Chronic intracranial hypertension in pathogenic *PTEN*: To shunt or not?

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Background

- Idiopathic intracranial hypertension also known as IIH is an uncommon neurological disorder in children.
- It is characterized by raised intracranial pressure (ICP) in the absence of a secondary cause such as hydrocephalus, brain parenchymal lesion, inflammatory process, vascular malformations, or central nervous system (CNS) infection [1].
- The diagnosis is made by an elevated opening pressure of cerebrospinal fluid or CSF in the absence of a secondary cause. Potential visual loss is the main concern.
- Management options includes conservative measurements such as weight loss, diuretics or surgical intervention in the form of CSF diversion or optic nerve sheath fenestration, depending on underlying cause and refractoriness of symptoms

Objective

- To report the case of a 3-year-old female with localization-related epilepsy , *PTEN* mutation, and idiopathic intracranial hypertension resulting in macrocephaly and sunsetting eyes
- When PTEN pathogenic variant is identified the literature suggests CSF diversion procedures are indicated. We present a PTEN associated IIH in an infant where medical therapy can be effective and suggest that such a conservative approach may be warranted.

Methods

- Neurological assessment and ophthalmological examination performed
- Diagnostic imaging with MRI to assess signs of intracranial pressure
- Lumbar puncture performed to measure intracranial pressure and analysis of CSF
- Acetazolamide administered to manage intracranial pressure and neurological symptoms
- Labs to monitor to metabolic acidosis

Results

- MRI revealed enlargement of the lateral ventricles and prominent fluid along the optic nerve sheath bilaterally
- Lumbar puncture showed an elevated opening pressure of 32 cm H20 , while CSF analysis was unremarkable.
- Following acetazolamide treatment, the patient experienced resolution of symptoms

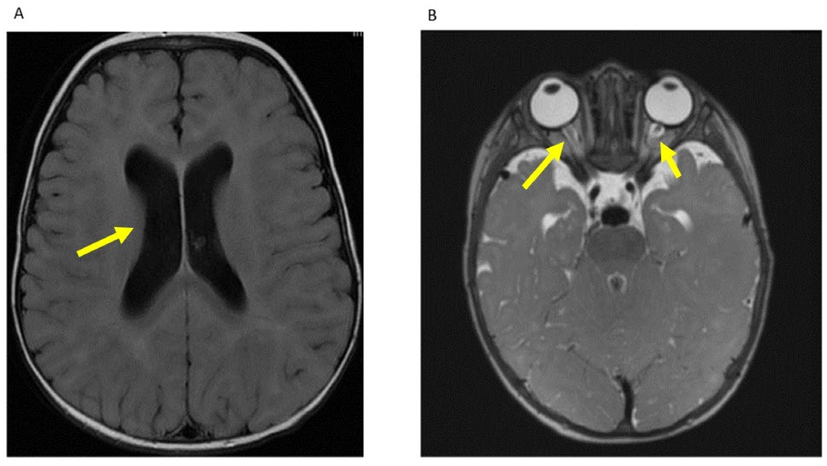

Figure 1. MRI brain and orbits with and without gadolinium contrast at 9 months of age. A. Axial cut, T1 sequence showing enlargement of the lateral ventricles. B. Axial cut, T2 sequence showing prominent fluid along the optic nerve sheath bilaterally (yellow arrows). No post enhancement noted (image not shown).

Discussion

- Idiopathic intracranial hypertension (IIH) is uncommon in infants and is almost always due to a choroid plexus papilloma or a result of genetic anomalies.
- IIH is potentially underrecognized in PTEN pathogenic variants
- Potential PTEN related pathophysiological mechanisms includes vasodilation by eNOS, increased CSF production, decreased CSF absorption in relation to PTEN loss and enhancement of PI3K/AKT signaling pathway.

Conclusions

- This case underscores the association between *PTEN* mutation, and symptomatic idiopathic intracranial hypertension
- Management of chronic IIH should be approached case by case
- Chronic causes of IIH may not always require surgical intervention such as in our case

References

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