“I sit to sleep”

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Pediatric Pulmonology Conference

8.7.15

Ruth

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Children’s Hospital

Case Presentation

- A.I. , 15-y-old boy, generally healthy.
- Presented to the pulmonary clinic with a 4-m history of recurrent productive cough only when lying supine.
- He sleeps in a sitting position, with an open mouth and snores without apneas.
- S/P adenoidectomy at the age of 4-y.
- No fever, fatigue, loss of weight or changes in the bowel movements.
- He is the 7^{th} child out of 8 children in his family.
- No history of asthma / allergy / smoking in his family.

Further work up

- ENT : Sinusitis →Augmentin; Allergy → Allergix.
- Physical Examination:

  Weight: 65 Kg, Height: 160cm, BMI: 25.4.

  BP: 120/60, P: 75, Saturation: 98% in room air.

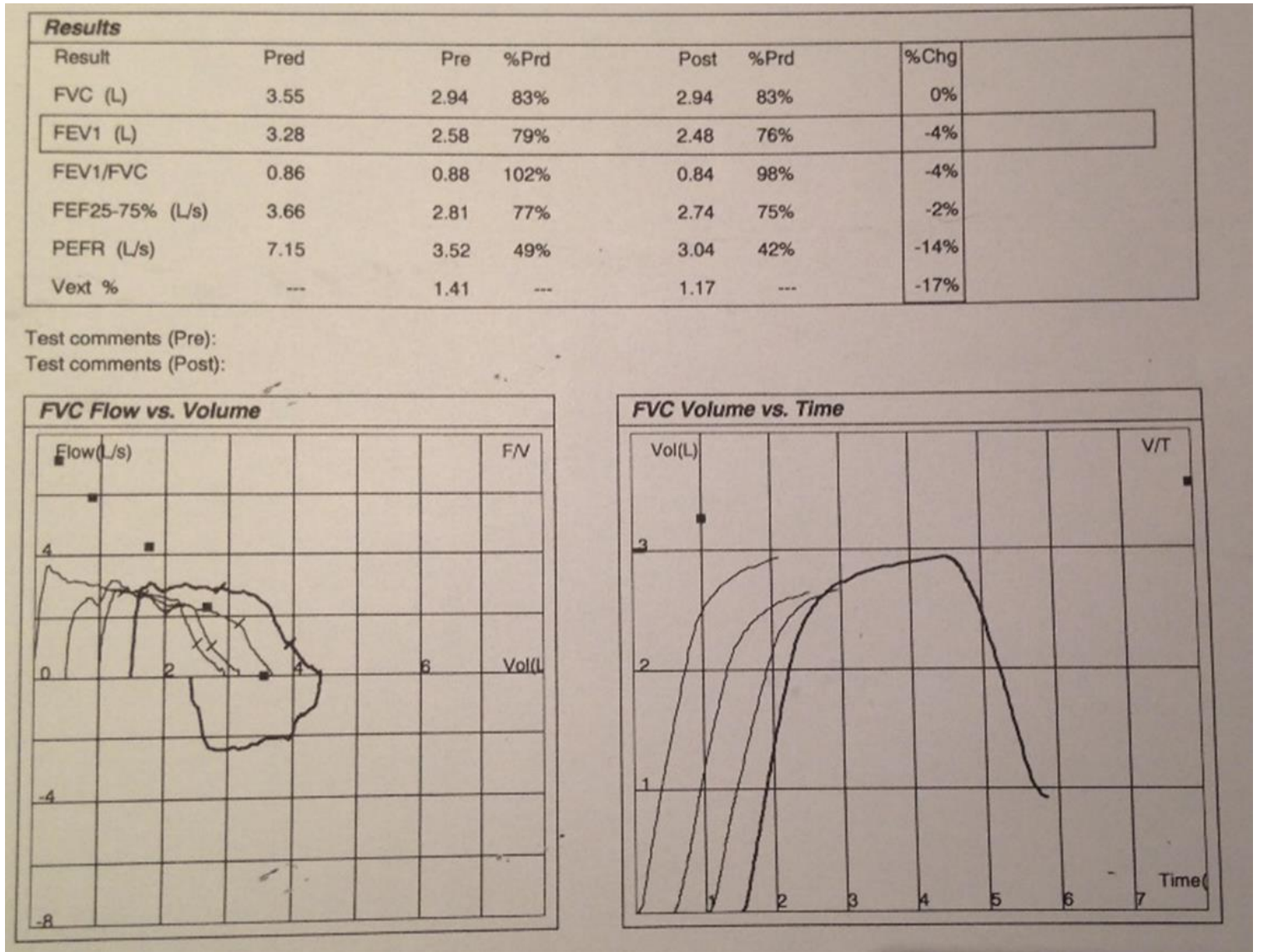
  Normal Breathing pattern without noise.

  Auscultation to the lungs and heart: normal.

  No Clubbing.
- Lab: WBC, Short chemistry- normal.
- CXR: normal.

Pulmonary Function Test

Fixed upper airway obstruction →



Results

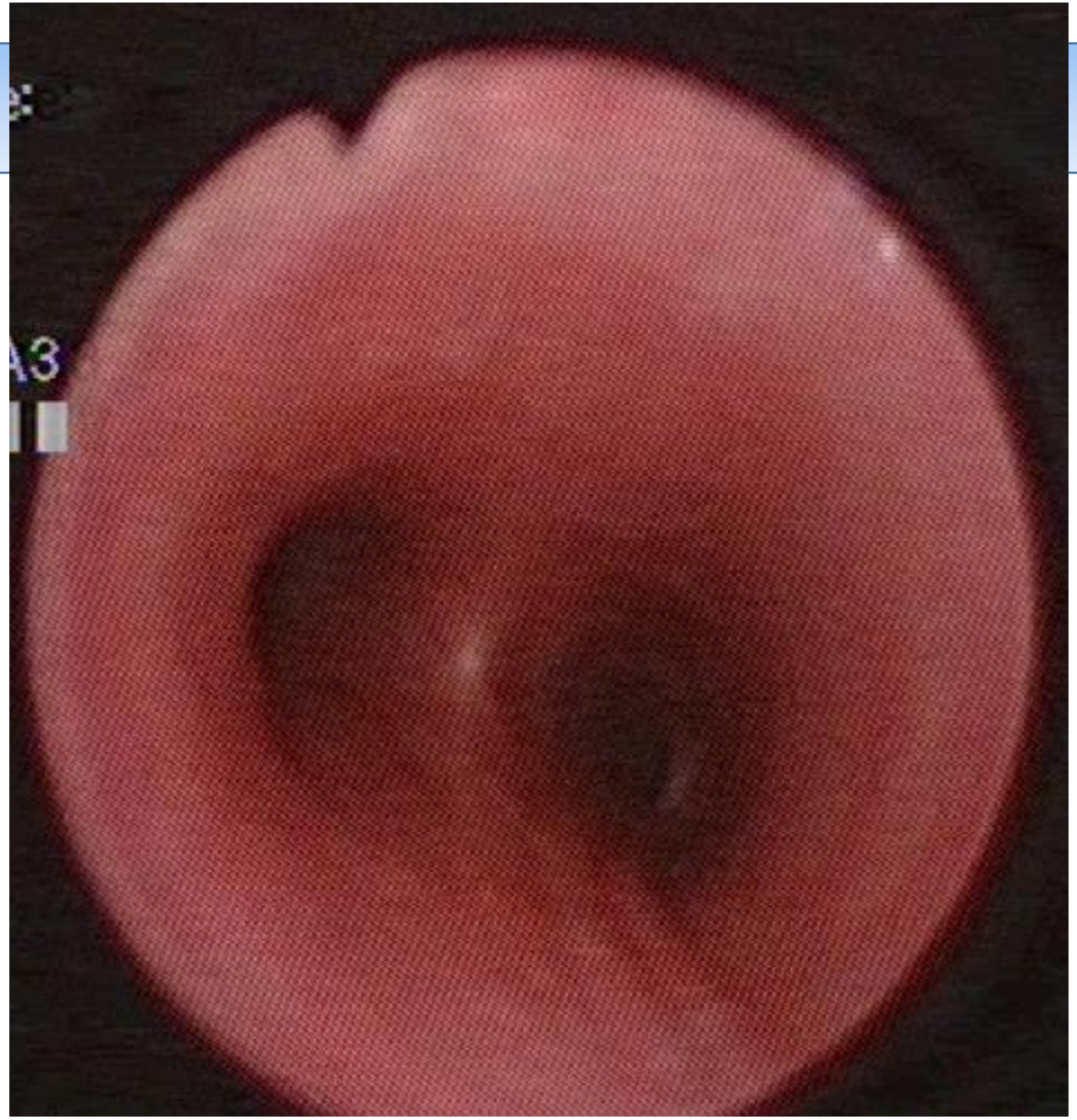
Result	Pred	Pre	%Prd	Post	%Prd	%Chg
FVC (L)	3.55	2.94	83%	2.94	83%	0%
FEV1 (L)	3.28	2.58	79%	2.48	76%	-4%
FEV1/FVC	0.86	0.88	102%	0.84	98%	-4%
FEF25-75% (L/s)	3.66	2.81	77%	2.74	75%	-2%
PEFR (L/s)	7.15	3.52	49%	3.04	42%	-14%
Vext %	---	1.41	---	1.17	---	-17%

Test comments (Pre):
Test comments (Post):

Fixed Upper Airway Obstruction-DD

- Congenital /acquired malformations
  - Vocal cord paralysis
  - Tracheomalacia /stenosis
  - Compression (vascular compression, dilated cardiac chamber, goiter)
- Infection- intrinsic /extrinsic airway narrowing
- Foreign body in airway or esophagus
- Trauma (post intubation/tracheostomy)
- Tumor (intrinsic/extrinsic)

Modified from Taussig 2nd edition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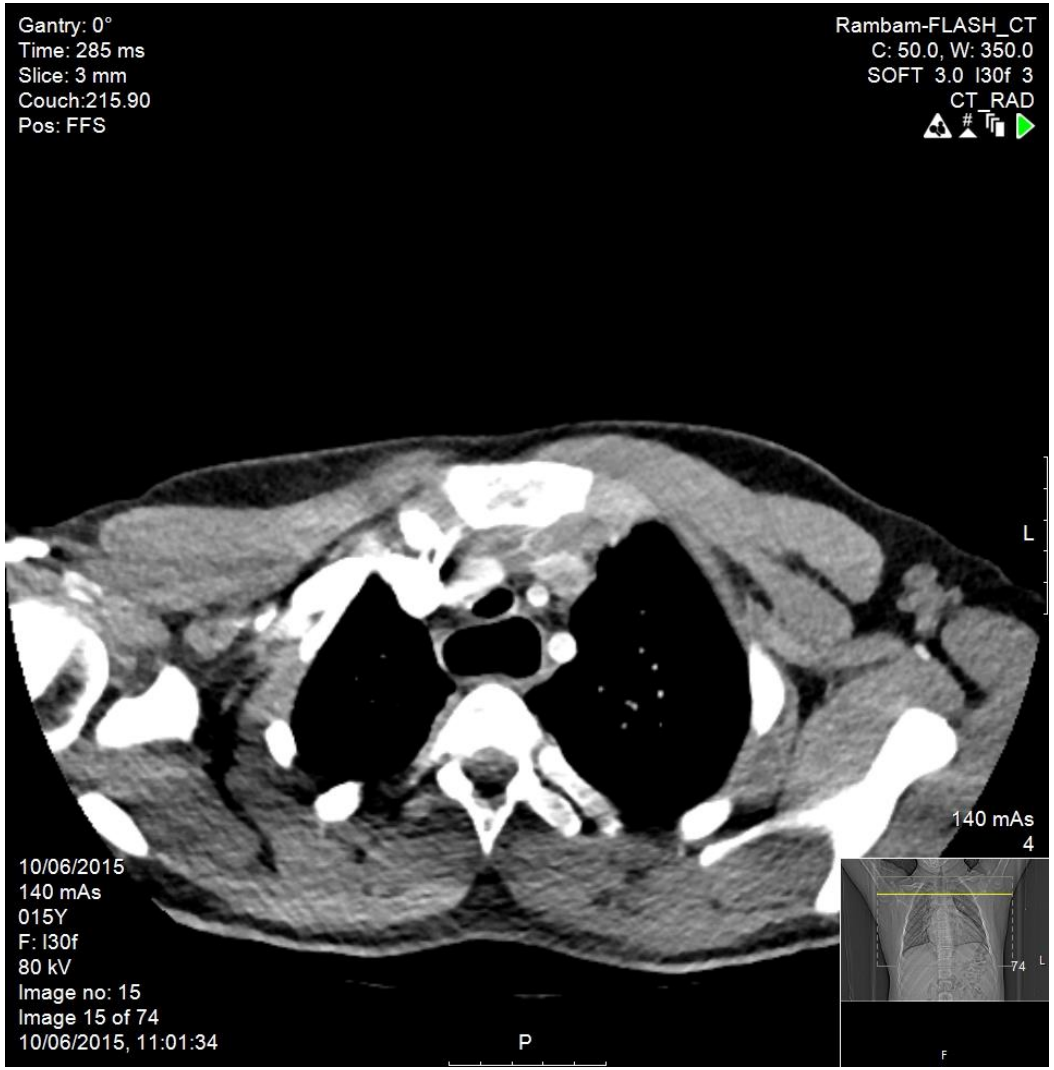
Heart Echo

- Normal Lt aortic arch.
- No vascular ring.
- Mild PHT (PAP = 39 mmHg).

Chest CT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Slice: 3 mm
Couch:215.90
Pos: FFS
Rambam-FLASH_CT
C: 50.0, W: 350.0
SOFT 3.0 I30f 3
CT_RAD
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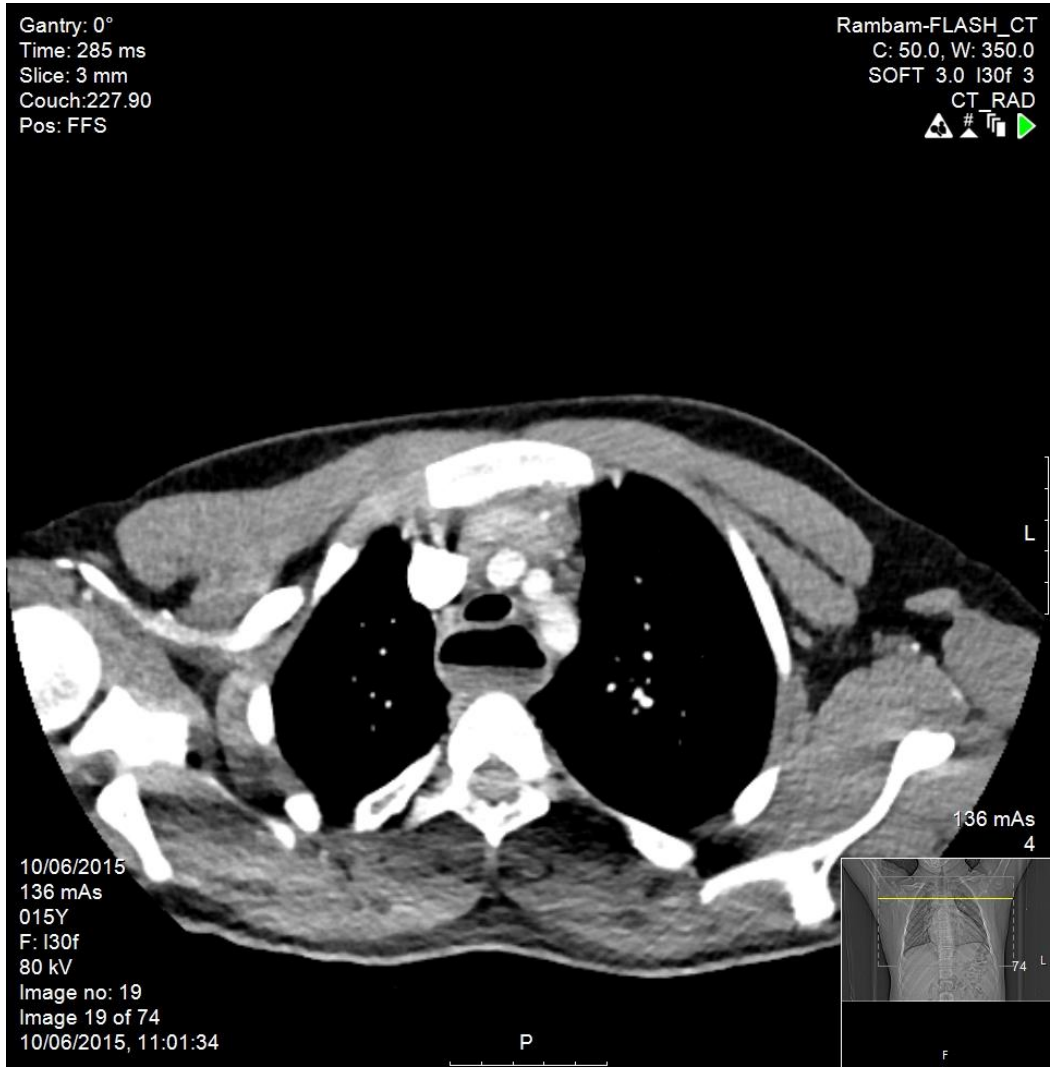
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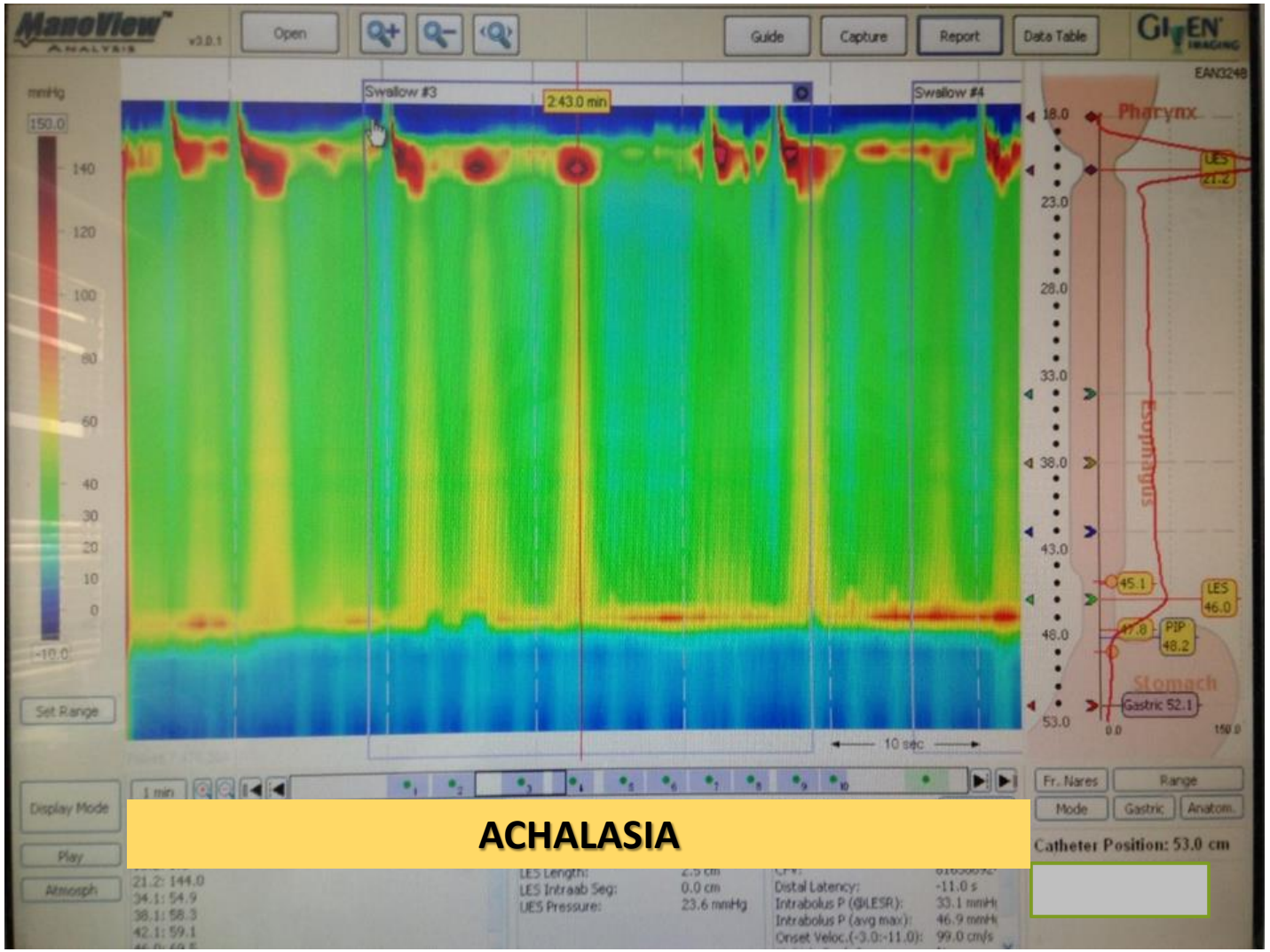
Panendoscopy

- Esophagitis in the lower half of the esophagus.
- Esophageal Dilatation with a lot of food content.
- No esophageal stenosis in the lower esophagus with an easy passage of the GEJ.

High Resolution Manometry



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Achalasia
A Systematic Review

John E. Pandolfino, MD, MSCI; Andrew J. Gawron, MD, PhD, MS

JAMA 2015; 313:1841-1852.

ACG Clinical Guideline: Diagnosis and Management of Achalasia

Michael F. Vaezi, MD, PhD, MSc, FACG[1], John E. Pandolfino, MD, MSCI[2] and Marcelo F. Vela, MD, MSCR[3]

Am J Gastroenterol 2013; 108: 1238-1249.

- Achalasia- a greek term that means "does not relax".
- Primary Achalasia is a disease of unknown etiology in which there is a loss of peristalsis in the distal esophagus and a failure of LES relaxation with swallowing.

Epidemiology

- An incidence of 1 in 100,000 individuals annually and prevalence of 10 in 100,000.
- Men=women.
- The peak incidence occurs between 30 and 60 years of age.
- **Onset before adolescence is rare** (0.11 in 100,000)**.**
- **Less typical symptoms (**recurrent pneumonia, nocturnal cough, aspiration, hoarseness, and feeding difficulties) and delayed diagnosis.
- Associated with Down syndrome, CCHS, Familial dysautonomia and Allgrove syndrome (Familial adrenal insufficiency and alacrima).

Etiology

- Functional loss of myenteric plexus ganglion cells in the distal esophagus and LES.
- The cause for an initial reduction of inhibitory neurons is unknown.
- May be an autoimmune process triggered by a viral infection (herpes, measles, HSV-1) in conjunction with a genetically susceptible host.
- Patients are more likely to have concomitant autoimmune diseases and the prevalence of serum neural autoantibodies is higher.

Pathophysiology

An inflammatory reaction (T-cell lymphocyte infiltrate) in the myenteric plexus

↓

Degeneration of inhibitory postganglionic neurons in the distal esophagus+LES

↓

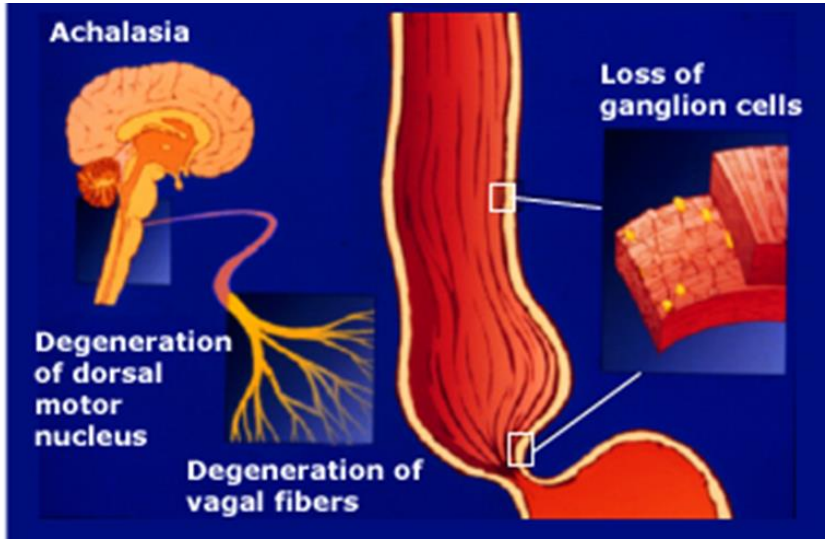
Imbalance between excitatory and inhibitory control

↓

Unopposed cholinergic stimulation

↓

Impaired relaxation of LES and aperistalsis in the distal esophagus.



Diseases associated with achalasia-like motility disorders

- Malignancy
- Chagas disease (Trypanosoma cruzi)
- Amyloidosis
- Sarcoidosis
- Neurofibromatosis
- Eosinophilic gastroenteritis
- Multiple endocrine neoplasia, type 2B
- Juvenile Sjogren's syndrome
- Chronic idiopathic intestinal pseudo-obstruction
- Anderson-Fabry disease

Symptoms and signs

Esophageal Symptoms

Dysphagia (90% of patients)

Heartburn (75% of patients)

Regurgitation or vomiting (45% of patients)

Noncardiac chest pain (20% of patients)

Epigastric pain (15% of patients)

Odynophagia (<5% of patients)

Other Associated Signs and Symptoms

Cough or asthma (20%-40% of patients)

Chronic aspiration (20%-30% of patients)

Hoarseness or sore throat (33% of patients)

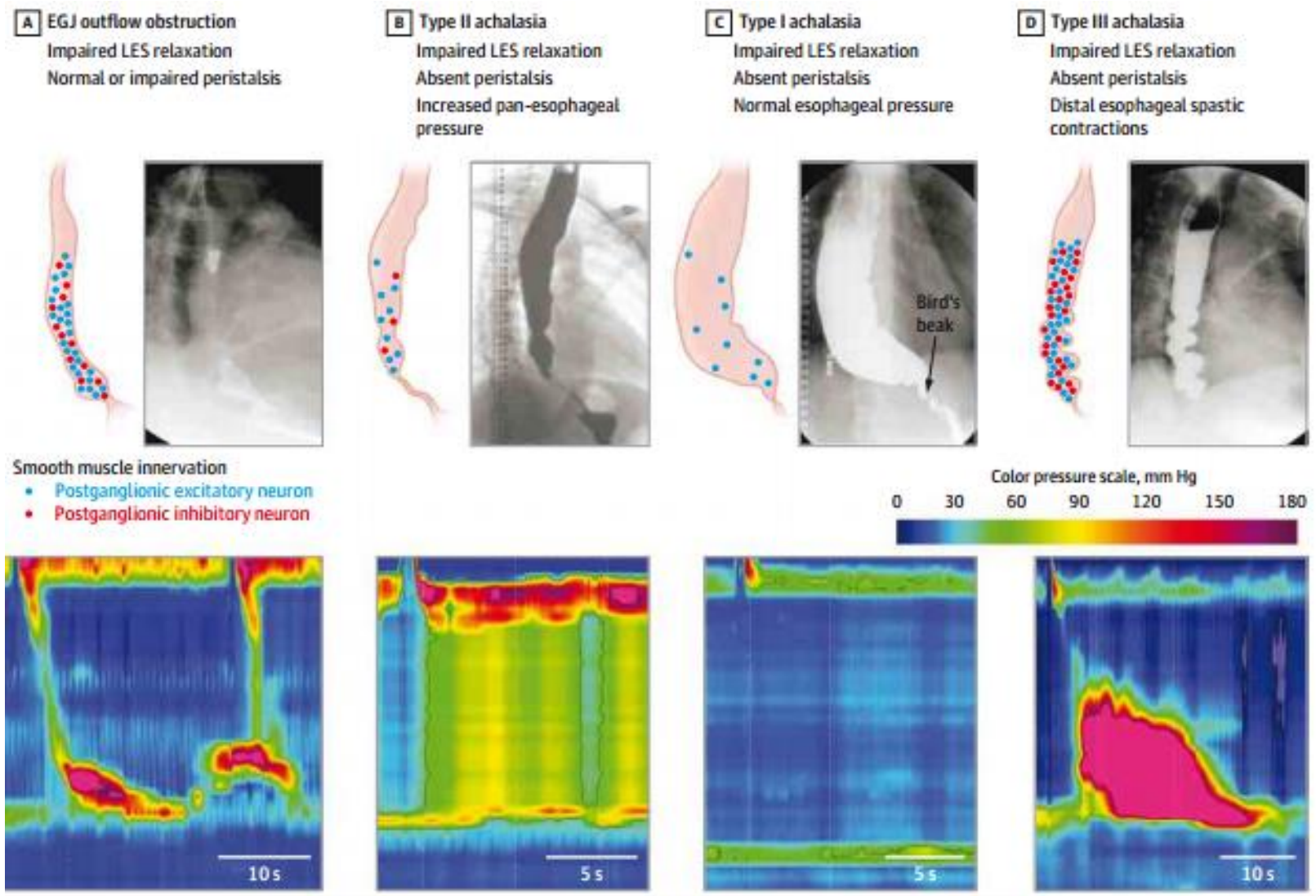
Unintentional weight loss (10% of patients)

[a] Data from Tsuboi et al,[33] Sinan et al,[34] and Ng et al.[35]

Diagnosis

- Esophagogastroduodenoscopy
- Barium Esophagram “bird’s-beak” appearance
- Esophageal Manometry

The Chicago Classification



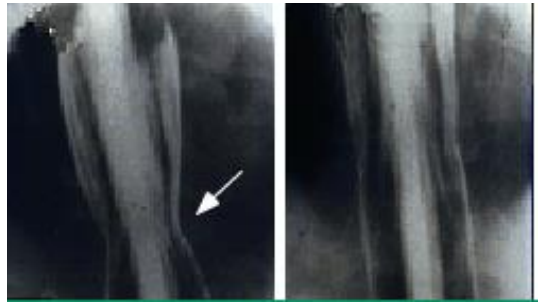
Pandolfino et al

Treatment

- Mechanical disruption of the muscle fibers of the LES (pneumatic dilation, surgical Heller myotomy or POEM).
- Biochemical reduction in LES pressure (injection of botulinum toxin, oral nitrates, or CCB).

➢Graded pneumatic dilation (70%-90% effective) or laparoscopic myotomy with partial funduplication (88%-95% effective).

➢Efficacy decreases over time and approximately 1/3-1/2 require repeat treatment within 10 years.



Follow-up

- Periodic evaluation by barium esophagram is the optimal approach.
- The risk of squamous carcinoma is higher, but no data to support routine endoscopic surveillance.
- Up to 6-20% of treated patients may have progressive dilation to megaesophagus or end-stage disease.
- Esophagectomy is reserved as a final option.

Acta Pædiatr 89: 356–64. 2000

CLINICAL OBSERVATIONS

Upper airway obstruction as a presenting sign of achalasia in childhood

A Kugelman[1], D Berkowitz[2], LA Best[3] and and L Bentur[4]

Respiratory & Upper AW Obstruction Mechanisms

- Extrinsic compression of the tracheo-bronchial tree by the dilated esophagus.
- Recurrent microaspiration.
- Hyper-reactivity of airways to the microaspirated food particles.
- The ‘pinchcock’ theory - cricopharyngeal muscle dysfunction or redundant esophageal folds create a one-way valve that allows air to enter the esophagus on inspiration, but prevents release of swallowed air.

Take Home Message

- Achalasia is rare in children.
- Presenting symptoms and esophageal contractile patterns may be inconsistent, resulting in delayed or missed diagnosis.
- Cough and unexplained upper AW obstruction can be the presenting symptoms of achalasia in childhood.

Don’t forget the esophagus!

תודה על ההקשבה!