A Case of Abducens Nerve Palsy and Subretinal Fluid Related to Idiopathic Intracranial Hypertension in Androgen Insensitivity Syndrome

Chan Young Yoon, MD
Department of Ophthalmology, Chuncheon Sacred Heart Hospital, Hallym University Medical Center, Chuncheon-si, Gangwon-do, Republic of Korea

Proprietary interest or competing interest: None of the authors have financial disclosure.

Corresponding author:
Juha Lee, MD
Department of Ophthalmology, Chuncheon Sacred Heart Hospital, Hallym University Medical Center, Chuncheon-si, Gangwon-do, Republic of Korea
Tel: 82-33-240-5176; E-mail: muyi709@gmail.com
Dear Editor,

Idiopathic intracranial hypertension (IIH) is diagnosed when the intracranial pressure (ICP) is elevated without a secondary cause.[1] ICP is determined by the production and absorption of cerebrospinal fluid (CSF). IIH is much more common in women and obesity.

A 21-year-old Asian woman (weight, 85.0 kg; height, 163.0 cm; body mass index (BMI), 31.99 kg/m2; blood pressure, 120/87 mmHg) visited our clinic for horizontal diplopia, eye pain, and photopsia. She presented with a 1-month history of headache and pulsatile tinnitus. She had visited the gynecologist eight years ago with ambiguous genitalia and amenorrhea. Pelvic magnetic resonance imaging (MRI) revealed an absence of the uterus and both ovaries. Hormonal test showed elevated 3.6 ng/mL of serum testosterone (the normal range for adolescent female, 0.20–0.38 ng/mL), and karyotype analysis showed 46, XY. She was diagnosed with partial androgen insensitivity syndrome (AIS) and had started to take conjugated estrogen. The ophthalmic examination revealed best-corrected visual acuities of 20/20 and intraocular pressure of 19 mmHg for both eyes. Her pupil sizes were equal and reactive without relative afferent papillary defect. Color vision tested with the Hardy-Rand-Rittler test was normal in both eyes. The alternating prism cover test revealed 10 prism dipters (PDs) esotropia in the primary position, 6 PDs esotropia in the left gaze, and 12 PDs esotropia in the right gaze. Ductions and versions showed a limitation in abduction in the left eye of grade -2. Fundus photography and optical coherence tomography (OCT) revealed bilateral optic disc swelling (Fig. 1A, 1B). The Humphrey visual field analyzer (Carl Zeiss, Dublin, USA) revealed enlarged blind spots in both eyes (Fig. 1E, 1F).

At the neurology department, Brain MRI and venography result normal. Diagnostic lumbar puncture revealed an opening pressure of >50 cm H2O (normal range, 10–25 cm H2O), colorless, a glucose level of 74 mg/dL (normal range, 50–75 mg/dL), and a total protein level of 16 mg/dL (normal range, 5–45 mg/dL) without red or white blood cells. She received 400 mL of 20% intravenous mannitol and 500 mg of oral acetazolamide for seven days after being diagnosed with IIH without discontinuing estrogen treatment. After seven days, she complained of metamorphopsia. Macular OCT showed subtle subretinal fluid (SRF) and interdigitation zone disruption in her right eye (Fig. 1I). After one month of acetazolamide, SRF, diplopia and lateral gaze limitation disappeared. Papilledema in both eyes decreased after seven months (Figs. 1C, 1D), and visual field tests returned to normal (Figs. 1G, 1H) on the same dose. Her weight and BMI remained unchanged throughout treatment. Even one month Revised Manuscript (Highlighted) after discontinuing the acetazolamide, no change was observed, but subsequent follow-up was lost.

Males account for about 9% of IIH cases.[2] Cases related to sex hormones have been reported. An obese man
with prostate cancer diagnosed with IIH during androgen deprivation therapy has been reported.[2] Nguyen et al. reported six cases of IIH during cross-sex hormone therapy, including five cases of female-to-male transition treated with testosterone and one case of male-to-female transition treated with conjugated estrogen, indicating that estrogen or testosterone use may cause IIH.[1]

Some mechanisms have been proposed to cause IIH. Serum estradiol levels are significantly influenced by the aromatization of androstenedione to estrone by adipocytes, which may explain the association between IIH and obesity.[3] Elevated estrone levels have been detected in the CSF of patients with IIH.[3] It is hypothesized that estrone stimulates the choroid plexus secretory cells to produce more CSF.[3] Some chorioretinal changes account for an incidence of approximately 7% in IIH.[4] A review revealed that choroidal folds were the most common manifestation (67%), followed by choroidal neovascular membrane (15%), and peripapillary SRF (10%).[4] Peripapillary SRF occurs due to disruption of the intermediary tissue of Kuhnt, allowing fluid to migrate between the retina and the retinal pigment epithelium.[5] In our case, macular SRF might have been caused by the contiguous extension of peripapillary SRF into the macula.[5]

In summary, IIH was successfully treated with acetazolamide treatment in AIS patients. Thus, it is necessary to check not only obesity, but also the history of taking exogenous sex hormones when treating patients with IIH. Further experimental research will be needed to determine how sex hormones affect the development of IIH.

Conflicts of Interest:
None.

Acknowledgments:
None.

Financial support:
None.
REFERENCES


Figure Legend

Figure 1. A, B. Fundus photographs of the patient at presentation showing bilateral papilledema at the initial examination. C, D. Fundus photographs of the patient seven months from the initial visit. Papilledema was almost resolved. E, F. Humphrey visual field test of the patient at the initial visit demonstrating bilateral enlarged blind spots. G, H. Enlarged blind spots resolved in both eyes after seven months. I. Macular OCT of the patient revealed SRF in her right eye. J. Macula improved to the normal state after seven months.