CASE REPORT

Case Report of A Young Girl with Persistent Hyperplastic Primary Vitreous and Psychosis

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ABSTRACT

Childhood- and adolescent-onset schizophrenia are rare. Along with positive, negative, affective and cognitive symptoms, eye-tracking dysfunction characterized by a disturbance in the smooth pursuit system has been the only associated visual impairment found in schizophrenia. This case report highlights a case of a child with persistent hyperplastic primary vitreous who had gradual onset of psychosis over two years duration. A final diagnosis of schizophrenia was considered and she was started on antipsychotics, to which she showed gradual response with mild extrapyramidal side effects.

Keywords: Schizophrenia, Psychotic disorders, Blindness, Child psychiatry

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INTRODUCTION

Early-onset schizophrenia (EOS) and very early-onset schizophrenia (VEOS) are uncommon but received great attention because they generally represent a more severe form of the illness (1) and are associated with more brain abnormalities and genetic risk factors (2). With developmental delay and poor academic performance, children with these two conditions are frequently misdiagnosed. Persistent hyperplastic primary vitreous, or also known as persistent foetal vasculature is a rare congenital eye abnormality which ccurred when the primary vitreous and hyaloid vasculature failed to regress. The aim of this case report is to highlight a rare case where both psychosis and visual defects exist in a young child.

CASE REPORT

A 12 year-old girl presented with persecutory delusion and became socially withdrawn for one year. She had visual hallucinations of dragons and dogs chasing her, and auditory hallucination as evidenced by hearing invisible voices threatening to kill her. She believed that she was controlled by ghosts that entered her body, and that that her parents were imposters trying to harm her. A month after the onset of psychosis, her parents sought treatment from a private psychiatrist who started an antipsychotic. She was well for a few months until the medication was stopped, and her symptoms gradually worsened a few weeks later.

As the symptoms worsened further, she was subsequently admitted to the psychiatric ward. In addition to the delusion, auditory and visual hallucinations, her speech was irrelevant and incoherent. She was irritable and aggressive towards her parents and herself. There were disorganized behaviours; pouring shampoo into the bath tub and playing with water in the bathroom. Her daily functioning was significantly impaired as evidenced by her poor self-care and to failure to attend school. Apart from the bilateral congenital partial blindness, she had no significant medical or surgical history. There was no significant family history of similar illness. Her developmental milestones were normal. She attended special school for the blind and her academic achievement was average.

On admission, previous medications from i.e. methylphenidate, escitalopram and bromazepam were stopped while risperidone 1 mg daily was switched to paliperidone 3 mg daily to reduce sedation. However, she responded poorly to paliperidone despite high dose at 12 mg ON for an optimal duration of 4 weeks. The medication was later changed to olanzapine 5 mg ON, after which she showed improvement. Mental state examination on admission revealed an uncooperative

young girl with poor rapport. Her speech was relevant and coherent but she screamed inappropriately during the interview. Her affect was restricted. There were visual hallucination, auditory hallucinations and persecutory delusions. She was orientated to time, place and person, but her attention and concentration were impaired. Her general knowledge was in keeping with her age. Mini mental state examination (MMSE) showed impaired orientation, attention and concentration, performing written command, writing sentence and copying pentagon. Considering that she was distracted by the hallucinatory voices and her visual impairment, the MMSE score at that time might not reflect her actual cognitive function.

Physical examination showed bilateral horizontal nystagmus with impaired visual acuity. There were no gross motor and sensory deficit but she was uncooperative for full cranial nerve examination and detailed eye examination. Her right and left visual acuity were 20/800 feet and 20/100 feet respectively, indicating seriously impaired vision. Eye examination under anaesthesia revealed hyaloid vasculature in posterior chamber that extended from the optic disc to the posterolateral aspect of the lens in both eyes. There were no cataract, glaucoma, and retinal detachment seen. MRI of brain and orbit showed persistent hyperplastic primary vitreous. There was no radiological signs of retinal detachment orbital mass (Fig. 1 & 2),

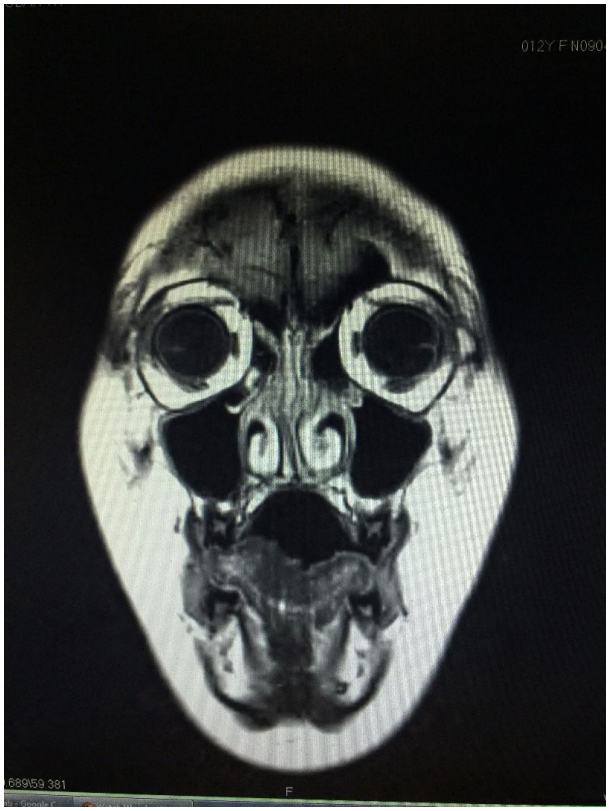

Figure 1: Coronal T1WI MRI orbit showing hyperintense signal of the fine linear structures in the posterior chamber of both orbits, extending from the head of the optic nerve posteriorly, to the posterolateral aspect of the lens anteriorly, which are suggestive of Cloquet's canal.

or brain parenchymal lesion. She was diagnosed with persistent hyperplastic primary vitreous and managed conservatively.

Upon discharge, her aggressive and disorganized behaviours resolved but she continued to have minimal psychotic symptoms. Apart from the ophthalmologist, she was also referred to a paediatric geneticist and neurologist for further assessment.

DISCUSSION

The presence of a congenital eye development problem along with psychosis in this girl had led to a few rare differential diagnoses including schizophrenia and Norrie disease. VEOS was among the differential diagnoses considered as the symptoms and course of her illness fulfilled the diagnostic criteria of schizophrenia outlined by the Fifth Diagnostic Statistical Manual (DSM-5) classification. Simultaneous presence of hallucinations, delusions and disorganized behaviour for more than a month fulfilled the criteria A of schizophrenia. Meanwhile, there was marked deterioration of function since the onset of her psychotic episode.

VEOS has been known to be associated with delay in motor, language and speech (1), social, and cognitive development, leading to misdiagnosis of intellectual disability, pervasive developmental disorder or global developmental delay. These delayed milestones characterize the premorbid period of schizophrenia, which is more pronounced in earlier onset of illness. In the case reported, there were academic limitations explained by her visual disability. Nevertheless, her academic decline paralleled to the cognitive pattern of VEOS in which the cognitive deficits are more severe than late-onset schizophrenia (2).

Psychotic Disorder due to Norrie disease (ND) was another differential diagnosis considered, as it is one of the causes of bilateral persistent hyperplastic primary

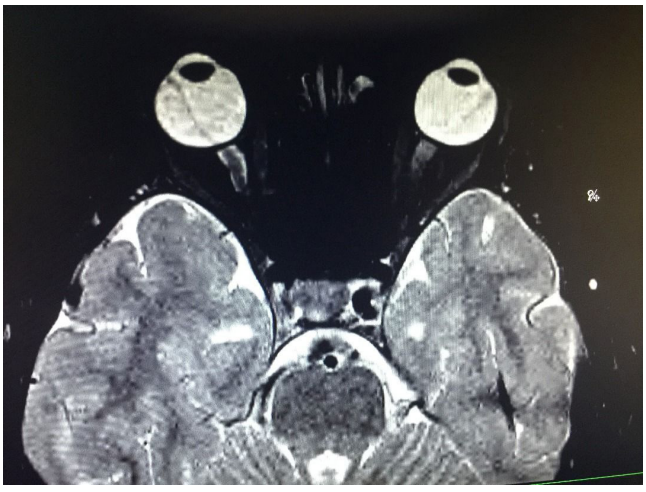

Figure 2: Axial T2WI MRI orbit showing hypointense signal of the structures in Figure 1

vitreous. This is a rare X-linked genetic disorder involving Norrie disease pseudoglioma (NDP) gene mutation which may occur spontaneously in an offspring of healthy parents causing abnormal retinal blood vessel formation. In NDP-related retinopathies, it has been reported that 30-50% of males do have developmental delay and psychotic-like feature (3). The child was referred to a paediatric geneticist, but ND was ruled out given the lack of clinical features and the absence of typical ophthalmic abnormality of pseudoglioma. Genetic testing was not able to be carried out due to lack of financial resource and NDP genetic testing facility. Another differential diagnosis considered was NMDA receptor encephalitis, which is a form of encephalitis characterized by psychotic symptoms such as delusion, auditory and visual hallucinations, behavioural change and seizures (4). However, there were no abnormal neurological findings, brain imaging or serological evidence of NMDA receptor antibodies to support this diagnosis.

The combination of psychosis with persistent hyperplastic primary vitreous in a young child is undeniably an extremely rare occurrence both in psychiatry and ophthalmology. Perhaps the most intriguing signs our case showed were blindness and persistent nystagmus, as the only visual problem that has been consistently documented in schizophrenia is the smooth pursuit eye movement dysfunction (5). VEOS is a rare occurrence and therefore its diagnosis should be made with caution. In this case, she fulfilled the symptom and duration criteria of VEOS, with significant impairment in her functioning. She was able to give history herself which was corroborated by her parents. The psychotic behaviour was also evident during the interview. Finally, the diagnosis of VEOS was made after excluding possible organic causes.

CONCLUSION

Psychotic features in children may present with a slightly different content due to their lower cognitive appraisal level but ultimately represent similar psychopathology as in adults. With such presentation and a high suspicion of EOS and VEOS, such children should further be assessed to exclude underlying organic causes. Nevertheless, it is acknowledged that the lack of confirmatory tests to rule out rare but possible diseases is a significant limitation in formulating a diagnosis.

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REFERENCES

1. Kyriakopoulos M, Frangou S. Pathophysiology of early onset schizophrenia. Int Rev Psychiatry. 2007 Aug; 19(4):315-24.
2. Rajji TK, Ismail Z, Mulsant BH. Age at onset and cognition in schizophrenia: metaanalysis. The British Journal of Psychiatry. Oct 2009. 195 (4) 286-293; DOI: 10.1192/bjp.bp.108.060723
3. Sims KB. NDP-Related Retinopathies. 1999 Jul 30 [Updated 2014 Sep 18]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1331/. Accessed January 18, 2018.
4. Helen Barry, Susan Byrne, Elizabeth Barret, Kieran C Murphy, David R Cotter. Anti-N-methyl-D-aspartate receptor encephalitis: review of clinical presentation, diagnosis and treatment. BJPsych Buletin 2015, 39, 19-23. Doi. 10.1192/pb.bp.113.045518.
5. Bolding M, Lahti A, White D, Moore C, Gurler D, Gawne TJ, Gamlin PD. Vergence Eye Movements in Patients with Schizophrenia. Vision research. 2014; 102:64-70. doi:10.1016/j.visres.2014.07.008.