CASE REPORT

A Rare Case of Idiopathic Intracranial Hypertension in Prepubertal Age

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ABSTRACT

A 2-year-old girl who was under genetic follow up for developmental delay and dysmorphism was accidentally found to have bilateral optic disc swelling during screening examination. She showed response to optokinetic drum examination and the anterior segment examination was unremarkable. Optic disc swellings were seen in both eyes. Lumbar puncture shows high opening pressure of 50 cm H2O with unremarkable CSF analysis. MRI of brain was done and showed features in keeping with mild cerebral atrophy, with no evidence of hydrocephalus or space occupying lesion. She was diagnosed with idiopathic intracranial hypertension and oral acetazolamide 125mg bd was commenced. However, papilloedema persist despite medical therapy. Ventriculoperitonel shunt was inserted to reduce the csf pressure. This case report highlights the importance of considering idiopathic intracranial hypertension as a cause of optic disc swelling in pre-pubertal children because delay in diagnosis and treatment may permanently affect visual function especially in children.

Keywords: Idiopathic intracranial hypertension, Paediatric, Pre-pubertal, Papilloedema, Disc swelling

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INTRODUCTION

We present a rare case of a 2-year-old girl with global development delay and dysmorphism, who developed idiopathic intracranial hypertension. To our knowledge and from literature search, such presentation is rare.

CASE REPORT

A 2-year-old girl with global developmental delay and dysmorphism, presented to us for a formal eye assessment. The parents did not notice any visual difficulties, but they noted that she needed to bring object closer to see for the past three months prior to seeing us. The child was otherwise well with no vomiting, behavioral change or body weakness. She was not on any medications. Her developmental milestones including fine motor were concurrent with a child eight to ten months old.

She was born at term by emergency lower segment caesarean section for fetal distress and breech presentation. Her birth weight was 2.55kg. She had an uneventful antenatal and postnatal history.

On general examination, she was noted to have tented upper lips, flat nasal bridge, anteverted nares, frontal bossing, large and low-set ears, hypotelorism, down slanting palpebral fissures and epicanthic folds. Metabolic screen /karyotype and DNA methylation for Prader-Willi Syndrome were negative. Based on Centers for Disease Control (CDC) chart, her height was less than 25th centile, weight less than 50th centile and occipitofrontal circumference more than 95th centile. Her Body Mass Index (BMI) was 13.71kg/m2.

On ocular examination, the child was orthophoric and anterior segment examination was normal. She responded to optokinetic drum indicating a reasonable vision. Cyclorefraction was +1.75DS OD and +1.50DS/-0.50 DC x1800 OS. No relative afferent pupillary defect was detected. Extraocular muscle movement was normal. Fundus examination showed bilateral established optic disc swelling with no macula star, cotton-wool spots or flame-shaped hemorrhages, no vitritis or vasculitis seen. (Fig. 1) Intraocular pressure was 13 mmHg OU. Other systemic examination including neurological examination were normal.

Blood investigation was normal for angiotensin converting enzyme, aquaporin 4, metabolic and infective screening. Lumbar puncture revealed elevated opening pressure at 50 cmH2O with unremarkable CSF analysis, ruling out sepsis and demyelinating disease. MRI of the brain showed features in keeping with mild cerebral atrophy, without any evidence of space occupying lesion, hydrocephalus or demyelinating

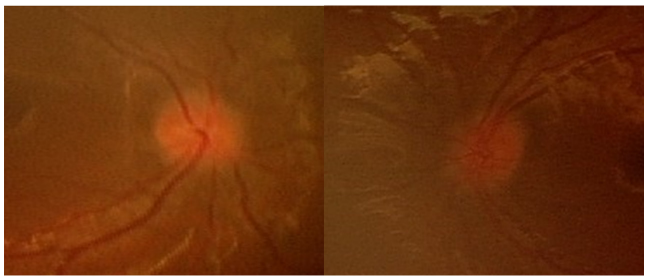

Figure 1: Fundus photo of OD and OS shows optic disc swelling at presentation

disease. The size of ventricles were normal.

A diagnosis of idiopathic intracranial hypertension was made and oral acetazolamide 125mg bd was commenced. Serial fundus examination and lumbar puncture for eight months after initial diagnosis showed persistent optic disc swelling and high intracranial pressure despite medical therapy. Investigations for sagittal sinus thrombosis and mastoiditis were negative. Medical therapy alone did not fully resolve the disc swelling and she demonstrated persistent high intracranial pressure as evident by an opening pressure of 29 cmH2O at lumbar puncture and incomplete resolution of disc swelling (Fig. 2 and 3). Ventriculoperitoneal shunt was inserted eight months after her first presentation.

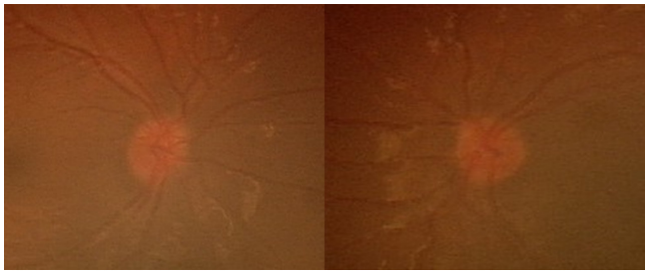

Figure 2: Optic disc of OD and OS at 3 months follow up showing improvement with of disc swelling with blurred margin at nasal part only

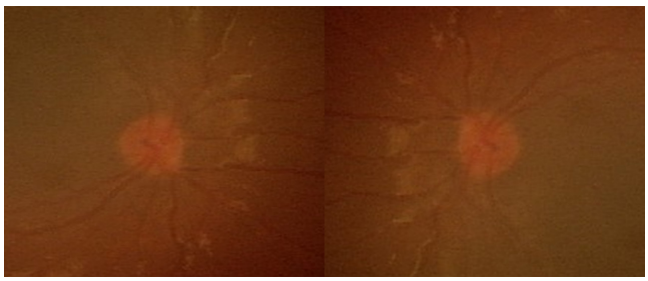

Figure 3: Optic disc at 6 months follow up prior to VP shunt insertion showing worsening of OD optic disc swelling and presistent blurred margin of the nasal area of OS

She continued to present with intermittent disc swelling of various degree despite being on acetazolamide and having a shunt. Patient was un-cooperative for vision assessment but according to the parents, her vision remained the same. The bilateral disc swellings did not fully resolve after 24 months of treatments. The revision of the shunt will be decided by the neurosurgeon if the intracranial pressure is not normalized as evident by persistent optic disc swelling.

DISCUSSION

Idiopathic intracranial hypertension (IIH) is a disease characterized by increased intracranial pressure with no identifiable cause (1) or with no clinical, radiological or laboratory evidence of intracranial pathology. Only a limited number of adult cases of IIH were reported in Asian countries. Prevalence of IIH is even lower in the pediatric population especially in pre-pubertal age. Recent study in United States estimated the incidence of primary and secondary intracranial hypertension of 0.63 and 0.32 per 100,000 children annually (2). The main effects of this condition when left untreated are progressive optic atrophy and blindness.

IIH in children is divided into prepubertal and pubertal groups. Studies found that obesity and female gender correlated with an increased risk of IIH in pubertal group and this is similar to adults IIH. Prepubertal patients on the other hand do not have any prominent risk factors. This is consistent with our case where our patient is at pre-pubertal age and was not obese. No genetic cause has been described, although familial cases have been reported.

Optic disc swelling can be an incidental finding during routine examinations as in our case and has become an important sign in younger children who have no symptoms. The commonest presentation in children is headache and is seen in up to 91% of cases. Children show more neurological signs and poorer visual prognosis if presented with no headache as they tend to present late (3). Other symptoms are diplopia, vomiting, neck stiffness and transient visual obscurations, irritability and ocular misalignment.

IIH is a diagnosis of exclusion. Because of the distinct clinical pictures compared to adult cases, Ko and Liu (4) proposed a special diagnostic criteria for IIH in pre-pubertal children. This criteria comprise of: 1) signs and symptoms of raised intracranial pressure (e. g. headache, vomiting, transient visual obscurations, or papilloedema),2) CSF opening pressure of more than 7.6 cmH2O in neonates and more than 28 cm H2O for 1-8 age years old.3)Normal CSF composition except in neonates who may have up to 19 WCC/mm3 (0-28 days old) and up to 9 WBC/mm3 (29 and 56 days old) and protein level may be as high as 150mg/dl, 4)No evidence of hydrocephalus, mass, structural or vascular lesion in MRI 5)Cranial nerve palsy is allowed 6)No other identifiable cause of intracranial hypertension.

The mechanism of raised intracranial pressure in IIH is not precisely known. An increased in cerebrospinal fluid production by the arachnoid villi or a decreased

absorption by the choroid plexus have generally believed to be the pathophysiology. Secondary causes are found in 53–77% of pediatric cases, however secondary causes for IIH are less frequently determined in adults. In this case, we did not find any secondary cause that could have contributed to the development of the condition.

Acetazolamide (carbonic anhydrase inhibitor) is the first-line agent in the treatment of IIH and acts by reducing CSF production. It is generally a safe medication and is typically used for 3 – 9 months until the resolution of the symptoms and disc swelling. Most pediatric cases of IIH resolve within 4.7 months of treatment, however up to 10% have permanent loss of visual acuity (5). Our patient failed to respond adequately to medical treatment and serial lumbar puncture and therefore requiring shunt insertion. We could not give her maximum dose of acetazolamide due to the difficulty in monitoring the side effects and this could explain why she did not show full resolution of disc swelling on medical treatment.

In a case of persistent high CSF pressure despite aggressive medical management, optic nerve fenestration (particularly if the vision is compromised) and shunting procedures are indicated. Ventriculoperitoneal shunt was inserted for our patient. VP shunts is shown to be effective in improving headache and vision in IIH patient (1).

CONCLUSION

This case report highlights the importance of considering idiopathic intracranial hypertension as a cause for optic disc swelling in pre-pubertal children. Managing paediatric IIH can proof to be challenging and delay in diagnosing and treating may permanently affect visual function especially in children.

AKNOWLEDGEMENT

We thank the patient's parent for their permission to publish this case.

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