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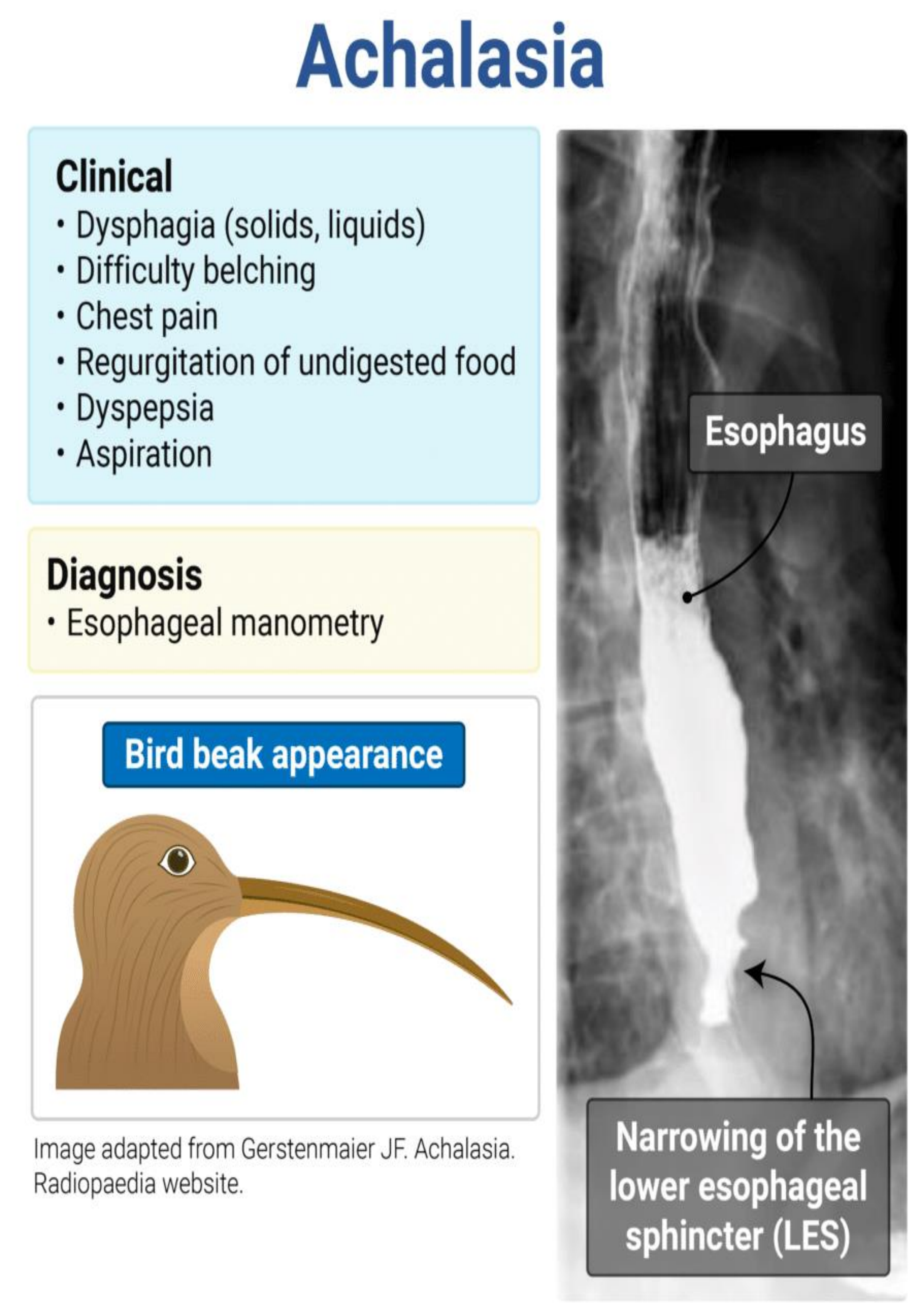

Image adapted from Gerstenmaier JF. Achalasia. Radiopaedia website.

Pediatric Achalasia: Understanding a Rare Esophageal Disorder

Achalasia is a rare esophageal motility disorder affecting children and adolescents. It's characterized by the failure of the lower esophageal sphincter (LES) to relax properly and the absence of normal peristalsis in the esophageal body.

This neurodegenerative disorder occurs less frequently in children than adults, with an annual incidence of only 0.11 per 100,000 children. Despite its rarity, achalasia can significantly impact a child's quality of life, growth, and development when left undiagnosed or untreated.

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Pathophysiology: The Underlying Mechanisms

Neurodegeneration

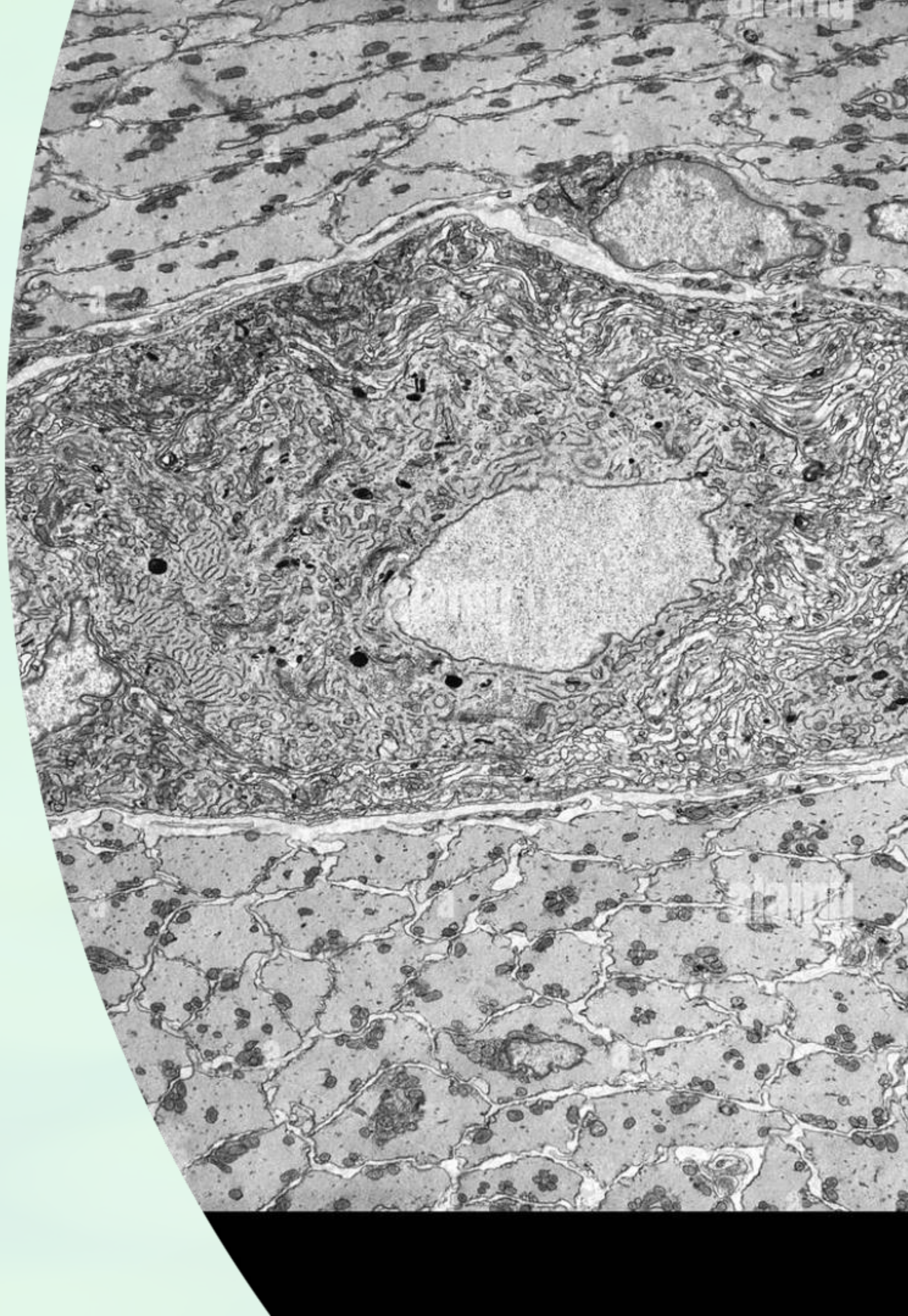
Loss of function in ganglion cells of the esophageal myenteric plexus, creating an imbalance between inhibitory and excitatory neurons

Sphincter Dysfunction

Failure of the LES to relax during swallowing, absence of esophageal body peristalsis, and increased LES resting pressures

Functional Impact

Impaired esophageal emptying leading to food retention, regurgitation, and progressive symptoms



Proposed Mechanisms of Neurological Disruption

Genetic Predisposition

Inherited factors that may increase susceptibility

Viral Infections

Herpes zoster, herpes simplex, measles, HPV

Autoimmune Attack

Cell-mediated and antibody-mediated pathways

Inflammatory Response

Eosinophils and mast cells causing fibrotic remodeling

Research has found varicella-zoster virus DNA in the saliva of a high percentage of achalasia patients, suggesting a potential viral trigger. The inflammatory response, particularly involving eosinophils and mast cells, may lead to increased cytokines and subsequent fibrotic remodeling of the esophageal wall.

Associated Conditions

Genetic Syndromes

- Trisomy 21 (Down syndrome)
- Familial dysautonomia
- AAA syndrome (achalasia, alacrima, ACTH insensitivity)

Neurological Disorders

- Congenital hypoventilation syndrome
- Autonomic dysfunction

Other Conditions

- Glucocorticoid insufficiency
- Eosinophilic esophagitis

The presence of these associated conditions can sometimes provide clues to the diagnosis of achalasia, especially in children with atypical presentations. Understanding these connections may help clinicians identify at-risk populations and initiate appropriate diagnostic workups earlier.

Clinical Manifestations in Children and Adolescents

Dysphagia

Progressive difficulty swallowing, reported in 84.8% of pediatric cases

Regurgitation

Undigested food returning to the mouth, present in 91.3% of cases

Weight Loss

Observed in approximately 26% of children before diagnosis

Chest Pain

Burning sensation or discomfort, affecting 47.8% of pediatric patients

Children with achalasia may also experience coughing or choking during eating (37% of cases), especially at night. These symptoms can significantly impact nutritional status, with many patients presenting with a low Body Mass Index (BMI) at diagnosis.

Atypical Presentations in Younger Children

Respiratory Symptoms

- Recurrent pneumonia
- Nocturnal cough
- Aspiration
- Hoarseness

These respiratory manifestations can often lead to misdiagnosis as asthma or other respiratory conditions, delaying proper treatment.

Feeding Difficulties

- Food refusal
- Prolonged feeding times
- Irritability during meals
- Failure to thrive

In infants and toddlers, these feeding issues may be mistakenly attributed to behavioral problems or other gastrointestinal disorders.

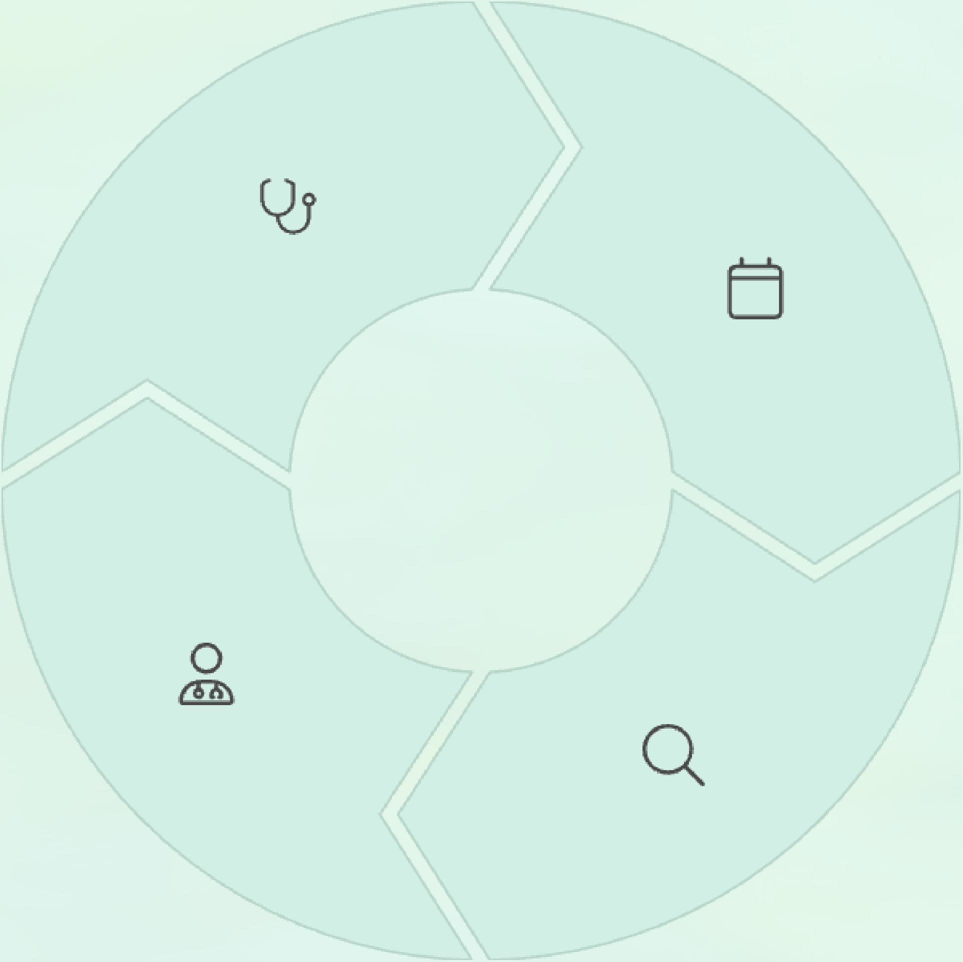
Diagnostic Challenges and Delays

Initial Misdiagnosis

Often mistaken for GERD, eating disorders, or asthma

Prolonged Timeline

Diagnosis delayed by 6-10 years in many cases



Clinical Awareness

Limited recognition due to the rarity of the condition

Symptom Variability

Inconsistent presentation across different age groups

The diagnostic journey for pediatric achalasia is often prolonged and frustrating for patients and families. The rarity of the condition combined with its variable presentation contributes to significant delays in proper diagnosis and treatment initiation.

Types of Achalasia: The Chicago Classification

Type I (Classic Achalasia)

Minimal esophageal pressurization

Type II (Achalasia with Compression)

Higher esophageal pressurization

Type III (Achalasia with Spasm)

Greatest maximal esophageal pressurization

In the pediatric population, Types I and II achalasia occur with similar frequency and share comparable clinical profiles. However, Type II tends to present with higher lower esophageal sphincter pressure and a less dilated esophagus compared to Type I. Type III is less common in children.

Treatment Response by Achalasia Type

Achalasia Type	Treatment Response	Distinguishing Features
Type I	Moderate response to therapies	Classic presentation, minimal pressurization
Type II	Best response to various therapies	Higher basal LES pressure, esophageal compression
Type III	Least responsive to standard treatments	Esophageal spasm, greatest pressurization

Understanding the specific achalasia subtype is crucial for treatment planning and prognostication. Type II patients generally have the most favorable outcomes across different therapeutic approaches, while Type III cases may require more aggressive or specialized interventions.

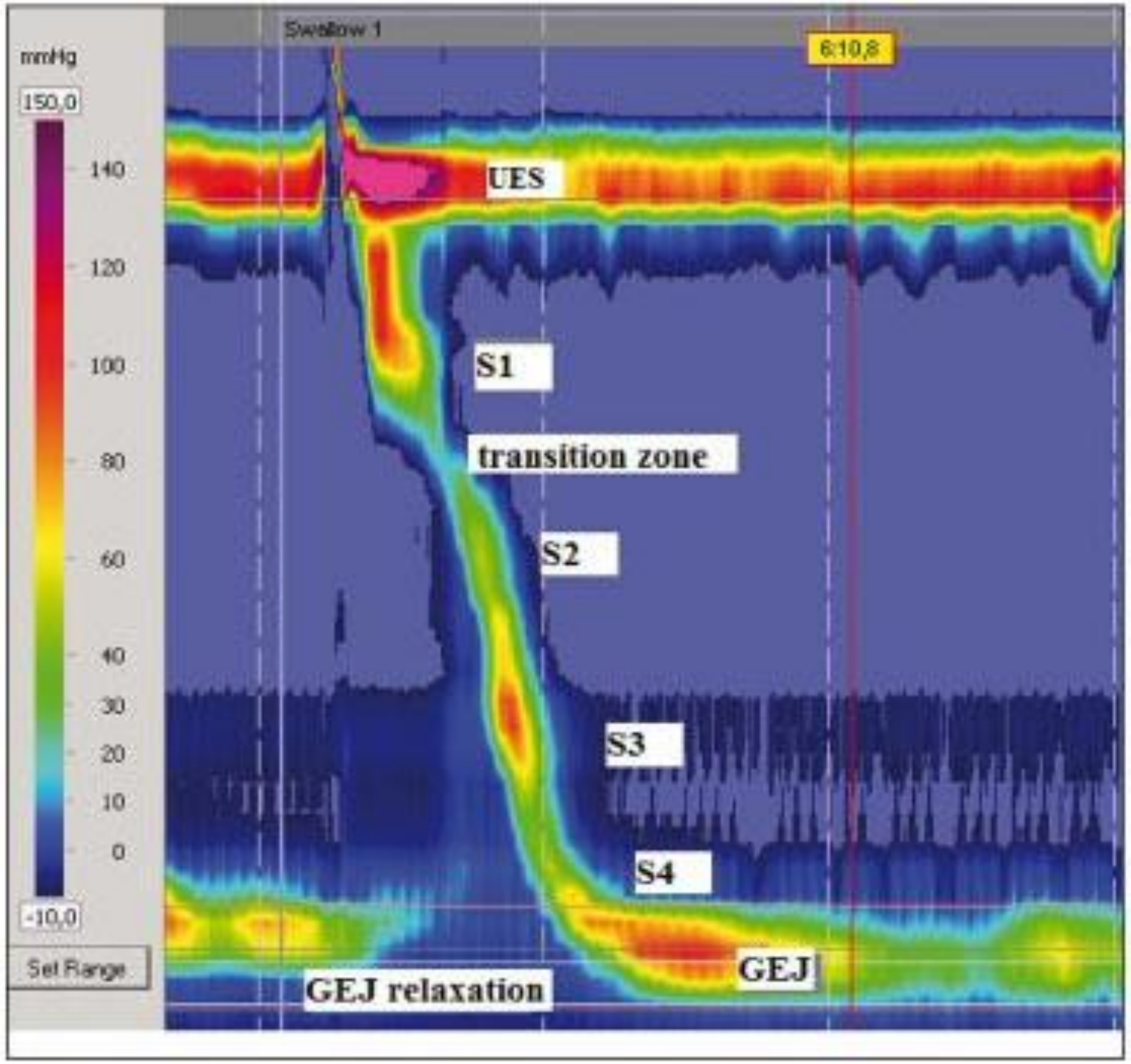
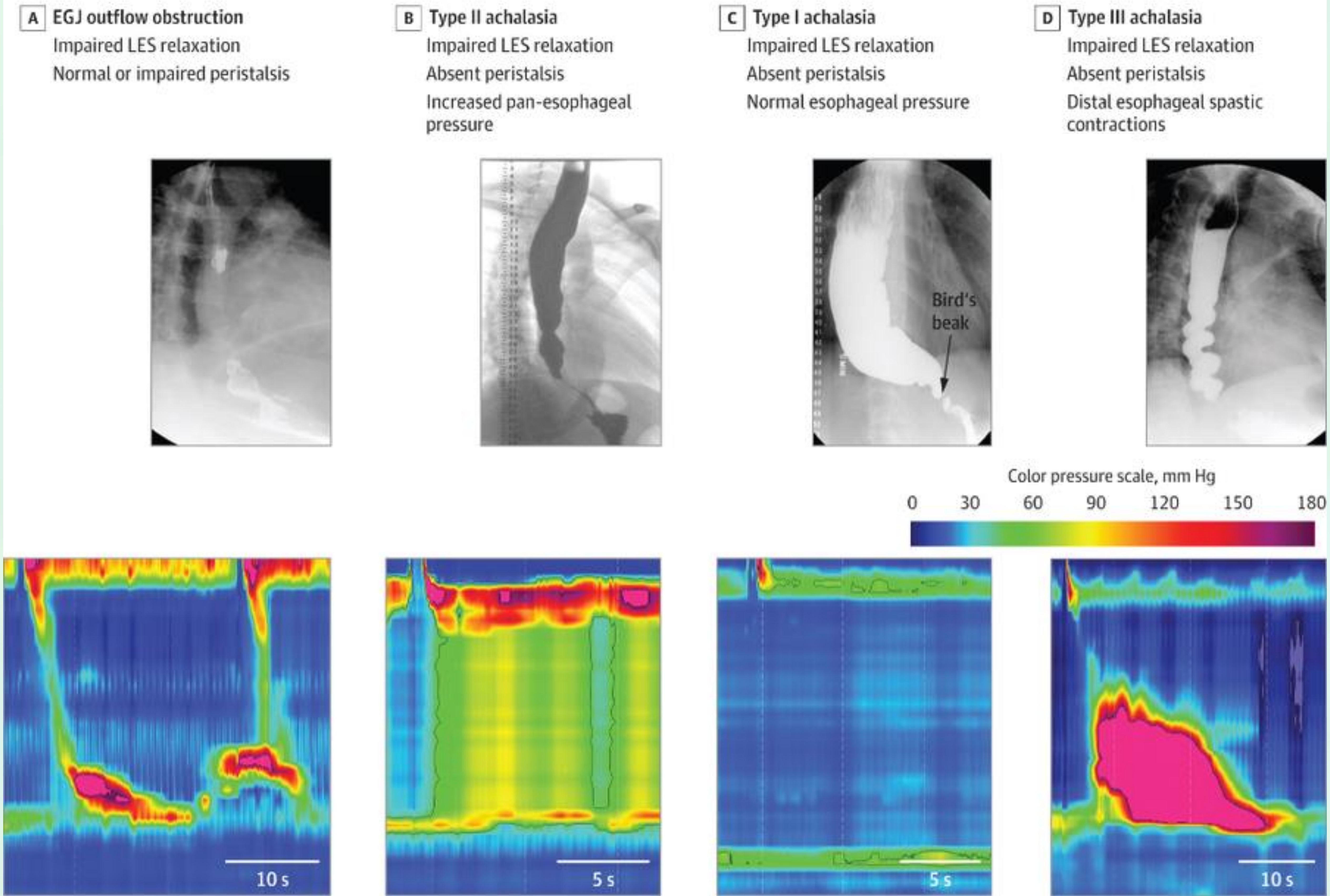
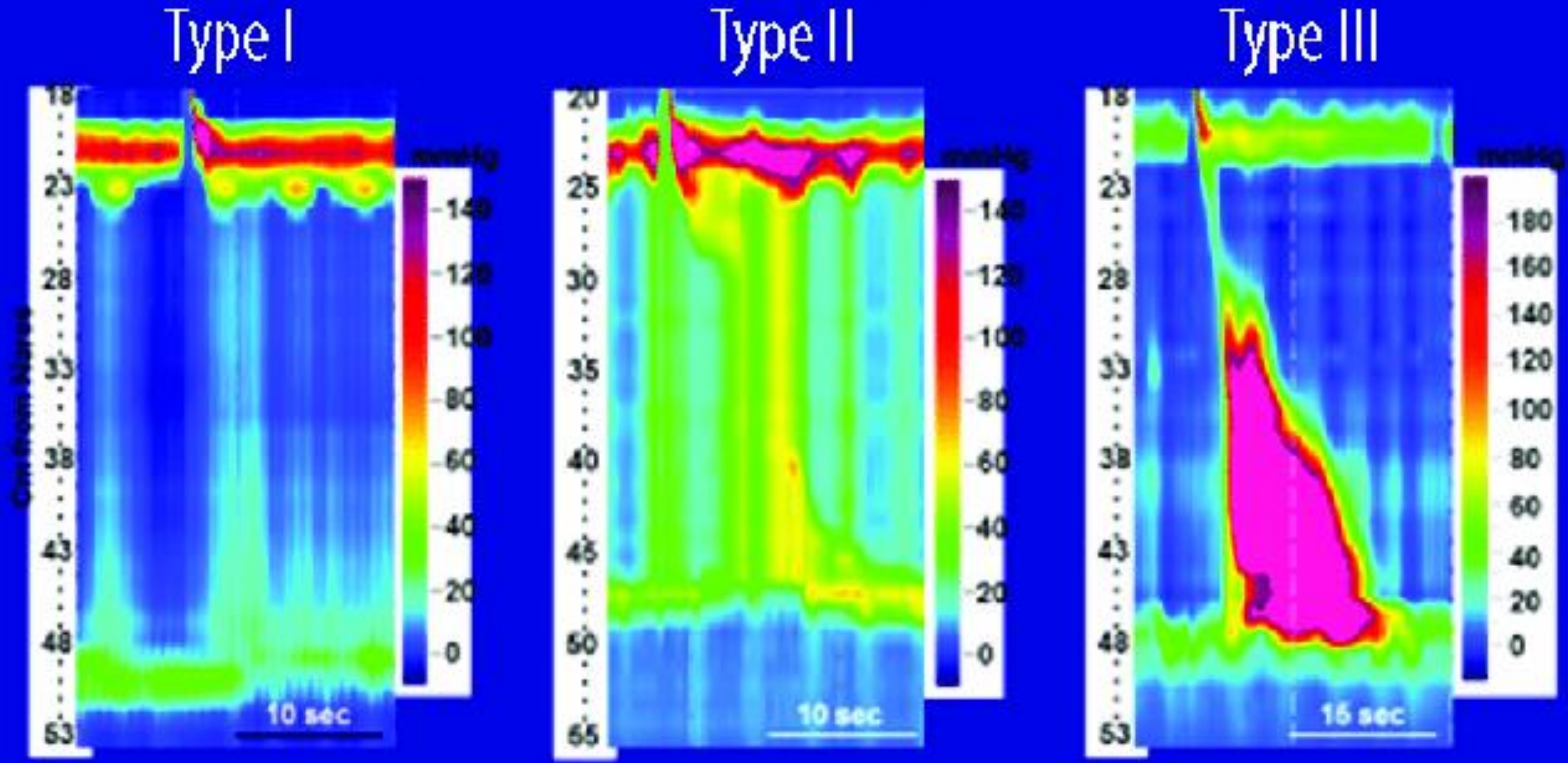
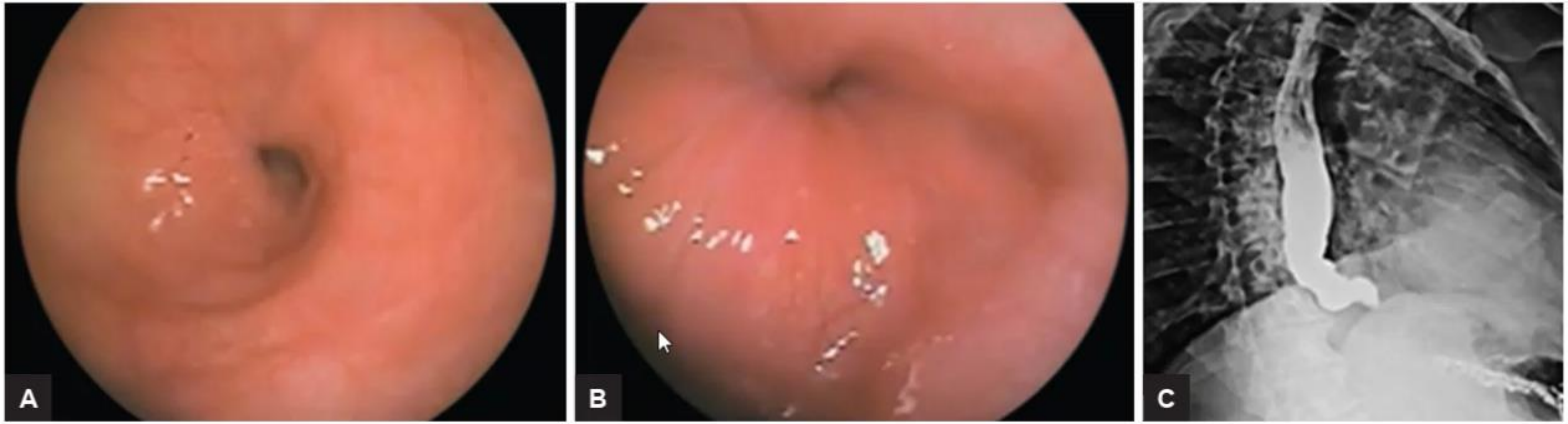


Fig. 1. *Normal peristaltic sequence.* EES: upper esophageal sphincter. UGE: gastro-esophageal junction. S1 is the first contractile segment representing the skeletal muscle component of the proximal esophagus, and extending from EES to the first esophageal pressure fall at the aortic arch region. This low-pressure segment represents the transition zone. The area corresponding to the lower two esophageal thirds is considered an area with predominant smooth muscle that may be further divided up into two overlapping neuromuscular segments (S2 and S3). The forth contractile segment (S4) represents the LES.

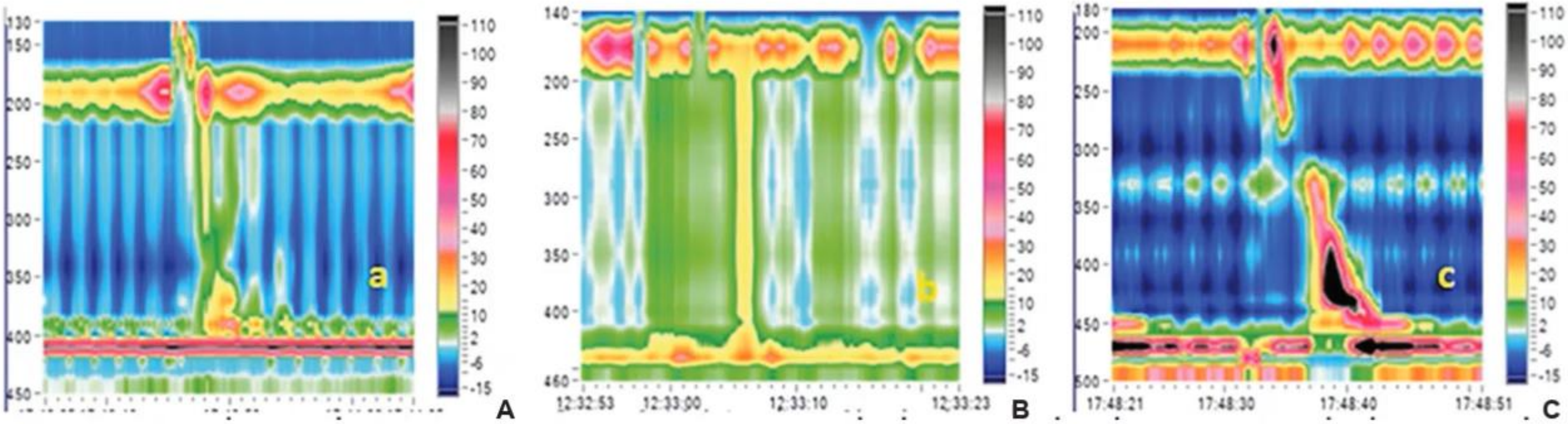

A EGJ outflow obstruction
Impaired LES relaxation
Normal or impaired peristalsis
B Type II achalasia
Impaired LES relaxation
Absent peristalsis
Increased pan-esophageal pressure
C Type I achalasia
Impaired LES relaxation
Absent peristalsis
Normal esophageal pressure
D Type III achalasia
Impaired LES relaxation
Absent peristalsis
Distal esophageal spastic contractions
Bird's beak
Color pressure scale, mm Hg
0
30
60
90
120
150
180
10 s
5 s
5 s
10 s

Achalasia clasification





Figs 1A to C: (A) Endoscopy showing dilated esophagus with liquid food residue; (B) tight gastroesophageal junction, not opening with air insufflation; and (C) barium swallow showing typical "birds beak appearance" of achalasia



Figs 2A to C: High resolution manometry: (A) Type I achalasia cardia; (B) type II achalasia cardia; and (C) type III achalasia cardia

Diagnostic Approach: Barium Swallow Study

Key Features

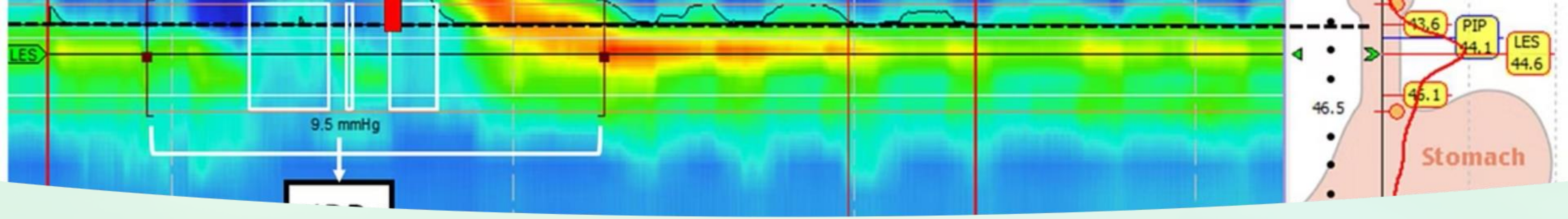
- Dilated esophagus
- "Bird's-beak" tapering of distal esophagus
- Delayed emptying of contrast
- Absence of primary peristalsis

The barium swallow is often the initial diagnostic tool used when achalasia is suspected. In children with advanced disease, this study alone may be sufficient for diagnosis due to the classic radiographic findings.

Advantages

- Non-invasive procedure
- Widely available
- Provides functional and anatomical information
- Well-tolerated by children

The barium swallow study offers a valuable initial assessment that can guide further diagnostic workup. It provides visual evidence of esophageal dysfunction that correlates with clinical symptoms.



Diagnostic Gold Standard: Esophageal Manometry

Elevated Resting LES Pressure
Higher baseline pressure at the lower esophageal sphincter

Incomplete LES Relaxation
Failure of the sphincter to properly relax during swallowing

Absent or Low-Amplitude Peristalsis
Lack of normal coordinated muscle contractions in the esophageal body

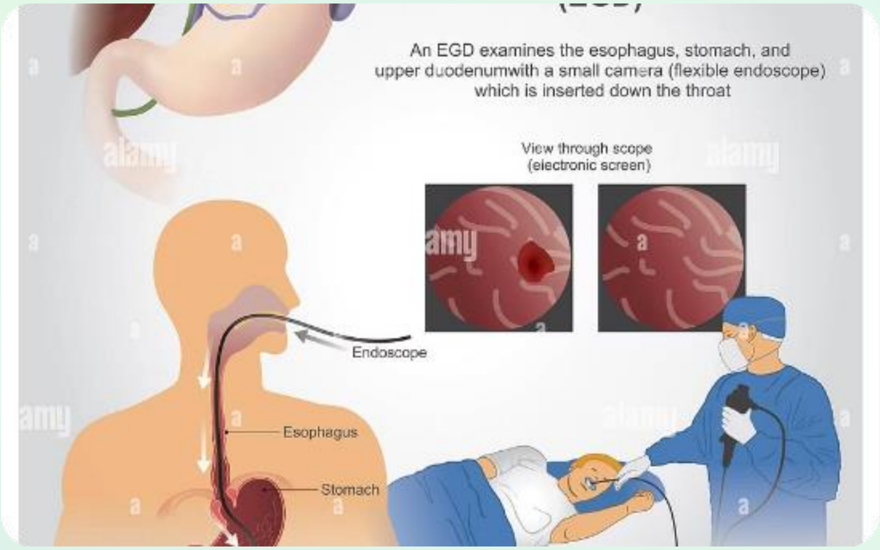
Subtype Classification
Determination of achalasia type based on pressure patterns

High-Resolution Manometry (HRM) is considered the definitive diagnostic test for achalasia. This sophisticated technology provides detailed pressure measurements throughout the esophagus and at the LES, allowing for precise diagnosis and classification according to the Chicago criteria.

Upper Endoscopy in Achalasia Diagnosis

Endoscopic Findings

Endoscopy may reveal a dilated esophagus with retained food or secretions. The gastroesophageal junction often appears tight but can be traversed with gentle pressure. The stomach typically appears normal.

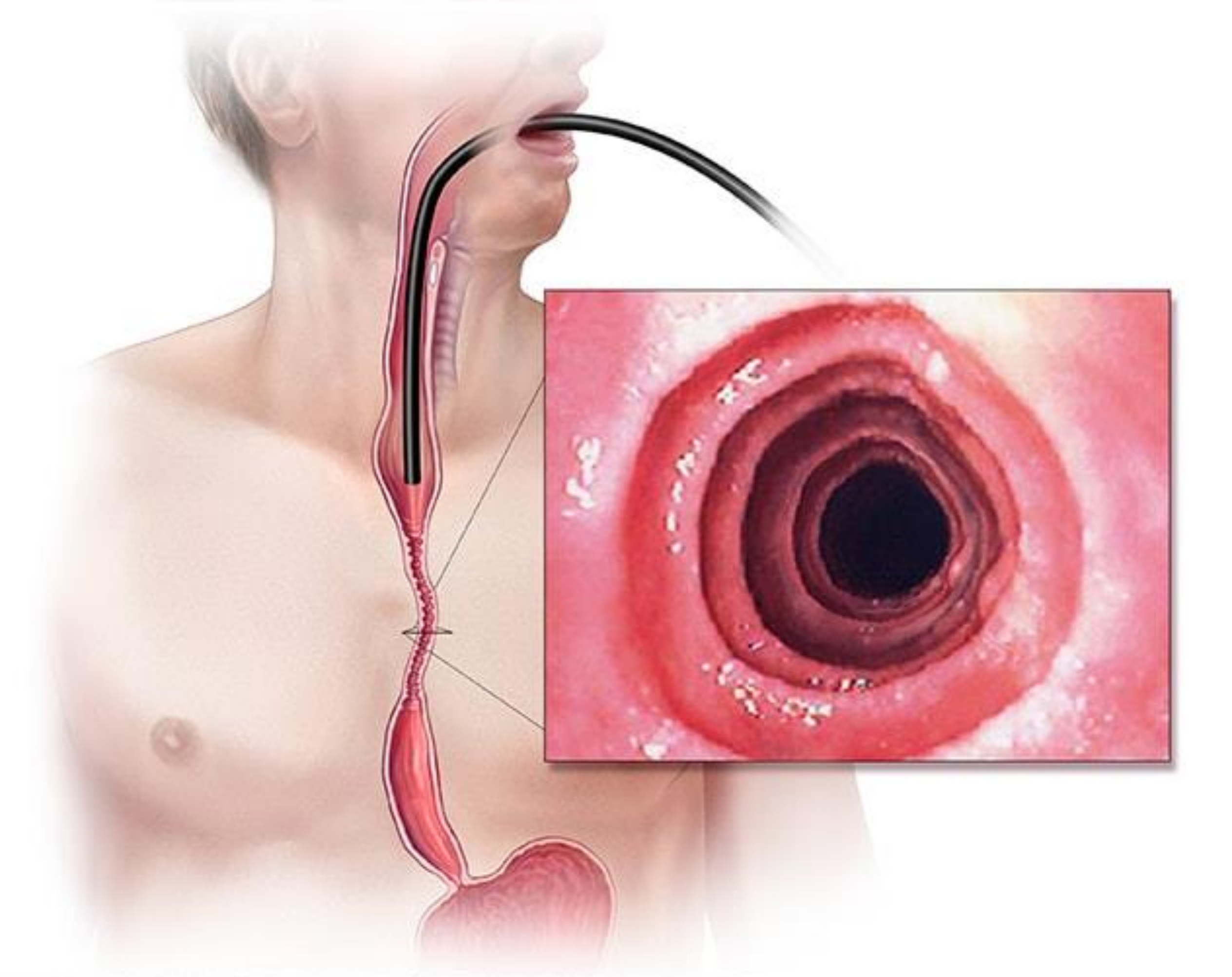


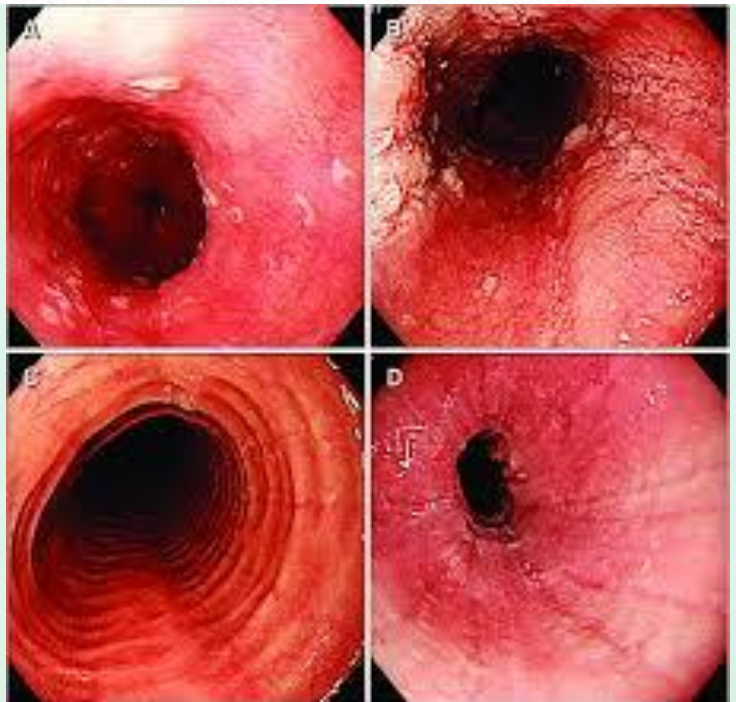
Biopsy Role

While routine in adults to exclude malignancy, esophageal biopsies are less frequently indicated in children with suspected achalasia. When performed, they help rule out other conditions like eosinophilic esophagitis.

Differential Diagnosis

Endoscopy helps exclude other causes of dysphagia such as esophagitis, strictures, or mechanical obstruction. It's essential for ruling out secondary causes of achalasia-like symptoms.





Ruling Out Secondary Causes

Infectious Etiologies

- *Trypanosoma cruzi* infection (Chagas' disease)
- Viral esophagitis
- Fungal infections

Inflammatory Conditions

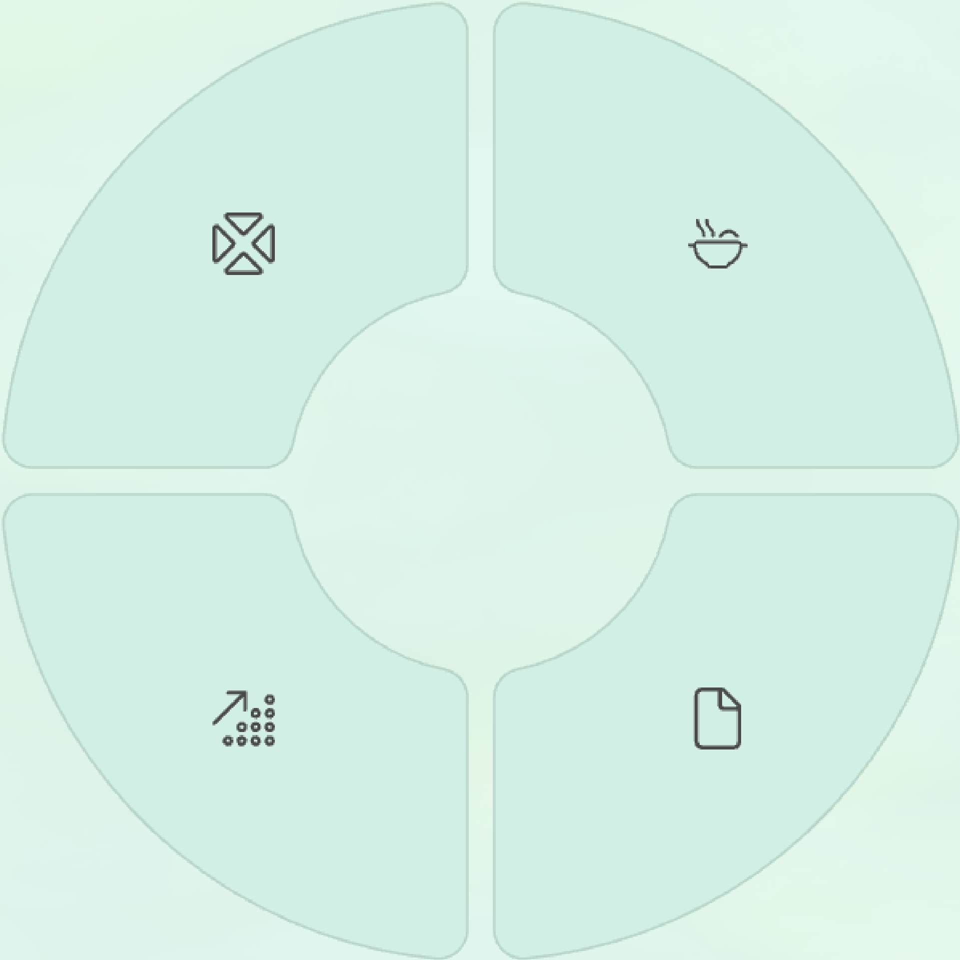
- Eosinophilic esophagitis
- Crohn's disease with esophageal involvement
- Sarcoidosis

Structural Abnormalities

- Esophageal strictures
- Vascular rings
- Malignancy (extremely rare in children)

A comprehensive diagnostic approach is essential to differentiate primary achalasia from conditions that can mimic its presentation. While malignancy is a significant concern in adults with achalasia-like symptoms, it is exceedingly rare in the pediatric population.

Treatment Goals and Approach



Reduce LES Pressure

Lower the resistance at the gastroesophageal junction

Improve Esophageal Emptying

Facilitate passage of food and liquids into the stomach

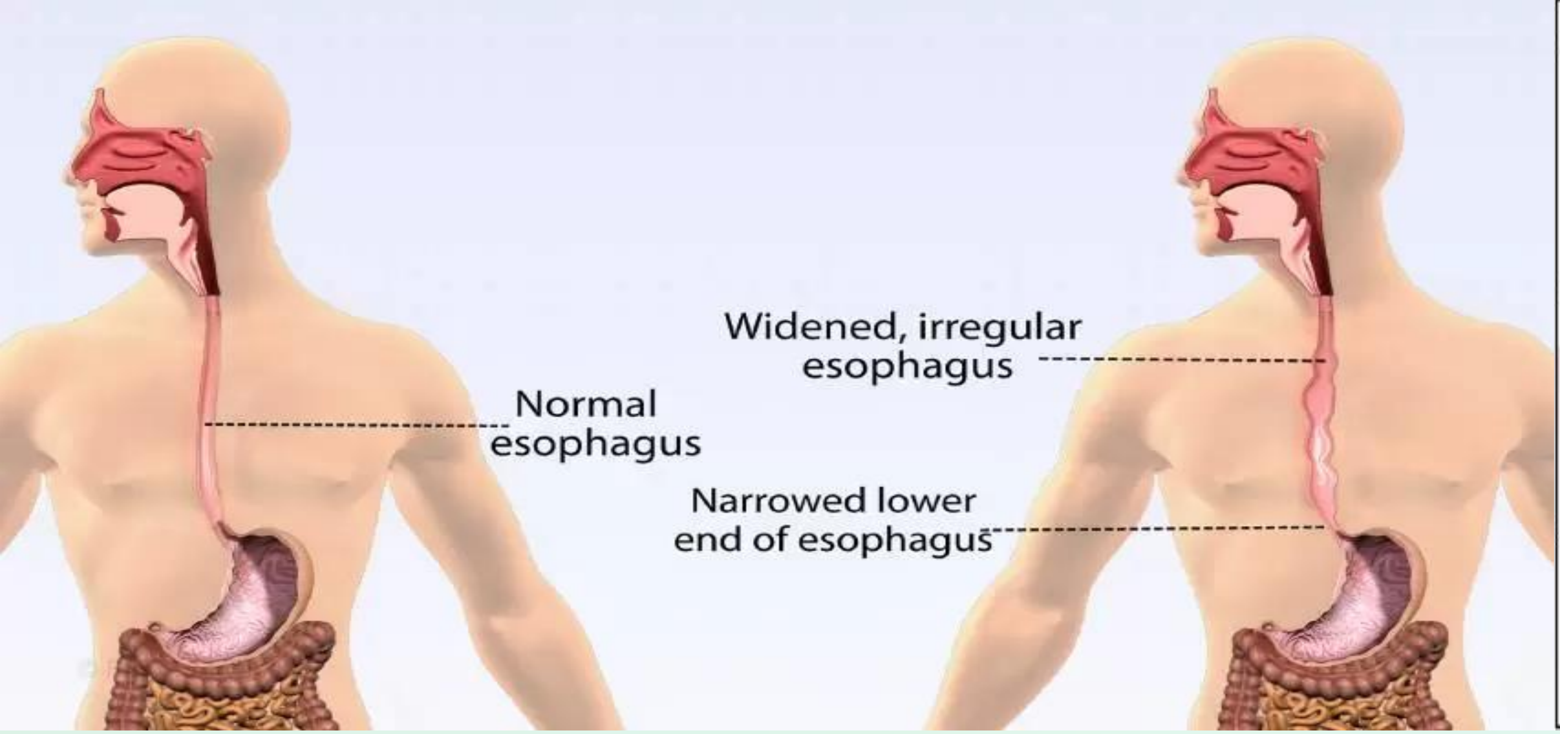
Restore Normal Growth

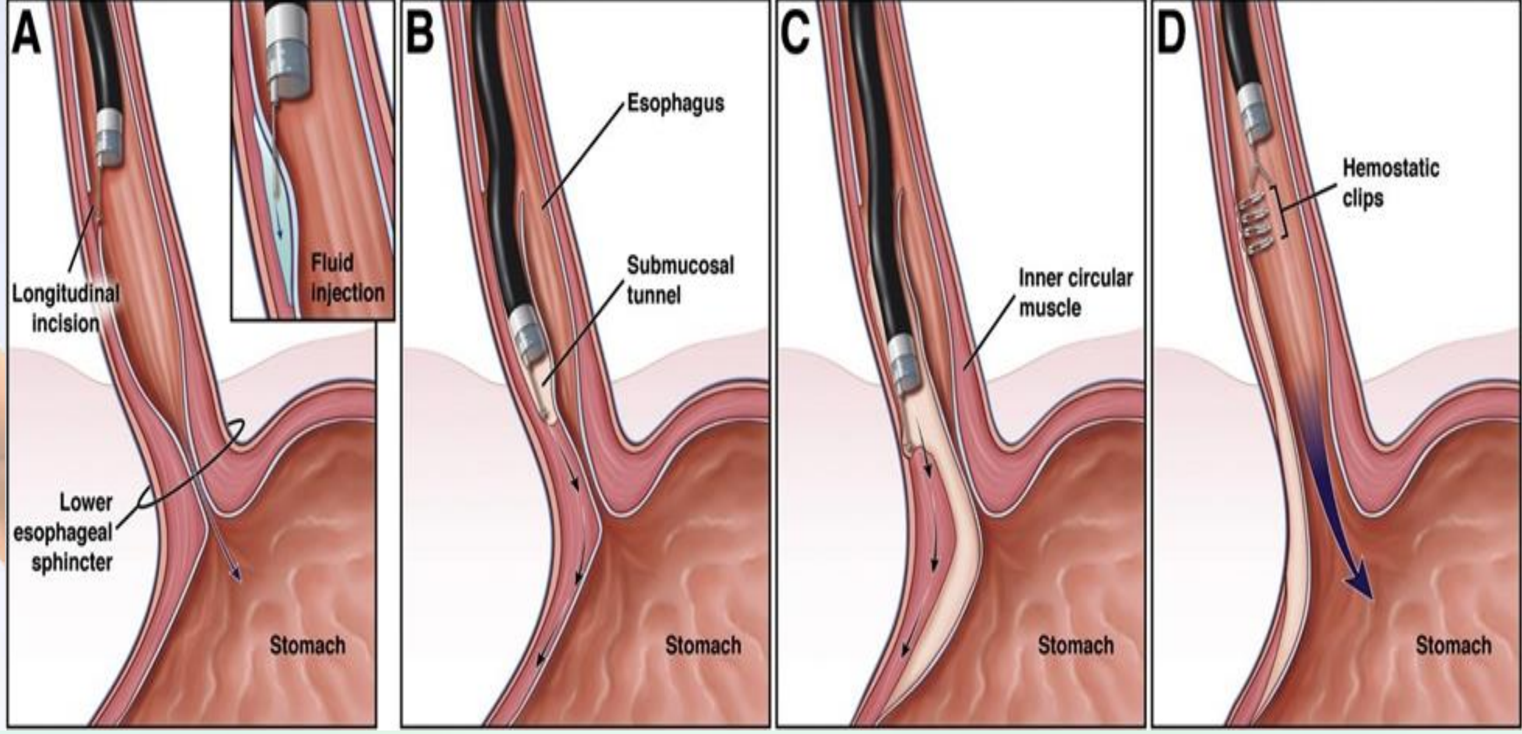
Improve nutritional status and weight gain

Alleviate Symptoms

Reduce dysphagia, regurgitation, and chest pain

Treatment for achalasia focuses on reducing the pressure at the lower esophageal sphincter to facilitate esophageal emptying. While the condition cannot be cured, effective management can significantly improve symptoms and quality of life for affected children.





Medical Management Options

Calcium Channel Blockers

Medications like Nifedipine work by relaxing the LES, but typically provide only temporary relief of symptoms

Nitrates

Can help reduce LES pressure but are limited by side effects and short duration of action

Limitations

Medical therapy generally fails to provid long-term symptom resolution and is considered a temporary measure

While medications may offer some relief, particularly for patients awaiting more definitive treatment, they rarely provide adequate long-term management of achalasia. The transient nature of their effects often leads to recurrent symptoms and progressive disease.

Because nitrates are short-acting, sublingual isosorbide dinitrate (5 mg) (where available) is administered 10 to 15 minutes before meals. Sublingual nitroglycerin 0.4 mg is an alternative if sublingual isosorbide dinitrate is unavailable and nitrate therapy is indicated [55].

Although 5-phosphodiesterase inhibitors (eg, sildenafil), anticholinergics (eg, atropine, dicyclomine, cimetropium bromide), beta adrenergic agonists (eg, terbutaline), and theophylline have been used to treat achalasia, there are very limited data to support their use. Sublingual Nitroglycerin Tablets or Spray:

Dosage:
0.3 to 0.6 mg per dose, administered sublingually (under the tongue)

Can be given 10–15 minutes before meals

Usually used 2–3 times per day depending on meals

While studies have described variable efficacy rates for treatment with calcium channel blockers for achalasia ,we avoid the use of short-acting nifedipine because of the risk of adverse effects including severe hypotension and ischemic complications

(Pediatric Dosage of Nifedipine for Achalasia (Off-label use)
Sublingual or oral dose:
0.25 to 0.5 mg/kg/dose, given 30–45 minutes before meals, typically 2–3 times daily.

Maximum single dose: Often capped at 10–20 mg depending on age and tolerability.)

Botulinum Toxin Injection

Mechanism of Action

Botulinum toxin is endoscopically injected into the LES, where it blocks the release of acetylcholine from nerve endings. This temporarily reduces muscle contraction and lowers sphincter pressure, allowing for improved esophageal emptying.

Limitations in Pediatrics

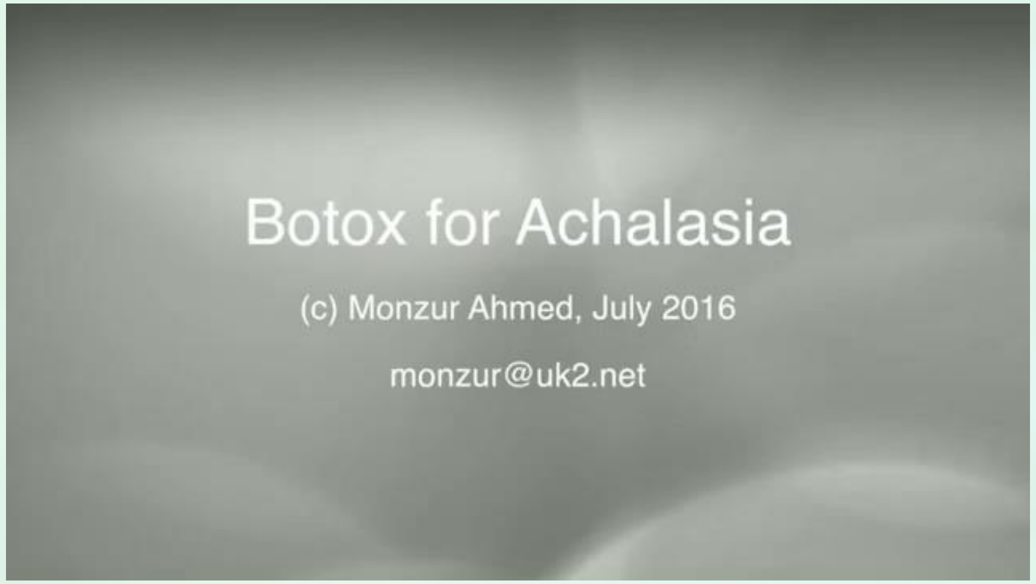
While this approach can provide symptom relief, the effects are typically short-lived, lasting only 3-6 months. Like medical therapy, botulinum toxin injection is generally considered a temporary measure rather than a definitive treatment for children with achalasia.

It may be most appropriate for patients who are poor candidates for more invasive procedures or as a bridge to definitive therapy.

Botulinum Toxin Injection: Technique

BOTOX.mp4

- Endoscopic injection of 100 units total, divided into 4 quadrants near LES
- Injection depth: submucosal layer
- Repeat injections are common



Botox for Achalasia

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Botulinum Toxin: Short-term Efficacy

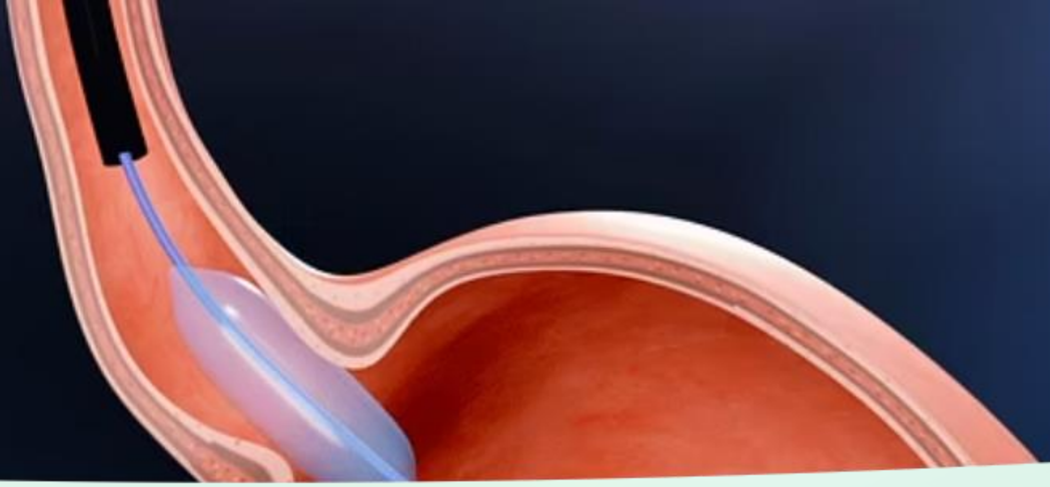
- Significant symptom relief in 70-90%
- Usually lasting for several months
- Less invasive, suitable for high-risk or pediatric patients

Botulinum Toxin: Long-term Outcomes

- Response diminishes over time
- Many patients require repeated injections
- Less durable compared to PD or surgery

Side Effects of Botulinum Toxin

- Mild chest discomfort
- Reflux symptoms
- Rare risk of mucosal ulceration or perforation
- No long-term adverse effects reported



Pneumatic Dilatation Procedure

Balloon Placement

An endoscope guides a deflated balloon to the LES

Controlled Inflation

The balloon is inflated to stretch and disrupt LES muscle fibers

Pressure Reduction

Disruption of the sphincter reduces resistance to esophageal emptying

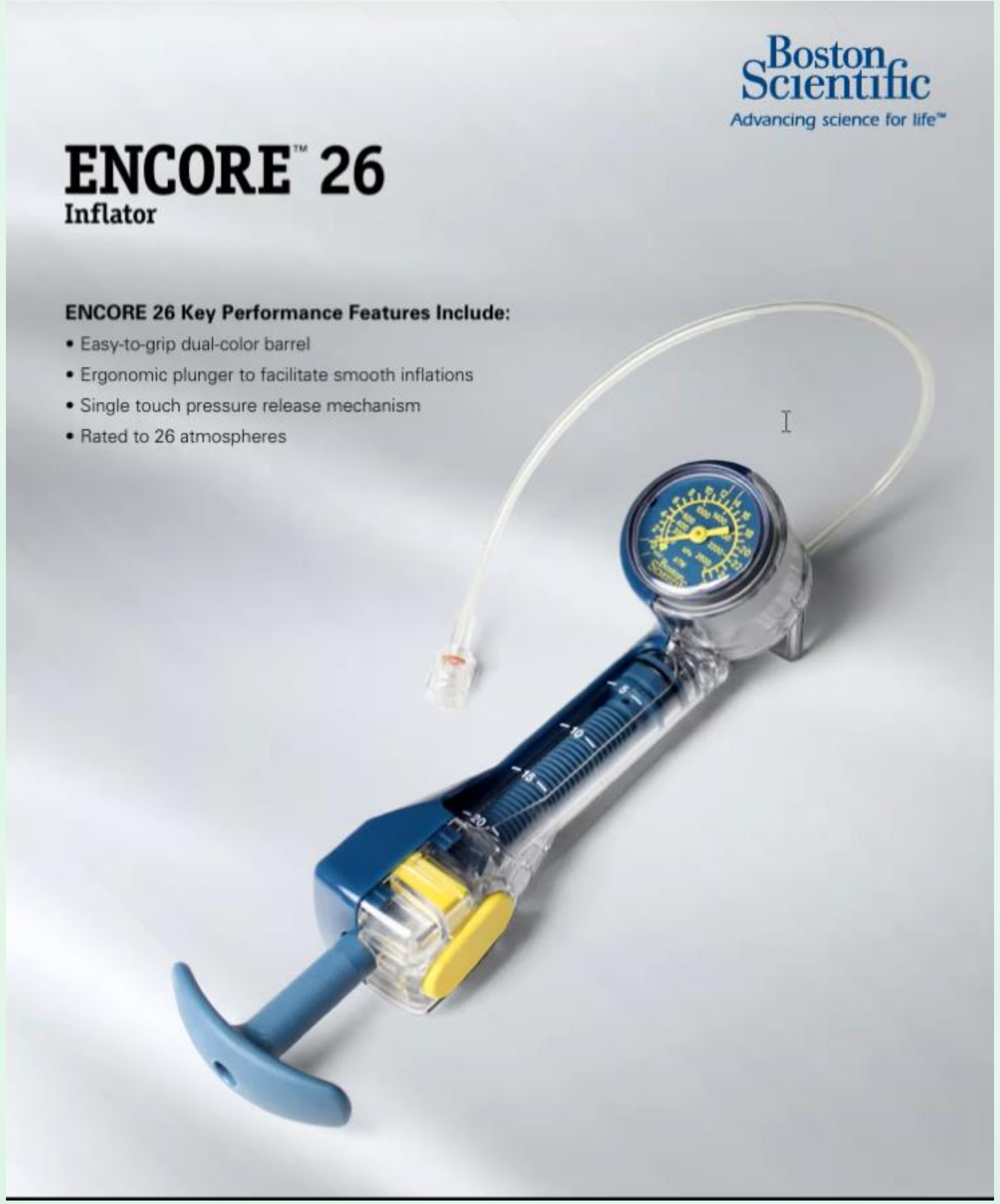
Repeat as Needed

Multiple sessions may be required for optimal results

Pneumatic dilatation is an endoscopic procedure that can be effective for some patients, but it carries a risk of esophageal perforation (approximately 2-5%). Success rates vary, and the procedure may need to be repeated as symptoms recur over time.

Pneumatic Dilation (PD): Technique

- Performed under fluoroscopy or endoscopy
- A deflated balloon (30-40 mm) is positioned across LES
- Balloon inflated to 7-15 psi for 60 seconds(20 psi)
- Usually repeated if necessary
- Goal: Tearing the muscular fibers to reduce LES pressure



- ***There is no consensus on the optimal method for performing PD.***

- The types of dilators used,
- The maximum diameter of the balloon (2.4 to 5 cm),
- The pressure to which the balloon is inflated (100 to >1000 mmHg),
- The rate of balloon inflation (rapid versus gradual),
- The duration of balloon inflation (several seconds to five minutes),
- The number of balloon inflations per dilating session (one to five)

The Rigiflex balloon,(USA)

Guidewire and via fluoroscopy

Three diameters (30, 35, and 40 mm).

The smallest balloon is typically used for the first dilation (30 mm).

If symptoms persist, larger balloons, "graded approach."

Uptodate:

We typically perform one balloon dilation per endoscopic session, with additional dilations being performed if symptoms persist or recur.

Fast for at least 12 hours

A liquid-only diet should be consumed for one or two days

Prior to the procedure, check for leaks or deformities.

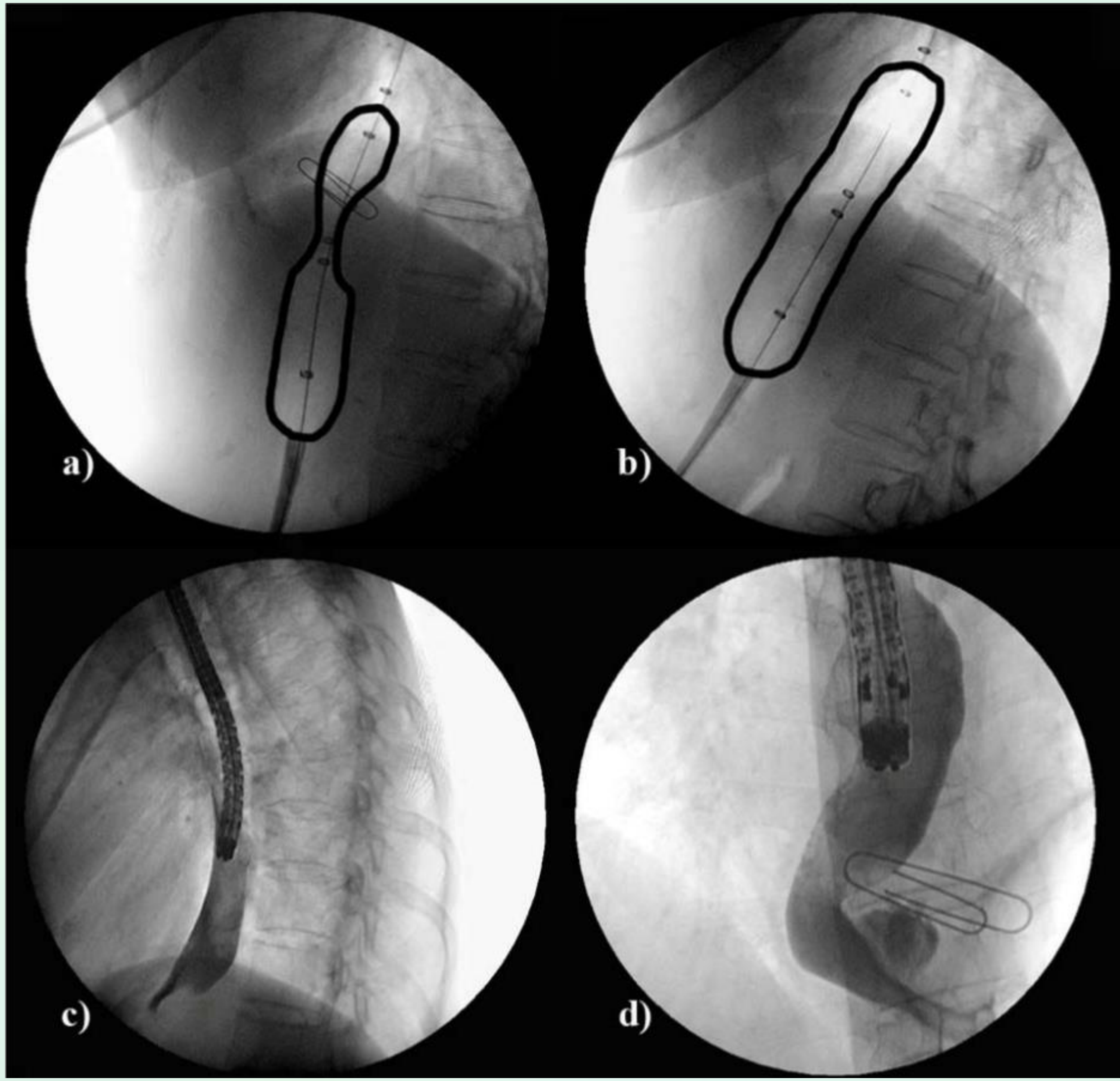
Prior to dilation: diagnostic endoscopic examination (cardia,Pseudoachalasia).

A guidewire is then passed through the biopsy channel of the endoscope into the stomach and the scope is withdrawn to the gastroesophageal junction. The distance between the incisors and the gastroesophageal junction should be noted using the markings along the length of the scope.

The endoscope is then removed, taking care to maintain the position of the guidewire in the stomach. To aid with initial balloon placement, a marker (such as paper tape) can be placed on the shaft of the dilating catheter corresponding to the previously noted distance between the incisors and the gastroesophageal junction. This distance should be measured from the middle of the balloon on the dilating catheter so that, when inserted, the middle of the balloon will be positioned across the LES.

Using fluoroscopy, the balloon is then gradually inflated with air, noting the position of the developing "waist" in the balloon

We often inject a small volume of dilute contrast into the balloon to assist in radiographic visualization.



After a satisfactory position is obtained, the balloon is fully inflated so that the waist is obliterated,
In our experience, approximately 7 to 15 pounds per square inch (psi) of pressure is required for waist obliteration, which usually requires approximately 120 mL of air.
To convert pounds per square inch (psi) to atmospheres (atm), multiply the psi value by 0.068046. For example, 15 psi is equivalent to approximately 1.02069 atm. The formula is atm = psi * 0.068046. This conversion is useful in various applications, including engineering and scientific fields, where pressure measurements need to be consistent across different units.

Inflation is maintained for 60 seconds, after which the balloon is rapidly deflated. We then perform a second full inflation for 60 seconds and again note the pressure required to obliterate the waist. This is usually at least 3 psi less than the initial pressure, thus indicating immediate improvement.

After the second inflation, the deflated balloon and guidewire are removed and the patient is transferred to the recovery area. The patient is observed for five to six hours

Perforation: (eg, tachycardia, persistent chest pain, subcutaneous emphysema)

ELSEVIER

Pneumatic Balloon Dilatation in Achalasia Cardia

video Journal & Encyclopedia GI Endoscopy

V Jayasekeran, B Holt, and MJ Bourke
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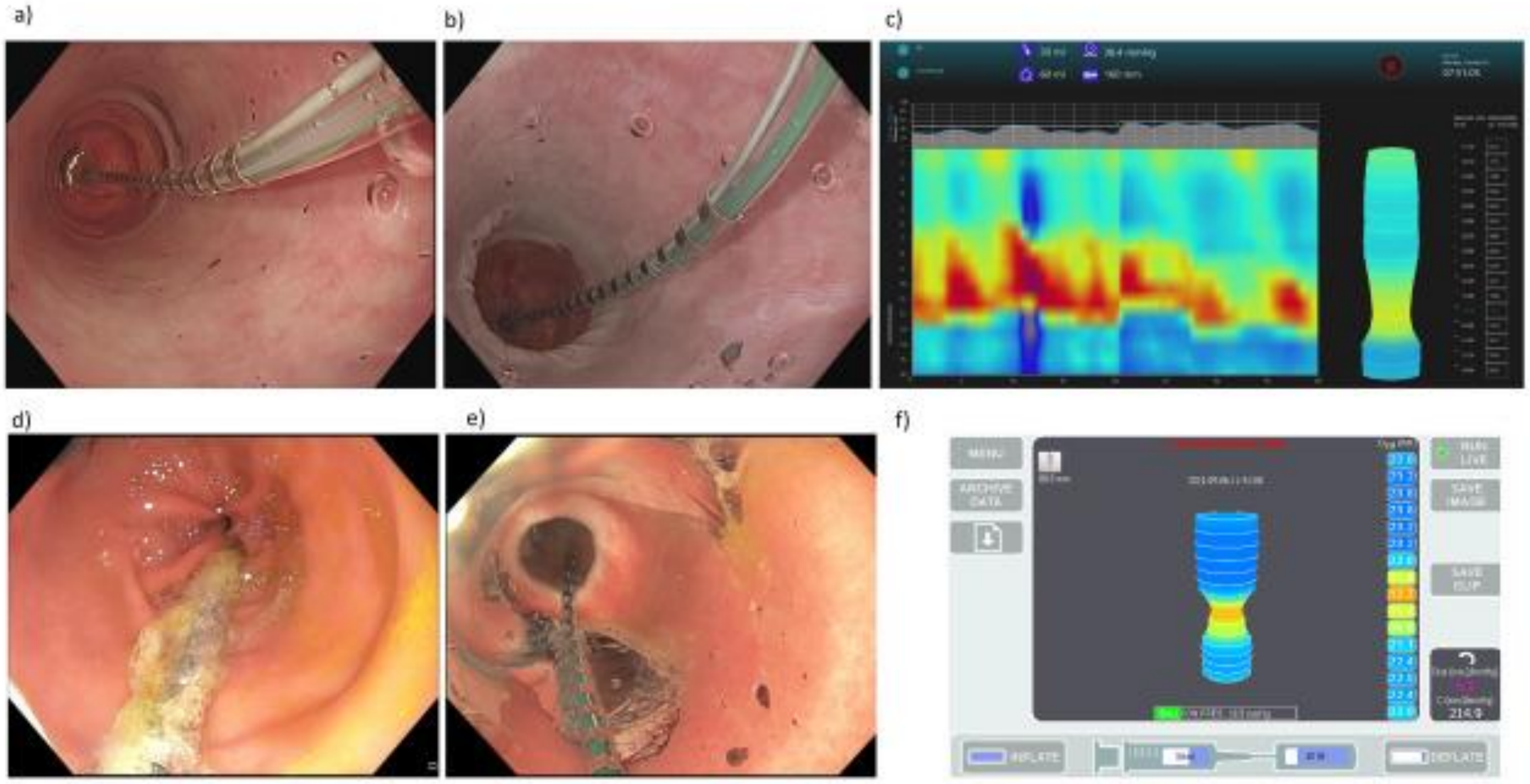
Sydney, NSW, Australia

Another tool for esophageal dilation has become available and consists of a hydraulically distended rigid balloon (EsoFLIP) built around a catheter that performs high-resolution impedance planimetry, similar to the one used for diagnostic procedures (EndoFLIP)

For achalasia dilations, the EsoFLIP ES-330 catheter can achieve dilations up to a maximum diameter of 30 mm [14]. After assembly, the EsoFLIP catheter is typically advanced alongside the endoscope over a guidewire for direct visualization of its position traversing the LES; thereafter the general principles of dilation are similar to PD. This technique can be used without fluoroscopy, relying instead on the use of modeling to convert the impedance-derived diameters into a visual display to identify the “waist” (high-pressure zone) and its subsequent obliteration with dilation. In addition,
impedance catheter can n
utility of these technical fe



Current Opinion in Pharmacology

Pneumatic Dilation: Efficacy

- Initial success rate in children: 70-90%
- Long-term success in 70% of cases
- Often considered first-line minimally invasive therapy
- Repeat dilations may be needed

Pneumatic Dilation: Complications

- Perforation rate: 2-5%
- Post-procedure reflux (less than 2%)
- Chest pain and discomfort
- Rarely, strictures or other complications

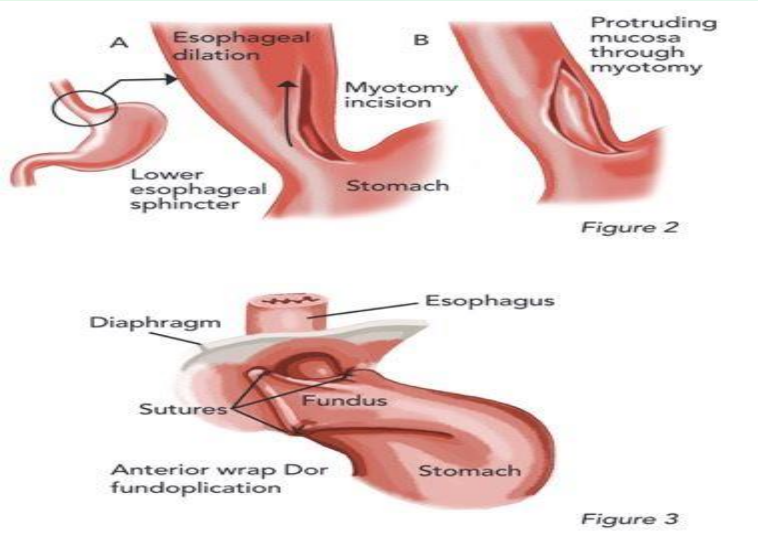
Factors Influencing PD Success

- Age: Younger patients may respond better
- Achalasia subtype: Type II responds best
- Initial LES pressure
- Degree of esophageal dilation
- Expertise of the operator

Surgical Myotomy: Types

- Classic Heller's myotomy
- Laparoscopic Heller Myotomy
- Per-Oral Endoscopic Myotomy (POEM)
- Choice depends on patient factors and operator expertise

Laparoscopic Heller Myotomy: The Gold Standard



Surgical Approach

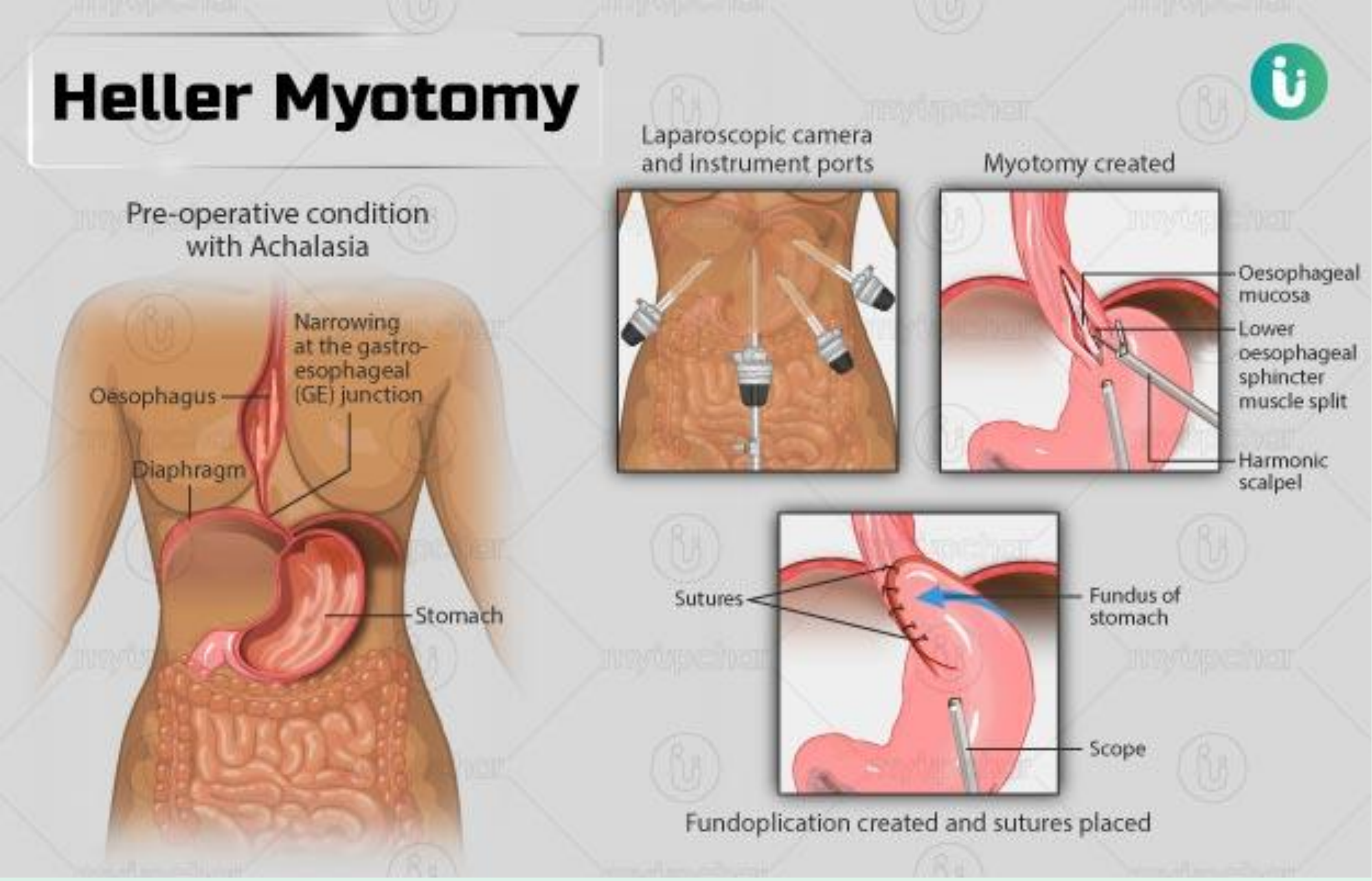
Laparoscopic Heller myotomy (LHM) involves making small incisions in the abdomen to access and cut the muscle fibers of the LES, permanently reducing its pressure. This minimally invasive approach offers excellent visualization and precision.

Anti-Reflux Component

LHM is typically combined with a partial fundoplication (wrapping the stomach around the lower esophagus) to prevent post-operative gastroesophageal reflux, which can occur when the LES is surgically weakened.

Pediatric Considerations

LHM is considered the treatment of choice and standard of care for children with achalasia. The procedure has demonstrated excellent long-term outcomes in the pediatric population with high success rates.



Laparoscopic Heller Myotomy (LHM)

- Minimally invasive surgical procedure
- Longitudinal incisions through the outer muscular layers of the distal esophagus and LES
- Often combined with fundoplication to reduce reflux

Peroral Endoscopic Myotomy (POEM): Emerging Option

Mucosal Incision

Creating an opening in the esophageal lining

Submucosal Tunnel

Forming a pathway to access the muscle layer

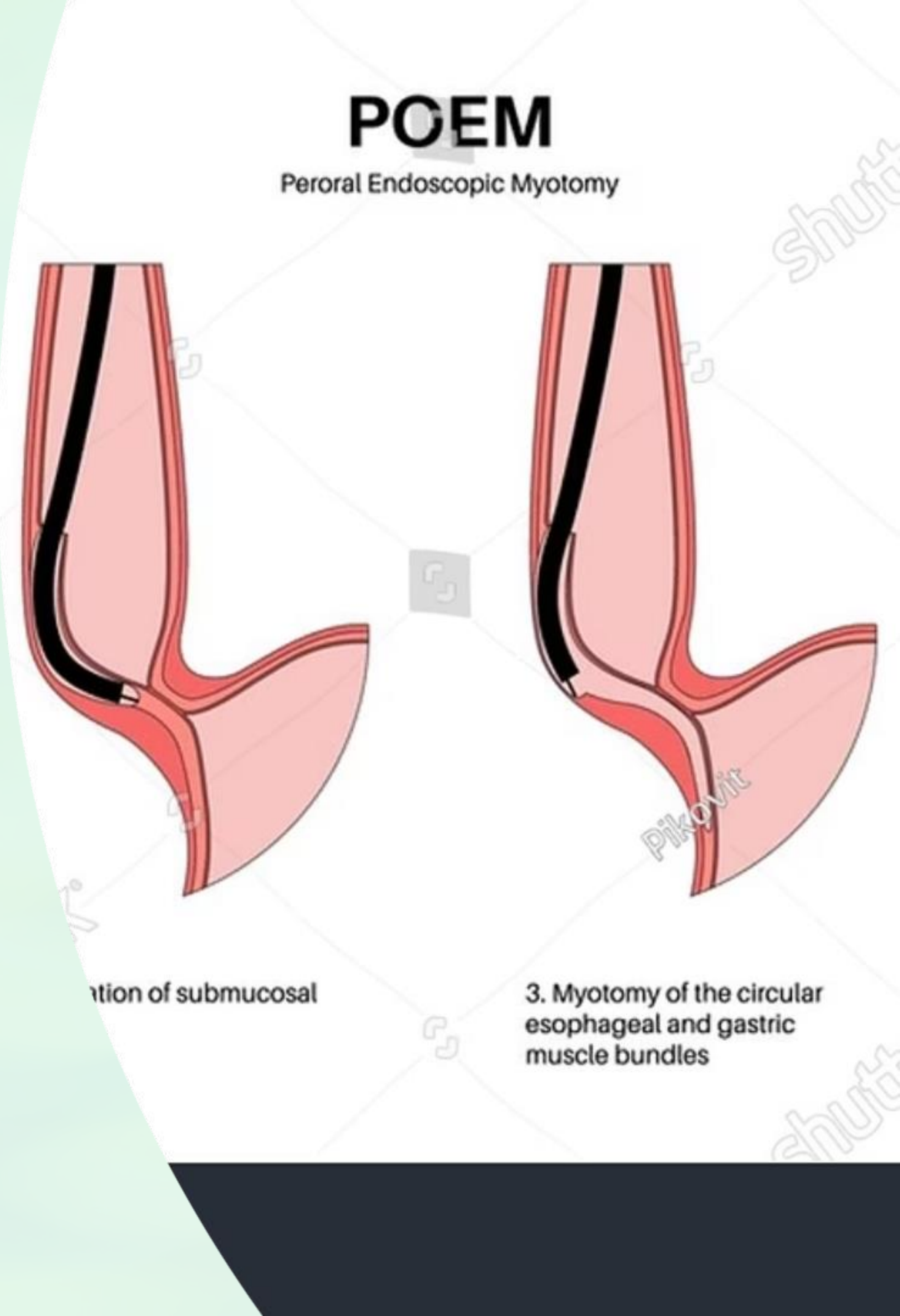
Myotomy

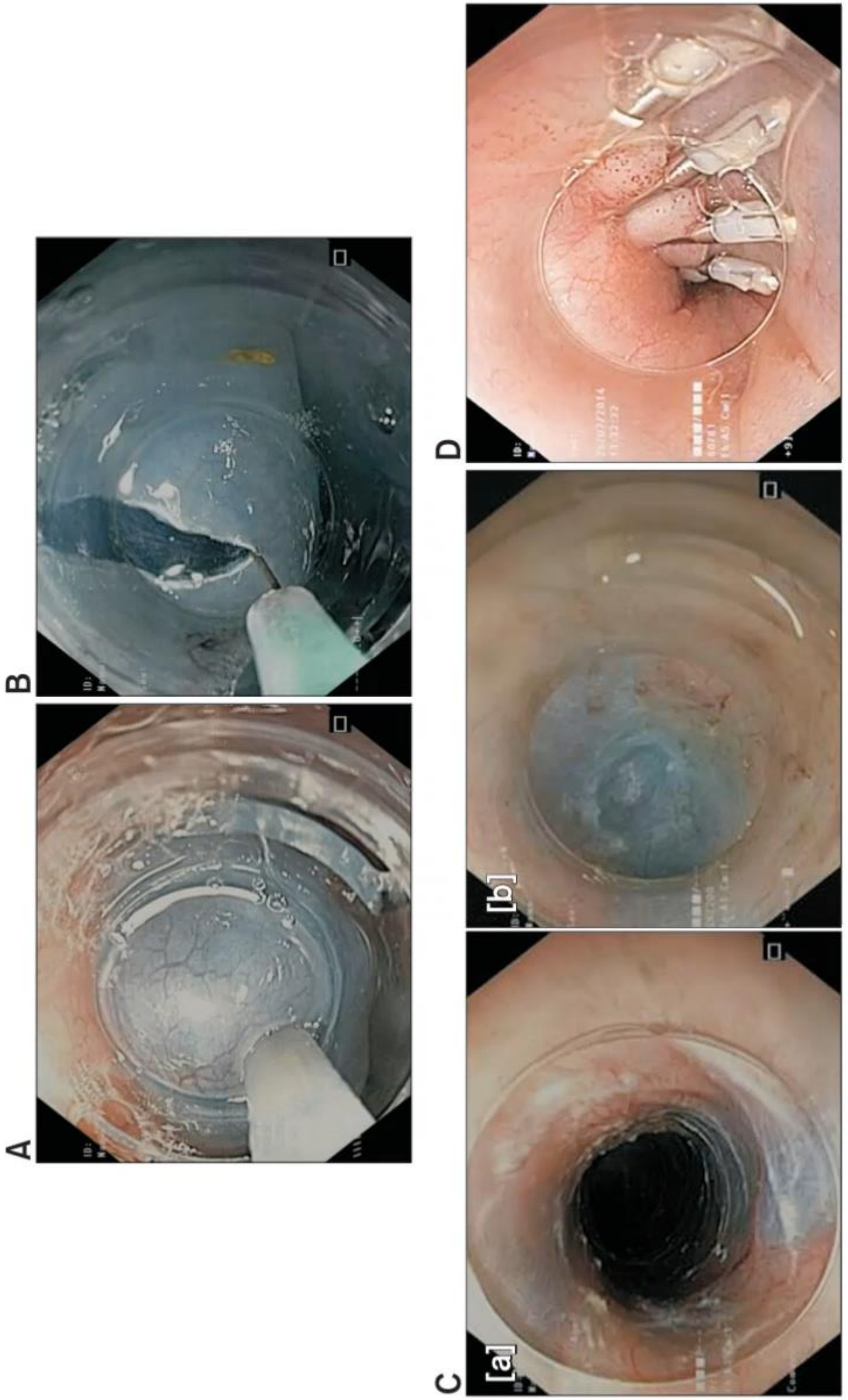
Cutting the circular muscle fibers of the LES

Closure

Sealing the mucosal entry point

POEM is a novel endoscopic technique that provides an incisionless approach to myotomy. While increasingly used in adults with promising results, more experience is needed to determine its safety, efficacy, and feasibility in the pediatric population. Early studies suggest it may be a viable option for children.





Comparing Treatment Options

Treatment	Efficacy	Durability	Invasiveness	Complications
Medical Therapy	Low	Temporary	Non-invasive	Minimal
Botulinum Toxin	Moderate	3-6 months	Minimally invasive	Low
Pneumatic Dilation	Moderate-High	Variable	Minimally invasive	Perforation risk
Heller Myotomy	High	Long-term	Surgical	Reflux, surgical risks
POEM	High	Under study	Endoscopic	Reflux, limited pediatric data

The selection of treatment approach depends on multiple factors including the child's age, achalasia subtype, severity of symptoms, local expertise, and family preferences. A multidisciplinary approach involving pediatric gastroenterologists and surgeons is essential for optimal management.

Choosing the Right Treatment

Factors to consider:

- Patient age and comorbidities
- Achalasia subtype
- Previous treatments
- Patient preference
- Center expertise

Post-Treatment Considerations

Gastroesophageal Reflux

Common after myotomy procedures, may require acid suppression therapy

Nutritional Rehabilitation

Addressing weight loss and optimizing growth parameters

Symptom Monitoring

Regular assessment for recurrence or persistence of symptoms

Long-term Follow-up

Ongoing care to ensure sustained treatment success

After treatment, children require careful monitoring for both immediate complications and long-term outcomes. While most experience significant symptom improvement, some may develop new issues like reflux or require additional interventions for persistent or recurrent symptoms.

Post-Treatment Follow-up

- Regular symptom assessment
- Manometry and barium swallow as needed
- PPI therapy for reflux
- Monitor for recurrence or complications

Pediatric vs. Adult Management

- Children often respond well to PD initially
- Surgery and POEM are options when less invasive methods fail
- Long-term follow-up essential in pediatric cases

Managing Recurrent or Persistent Symptoms

- Repeated dilation or injections
- Consider surgical intervention if refractory
- Evaluate for other causes of dysphagia

Reinforcement via Re-dilation or Repeat Injections

- Repeated PD and botulinum toxin injections are common
- Persisting symptoms warrant surgical referral

Risks and Complications

- Perforation, bleeding, infection
- Reflux esophagitis
- Esophageal rupture (rare)
- Need for appropriate management strategies

Nutritional Management

Pre-Treatment Challenges

- Difficulty consuming adequate calories
- Avoidance of certain food textures
- Prolonged meal times
- Weight loss or growth faltering

Before effective treatment, children with achalasia often develop adaptive eating behaviors and may restrict their diet to foods they can more easily swallow, potentially leading to nutritional deficiencies.

Post-Treatment Approach

- Gradual reintroduction of varied textures
- Caloric optimization for catch-up growth
- Monitoring of weight gain trajectory
- Addressing eating behaviors and anxiety

After treatment, a structured nutritional rehabilitation program helps restore normal growth patterns and healthy eating habits. Some children may benefit from consultation with a pediatric dietitian.

Long-Term Outcomes and Prognosis

85-95%

Symptom Improvement

Percentage of children experiencing significant symptom relief after Heller myotomy

10-15%

Symptom Recurrence

Proportion of patients who may develop recurrent symptoms over time

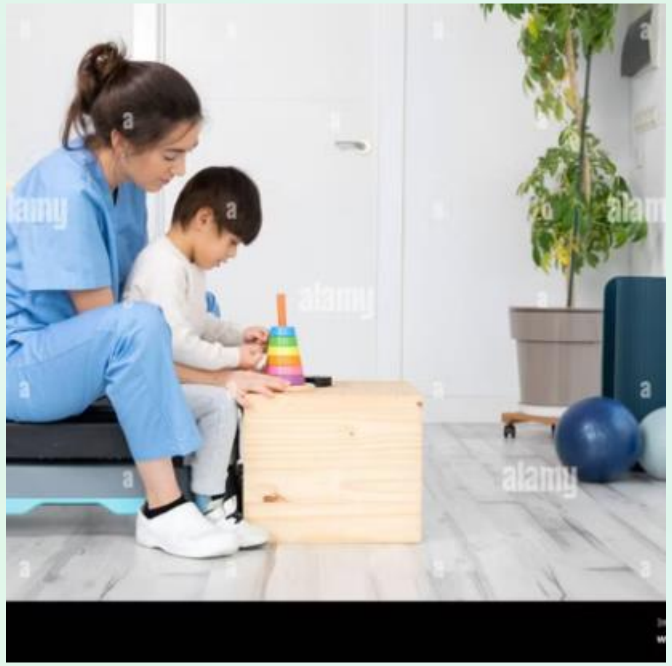
20-30%

Post-Procedure Reflux

Approximate rate of reflux symptoms following myotomy procedures

The long-term prognosis for children with achalasia is generally favorable with appropriate treatment. Most achieve substantial improvement in swallowing function and quality of life. Regular follow-up is important to monitor for late complications or symptom recurrence, which may necessitate additional interventions.

Psychosocial Impact and Support

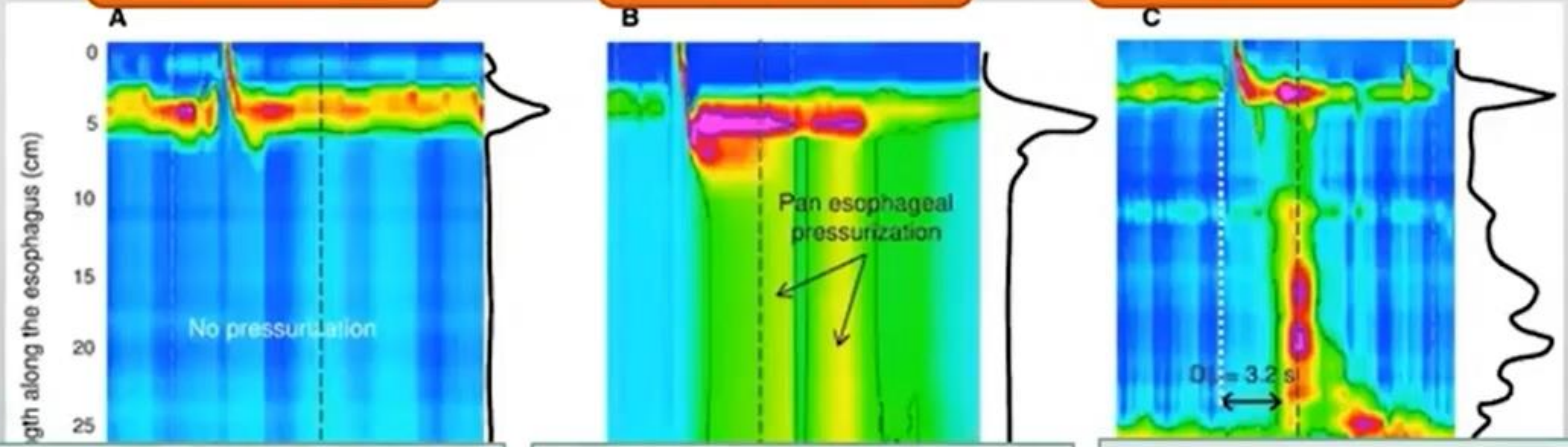


The psychosocial impact of achalasia on children and adolescents can be significant. Difficulties with eating may lead to embarrassment in social situations, anxiety around mealtimes, and feelings of being different from peers. School attendance and participation in normal activities may be affected by symptoms and medical appointments.

Comprehensive care should address these psychosocial aspects through appropriate support services, including psychological counseling when needed. Parent and family support is crucial, as is education of school personnel about the condition.

Chicago Classification Subtypes of Achalasia

Type I Achalasia	Type II Achalasia	Type III Achalasia



Type I Achalasia	Type II Achalasia	Type III Achalasia
Dilated esophagus associated with poor treatment response	Best response to treatment Any treatment	Worst prognosis POEM approach allows for a long myotomy

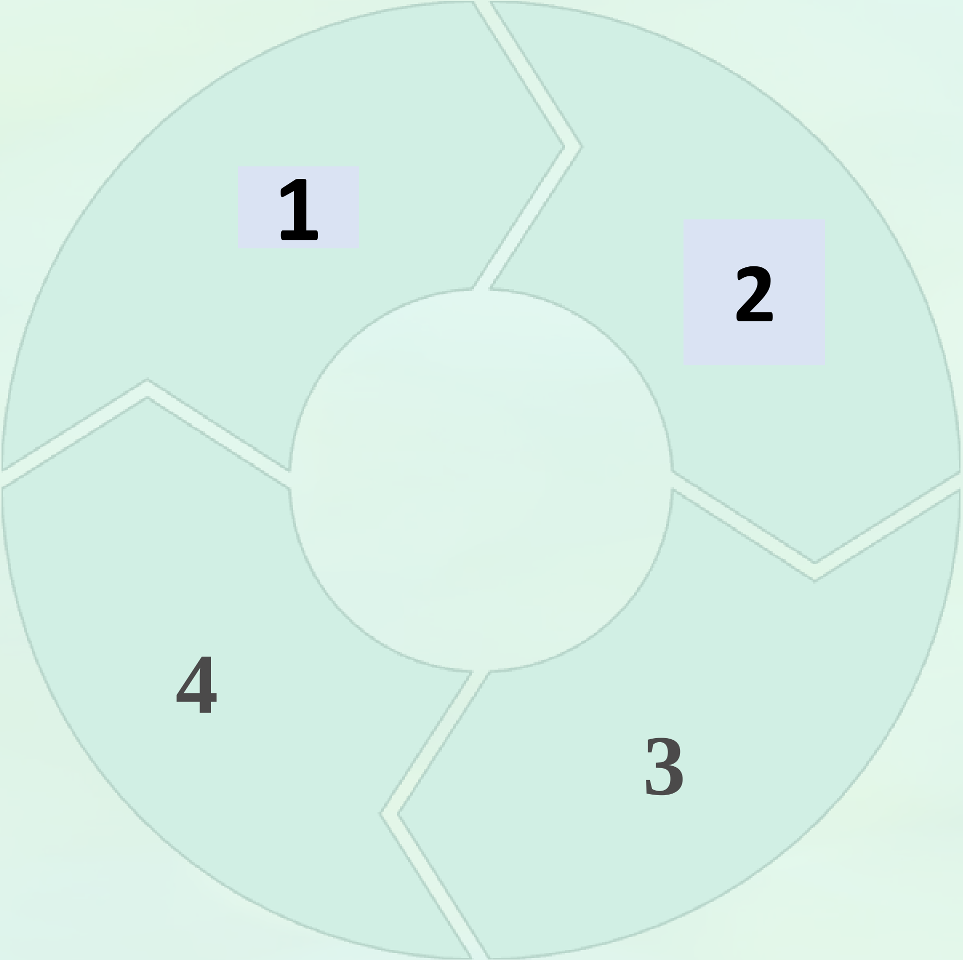
Future Directions in Pediatric Achalasia

Pathophysiology Research

Deeper understanding of underlying mechanisms

Genetic Studies

Identifying potential genetic markers and risk factors



Pediatric-Specific Guidelines

Development of standardized management protocols for children

Novel Techniques

Refinement of POEM and other minimally invasive approaches

Ongoing research aims to improve our understanding of achalasia's causes and to develop more effective, less invasive treatments. Pediatric-specific studies are particularly important, as findings from adult populations may not always apply directly to children. Collaborative registries and multicenter studies will help advance knowledge of this rare condition.


Thanks
Write
what should NOT be
forgotten.