Localized Ocular Amyloidosis: Case Series

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Financial Disclosures

- No financial disclosures
Localized Ocular Amyloidosis

- Background
- Four patient cases: A, B, C, D
- Workup of systemic amyloidosis (AL)
- Discussion
- Follow-ups
BACKGROUND:
Ig Light Chain Amyloidosis (AL)

- Clonal plasma cell neoplasm; clonal Ig light chains, either \( \lambda \) or \( \kappa \), misfold into amyloid and deposit in tissues
- Pathogenesis depends on the degree of systemic deposition into vital organs
- Rarely, amyloidosis is localized
Case: Patient A

Case: Patient A

- **Clinical:** 31yo F with left ptosis, watery eyes, a left inner eyelid lesion and a foreign body sensation (FBS) in her left eye

- **Physical exam:** left eye ptosis

- **Bx:** Congo red + staining on left upper lid with positive AL (lambda) type amyloid deposition
Case: Patient B

Case: Patient B

- **Clinical**: 49yo F with L proptosis, L FBS, limited eye movement and binocular horizontal diplopia
- **Past MRI**: enlargement of left EOM; thought to be secondary to orbital pseudotumor; tx with prednisone; no improvement
- **Physical exam**: unremarkable except for left eye proptosis, diplopia and enlarged EOM on left
- **Bx**: anterior orbitotomy with medial rectus; Congo red +; positive for AK (kappa) type amyloid
Case: Patient C

Case: Patient C

- **Clinical:** 69yo M with L ptosis for the past month with a change in vision
- **Physical exam:** unremarkable except for left upper lid ptosis with significant visual field changes
- **Bx:** L orbicularis muscle and full-thickness wedge left upper lid excision; immunohistochemistry suggestive of AA, weak staining for $\lambda$ and $\kappa$ was also present
Case: Patient D

Case: Patient D

- **Clinical**: 66yo M with new floaters/flashes in L eye with restricted R EOM
- **Physical exam**: unremarkable
- **Bx**: R inferior rectus mass; Congo red +; positive for AL (lambda) type amyloid
AL Workup

- Heart
  - NT-proBNP or BNP
  - Troponin

- Kidney
  - Serum creatinine
  - Serum albumin
  - 24-hour urine protein
  - Uric acid

- Liver
  - LDH
  - Alkaline phosphatase

- Plasma cell-specific
  - Serum immunofixation (Patient A and B only)
  - M spike
  - Serum kappa/lambda light chain
  - FISH study (for Patient B only)

AL Systemic Workup

- All four of our patients **within normal limits** with the following exception:
- **Patient B**: mildly elevated kappa levels in serum

<table>
<thead>
<tr>
<th>Patient</th>
<th>Age at Dx</th>
<th>Sex</th>
<th>Location</th>
<th>Amyloid Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>31</td>
<td>F</td>
<td>conjunctiva</td>
<td>AL lambda</td>
</tr>
<tr>
<td>B</td>
<td>49</td>
<td>F</td>
<td>L medial rectus</td>
<td>AL kappa</td>
</tr>
<tr>
<td>C</td>
<td>69</td>
<td>M</td>
<td>L orbicularis, upper eyelid</td>
<td>AA (no mass spec)</td>
</tr>
<tr>
<td>D</td>
<td>66</td>
<td>M</td>
<td>R inferior rectus</td>
<td>AL lambda</td>
</tr>
</tbody>
</table>
Discussion

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REVIEW

Ocular adnexal and orbital amyloidosis: a case series and literature review

Eduardo R. Mora-Horna · Rubí Rojas-Padilla · Vianhi G. López · Martín J. Guzmán · Ariel Ceriotto · Guillermo Salcedo
## Demographics

<table>
<thead>
<tr>
<th></th>
<th>Present study $N = 53$</th>
<th>Demirci et al. [25] $N = 50$</th>
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</thead>
<tbody>
<tr>
<td>Male/female</td>
<td>19/34</td>
<td>21/29</td>
</tr>
<tr>
<td>Mean age (years)</td>
<td>54.1</td>
<td>46.7</td>
</tr>
<tr>
<td>Bilateral involvement</td>
<td>14 (26.4 %)</td>
<td>19 (38 %)</td>
</tr>
<tr>
<td>Signs and symptoms</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mass</td>
<td>45 (84.9 %)</td>
<td>42 (84 %)</td>
</tr>
<tr>
<td>Ocular surface symptoms</td>
<td>25 (47.2 %)</td>
<td>–</td>
</tr>
<tr>
<td>Ptosis</td>
<td>16 (30.2 %)</td>
<td>15 (30 %)</td>
</tr>
<tr>
<td>Subconjunctival hemorrhage</td>
<td>9 (17 %)</td>
<td>16 (33 %)</td>
</tr>
<tr>
<td>Systemic amyloidosis</td>
<td>8 (15.1 %)</td>
<td>3 (6 %)</td>
</tr>
</tbody>
</table>

Table 2. Comparison of the main features of superficial amyloidosis
Clinical features

- **Conjunctival**
  - Eyelid edema
  - Papules
  - Yellow subconjunctival plaques
  - Eyelid ptosis

- **Superficial amyloidosis**
  - Alteration in lacrimal film
  - Superior palpebral conjunctiva/levator muscle involvement
  - Ptosis

- **Deep amyloidosis**
  - EOM, orbital fat, lacrimal gland
  - Limited movement
  - Proptosis
  - Ocular displacement
  - Ptosis
Limitations of the Study

- Not focused solely on AL (also AA)
- Some pts have underlying bone marrow malignancies that can complicate diagnosis
  - Multiple Myeloma
  - MALT lymphoma
  - Solitary osseous plasmacytoma
  - Waldenström macroglobulinemia
- One patient with systemic amyloidosis
Patient A: total resection of ocular lesions; close observation and follow-up

Patient B: offered systemic rituxumab or radiation therapy; refused treatment

Patient C: total resection of ocular lesions; close observation and follow-up

Patient D: close observation and follow-up
Take Home Points

- Patient with **unilateral ptosis**, consider systemic amyloidosis on differential diagnosis
- Patients with localized ocular amyloidosis in the conjunctiva and/or extraocular muscles **have very low risk** of developing systemic involvement
- A **watchful waiting** approach appears acceptable in the absence of symptoms
References


QUESTIONS?