PLG|D CONNECTIONS

HYDROCEPHALUS IN A CHILD WITH HISTORY OF LIGNEOUS CONJUNCTIVITIS

Literature-Based Case Study: 10-year-old female child (plasminogen activity, 41.5%)§

This child had a history of chronic ligneous conjunctivitis with multiple surgical interventions followed by pseudomembrane regrowth. Hydrocephalus was initially identified at 6 months of age and was thought to be nonprogressive by 3 years of age. Hospital admission at 10 years of age revealed serious systemic manifestations. The patient was the third child of first degree consanguineous parents.

AGE

6 months

Initial diagnostic
Hydrocephalus

3 years

Diagnostic
Nonprogressive hydrocephalus, shunt operation not necessary

Surgical interventions
Multiple excisions of the conjunctival pseudomembranous lesions followed by re-growth

10 years

Physical examination
Corneal opacity and leukocoria on the right eye, ligneous conjunctivitis of the left eye, proptosis, blepharitis, and ectropion on the left eye, ligneous gingivitis, macrocephaly (head circumference greater than 97th percentile), frontal bossing

Neurological examination
Conscious and cooperative, full range of eye movement

Lab test results
Prothrombin time: 14 sec, activated partial thromboplastin time: 26.2 sec, INR: 1.08, prothrombin time activity: 89%

Plasminogen level lab results**
Patient: 41.5%
Affected sister (gingival hypertrophy and eye infections without any complications): 41%
Mother: 147.6%
Father: 117.4%

Cranial magnetic resonance imaging
Cerebellar and vermian atrophy, dilatation of the third and lateral ventricals, diffuse cerebral parenchymal atrophy

Development and psychometric tests (Standford-Binet)
Mild degree mental retardation

Electromyography examination
Axonal lesion of peroneal branch of the left sciatic nerve with a few regeneration signs

Genetic testing
No PLG gene mutations but 3 homozygous polymorphisms were identified

Diagnostic inference
Type I Plasminogen Deficiency (PLGD)

Bronchoscopy examination
Mucosal irregularities of pharynx, trachea, larynx, and two main bronchi

*** Normal levels of plasminogen activity: 70-130%¥*

References:
§ Aslan et al. Neuropediatrics. 2005;36(2):108-111.
¥ Rare bleeding disorders. http://www.rarecoagulationdisorders.org/diseases/plasminogen-deficiency/laboratory-evaluation. Accessed April 21, 2017.

KEY TAKEAWAYS

- Hydrocephalus is a potentially life-threatening complication of Plasminogen Deficiency (PLGD).
- PLGD is an ultra-rare genetic disease that can have devastating effects on multiple organ systems.
- Authors recommend to test for PLGD in patients with hydrocephalus as obstruction of the ventriculoperitoneal shunts is possible when such condition is overlooked.