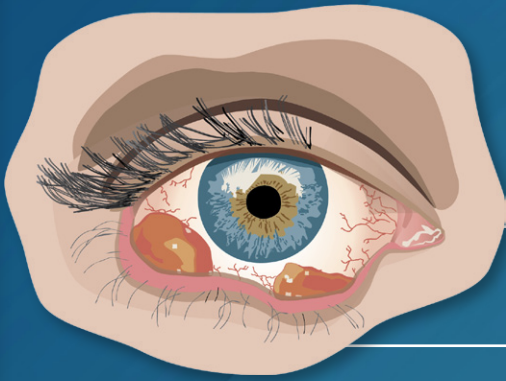




# PLASMINOGEN DEFICIENCY TYPE 1:

## The Patient Journey



**Contributing Faculty:** Charles Nakar, MD  
Angela Y. Zhu, MD, DABO

### WHAT IS PLGD-1?

Plasminogen deficiency type 1 (PDGD-1) is a condition in which the body does not produce enough of a coagulation factor called plasminogen. This disorder is characterized by low plasminogen activity and antigen levels that may cause an accumulation of fibrin material in the eyes, mouth, airways, gastrointestinal and genitourinary tracts, and other areas of the body.

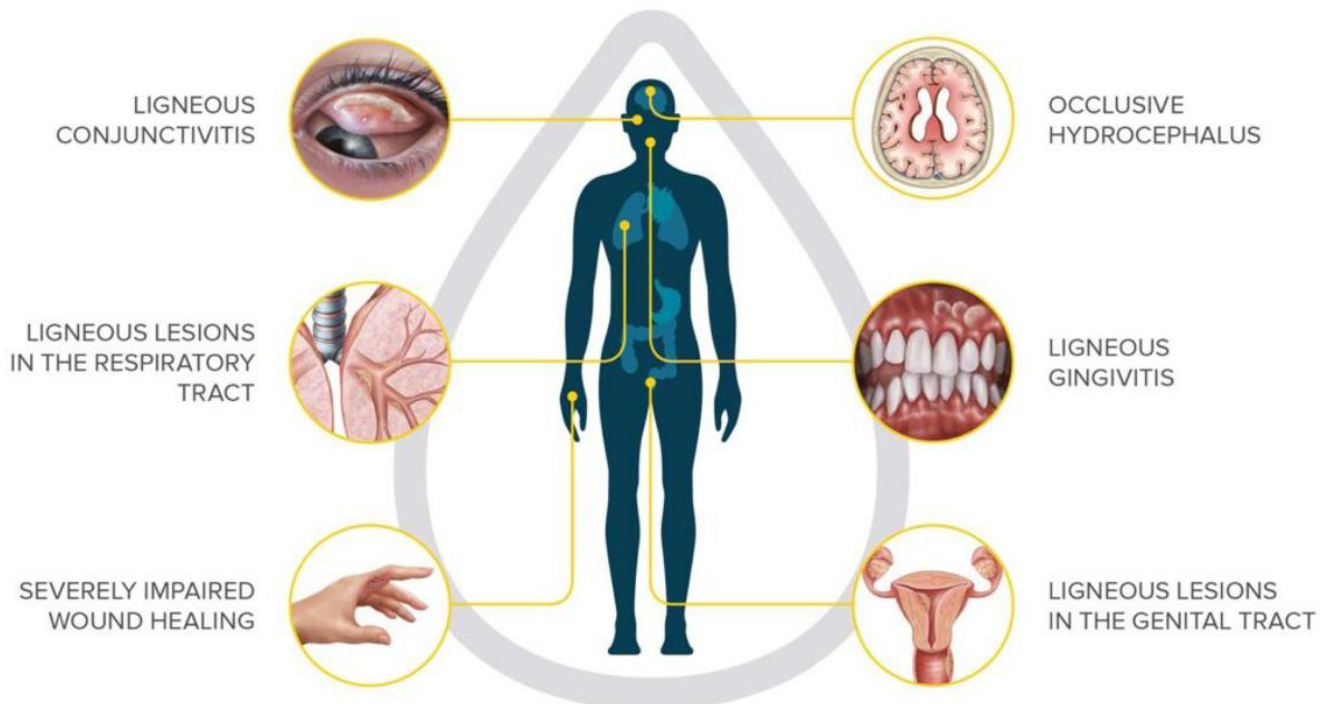
## Initial Presentation

- **Symptoms: chronic conjunctivitis, oral lesions, and respiratory signs**

- » Multisystem involvement
- » Eventual fibrin accumulation

- **First point of contact:**

- » Primary Care Provider or emergency department
- » Ophthalmologist
- » Dentist



# Historical Management

- **Initial therapies may include:**

- » Topical antibiotics and/or steroids
- » Surgical lesion removal (may lead to rapid regrowth)
- » Other symptomatic treatments

| Therapy                       | Systemic | Topical | Surgical |
|-------------------------------|----------|---------|----------|
| Fresh frozen plasma           | ✓        | ✓       |          |
| Plasminogen concentrate       | ✓        | ✓       |          |
| Anticoagulants                | ✓        | ✓       |          |
| Immunosuppressants            | ✓        | ✓       |          |
| Hormone therapy               | ✓        | ✓       |          |
| Oral contraceptives           | ✓        |         |          |
| Antihistamines                | ✓        | ✓       |          |
| Antibiotics                   | ✓        | ✓       |          |
| Thrombolytics                 |          | ✓       |          |
| Hyaluronidase                 |          | ✓       |          |
| Alpha chymotrypsin            |          | ✓       |          |
| Mast cell stabilizer          |          | ✓       |          |
| Membrane stripping            |          |         | ✓        |
| Amniotic membrane application |          |         | ✓        |

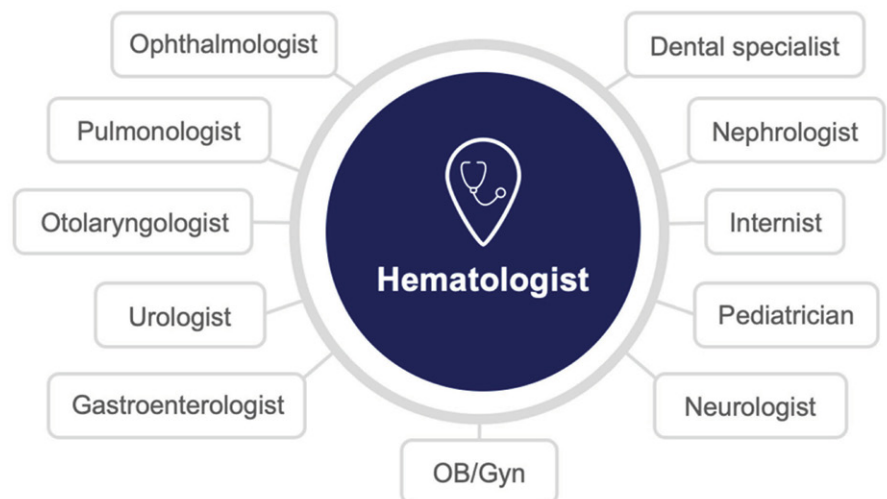
## Referral to Specialist

- **Triggers for referral:**

- » Recurrent or treatment-resistant conjunctivitis
- » Suspicious oral, respiratory, or mucosal lesions

- **Specialists involved:**

- » Hematologist (ultimately required for systemic treatment)
- » Ophthalmologist
- » Dentist/oral surgeon
- » Pulmonologist
- » Obstetrics and Gynecology
- » Others based on organ/system involvement



# Diagnosis

- **Clinical suspicion:**

- » Ligneous conjunctivitis (most common manifestation)
- » Recurrent lesions on mucosal surfaces

- **Common misdiagnoses:**

- » Bacterial/viral/allergic conjunctivitis, pyogenic granuloma

- **Diagnostic tests:**

- » **Labs:** Low plasminogen activity and antigen levels
- » **Pathology:** Fibrin-rich, hyaline-like pseudomembranes
- » **Genetic testing:** PLG gene mutation confirmation
- » **Imaging:** To detect systemic involvement

## Multidisciplinary Management Plan

- **Treatment planning by Hematology and Ophthalmology:**

- » IV plasminogen infusion (eg, plasminogen, human-tvmh)
  - ▷ Typical starting schedule: every 2-4 days
  - ▷ May taper to weekly or individualized dosing based on PLG activity levels
- » Surgical interventions if needed (eg, airway management, lesion excision)

- **Site of care:**

- » Infusions often begin in a supervised setting (eg, Hospital, Hemophilia Treatment Center)
- » Transition to home infusions as appropriate

## Follow-Up and Monitoring

- **Ongoing monitoring by:**

- » Hematologist (systemic disease and infusion therapy)
- » Subspecialists based on organ involvement

- **Frequency:**

- » Infusion schedule monitoring (eg, every 3-4 days)
- » Multidisciplinary team follow-up every 3-6 months

- **Key challenges:**

- » Venous access (especially in pediatric patients)
- » Managing potential side effects with initiation of therapy (tissue sloughing and bleeding)
- » Managing other potential side effects (allergy and antibody development)

# Long-Term Management Goals

Stabilization of lesions

Prevention of recurrence

Home therapy education and self-management

Quality-of-life improvement

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