Mostly, achalasia has an insidious onset and the patients often had an odyssey of consultations of physicians and medical specialists before the correct diagnosis is made. It is all the more important to discuss achalasia in university and to remember it in the right situation - be it when examining patients or during exams. Below is a discussion of all the important information concerning achalasia.

Definition of Achalasia

Achalasia as a functional disorder of the esophagus
Achalasia is a functionally caused narrow position of the muscles of the lower esophageal parts. It also shows decreased peristalsis and absent physiological slackening of the lower esophageal sphincter at swallowing. If not treated, achalasia progresses from stage I (hypermotile form) to stage II (hypomotile form) and stage III (amotile form).

Epidemiology of Achalasia

Achalasia as an exceptional disease

With an incidence of 1/100,000 inhabitants, achalasia is a rather rare disease. It affects men and women with the same frequency, mostly in the third to fifth decade of life. Achalasia is even scarcer in the context of the autosomal-recessive congenital disease triple-A-syndrome (achalasia, alacrimia, and adrenal insufficiency).

Etiopathogenesis of Achalasia

Features of achalasia

To date, the causes of primary achalasia are unknown. Due to an unclear genesis, degeneration of the inhibitory ganglia cells of the myenteric plexus (Auerbach’s plexus) occurs in the lower esophagus. Normally, these neurons make for a relaxation of the esophagus, especially during swallowing. Their absence results in deficient slackening in the course of the swallowing reflex and an increased resting pressure of the lower esophageal sphincter. One also speaks of a coordination disorder of the esophagus motility.

Since food now often bottles up in front of the narrowed lower esophageal sphincter, a dilatation of the esophagus directly in front of the narrow passage develops in the further course. In very distinct findings, a ‘megaesophagus’ results.
If achalasia occurs in the context of another disease, this is referred to as secondary achalasia. Amongst others, Chagas’ disease (Trypanosoma cruzi infection) or tumor diseases can be the cause for it.

**Symptoms of Achalasia**

**Dysphagia and regurgitation at achalasia**

Primarily, the patients complain about having dysphagia for several years. Often, they have to drink a lot of fluids after food intake to be able to completely swallow solid food components. However, even swallowing fluids is considered hard in later stages.

Especially in lying position, regurgitation of food occurs, which is associated with the complications of aspiration pneumonia and esophagitis. This esophagus inflammation called retention esophagitis increases the carcinoma risk to the 30-fold compared to the normal population. In the course of increased *regurgitation, foetor ex ore* can often be detected.

Furthermore, the patients often describe bloating. Very rarely, convulsive pain (*hypermotile achalasia*) is also present, especially in the beginning of the disease. It decreases in the further course.
Diagnosis of Achalasia

Anamnesis of achalasia

Concerning anamnesis, one has to especially ask if the complaints occurred suddenly or within very little time and if they are rapidly progressive. This would rather suggest a carcinoma as a suspected diagnosis. However, the complaints mostly existed for many years, and the patient often presents with a long medical history in case of achalasia.

Manometry of achalasia

Manometry (physical pressure measurement) is the gold standard for detection of achalasia. Pressure measurement occurs in different parts of the esophagus (the normal resting pressure is at ca. 20 mmHg).

Three characteristic features can be detected: absent slackening during the swallowing reflex, pathological peristalsis, and an increased resting pressure. In the course of manometry, classification of the severity of achalasia is mostly made at the same time:

<table>
<thead>
<tr>
<th>Stage</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stage I</td>
<td><strong>Hypermotile</strong>: The muscles of the esophagus actively contract to pass the narrowed lower esophageal sphincter.</td>
</tr>
<tr>
<td>Stage II</td>
<td><strong>Hypomotile</strong>: The distal esophagus dilates at a constant contraction of the lower esophageal sphincter.</td>
</tr>
<tr>
<td>Stage III</td>
<td><strong>Amotile</strong>: The esophageal muscles do not contract anymore. Only the esophageal sphincter is still narrow; increasing dilatation of the preceded esophageal part.</td>
</tr>
</tbody>
</table>
Esophagus barium swallow at achalasia

At an esophagus barium swallow, the so-called ‘champagne glass shape’ with prestenotic dilatation (megasophagus) can be seen in an x-ray. Peristalsis is disturbed. With a delay, the contrast agent drains from the esophagus into the stomach.

**Note:** In exams, x-ray barium swallow images are often shown. You should then recognize the ‘champagne glass shape’ and be able to associate it with achalasia.

Endoscopy with biopsy at achalasia

Esophagoscopy with biopsy is obligatory to exclude the presence of a carcinoma. Due to the risk to develop a carcinoma in the further course of the disease, the procedure should be repeated every one to two years. In the context of achalasia, endoscopy typically shows a widened esophagus and a lower esophageal sphincter that can easily be passed through the endoscope.

**Note:** Due to the increased risk for carcinomas, regular endoscopic check-ups are necessary.

Differential Diagnosis

Alleged achalasia

The differential diagnoses of esophagus achalasia especially include the esophageal carcinoma, which is mostly characterized by a distinctly more rapid progression. In the event of B symptoms, you should absolutely perform biopsy! Further motility disorders, which are differential diagnoses of esophagus achalasia, are:

- **Esophagus spasm:** The esophagus spasm is a rare, also neuromuscular functional disorder of the esophagus of unknown genesis. **Paroxysmal retrosternal pain** and **dysphagia** occur. 24 h manometry is the leading diagnostics and shows primarily reinforced and continuous esophagus contractions. In milder cases, calcium antagonists are administered. In severe cases, botulinum toxin is injected or it is surgically intervened.

- **Hypercontractile esophagus ('nut cracker esophagus'):** This motility disorder is especially characterized by non-physiologically long lasting contractions (> 5 seconds) of the esophageal muscles and particularly great pressure amplitudes (individual amplitudes are partial > 200 mmHg). This can be measured in manometry. The function of the lower esophageal sphincter is not impaired at this motility disorder.

Therapy of Achalasia

Pharmacological therapy of achalasia

Unfortunately, the long-term results are bad. However, a therapy attempt with a calcium antagonist, e.g., **nifedipine** should be tried, especially in the hypermotile stage. Nifedipine decreases the pressure of the lower esophageal sphincter. It should be applied half an hour before meals.
Interventional therapy at achalasia

**Pneumatic balloon dilatation**: In the course of esophagoscopy, the cardia muscles are dilated or controllingly torn during this procedure. Often, one session is not enough, and the patients have to be treated several times. The success rate amounts to ca. 60%. Due to the risk of perforation (2—5 %), an esophagus barium swallow should always be performed after the intervention for control.

**Attention**: At suspicion of perforation, barium must not be used for this procedure (risk of mediastinitis or peritonitis!), but a water-soluble contrast agent. Another alternative is the endoscopic injection of botulinum toxin. It is injected directly into the lower esophageal sphincter to react its dilatation. Still, the procedure mostly has to be repeated after half a year. Due to the short duration of effect, this method is, thus, for patients, who cannot undergo other interventions due to distinct comorbidities.

Surgical interventions at achalasia

As therapy alternatives at insufficient success of the mentioned methods, surgical procedures are used. The success rate is greater than 90%. However, one has to consider the complication of the post-surgical reflux disease. Primarily, two surgery techniques are used:

1. **Extramucous cardia myotomy according to Gottstein/Heller (laparoscopic or transabdominal)**: The muscles of the stomach entry (cardia) are split in their longitudinal orientation in a ca. 6 cm long line. The mucosa is not affected. To avoid the reflux disease, Nissen’s fundoplication is subsequently performed in most cases.

2. **Transgastric esophagus fundostomy according to Prager**: During this procedure, a left-sided side-to-side esophagus fundostomy (TSE) is performed transgastrically, during which the gastrostomy is sealed. In addition, partial fundoplication is mostly also performed.

In the event of failure of all treatment approaches, esophagectomy with a small intestine interponate has to be considered as a last-resort measure.

**Note**: Balloon dilatation is first-resort therapy.

Review Questions

The answers are below the references.

1. **Which answer most likely describes achalasia?**
   - A. A congenital malformation of the esophagus.
   - B. The disturbed slackening of the lower esophageal sphincter.
   - C. An overshoot in ganglia cells of the esophageal Auerbach’s plexus.
   - D. A decreased resting tone of the lower esophageal sphincter.
   - E. None of the mentioned answers are correct.

2. **Which treatment measure is most likely indicated in a 32-year-old patient with achalasia in the amotile stage?**
   - A. Nissen’s fundoplication
   - B. Esophagectomy with small intestine interponate
   - C. Pharmacological therapy with nifedipine
D. Pneumatic balloon dilatation
E. Botulinum injection into the lower esophageal sphincter

3. A 40-year-old patient presents with long-existing dysphagia in your general medicine practice. He describes that he now has even troubles with swallowing fluids. Especially at night, chronic cough and regurgitation agonize him. Which of the following diagnostic measures proves the suspected diagnosis best?

A. Long-term pH-metry
B. MRI of the neck and thorax
C. H2-breathing test
D. Chest x-ray
E. Esophagus manometry

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Correct answers: 1B, 2D, 3E

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