

# DYSPHAGIA AND OPMD: MORE THAN AN OCULOPHARYNGEAL PROBLEM?

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# OVERVIEW

- Background
- Normal swallowing review
- Symptoms of OPMD dysphagia (difficulties swallowing)
- Tests and evaluation
- Treatment options



# BACKGROUND

## Late onset hereditary myopathy

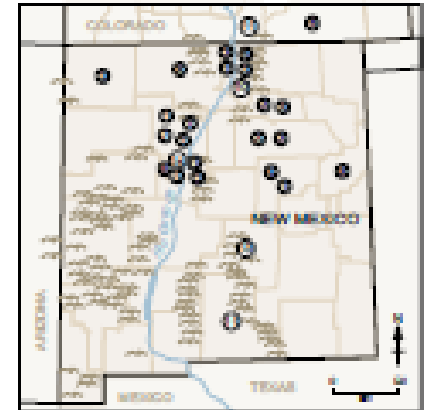
- Inherited disease (passed from generation to generation)
- Characterized by progressive ptosis (weakness of the eyelids)
- Dysphagia (difficulties swallowing from weakness of swallowing muscles)
- Limb weakness
- Doesn't shorten life but may change the way people live



# BACKGROUND

## OPMD in New Mexico

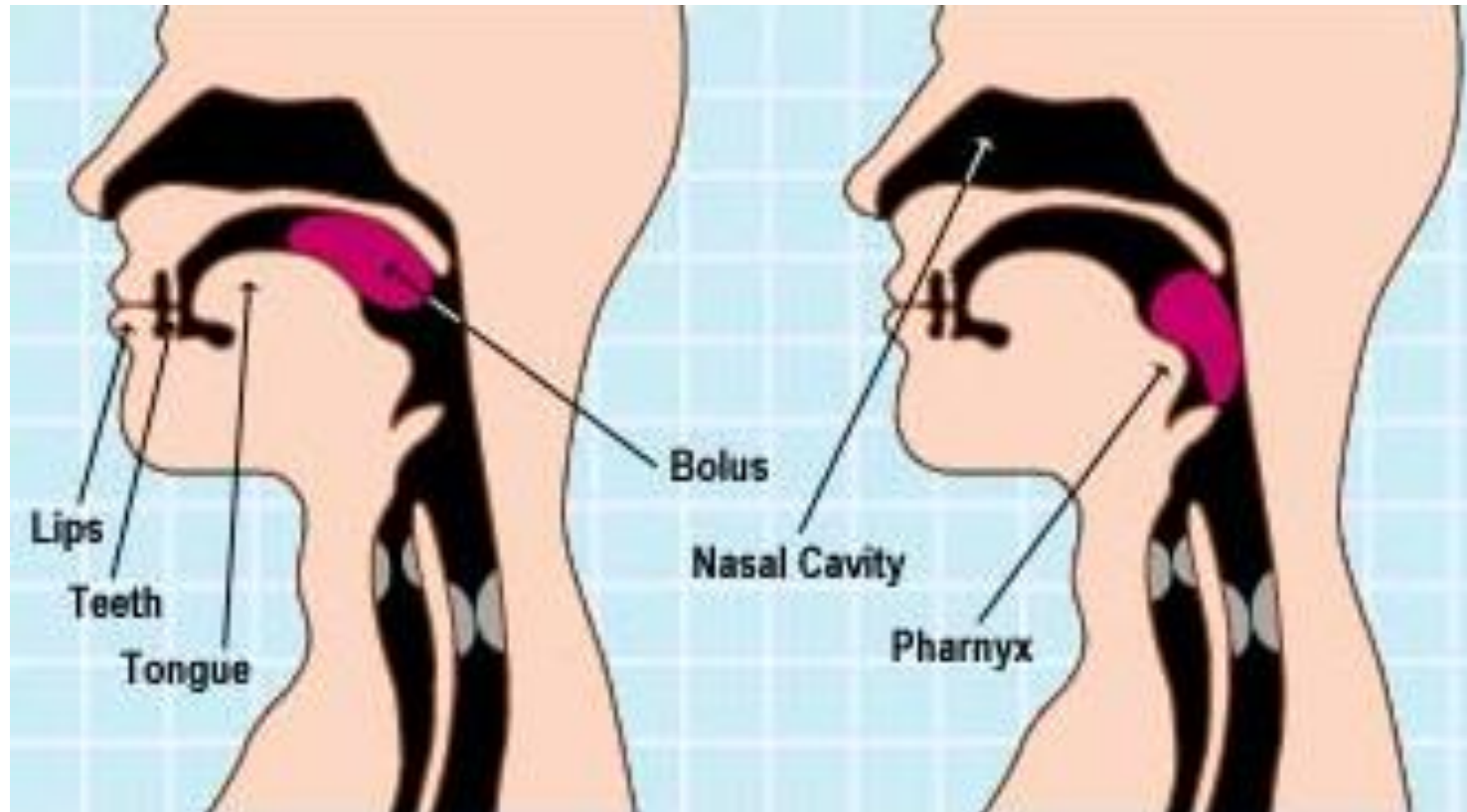
- 216 patients
- Symptoms
  - Ptosis: 190 (88%)
  - Dysphagia: 127 (59%)
- Onset
  - Ptosis before dysphagia: 20 (43%)
  - Ptosis and dysphagia together: 20 (43%)
  - Dysphagia before ptosis: 7 (14%)
- Mean onset of symptoms
  - Ptosis: 52 years of age
  - Dysphagia: 54 years of age
  - Proximal weakness: 63 years of age



# WHAT HAPPENS WHEN WE EAT?

- Ingest food into our mouth and hold it
- Initiate a swallow and move the food to the back of the throat
- A flap/epiglottis covers the wind pipe/trachea to prevent food from entering the trachea and lungs (aspiration)
- A valve at the top of the esophagus (upper esophageal sphincter) opens to allow food into the esophagus





# DYSPHAGIA – CLINICAL FEATURES

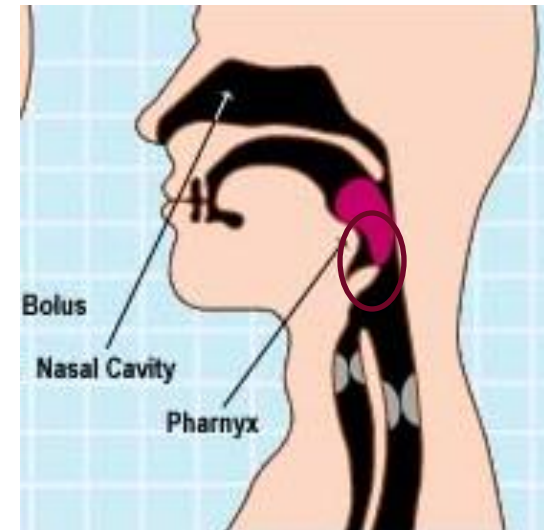
- Progressive oropharyngeal muscle weakness
  - Manifests with increased time to eat meals and avoidance of dry and solid foods
  - With progression, fluids may become difficult to swallow
  - End stage: characterized by aspiration, malnutrition, and weight loss
- Oral
  - Tongue weakness if observed
  - Muscles too weak to hold food or push bolus to back of throat



# DYSPHAGIA – CLINICAL FEATURES

## Pharynx

- Muscles may be too weak to get food into esophagus and food may “pool” in little pockets
- Muscles of flap may be too weak to protect voice box and trachea (aspiration)
- Soft palate may not keep food out of nasal cavity

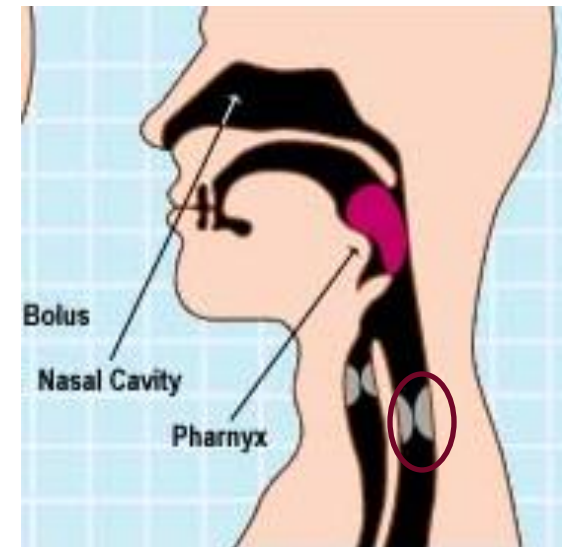




# DYSPHAGIA – CLINICAL FEATURES

## Esophagus

- The valve at the top end of the esophagus (upper esophageal sphincter) may be too thick and may not open to let food pass into the esophagus
- Older studies suggest that esophageal motility is impaired
- UNM retrospective study: self reported heartburn

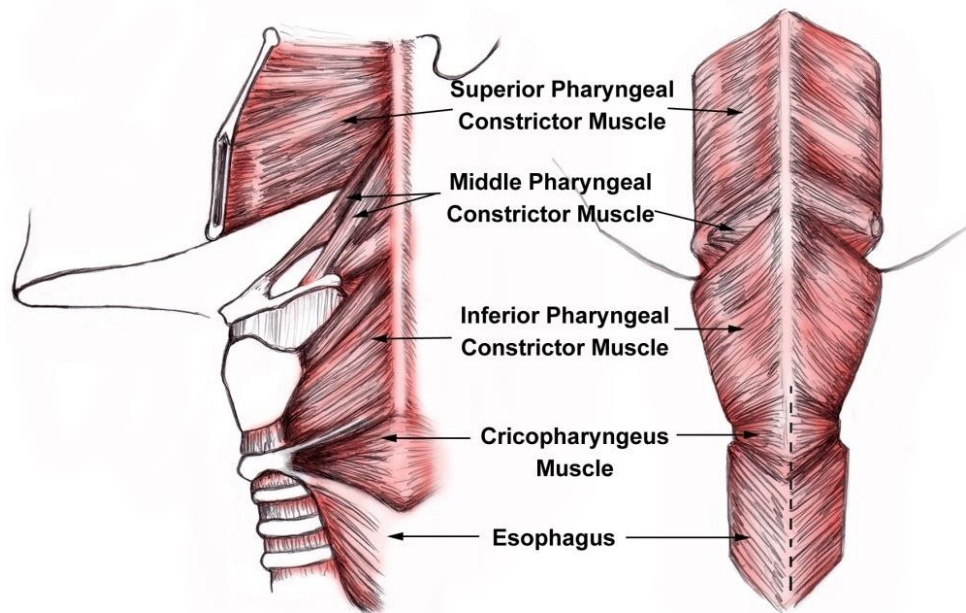


<sup>1</sup> Timony et al. Esophageal smooth muscle dysfunction in OPMD. *Dig Dis Sci* 1996; 41: 1350-1354.

<sup>2</sup> Castell et al. Manometric characteristics of the pharynx, upper esophageal sphincter, esophagus, and lower esophageal sphincter in patients with OPMD. *Dysphagia* 1995; 10: 22-26.

# UPPER ESOPHAGEAL SPHINCTER

- Inferior pharyngeal constrictor
- Cricopharyngeus muscle
- Cervical esophagus



# TESTS FOR SWALLOWING

- Speech Pathology Video Barium Swallow
- UGI Endoscopy
- Esophageal manometry/motility with impedance



# BARIUM SWALLOW



# BARIUM SWALLOW

- First test for dysphagia
- Can identify transfer problems
- Can tell if food goes down the trachea
- Can tell if the upper esophageal sphincter doesn't relax to allow food into the esophagus
- Look for blockage



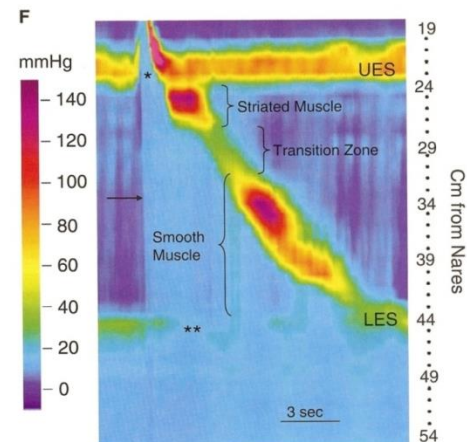
# BARIUM SWALLOW – COMMON FINDINGS

- Barium/food leaks from mouth or nose (nasal/oral regurgitation)
- Multiple swallows required to move barium/food from the mouth to the throat and esophagus
- Barium/food stays (“pools”) in throat and doesn’t get into esophagus
- Throat muscles seem weak
- Barium/Food gets past flap into voice box or trachea
- Muscles of the upper esophageal sphincter are too thick and don’t allow food to pass



# ESOPHAGEAL MANOMETRY AND IMPEDANCE

- Measures the pressures in the throat and esophagus
- Usually done without sedation



# TREATMENT

- Treatment (alteration of the cricopharyngeus muscle anatomy and function)
  - Cricopharyngeal myotomy (surgery)
  - Botox injection (powerful nerve toxin injected to induce muscle relaxation)
  - Esophageal dilation
- Dietary changes:
  - Smaller meals
  - Soft, ground diet
  - Liquids via cup
  - Allow more time to swallow
  - Alternate solids and liquids
  - Eat sitting upright
  - Remain upright after meals 30-60 minutes
  - Head flex/Chin tuck





# CRICOPHARYNGEAL MYOTOMY

- First reported by Peterman in 1964
- Fully described by Montgomery and Lynch in 1971
- Technique: division of entire inferior pharyngeal constrictor muscle, cricopharyngeus muscle, and the upper part of the circular fibers of the cervical esophagus
- Hypothesis: remove obstruction made by constrictive UES that cannot be overcome by decreased pharyngeal propulsion
- Retrospective study of 37 patients from 1980 to 1995
  - Mean follow-up 6.2 years:
    - Totally relieved or rarely occurring symptoms: 18/37 (49%)
    - Moderate symptoms/partial: 12/37 (32%)
    - Severe symptoms/failure: 7/37 (19%)
    - Follow-up at 8 years: Nearly all patients had recurrence of swallowing and tracheobronchial symptoms



# CRICOPHARYNGEAL MYOTOMY

Retrospective study of 22 patients from 1987 to 1995

- 12 patients underwent cricopharyngeal myotomy
  - Mean follow-up 29.6 months
  - Improvement: 10 patients
  - Partial improvement: 1 patient
  - No improvement: 1 patient
- Factors associated with favorable outcome were residual pharyngeal propulsion and no weight loss
- Conclusion:
  - Cricopharyngeal myotomy is an effective treatment of dysphagia with adequate residual propulsion but does not modify the final prognosis and is contraindicated in cases with pharyngeal aperistalsis



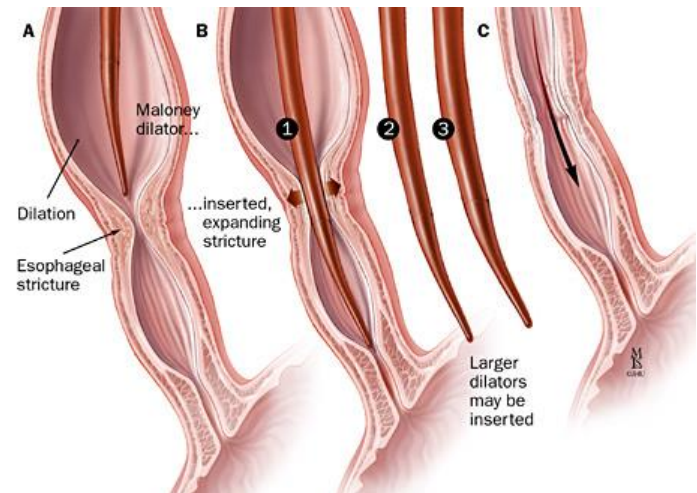
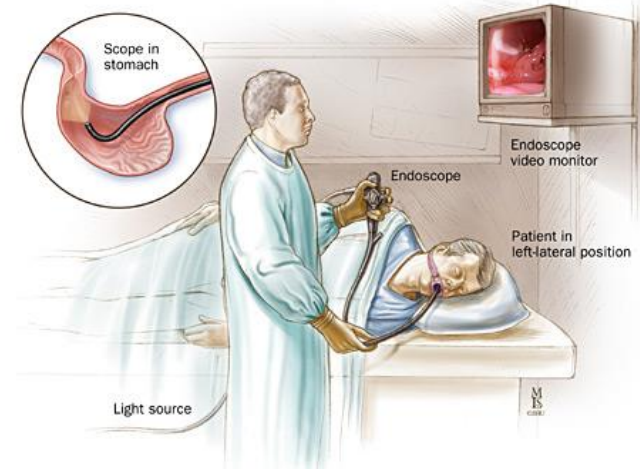
# BOTOX

- Injection of powerful neurotoxin produced by *Clostridium botulinum*
- Limited to cases
- Limitations/side effects:
  - Temporary
  - Dysphonia
  - Aspiration



# DILATION PROCEDURE

- Conscious sedation: light sleep
- Endoscopy evaluation of esophagus, stomach, and small intestine
- A wire is passed through the scope and positioned in the stomach. The scope is removed and exchanged with the wire
- A Savary dilator is passed over the wire to stretch the cricopharyngeus muscle



# RECENT DILATION STUDY

- Retrospective study from 1995-2007
- 9 patients
- Dilation performed using 54Fr Savary-Gilliard bougie
- Symptom severity prior to dilation and at follow-up (1, 4, and 12 months) was evaluated using the Sydney Swallow Questionnaire (SSQ)
- Median total treatment dilation period: 13 years
- Median number of dilatations per patient: 7.2
- Median interval between treatments: 15 months



# RECENT DILATION STUDY

- Mean SSQ prior to dilation: 1108.11
- Mean SSQ at last follow-up: 297.78 (73% decrease);  $p= 0.0001$ 
  - Interview was performed an average of 4.57 months after the most recent dilation (range 3-8 months)
- Conclusion:
  - Repeated cricopharyngeal dilation is a safe, effective, well-tolerated and long-lasting treatment for dysphagia in OPMD



# PRIOR UNM STUDY

- Retrospective study
  - OPMD patients seen in UNM GI, ENT, Neurology
  - Diagnosis of OPMD with or without genetic confirmation
- Results:
  - 100 patients
  - Mean age 59
  - Dysphagia: 78%
    - Mean age of onset 55
    - Progressive: 83%
    - Weight loss: 21%
  - Heartburn – self-reported in 45%



# PRIOR UNM STUDY

## ○ Results:

- Prolonged meals and increased symptoms with solids: 90%
- Choking spells: 75%
- Pill dysphagia: 25%

## ○ Dysphagia treatment:

- Savory dilation: 20 patients
  - 2 minor complications – dyspnea, epigastric pain
- Botox: 12 patients
  - 5 minor complications – dysphonia, hoarseness, soreness
- Savory dilation and Botox: 8 patients





# PRIOR UNM STUDY

- Results:
  - 82% improved with dilation
  - 66% improved with Botox treatment
  - No significant difference between the treatments ( $p=0.4$ )
  - No significant difference in complications between the treatments ( $p=0.09$ )



# PROSPECTIVE UNM STUDY

- Assess esophageal dysphagia via modern high-resolution manometry
  - Are there esophageal disorders we should be treating?
- We plan to combine manometry with pH/impedance to determine if patients experience acid or non-acid reflux
  - Do OPMD patients benefit from treatment of GERD
- We hope to obtain more information regarding the role of dilation for OPMD patients
  - 54Fr dilation followed by 60Fr dilation if no improvement or recurrence of symptoms



# REFERENCES

- Becher et al. Oculopharyngeal Muscular Dystrophy in Hispanic New Mexicans. *JAMA*. 2001; 286: 2437-2440.
- Manjaly et al. Cricopharyngeal dilatation for the long-term treatment of dysphagia in OPMD. *Dysphagia* 2011 Jul 30: Epub ahead
- Tiomny et al. Esophageal smooth muscle dysfunction in OPMD. *Dig Dis Sci* 1996; 41: 1350-1354.
- Bender MD. Esophageal manometry in oculopharyngeal dystrophy. *Am J Gastroenterol*. 1976 Mar; 65(3):215-21.
- Duranceau et al. Oculopharyngeal dysphagia in patients with OPMD. *Can J Surg*. 1978 Jul; 21 (4):326-9.
- Castell et al. Manometric characteristics of the pharynx, UES, esophagus, and LES in patients with OPMD. *Dysphagia*. 1995 Winter; 10(1):22-6.
- Fradet et al. Upper esophageal sphincter myotomy in OPMD: long-term clinical results. *Neuromusc Disorders* 7 Suppl 1997; S90-S95.
- Perie S et al. Dysphagia in OPMD: a series of 22 French cases. *Neuromuscul Disord* 1997 Oct; 7 Suppl 1: S96-9.
- Taillefer et al. Manometric and radionuclide assessment of pharyngeal emptying before and after cricopharyngeal myotomy in patients with OPMD. *J thorac Cardiovasc Surg* 1988; 95: 868-875.



# QUESTIONS?

