A FATAL CASE OF TYPE I PLASMINOGEN DEFICIENCY (PLGD) IN AN INFANT WITH MULTIPLE AFFECTED ORGAN SYSTEMS

**Literature-Based Case Study: 1-month-old male child (plasminogen activity, 2.2%)**

This patient was diagnosed with Type 1 Plasminogen Deficiency (PLGD) when hospitalized at 28 days of age based on nonresponse of conjunctivitis to antibiotics, signs of hydrocephalus, and repeated vomiting. During hospitalization, respiratory distress with pseudomembranous obstruction of the bronchi and partial lung collapse occurred, leading to fatal respiratory failure 10 days following discharge. The patient was the only son of first cousin Turkish parents.

**AGE**

- **3 days**
  - Physical examination
    - Bilateral conjunctivitis
  - Initial diagnosis consideration
    - Gonococcal or chlamydial conjunctivitis
  - Treatment
    - Erythromycin, topical antibiotics, and corticosteroids

- **28 days**
  - Physical examination
    - Conjunctivitis (did not respond to the initial treatment), vomiting for 7 consecutive days, head circumference above the 97th percentile, bulging large anterior fontanelle, open posterior fontanelle and cranial sutures, both eyes were almost completely closed by ligneous pseudomembranes.
  - Lab test results
    - Prothrombin time: 12 sec, activated partial thromboplastin time: 27 sec, fibrinogen level: 263 mg/dl, D-dimer level <0.2 lg/ml, haemoglobin level: 11.5 g/dl, leukocyte count: 14,600/mm³, platelet count: 426,000/mm³, sedimentation rate: 76 mm/h, C-reactive protein level: 0.52 mg/dl
  - CT Scan: Enlargement of all four ventricles
  - Treatment (conjunctivitis)
    - Intravenous ceftriaxone and clarithromycin, topical ofloxacin and corticosteroids

- **35 days**
  - Clinical manifestations
    - Persistent vomiting and progressive hydrocephalus
  - Subsequent clinical manifestations
    - Inspiratory stridor, respiratory distress
  - Subsequent procedures
    - Ventriculo-peritoneal shunt
  - Chest X-ray
    - Atelectasis at the upper segment of the right lung

- **45 days**
  - Bronchoscopy
    - Tracheal pseudomembranes (pseudomembrane obstructing the bronchus of the upper segment of the right lung removed)
  - Chest X-ray (post-bronchoscopy)
    - No pulmonary atelectasis
  - Subsequent clinical manifestations
    - Occasional respiratory distress with one time expectoration of a thick, white, wood-like material followed by disappearance of respiratory distress
  - Other clinical manifestations
    - Progressive bilateral hydrocele, unilateral inguinal hernia requiring surgery
  - Mucosal biopsies (conjunctiva and trachea)
    - Massive exudation of fibrin with inflammatory cellular infiltration, disrupted epithelium, and areas containing amorphous, eosinophilic, hyaline material
  - Plasminogen lab results**
    - Plasminogen activity: 2.2%
    - Plasminogen antigen: <1.24 mg/dl
  - Diagnosis
    - Type I Plasminogen Deficiency (PLGD)
  - Genetic testing
    - Homozygous mutation in PLG gene. Both of his parents, who were asymptomatic, were heterozygous for this mutation.

- **3 months**
  - Recommended inpatient treatment (denied by the parents)
    - Systemic purified plasminogen replacement therapy
  - At parent’s request, the patient was discharged with topical corticosteroid treatment. He died from respiratory failure 10 days later.

**References:**


**KEY TAKEAWAYS**

- PLGD is an ultra-rare genetic disease that can have devastating effects on multiple organ systems.
- This patient developed ligneous conjunctivitis, respiratory distress and hydrocephalus, as well as bilateral progressive hydrocele and unilateral inguinal hernia.
- Authors note that various therapeutic approaches for ligneous conjunctivitis including hyaluronidase eye drops, corticosteroids, cyclosporine, and antiviral agents as well as surgical debridement of pseudomembranes yielded disappointing results in multiple cases.