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Clinical Course of Two Children with Congenital Plasminogen Deficiency Type 1

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Introduction: Type I plasminogen deficiency is an autosomal recessive hereditary disease. Severe forms of plasminogen deficiency are a rare disease characterized by chronic mucosal pseudomembranous lesions in mucosaes such as ligneous conjunctivitis, mouth, nasopharynx, tracheobronkoalveolar tree, and female genital tract. In this report, we evaluated the clinical course of two patients with type 1 congenital plasminogen deficiency.

Case 1: An 11-year-old girl was first admitted to our clinic with a wheezing and cough at the age of 35 months. First-degree cousin marriage, ligneous conjunctivitis, respiratory problems, presence of fibrin membranes during bronchoscopy, low levels of plasminogen activity and genetic analysis were diagnosed as Congenital Type 1 plasminogen deficiency. First shortness of breath, wheezing episode was 2 years old. The first hospitalization due to respiratory problems while it was 36 months. During her follow-up, she was hospitalized with pneumonia 10 times in our clinic and 2 times in the other centers. The most recent high-resolution lung tomography (HRCT) showed nodular areas on both lungs with diffuse ground glass density and calcific, polypolid lesions and secretions extending from the trachea to the inferior carina and continued until both bronchial levels.

Case 2: A 7-year-old boy patient was admitted to our clinic with cough at the age of 4 months. He had undergone ventriculoperitoneal shunting due to hydrocephaly when he was 24 days old. First-degree cousin marriage, ligneous conjunctivitis, respiratory problems, a 10-month-old brother who died due to the same disease history, low plasminogen activity and genetic analysis was diagnosed Congenital Type 1 plasminogen deficiency. The first hospitalization due to respiratory problems while it was 15 months. She was hospitalized in a pediatric intensive care unit for 2 times and 22 times to hospital inpatient services mainly because of respiratory problems. In the most recent HRCT, polypoid soft tissue densities were observed in the tracheal bronchial tree, including calcification. Atelectasis and bronchiectasis were more prominent in both lungs.

Conclusion: Congenital plasminogen deficiency is characterized by chronic mucosal pseudomembranous lesions consisting of subepithelial fibrin deposition and inflammation. It may cause mucosal involvement in the body and cause pseudomembrane formation and common clinical findings. Currently, sys-plasminogen eye-drops have been using to stop the progression of eye symptoms or even provides regression. Systemic plasminogen concentrate is likely to be used soon. It is possible that patients have hope for improvement of their systemic symptoms.

Keywords: Ligneous conjunctivitis, plasminogen deficiency, respiratory distress