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Title

Plasminogen deficiency: Case report and review

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Introduction

Plasminogen deficiency is a rare autosomal recessive disorder characterized by impaired fibrinolysis and fibrin accumulation. Ligneous conjunctivitis is a hallmark ocular manifestation of this deficiency. We present a case of plasminogen deficiency in a Saudi girl with ligneous conjunctivitis and occlusive hydrocephalus.

Methodolgy

We report a 26-year-old female with a devastating history that started when she was 1 month old with recurrent attacks of redness and tearing in her bilateral eyes, for which she was diagnosed as having conjunctivitis. Both local and systemic treatments failed to improve the recurrences. Following this inflammation, the family noted growing lesions on both eyes, and medical attention was sought. During her evaluation, membrane deposition on the palpebral conjunctivae was noted, and she was kept under observation.
Thereafter, in her 2nd month of life, she developed signs of increased intracranial pressure, and she started to have projectile vomiting and abnormal posturing. She was diagnosed with occlusive hydrocephalus and had a Ventriculoperitoneal Shunt (VP) performed. This became occluded, and she had a repeat of her VP shunting performed. The constellation of these symptoms dictated further evaluation, which included a comprehensive genetic history and laboratory evaluation. On further investigation, she was proven to be a homozygous carrier of plasminogen deficiency, which resulted in occlusive hydrocephalus as well as ligneous conjunctivitis.

Results

Plasminogen deficiency (PLG) is considered to be an ultra-rare, underdiagnosed autosomal recessive, multisystem disorder caused by mutations in the PLG gene resulting in 2 types, congenital type I plasminogen deficiency (hypoplasminogenemia) and congenital type II plasminogen deficiency (dysplasminogenemia). The disease prevalence is estimated to be around 1.6 per million and it exhibits a subtle female to male predominance. The spectrum of manifestation is based on the fact that plasminogen deficiency leads to the accumulation of the fibrin-rich matrix in various parts of the body leading to the classical presentation of Ligneous conjunctivitis, where the patient presents with chronic tearing, redness following the presence of pseudomembranes of “wood-like” pedunculated white, yellow-white, or red masses on the conjunctiva. The treatment of PLG deficiency is by restoration of the normal level by administering a human plasma-derived plasminogen; by replacing it, the body regains its fibrinolysis ability and resolution of the formed ligneous lesions ensues in the affected parts with regaining of the normal function. In 2021 a breakthrough in the management of PLG deficiency was represented by approval of the Food and Drug administration of Purified plasminogen derived from human plasma administered every 2-4 days at a dose of 6.6 mg/kg of body weight.

Conclusion

Plasminogen deficiency is considered to be an ultra-rare underdiagnosed autosomal recessive, multisystem disorder caused by mutations in the PLG gene, resulting in hypoplasminogenemia or dysplasminogenemia. The spectrum of manifestation includes congenital hydrocephalus, ligneous conjunctivitis, ligneous gingivitis, ligneous cervicitis and infertility. Purified plasminogen derived from human plasma is approved for the treatment of PLG deficiency, other less optimal treatments includes surgical excision, oral estrogen, glucocorticoids, cyclosporine, and azathioprine; were used with only limited benefit.