**New Adult Clinical Autoinflammatory Disease Case**

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**Case presentation**

- A 45-year old white F
- Recurrent erythematous rash 1 yr
- Rash lasted 3 to 7 days and disappeared for 1 to 2 days
- Photo: erythematous plaques and patches
Spongiotic dermatitis
Case, cont’d

- Recurrent morning swelling, pain, and weakness in the hands and feet, rarely forearms and knees
- Recurrent blurred vision
- 3 to 7 days/episode. Severe abdominal pain lasting about 2 hours AM
- She denied fevers.

Case, cont’d

- ROS: wt loss, night sweats, and dry mouth, IBS type.
- FH: negative
- Born in Wales and married with healthy 2 children.
- PE: skin changes, otherwise negative
**Labs**

- CBC, CMP, UA all wnl
- ESR, serology, CK and aldolase wnl
- EGD and Colonoscopy negative
- Ophthal exam: normal
- Lip minor salivary gland bx: negative

**DDX**

- FMF
- TRAPS
- Blau’s syndrome
- NAID
Genetic testing

- **NOD2** gene mutation: heterozygous IVS8+158, R702W

- TNFRSF1A for TRAPS: negative

**NOD2-associated autoinflammatory disease (NAID)**

- Newly reported disease

- White adult onset mostly

- Phenotype:
  - Periodic fever, dermatitis and polyarthritis
  - Features: Spongiotic dermatitis and pedal swelling

- Genotype: NOD2 variants: IVS8+158, R702W and R703C
### Characteristics of Cumulative Clinical and Laboratory Manifestations of NAID Patients

<table>
<thead>
<tr>
<th>Variable</th>
<th>Positive NOD2 patients (n=22)</th>
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</thead>
<tbody>
<tr>
<td>Gender (Female/Male)</td>
<td>13/9</td>
</tr>
<tr>
<td>Mean age at diagnosis (range, years)</td>
<td>40.1 (17-72)</td>
</tr>
<tr>
<td>Mean disease duration (range, years)</td>
<td>4.7 (1-13)</td>
</tr>
<tr>
<td>Ethnicity (White)</td>
<td>22/22 (100%)</td>
</tr>
<tr>
<td>Familial</td>
<td>3/22 (13.6%)</td>
</tr>
<tr>
<td>Weight loss</td>
<td>13/22 (59.1%)</td>
</tr>
<tr>
<td>Fever</td>
<td>13/22 (59.1%)</td>
</tr>
<tr>
<td>Skin disease</td>
<td>19/22 (86.4%)</td>
</tr>
<tr>
<td>Arthritis/arthralgia</td>
<td>20/22 (90.9%)</td>
</tr>
<tr>
<td>Gastrointestinal</td>
<td>13/22 (59.1%)</td>
</tr>
<tr>
<td>Serositis/chest pain</td>
<td>5/22 (22.7%)</td>
</tr>
<tr>
<td>Sicca-like</td>
<td>9/22 (40.9%)</td>
</tr>
<tr>
<td>Pulmonary</td>
<td>3/22 (13.6%)</td>
</tr>
<tr>
<td>Uveitis</td>
<td>0/22 (0%)</td>
</tr>
<tr>
<td>Raised ESR/CRP</td>
<td>9/20 (45.0%)</td>
</tr>
</tbody>
</table>

ESR, erytherocyte sedimentation rate; CRP, C-reactive protein
NOD2 gene
Pathogenesis of NAID

- Interaction: gene mutations+environment (GI stress, trauma)

- Cytokines: Th1/Th2 might not play a role. IL-17 and some type of T cells may contribute

- Under investigation
Therapy and Prognosis for NAID

Depends on signs and symptoms
- Topical steroids
- Small dose of prednisone
- Sulfasalazine
- Biologics

- Patients appear to run benign and intermittent course; some have nearly persistent polyarthritis, fatigue, rash, and fever
References


