CASE PRESENTATIONS

UCSF CME: Interstitial Lung Disease
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Disclosures

• I have nothing to disclose
Multidisciplinary Panelists

- Brett Elicker, MD – Radiology
- Kirk D. Jones, MD – Pathology
- Christine Garcia, MD, PhD – Pulmonology, Genetics
- Brett Ley, MD – Pulmonology
- Rupal Shah, MD – Pulmonology
- Paul Wolters, MD – Pulmonology

Case 1
Case 1: HPI

• 69 year old man presenting with several months of a non-productive cough and dyspnea on exertion
• PMH: atrial fibrillation
• Medications: apixaban
• FH: No family history of ILD, cryptogenic cirrhosis, aplastic anemia/myelodysplastic syndrome/leukemia, or neonatal respiratory distress
• Exposures:
  • Never smoker
  • No occupational exposures
  • Sleeps with down comforter
  • Moldy smell from crawl space that communicates with bedroom

Case 1: Exam and Diagnostic Evaluation

• Exam: bibasilar inspiratory crackles, white hair
• PFTs:
  FVC 67%
  TLC 66%
  DLCO 35%
• Labs:
  • Extensive panel of autoimmune serologies – negative
  • HP panel – A fumigatus mildly positive
Case 1:
Chronic Hypersensitivity Pneumonitis

Case 2
Case 2: HPI

• 79 year old woman with a h/o sudden onset SOB
  • Symptoms improved on 60mg of prednisone followed by a 6 week taper.
  • Presenting 2 months later with recurrent symptoms → prednisone restarted
• PMH: Urethral strictures, q6 week dilations and frequent UTIs
• Medications: Prednisone, Gabapentin, Nitrofurantoin
• FH: Mother started graying in her 20s
• Exposures:
  • Retired RN
  • Former smoker, 36 pack-years
  • Down comforter and pillows
• ROS: joint stiffness, low back pain, neuropathy

Case 2: Exam and Diagnostic Evaluation

• Exam: Facial swelling, mild inspiratory crackles, Heberdens nodes, DIP deformities
• PFT Trend

![Graph showing PFT Trend]

• Labs: ANA 1:40 speckled, CK 36, RF and anti-CCP negative
Case 2:
Drug-Induced ILD

Case 3
Case 3: HPI

• 51 year old Mexican man presenting with several years of cough and frequent throat clearing
• PMH: allergic rhinitis, GERD, HTN
• Medications: loratadine, fluticasone nasal spray, pantoprazole, losartan
• FH: No family history of premature graying, cryptogenic cirrhosis, aplastic anemia/myelodysplastic syndrome, neonatal respiratory
• Exposures:
  • Works in a shipping warehouse, no known exposures
  • Former smoker, 5 pack year history
  • Denies exposure to down, birds, mold/mildew

Case 3: Family History

• Mother – pulmonary fibrosis, deceased at age 77
• Brother – pulmonary fibrosis, deceased at age 49
• Brother – pulmonary fibrosis, deceased at age 64
Case 3: Exam and Diagnostic Evaluation

- Exam: bibasilar inspiratory crackles
- PFTs:
  - FVC 75%
  - FEV1/FVC 86%
  - TLC 77%
  - DLCO 100%
- Labs:
  - Extensive panel of autoimmune serologies – negative
  - CBC normal, MCV 92
Case 3: Clinical Course

- Nintedanib initiated
- Referred to genetic counseling

<table>
<thead>
<tr>
<th>Gene</th>
<th>Variant</th>
<th>Zygosity</th>
<th>Variant Classification</th>
</tr>
</thead>
<tbody>
<tr>
<td>PARN</td>
<td>c.272A&gt;G (p.Tyr91Cys)</td>
<td>heterozygous</td>
<td>Uncertain Significance</td>
</tr>
</tbody>
</table>

The following genes were evaluated for sequence changes and exonic deletions/duplications:

DKC1, NHP2, NOP10, PARN, RTEI1, TERC, TERT, TINF2

Results are negative unless otherwise indicated

Benign, Likely Benign, and silent and intronic variants with no evidence towards pathogenicity are not included in this report but are available upon request.

Case 3:
Familial Pulmonary Fibrosis
Case 4

Case 4: HPI

- 50 year old woman presenting with 6 weeks of non-productive cough and progressive dyspnea on exertion, hospitalized for hypoxemic respiratory failure
- PMH/Medications: none
- FH: No family history of ILD or autoimmune disease
- Exposures:
  - Personal trainer
  - Never smoker
  - Denied exposure to mold, birds, down
- ROS: New pain, stiffness and swelling of hands
Case 4: Clinical Course

Onset of cough, dyspnea, hand swelling and stiffness

Hospitalized for acute respiratory failure. Dx: ARDS. Intubated. Proned. Antibiotics

No clinical improvement. Negative infectious workup, including BAL.

High dose IV corticosteroids started.

Extubated and discharged on steroid taper.

JULY AUGUST SEPTEMBER

Rehospitalized for hypoxic respiratory failure.

Case 4: Diagnostic Evaluation

- PFTs (after 2 months of corticosteroids)
  - FVC 55%
  - FEV1/FVC 89%
  - TLC 60%
  - DLCO 40%

- Serologies
  - ANA negative
  - Rheumatoid Factor negative
  - SSA weakly positive
  - SSB negative
  - ANCA negative
  - CCP negative
Case 4: Diagnostic Evaluation (continued)

- Additional Serologies
  - Scl 70 negative
  - CK 84
  - Aldolase 9.4 (<8.1)
  - Sm negative
  - RNP negative
  - SSA-Ro-60 negative
  - SSA-Ro-52 163 (<20)
  - Jo-1 5.8 (<1.0)
  - Additional negative studies: PL7, PL12, Mi2, Ku, EJ, OJ, SRP, PM-Scl
Case 4: Clinical Course (continued)

- Diagnosis: Antisynthetase syndrome/Inflammatory myositis-associated ILD
- Treatment: prednisone and mycophenolate → mycophenolate 3g qd
- PFT Trend

![Graphs showing FVC and DLCO trends over time.](image)