /f_i cation 4.0. The broad categories of ‘disorders of oesophagogastric junction (OGJ) out /f_l ow’ and ‘disorders of oesophageal peristalsis’ are differentiated by the integrated relax ation pressure (IRP). FLIP , functional lumen imaging planimetry; IBP , intrabolus pressurisation; LOS, lower oesophageal sphincter; MRS, multiple rapid swallow; OGJOO, oesophagogastric out /f_l ow obstruction; POP , pan-oesophageal pressurisation; RDC, rapid drink challenge; TBO, timed barium oesophagram. Table 66.1 Clinical scoring system for achalasia (Eckardt score). Score Symptom Weight loss (kg) Dysphagia 0 None None 1 <5 Occasionally 2 5–10 Daily 3

> 10 Each meal Disorders of peristalsis Abnormal median IRP Yes No Step 2: Wet swallows in secondary position + MRS/RDC Yes Elevated LOS IRP in varying positions ± elevated IBP/POP No No Yes Absent 100% failed peristalsis contractility No Distal ≥ 20% swallows with Yes oesophageal premature contractions spasm No ≥ 20% swallows with Hypercontractile Yes hypercontractility oesophagus No No evidence of OGJ Ineffective 70% ineffective or ≥ 50% Yes out /f_l ow obstruction oesophageal failed swallows motility No No evidence of disorder of peristalsis Consider meal challenges based on symptom

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Retrosternal pain Regurgitation None None Occasionally Occasionally Daily Daily Each meal Each meal

Botulinum toxin Botulinum toxin is a potent presynaptic inhibitor of acetylcho line release from nerve endings. When injected endoscopically into the LOS, it interferes with the LOS cholinergic excitatory neural activity and paralyses the sphincter muscle. The reported symptom relief decr eased from 70% in 3 months to around 40% in a year. The injection usually has to be repeated after a few months. Because the e ff ect is temporary , it is sometimes used when the diagnosis of achalasia is in doubt. Repeated Ernst Heller , 1877–1964, surgeon, St George's Krankenhaus, Leipzig, Germany . injection may result in scarring, making subsequent treatments more di ffi cult. It should not be o ff ered as first-line treatment in patients who are suitable for myotomy or pneumatic dilatation and its indication is usually r estricted to elderly patients with comorbidities. Pneumatic dilatation This involves stretching the LOS with a non-compliant balloon to disrupt the sphincter muscle and render it less competent. Plastic (polyethene) balloons with a precisely controlled exter - nal diameter are used. If the pressure in the balloon is too high, the balloon is designed to split along its length rather than expanding further. Balloons of 30–40 /uni00A0 mm in diameter are available and are inserted over a guidewire. There is no standardised dila tation protocol. Generally , it is preferred to have serial dilatations in a graded manner, from 30 /uni00A0 mm to 35 /uni00A0 mm and 40 /uni00A0 mm. Serial pneumatic dilatation has similar e ffi cacy to surgical myotomy in selected patients. Features that predict optimal response ar e: patients older than 45 years, female, those with an undilated oesophagus, those who have responded to first dil - atation and those with type II achalasia. Perforation is uncom - mon; the reported incidence averaged about 1.9% (0–16%). With a 30-mm balloon, the chance of perforation should be less than 0.5%. The risk of perforation incr eases with big - ger balloons, which should be used cautiously for progressive - dilatation over weeks. It is important to have an experienced endoscopist performing the procedure and surgical back-up in case of perforation. Heller's myotomy This involves cutting the muscle of the lower oesophagus and gastric cardia (Figure 66.26).Typically , anterior myotomy is

Figure 66.24 Pseudo-achalasia in a patient with cancer of the oesophagogastric junction. The patient was referred as having possible achalasia based on a barium contrast study the obstruction, prompting a computed tomography scan surgical specimen (c) . Figure 66.25 Barium contrast study showing the typical 'rat's tail' appearance of achalasia. (a) . Endoscopy could not get past (b) , making a diagnosis of cancer. The resected

and 2–3 /uni00A0 cm distally into the gastric cardia. Transabdominal or transthoracic approaches have been advocated. Currently , the standard procedure is a laparoscopic approach. The major complication is GORD, which can occur in up to 40% of patients. The addition of a partial fundoplication (anterior Dor or posterior Toupet) has been shown to be e ff ective in reducing the incidence of GORD. A complete 360° fundoplication (Nissen) is considered contraindicated because the increase in outflow resistance against an aperistaltic oesophageal body will probably result in postoperative dysphagia. Laparoscopic myotomy is superior to single pneumatic dilatation in e ffi cacy and durability . The surgical outcome is better in types I and II achalasia than in type III. For the latter, a longer extended proximal myotomy is often needed for adequate treatment. Peroral endoscopic myotomy Peroral endoscopic myotomy (POEM) involves opening the mucosa at a short distance proximal to the intended myotomy site. Entrance is gained into the submucosal plane, which is extended distally to about 2–3 /uni00A0 cm into the gastric cardia. The circular +/– longitudinal muscles are then cut using ESD instruments. Typically , the myotomy extends a

minimum of 6 /uni00A0 cm in the oesophagus proximally and 2 /uni00A0 cm into the gastric cardia distally (Figure 66.27). The mucosal opening is then closed with endoclips. In type III achalasia, there is a spastic component at the distal oesophagus that responds less well to pneumatic dilatation and Heller’s myotomy . POEM has the advantage in that it can extend the length of the myotomy proximally , tailored to preoperative HRM and barium swallow parameters. POEM can also be utilised to treat other types of ‘spastic’ oesophageal motility disorders such as distal oesoph ageal spasm and hypercontractile oesophagus. Randomised controlled trials have demonstrated similar e ffi cacy of POEM to pneumatic dilatation and Heller’s myotomy in relieving dysphagia. Without any antir eflux procedure, the incidence of GORD is expectedly higher in POEM compared with Heller’s myotomy with partial fundoplication. The incidence of oesophagitis at 3 months after POEM can be as high as 57%, which may subject patients to lifelong acid suppression therapy or subsequent antireflux operation. Oesophagectomy Oesophagectomy is reserved only for the treatment of patients with ‘end-stage’ achalasia with a sigmoidal or megaoesopha gus that is not responding to other methods (Figure 66.28 Depending on the chronicity of the disease, the symptoms of achalasia may be tolerated. However, a grossly dilated oesoph agus pr edisposes to regurgitation and aspiration pneumonia. Balancing the risk of an oesophagectomy with the patient’s quality of life and risk of aspiration complication, surgery can be a reasonable option for surgically fit patients. Follow-up Treatment success is usually defined by symptom relief. The Eckardt score is quantified and compared with the preoperative score. Patients should be counselled on a post-treatment diet as the oesophageal body motility remains defective. Ideally , - -). - HRM, barium contrast study , endoscopy and 24-hour pH monitoring should be performed postoperatively to objectively assess LOS function, bolus retention, response to treatment, presence of oesophagitis and acid reflux. This depends on the availability of resources and patients’ preference.

(b) (c) Figure 66.26 Laparoscopic myotomy and Dor hemi-fundoplication. (a) The lower oesophageal myotomy extending onto the stomach for at least 2 cm. (b) Completion of the myotomy; the light of the endo

scope can be seen shining through the thin mucosa. (c) Completion of the Dor anterior fundoplication.

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Figure 66.27 The procedure of peroral endoscopic myotomy (POEM). mucosa and the muscle layer down to the stomach. (c) Myotomy starts a short distance below the mucosal opening. into the proximal stomach. (e) The mucosal opening is closed by endoclips. Achalasia is the most common oesophageal motility disorder A normal endoscopy does not exclude the diagnosis of achalasia Beware of pseudo-achalasia HRM is the gold standard for the diagnosis of oesophageal motility disorder Laparoscopic myotomy, pneumatic balloon dilatation and POEM are effective treatments of achalasia Type III achalasia may be better treated with long myotomy by POEM

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