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Papilledema in Facio-Audio-Symphalangism Syndrome: Rare Presentation of Rare Syndrome

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Abstract | **Case Report**

In this case report, a 10-year-old girl known to have speech delay, abnormal behavior, mental sub-normality comes with headache, photophobia and decrease in vision.

Keywords: Papilledema, headache, visual acuity loss, photophobia, facio-audio-symphalangism syndrome (FASS).

INTRODUCTION

- Papilledema is defined as optic disc swelling secondary to high intracranial pressure [1]
- The occurrence of papilledema is rare in pediatric patients and varies depending on the underlying etiology [1]
- This is the first reported case globally of facio-audio-symphalangism syndrome presenting with papilledema.

CASE PRESENTATION

This is a 10 years old female child, known to have speech delay, abnormal behavior, mental sub-normality following with multiple clinics. She was firstly seen in neurosurgery clinic on March 2024 due to CT brain finding of incidental Bilateral optic calcification involving optic disc.

With this CT brain finding, patient was referred to ophthalmology and was diagnosed with papilledema grade 1. During her follow ups with ophthalmology, she was diagnosed on Dec 2024 with papilledema grade 2 and became symptomatic with decrease in visual acuity and headache. Patient was referred to neurology clinic due to symptomatic papilledema in form of 3 weeks history of on/off occasional headache associated with photophobia, decrease in vision acuity and bilateral decrease of hearing.

On examinations patient looked failure to thrive, with dysmorphic features in form of (Frontal bossing, large long ears, small triangular face, hemicylindrical nose, short philtrum, small chin) on hearing aid with speech delay, and weight below 3rd centile. Vital signs all were within normal, central nervous system examinations revealed Mild hypotonia with scoliosis and restriction of neck extension, normal cranial nerve examination. Visual acuity examination with Correction was 20/50 and 20/70, with no relative Afferent Pupillary Defect, intact full extraocular movement and other examinations unremarkable.

Whole exome sequence was sent for this child which revealed a heterozygous pathogenic variant identified in the NOG gene consistent with the genetic diagnosis of autosomal dominant facio-audio-symphalangism syndrome (FASS).

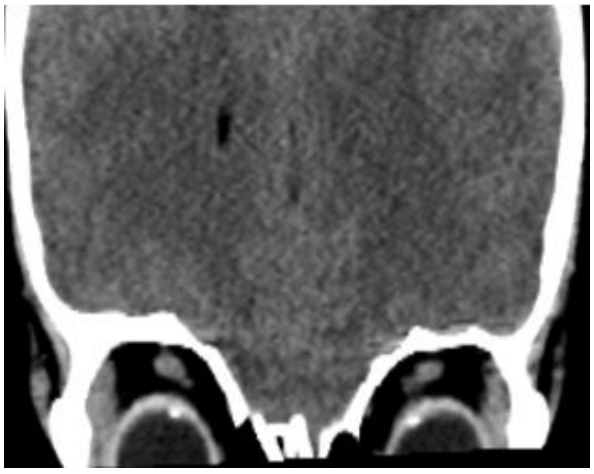

Figure A non-contrast coronal view CT brain

DISCUSSION

The facio-audio-symphalangism syndrome (FASS) was first described in 1985 by Hurvitz SA *et al.,*

It is a rare autosomal dominant syndrome affecting novel nonsense mutation in the NOG gene which encodes the noggin polypeptide, located on chromosome 17q22.

It contributes in skeletal development by binding and inhibiting BMP4 (Bone Morphogenetic Proteins 4). [3]

During embryonic development, bone Morphogenetic Proteins have an important function in pattern formation and tissue specification.

In this syndrome the anomaly arise around 8th week of gestation, during differentiation of the interzones of finger joints, prior to joint cavity formation.

As development progresses, vascularization of the surrounding connective tissue promotes the formation of synovial cavities, which separate the cartilaginous surfaces of the adjacent bones.

Mutations in the NOG gene during this stage may disrupt normal joint development, potentially leading to progressive joint fusion over time [3].

The facio-audio-symphalangism syndrome (FASS) characterized by facial dysmorphism with a broad hemicylindrical nose, lack of alar flare, associated with proximal symphalangism of the fingers, carpal and tarsal fusion and congenital conduction hearing loss [2,3].

Symphalangism has been reported with a numbers of other syndromes such as Apert syndrome, Pfeiffer syndrome, diastrophic dwarfism and metatrophic dysplasia [3].

Previous literature has not reported any association between facio-audio-symphalangism syndrome (FASS) and raised intracranial pressure or optic nerve involvement.

In our case, the patient presented with symptoms suggestive of intracranial hypertension and was diagnosed via fundoscopic examination with bilateral papilledema. Although papilledema most commonly associated with etiologies like idiopathic intracranial hypertension, mass lesions, or cerebral venous sinus thrombosis, these causes were ruled out in our case based on comprehensive neuroimaging and diagnostic work-up. [4]

This finding of papilledema raises the potential that it could be an unrecognized clinical feature of facio-audio-symphalangism syndrome (FASS), or possibly reflect an underlying pathophysiological process that has not yet been identified. To the best of our knowledge, this is the first reported case of facio-audio-symphalangism syndrome associated with papilledema, highlighting the need for further research and awareness of possible neurologic and ophthalmic manifestations in this rare condition.

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