Management of pediatric diffuse lung disease in Switzerland

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chILD

Rare disease

Low prevalence (< 1/100’000)

American guidelines for classification, evaluation and management  
Kurlang G, Amer J Respir Crit Care Med 2013;188

Pan european registry to harmonise diagnostic approaches and treatment protocols  
Bush, Thorax 2013;68

Australasian Registry Network of Orphan Lung diseases  
Casamento, Orphanet Journal of Rare Disease 2016;11
Survey Monkey questionnaire

Sent by email to 68 paying members of the SGPP (32 pediatric pulmonologists)

4 bounced, 2 opted out
13 answered (21%), 11 pediatric pulmonologists (34%)

43 questions

Demographics (5)
Generic (11)
Specific to patients, work up and treatment (27)

Demographics

77% between 35-55 years
Equal distribution of new-confirmed-founding members
77% teaching hospital
Patients

54% follow at least one ILD patient

- 1 patient 15.4%
- 2-5 patients 30.8%
- >5 patients 7.7%

16 ILD patients total
Mean age 6.5 years, median 6, range 2-14

11 diagnosed in the last 18 months
Mean age 6.1 years, median 3.5, range 2-14

Time frame before diagnosis
Mean 3.7 months, range 1-12
# Diagnosis

| Diagnosis                                                | # cases (age yrs) |
The|----------------------------------------------------------|------------------|
<table>
<thead>
<tr>
<th></th>
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</tr>
</thead>
<tbody>
<tr>
<td>Desquamative interstitial pneumonia</td>
<td>2 (5-10)</td>
</tr>
<tr>
<td>Hypersensitivity pneumonitis</td>
<td>2 (10-8)</td>
</tr>
<tr>
<td>Neuroendocrine cell hyperplasia</td>
<td>2 (2-2)</td>
</tr>
<tr>
<td>ABCA3 deficiency</td>
<td>2 (&lt;1-8)</td>
</tr>
<tr>
<td>Idiopathic pulmonary hemosiderosis</td>
<td>1 (6)</td>
</tr>
<tr>
<td>Pulmonary interstitial glycogenosis</td>
<td>1 (3)</td>
</tr>
<tr>
<td>Bronchiolitis obliterans (?)</td>
<td>1 (14)</td>
</tr>
<tr>
<td>Follicular bronchiolitis with veno occlusive disease</td>
<td>1 (2)</td>
</tr>
<tr>
<td>Congenital kystic lymphangioma</td>
<td>1 (10)</td>
</tr>
<tr>
<td>Unspecific ILD</td>
<td>2 (2-14)</td>
</tr>
<tr>
<td>Unknown</td>
<td>1 (2)</td>
</tr>
</tbody>
</table>
Usual investigations

80% have access to skilled pediatric radiologists and surgeons

All have access to an endoscopy suite
Usual investigations

Blood work

- Immune function: always
- Autoantibodies: 75%
- Environnemental organic dust exposure: 58.3%
- ACE inhibitor: 25%
- Genetic studies in the child: 41.7%
- Genetic studies in the parents: 8.3%

Thoracic CT scan

- HRCT insp/exp: 77%
- Insp/exp: 15.4%
- With contrast: 54%

Pulmonary function tests: 85%
Other investigations

**Bronchoscopy + BAL**
- Complete microbiological studies: 100%
- Total cell count: 100%
- Lymphocyte subsets: 100%
- Lipid laden macrophage index: 100%
- Iron staining: 67%
- PAS staining: 92%
- CD1a: 50%

**Transbronchial biopsy**: 8.3%

**Lung biopsy**
- Prior to starting steroids: 87.5%
- According to specific SOP: 75%
- Pathology expert reading: 91%
### First treatment option

<table>
<thead>
<tr>
<th>Treatment</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Oral prednisolone</td>
<td>80%</td>
</tr>
<tr>
<td>IV Methylprednisolone</td>
<td>10%</td>
</tr>
<tr>
<td>Hydroxychloroquine</td>
<td>10%</td>
</tr>
</tbody>
</table>
Perspectives

Are swiss chILD guidelines needed?  63.6% NO
Proposed flow chart for investigating chILD

Bush, Thorax 2015;0
Proposed general diagnostic approach in infancy

A

- Specific Diagnosis Established
  - Persistent Disease or Disease Out of Proportion to Presumed Diagnosis
  - Normal or Other Specific Diagnosis

B

- Specific Diagnosis Established
  - Persistent Disease or Disease Out of Proportion to Presumed Diagnosis
  - Normal or Other Specific Diagnosis

CVHRCT

Genetic Testing

See Figure 4

NEGATIVE FAMILY HISTORY

- Specific diagnosis ruled out
- chILD Syndrome
  - NEGATIVE FAMILY HISTORY
  - Rapid Deterioration or Progression

NEGATIVE

- DIAGNOSTIC: Specific Diagnosis Established

LUNG BIOPSY WITH ELECTRON MICROSCOPY

ATS Clinical Practice Guidelines, AJRCCM 2013;188
Proposed general diagnostic approach in childhood

ATS Clinical Practice Guidelines,
AJRCCM 2013;188
Perspectives

Are swiss chILD guidelines needed?  63.6% NO

91% know about chILD-EU

63.6% have already included cases

91% willing to include their cases/ use chILD-EU SOP

70% in favour of having a SGPP representative in chILD-EU (3 potential volunteers!)
Conclusion

chiLD is a group of rare diseases
Patients are receiving specialised care
There seems to be a consensus regarding management

What’s next?

THANK YOU!