

Plasminogen Deficiency, Type 1: For Primary Care



Overview of Plasminogen Deficiency (PLGD)

- PLGD Type 1: a quantitative protein deficiency, with decreased plasminogen activity and decreased antigen
 - Due to a genetic mutation in the gene for plasminogen; > 50 different mutations have been identified; Autosomal Recessive inheritance
 - Most common presenting symptom is ligneous conjunctivitis, but multi-organ, systemic disease that can be life-threatening
- PLGD Type 2: reduced functional activity of plasminogen, but normal antigen levels; patients are asymptomatic

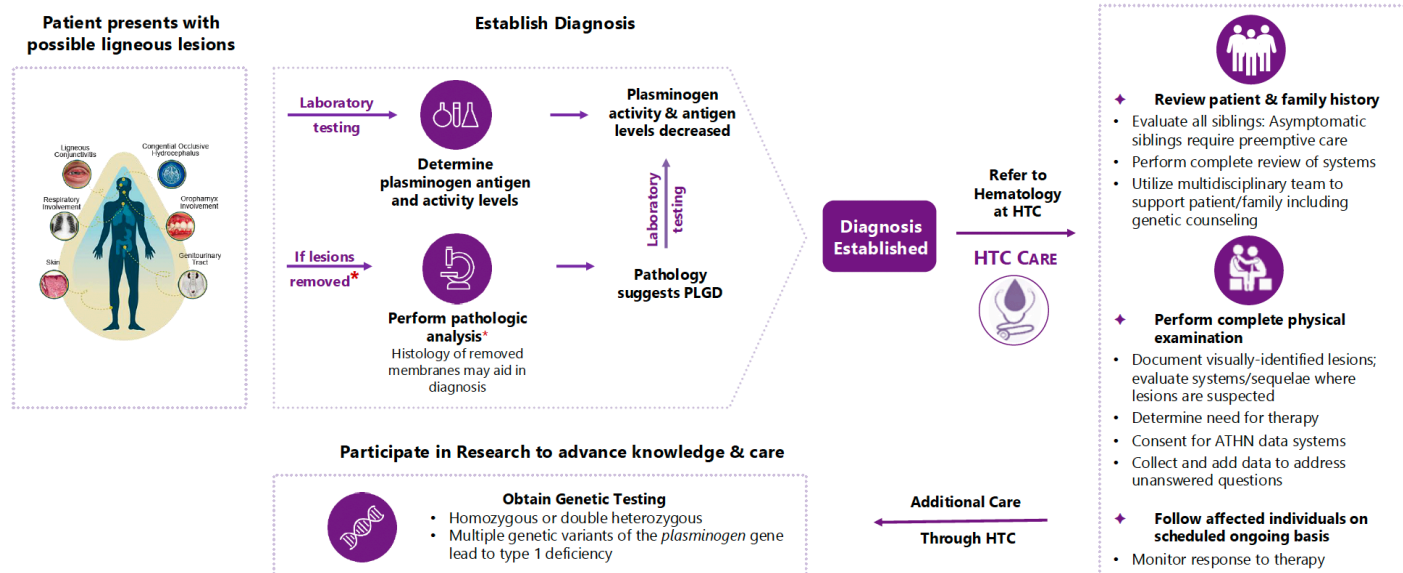
	Normal	PLGD Type 1	PLGD Type 2
Plasminogen Activity	70-130%	Decreased	Decreased
Plasminogen Antigen	6-25 mg/dL	Decreased	Normal
For Patients: My Plasminogen Activity			

Diagnosis

- Complicated by heterogeneous symptoms; symptoms can wax and wane
- Mucosal surfaces of the eyes, ears, nose, gums, airways, lungs, GI tract, kidneys, GU tract, CNS, and skin can all be affected
- Initial point of medical contact therefore includes many disciplines

Treatment

- Ryplazim (plasminogen, human-tvmh) given by IV infusion leads to resolution of lesions
- Surgical removal of lesions, though initially helpful, leads to accelerated regrowth
- Referral to a Hemophilia Treatment Center (HTC) to serve as medical home, and:
 - Educate on product use
 - Ongoing symptom monitoring, outcomes, safety
 - Administer doses, determine dosing schedule
 - Teach home infusion



*Lesion removal prior to diagnosis not recommended

Primary Care Specific Diagnostic Challenges

- ◆ Diagnosis complicated by
 - Rarity
 - Heterogenous symptomology → Initial point of medical contact includes many disciplines (e.g., ophthalmology, dental, ENT, primary care provider)
 - Waxing/waning symptoms
- ◆ Examples of symptoms



Ligneous conjunctivitis



Airway obstruction

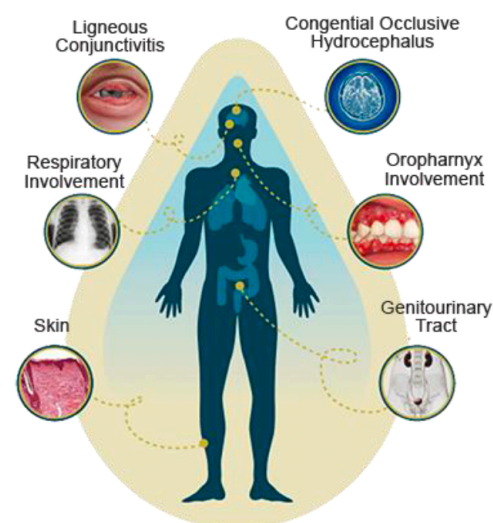


Photo on left: Schuster et al, *Survey of Ophthalmology*, vol 48, Ligneous conjunctivitis, p 369-88, 2003 [Used with permission from Elsevier]; Middle photo: Cohen et al, *Pediatric Pulmonology*, p 2, 2012 [Used with permission from John Wiley and Sons]; Image on right from <https://www.plgdeficiency.com/> Accessed 27 January 2023

Primary Care Specific Treatment Considerations

- Ligneous conjunctivitis is the most common manifestation of PLGD
- Ligneous gingivitis / periodontitis is the second most common manifestation of PLGD
- New or suspected diagnosis:
 - Obtain diagnostic blood test (plasminogen activity level) or refer to hematologist to order
 - If confirmed, refer to HTC to establish care and to perform thorough review of systems
 - Patients may have more than one system affected at presentation or occurring over time
- Confirmed diagnosis:
 - Coordinate clinical care and collaborate closely with existing care team at HTC
 - Send clinical notes and photos
- Be suspicious of common diagnoses in PLGD patients, as they may result from the condition:
 - Chronic hoarseness due to edema or lesions of the vocal cords
 - Ligneous conjunctivitis (often the first noticeable symptom of PLGD)
 - Nasal cavity lesions leading to irritation, epistaxis, and obstruction
 - Chronic sinusitis
 - Recurrent ear infections with frequent malfunction of myringotomy tubes
 - Cholesteatoma-like complications of the middle ear
 - Recurrent tonsillitis, with or without tonsil stones

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