Diagnostic Approach to chILD, Treatment and Future Directions

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Courtesy of chILD Foundation
First Steps

- **Recognizing** ‘chILD’ is important and not always easy
- Diagnosis often delayed or missed
- Symptoms overlap with other respiratory diseases
- Look for **respiratory symptoms OUT OF PROPORTION** to more common or known underlying disease

A Boston Children’s Hospital patient
Diagnostic Team

- Multi-disciplinary team with knowledge of chILD
- Individual patient & family centered approach

Boston Children’s Hospital chILD Program Team

A Boston Children’s Hospital patient
National Team: Children’s Interstitial Lung Disease Research Network

* chILD ‘Center of Excellence’
* Satellite

Slide courtesy of Robin Deterding
Team Objectives

• Primary role of diagnostic process is to answer a clinically relevant question

• Team’s responsibility to weigh risk and benefit appropriately
Diagnostic Approach

• Guided by team experience and chILD expert guidelines
• Rule out other common causes of diffuse lung disease
• Radiographic imaging
• Lung function testing
• Bronchoscopy
• Genetic testing
• Lung biopsy
• Look for coexisting complications: infection, immune dysfunction, aspiration, environmental exposures, autoimmune disease, pulmonary hypertension, and other
• Consultation with other subspecialists
Expert Guidelines: Diagnostic Approach

RULE OUT/TREAT UNDERLYING DISEASE:
- Infection/Pneumonia: Bronchoscopy/BAL
- CHD/ Pulmonary Hypertension: EKG; Echocardiogram
- HIV/Immunodeficiency: CF; Recurrent Aspiration

NEGATIVE EVALUATION OR PERSISTENT DISEASE DESPITE TREATMENT

CVHRCT
- Suggests alveolar proteinosis (diffuse ground glass)

GENETIC TESTING
- see Fig. 4

DIAGNOSTIC
- NEGATIVE

BIOPSY WITH E.M.

Clinical Practice Guidelines
AJRCCM, August 2013
Pulmonary Function Testing

• A series of breathing tests to determine how much, and how well, the lungs take in and expel air

• Exercise testing to determine exercise tolerance and oxygen levels during exercise

http://www.chp.edu/our-services/pulmonology/patient-procedures/infant-pulmonary-function-testing
Radiographic Imaging: Chest Xray

Courtesy of Geoff Kurland
Radiographic Imaging: Chest CT

- Challenges in children
- May need anesthesia
- Radiation considerations
- Some diagnostic
- Biopsy planning

http://www.childrenshospital.org
Bronchoscopy

• Examination of airways to look for:
  – Structural abnormalities
  – Infection
  – Signs of Inflammation
  – Bleeding
  – Aspiration
  – Other
Genetic Testing

- Some chILD diseases have known genetic causes
- Various genetic tests on blood and other tissue to detect
  - Specific disorders
  - Panels
  - Whole exome/whole genome

Courtesy of Larry Nogee
Lung Biopsy

- Involvement may be patchy
- Try to avoid when sick with intercurrent infection or on therapy that may change results
- Thoracoscopic if possible
- Need to collapse the lung to obtain with scope
- Minimize risk

Courtesy of Craig Lillehei
Pathology Review

Courtesy of Sara Vargas

Dishop, PAIP 2010

Courtesy of Geoff Kurland

Dishop, PAIP 2010
ChILD Treatment

Supportive Care

• Oxygenation
• Ventilation
• Nutritional support
• Immunizations
• Avoid Smoking
• Bronchiectasis therapy
• Exercise therapies/pulmonary rehab
• Referral for transplant

Anti-Inflammatory Therapies

• Steroids
• Azithromycin
• IVIG
• Hydroxychloroquine
• Steroid sparing anti-inflammatories
• Rescue therapies
Respiratory Support

Breathing Support

• Supplemental oxygen
• Non-invasive Ventilation
  – CPAP
  – BiPAP
• Invasive Ventilation
  – Intubation
  – Tracheostomy
  – ECMO
  – Artificial lung devices

A Boston Children’s Hospital patient
Nutritional Support

• Children with ILD often work harder to breathe and need to consume more calories

• Nutritional support often needed:
  • High calorie foods
  • Fortified liquids
  • Caloric supplements
  • NG or NJ tubes
  • G or GJ tubes
  • TPN

• Difficulty swallowing/oral aversion can be present and requires expertise

A Boston Children’s Hospital patient
Other Supportive Care

• Prevent further damage to the lungs
  – Avoid infections as much as possible
  – Keep all vaccines up to date (this includes other family members)
  – No smoke exposure

• Monitor for problems in other organ systems
Treatment Challenges

- Lack of randomized controlled trials
- Individual/network experience
- Based on adult treatment
- Treat primary and coexisting conditions
- Multi-disciplinary team needed
- More options needed with data to support use
Lung Transplant for ChILD

• ChILD 2\textsuperscript{nd} most common reason for pediatric lung transplant
  – Pulmonary fibrosis, interstitial lung disease, surfactant abnormalities, non-lung transplant BO, & lung growth abnormalities

• Younger patient population of the pediatric transplant recipients

• Similar outcomes to other populations
  – May be less rejection (younger vs. disease)

\textit{Ped Pulm} 48:490
ChILD Research Network

Because disorders are rare, collaboration between multiple centers and families is imperative

Interdisciplinary and Multi-Center Collaboration:
Improve care for children and families
Finding new treatments and ultimately cures
- Clinical Care
- Research
- Education
- Advocacy

ChILD
CHILDREN’S INTERSTITIAL AND DIFFUSE LUNG DISEASE FOUNDATION
ChILD Research Network

- Collaboration chILD Foundation
- Monthly phone meetings
- Conferences
- Patient data registry
- Multiple chILD publications
- Grants
- New research
- Educational materials/events
- Planning treatment/interventional studies

Courtesy of chILD Foundation
ChILD Partnership

• Families can help by:
  – Participating in chILD foundation activities
  – Supporting each other
  – Asking questions
  – Raising awareness
  – Enrolling in studies
  – Fundraising
  – Participate in the #CartwheelsForKids Challenge!

Research Network Family Member
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