“Pediatric Aero-Digestive Disorders in the New Century”

A Valley-Mount Sinai Kravis Children's Hospital educational symposium.

Valley Health System

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CHILDREN’S HEALTH

Rheumatologic Conditions Affecting the Esophagus in Children

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Faculty disclosure

1. There are no commercial products or services being discussed
2. No financial disclosures
3. No unlabeled use of a product is being discussed
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Objectives

- Understand what rheumatic conditions affect the esophagus and airway in children

- Identify children who require intervention and/or referral to pediatric rheumatology for rheumatologic diseases affecting the esophagus and airway

- Understand basic treatment concepts and approaches for children rheumatologic disease affecting the esophagus and airway

Overview

- Multiple rheumatic diseases in children are known to affect the esophagus and airway
  - These are often heterogeneous systemic diseases with variable manifestations
  - So aerodigestive system not ALWAYS affected, but can be
- Will go through the ‘most common’ ones
  - What is it?
  - Aerodigestive manifestations
  - What else should we look for?
  - Workup and basic principles of management

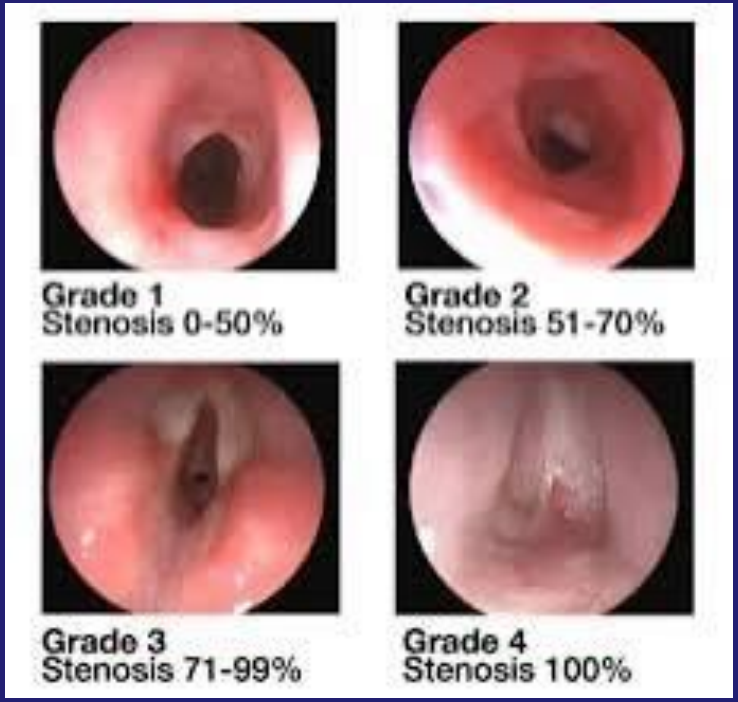
Granulomatosis with Polyangiitis (GPA)

GPA: What is it?

- Variable vessel vasculitis
  - Especially small vessels
- ANCA-associated
  - Most commonly anti-PR3 or c-ANCA
  - Rarely anti-MPO or p-ANCA
- Pathology is notable for granulomas
  - ****

GPA: Aerodigestive manifestations

- Subglottic stenosis
  - Tracheal stenosis
  - Laryngeal stenosis
- This is more common in children than adults
  - Can be isolated manifestation



GPA: Additional manifestations

- Commonly a pulmonary-renal syndrome
- Constitutional symptoms
  - Fever
  - Fatigue
  - Weight loss
- Less commonly:
  - Sinus disease
  - ENT disease – vision/hearing loss, ulcers
  - Arthritis
  - Rash

GPA: Workup

- Thorough history and physical examination
- Bloodwork
  - Inflammatory markers
  - ANCAs
  - STUDIES TO RULE OUT OTHER POTENTIAL DIAGNOSES
- Imaging as appropriate
- Biopsy
  - Skin when available
  - Other involved areas where appropriate

GPA: Principles of management

- Immunosuppressive treatment
- Initially steroids
  - Pulse dose vs. high dose
- Rituximab often used for induction of remission
  - Though less intensive therapies used for mild disease
- Many options for maintenance of remission
  - Usually for ~2 years
  - Then trial taper
- Increasing role for Avacopan
  - C5a inhibitor

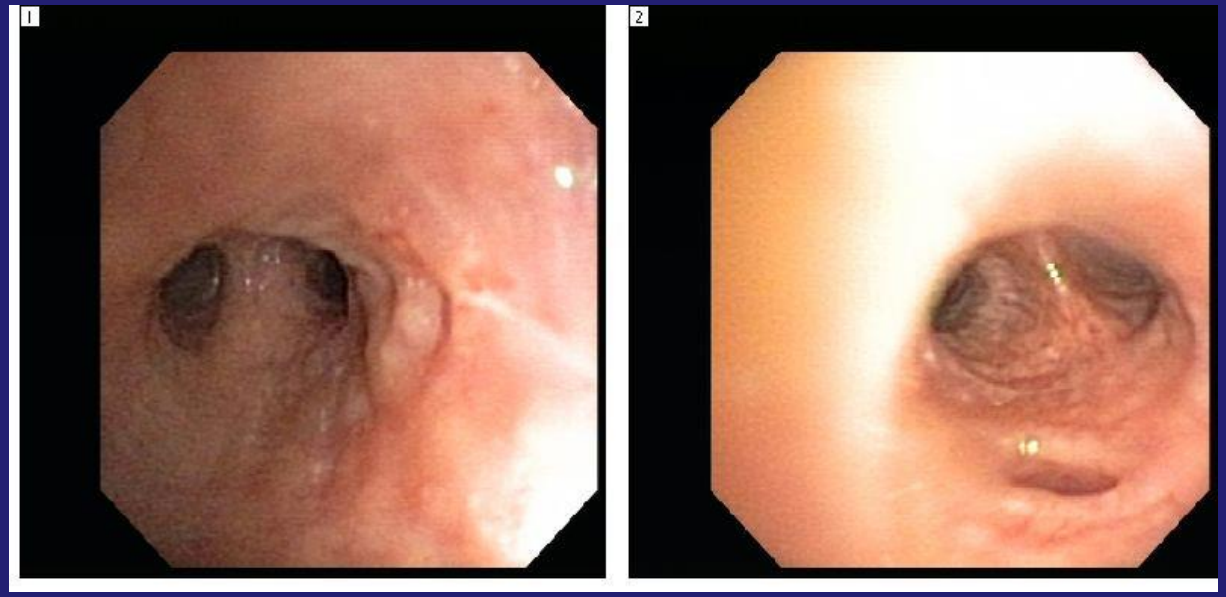
Sarcoidosis

Sarcoidosis: What is it?

- Growth of collections of inflammatory cells in different parts of the body
- Associated with NOD2 mutations
- Pathology is notable for granulomas
- Heterogeneous disease with varying manifestations in children and adults
  - Blau syndrome
  - Early-onset sarcoidosis

Sarcoidosis: Aerodigestive manifestations

- Nodules/granulomas in airway
- Can lead to:
  - Dysphagia
  - Laryngeal paralysis
  - Upper airway obstruction



Sarcoidosis: Additional manifestations

- Most commonly pulmonary syndrome
- Can also involve
  - Lymph nodes – proliferation and enlargement
  - Constitutional symptoms
  - Skin – rashes
  - Eyes – panuveitis
  - Joints – arthritis
- Adults can have Lofgren syndrome
  - Triad of fever, erythema nodosum, and hilar adenopathy
- Children can have Blau syndrome
  - Triad of rash, arthritis, and uveitis

Sarcoidosis: Workup

- Thorough history and physical examination
- Bloodwork
  - Inflammatory markers
  - ACE, lysozyme, and soluble IL-2 receptor
  - STUDIES TO RULE OUT OTHER POTENTIAL DIAGNOSES
- Imaging as appropriate
- Biopsy
  - Skin when available
  - Other involved areas where appropriate

*Can be particularly difficult to distinguish from TB

Sarcoidosis: Principles of management

- Immunosuppressive treatment
- Initially steroids
  - Pulse dose vs. high dose
- Many options for continued steroid-sparing treatment, if needed
  - Methotrexate
  - Azathioprine
  - Cyclophosphamide
  - Anti-TNF agents
- Treatment duration can be very short OR up to 2 years with trial of taper

Juvenile Dermatomyositis (JDM)

JDM: What is it?

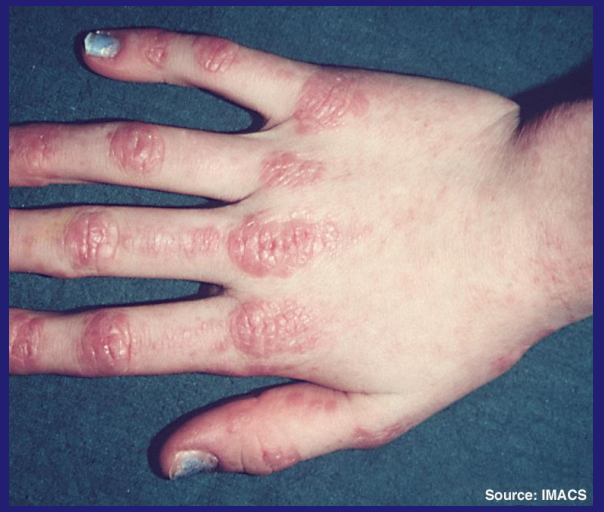
- Rare autoimmune inflammatory myositis
  - With vasculitis component
- Seasonal clustering
  - Suggests role of infections as triggers
- In adults, almost always a paraneoplastic phenomenon
  - However, stand-alone entity in children

JDM: Aerodigestive manifestations

- Primarily caused by muscle weakness
- Can lead to:
  - Dysphagia
  - Difficulty breathing
- Can also have abdominal symptoms
  - Secondary to vasculitis

JDM: Additional manifestations

- Most commonly symmetric proximal muscle weakness + rash
- Muscle involvement:
  - Can involve any muscle
- Rashes
  - Heliotrope
  - Gottron's papules
  - Malar, erythroderma
- Many other possible findings
  - Lung disease, cardiac disease, ocular disease
- Calcinosis



JDM: Workup

- Thorough history and physical examination
  - Muscle strength testing + rashes
- Bloodwork
  - Inflammatory markers
  - Muscle enzymes: CPK, AST/ALT, aldolase, LDH
  - Myositis specific antibodies (MSSAs)
  - STUDIES TO RULE OUT OTHER POTENTIAL DIAGNOSES
- Imaging as appropriate
- Biopsy
  - Skin when available
  - Muscle if needed

JDM: Principles of management

- Immunosuppressive treatment
- Initially steroids
  - Pulse dose vs. high dose
  - Slowly taper over one year
- Many options for continued steroid-sparing treatment
  - IVIG
  - Methotrexate
  - Cellcept
  - Rituximab
  - JAK inhibitors

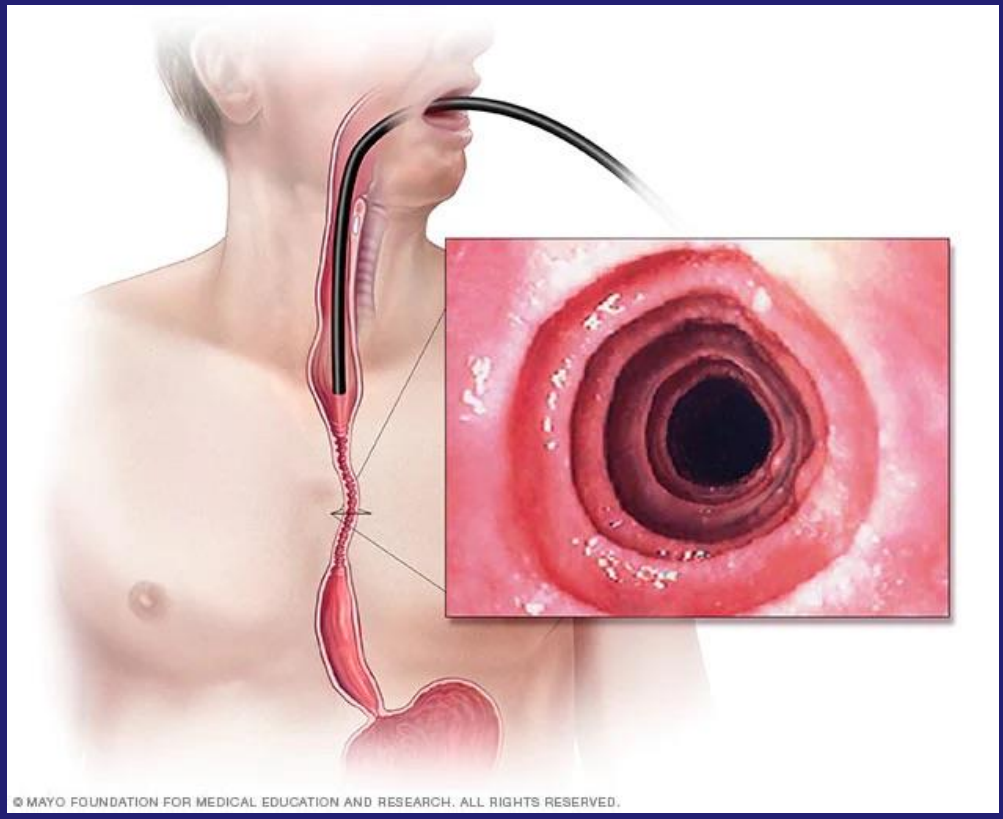
Polyarticular JIA (pJIA)

pJIA: What is it?

- One of the types of juvenile idiopathic arthritis
- Adult correlate: rheumatoid arthritis
  - Suggests role of infections as triggers
- Strong female predominance
- Most commonly consists of symmetric small joint arthritis
  - Other joints can be involved
  - Wrists, large joints, mandible, C1/C2

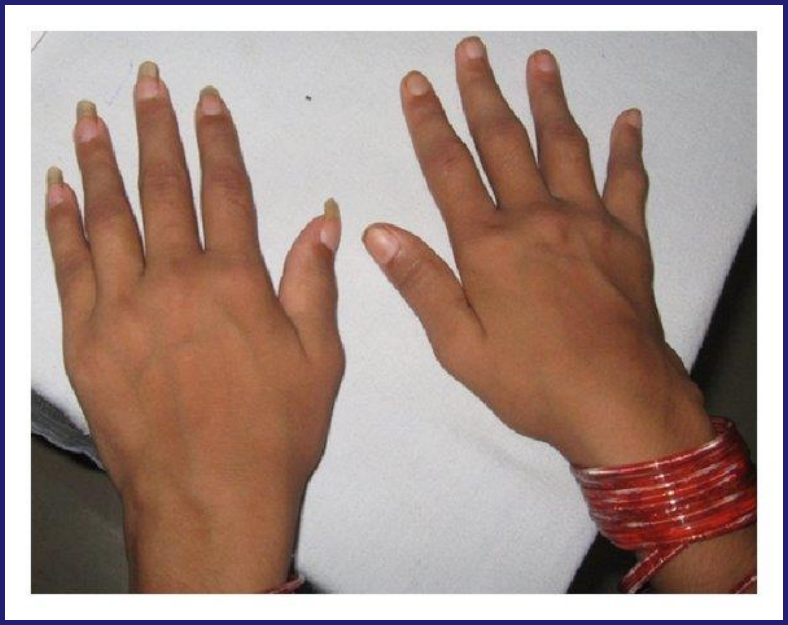
pJIA: Aerodigestive manifestations

- Esophageal dysmotility
- Esophagitis
- Dysphagia
  - Secondary to TMJ or C-spine arthritis
- Increased occurrence of IBD



pJIA: Additional manifestations

- Symmetric small joint arthritis
- Can also involve:
  - Constitutional symptoms
  - Lung disease
  - HSM
  - Rheumatoid nodules



pJIA: Workup

- Thorough history and physical examination
  - Evaluation for arthritis
- Bloodwork
  - Inflammatory markers
  - Rheumatoid factor (RF)
  - Anti-citrullinated peptide (CCP)
  - STUDIES TO RULE OUT OTHER POTENTIAL DIAGNOSES
- Imaging as appropriate

pJIA: Principles of management

- Immunosuppressive treatment
- Rarely require steroids
- Many options for treatment
  - Methotrexate
  - Anti-TNF therapy
  - IL-6 blockade
  - Abatacept
  - Rituximab
  - JAK inhibitors

Sjogren’s disease

Sjogren's: What is it?

- Rare autoimmune disease
  - Can have co-incidence with SLE and RA
- Most commonly leads to sicca syndrome
  - Dry eyes
  - Dry mouth
- However, can have many other manifestations

Sjogren's: Aerodigestive manifestations

- Dysphagia
- Increased incidence of celiac disease
- Can also have liver dysfunction

Sjogren's: Additional manifestations

- Sicca syndrome
  - Loss of taste, dental cavities, ulcers
- Constitutional symptoms
- Arthritis
- Enlarged lymph nodes and salivary glands
- Rashes
- Many other components
  - Lung disease, cardiac disease, ocular disease

Sjogren's: Workup

- Thorough history and physical examination
- Bloodwork
  - Inflammatory markers
  - Antibodies: anti-Ro/La, RF
  - Quantitative Ig's – especially IgG
  - STUDIES TO RULE OUT OTHER POTENTIAL DIAGNOSES
- Imaging as appropriate
- Biopsy
  - Skin when available
  - Other involved areas as needed

Sjogren's: Principles of management

- Topical treatment for dry eyes/mouth
- Additional agents:
  - Plaquenil
  - Steroids
  - MTX
  - Infliximab
  - Rituximab
- *GI symptoms will generally resolve with treatment of underlying disease
  - Aside from concurrent autoimmune disease

Summary

Summary

- Rheumatologic disease is rare
- However, can have important aerodigestive manifestations
- Important to evaluate thoroughly when something seems ‘off’
- Start with a history and physical examination and you will gain a lot
- Then use laboratory analysis, imaging, and biopsy to supplement this, as appropriate

Questions?

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