Case Report

Situs inversus with atrial septal defect and pulmonary stenosis presenting as cortical blindness

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ABSTRACT

Cortical blindness is a rare form of blindness characterized by the normal anterior visual afferent system. Cerebrovascular diseases, tumor involving bilateral occipital lobe, cardiac surgery or cerebral angiography are common causes for cortical blindness. Some cases may be associated with denial of blindness when it is called Anton syndrome. We came across a 9-year-old male child presenting to us for evaluation of blindness which turned out to be cortical blindness after the examination. EEG and VEP were consistent with cortical blindness with MRI showing parieto-occipital lobe signal intensity alterations. Evaluation for the cause of cortical blindness led to the diagnosis of situs inversus in the patient with an atrial septal defect. The patient had an episode of dehydration due to gastroenteritis which resulted in vascular compromise thereby leading to hypoxic damage to the brain and cortical blindness. Presentation of cortical blindness in a patient of situs inversus is unique and hence being reported.

Keywords: *Anton syndrome, Atrial septal defect, Cortical blindness, Situs inversus.*

The heart or great vasculature related anomalies are observed in around 1% of live births. The important cause of childhood stroke is congenital cyanotic heart disease with a right to left shunt [1]. Central nervous system manifestations are commonly encountered in congenital cyanotic heart diseases.Children with cardiac disorders often harbour hemostatic abnormalities. It is more reported in congenital cyanotic heart diseases [2].

Cortical blindness is a rare form of blindness characterized by the normal anterior visual afferent system. Cerebrovascular disease is the most common cause of cortical blindness [3]. It can also be caused by tumor involving bilateral occipital lobe, cardiac surgery or cerebral angiography [4]. Some cases may be associated with denial of blindness when it is called Anton syndrome. We report a unique case of cortical blindness which after detailed evaluation turned out to be due to atrial septal defect, severe pulmonary stenosis, and situs inversus.

CASE REPORT

A 9-year-old male child presented to us with a complaint of bilateral loss of vision for the past 10 days. The patient had developed acute onset loose stools of around 10-12 episodes per day for 2 days after which the patient became drowsy and then lethargic and was admitted for diarrhea in a nearby hospital. On the day of admission, the patient had developed one episode of generalized tonic-clonic seizure for 1-2 minutes. After 2 days of treatment in the hospital for diarrhea and dehydration, the patient's condition improved and became fully conscious. As the patient regained consciousness, parents noticed that he was not able to see from both eyes. His blindness persisted despite improvement in all other symptoms.

He was referred to us for evaluation of blindness. When the patient presented to us, he had no complaints other than persistent blindness in both eyes. There was no history of any visual complaints prior to the onset of diarrhea. There was no history of similar illness in the past, any focal weakness, sensory loss or incontinence once the patient was fully conscious. Also, there was no history of any fever or any pain, redness, excessive tears or swelling in either of the eyes. There was no significant family history.

On examination, the patient was alert, conscious, oriented and well-nourished. His higher mental function and vitals were normal. In the cranial nerve examination, the patient had no perception of light in both eyes. However, the patient denied the fact that he is unable to see and was very eager to prove himself. He never said he cannot see and used to confabulate to the tests we did. Apart from the inability to see, bilateral pupils were of normal size and shape with normal reaction to light. There was no relative afferent pupillary defect and fundus examination was normal bilaterally. Other cranial nerves were also normal. Also, the motor and sensory system examinations were normal. On a cardiovascular examination, the apical impulse was in the right fifth intercostal place just lateral to the midclavicular line. First heart sound was widely split and fixed. Rest of the cardiovascular examination was normal with no cyanosis. Also, respiratory examination was normal with no organomegaly in per abdomen examination.

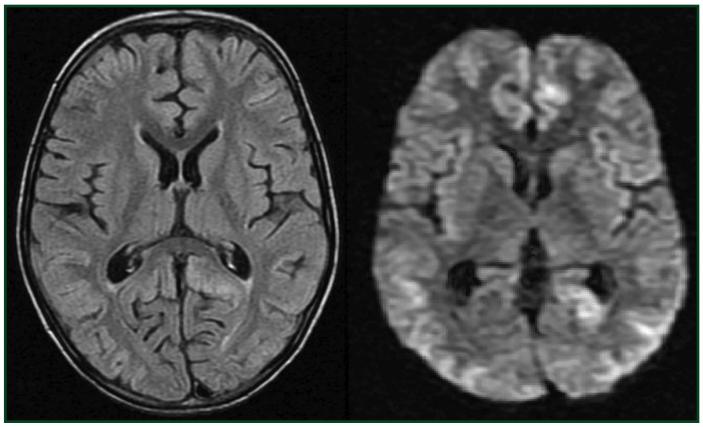

Figure 1: MRI showing gyral swelling with signal intensity alterations in cortical and subcortical region of bilateral parieto-occipital and frontal lobes with no gadolinium enhancement consistent with hypoxic brain damage.

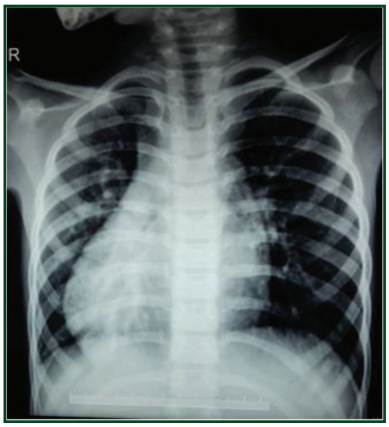

Figure 2: Chest radiograph showing dextrocardia.

The patient was approached as a case of cortical blindness with Anton syndrome with dextrocardia and possibility of the atrial septal defect. Routine blood investigation including complete blood count, liver function tests, renal function test, serum lactate, and ammoniawere within normal limits. Visual evoked potential (VEP) recordings were absent. Electroencephalogram (EEG) revealed the absence of alpha reactivity and lack of posterior predominance. Magnetic resonance imaging (MRI) of the brain was done to look for the cause of cortical blindness. MRI revealed gyral swelling with signal intensity alterations in the cortical and subcortical region of bilateral parieto-occipital and frontal lobes with no gadolinium enhancement consistent with hypoxic brain damage (Fig. 1).

Routine cerebrospinal fluid analysis along with viral markers was normal. Chest radiograph showed dextrocardia (Fig. 2). Cardiac evaluation (2D-ECHO) revealed situs inversus, dextrocardia, atrial septal defect with the bidirectional flow along with severe valvular pulmonary stenosis. Ultrasound and MRI abdomen also revealed situs inversus with no other abnormality (Fig. 3).

The cause of cortical blindness in this patient was due to hypoxia secondary to atrial septal defect and pulmonary stenosis. The episode of diarrhea led to dehydration and hypovolaemia. This hypovolaemia led to decompensation of atrial septal defect leading to impaired circulation and hypoxia to the brain thus resulting in cortical blindness. At the end of 4 weeks, the patient was able to count finger at one meter. The patient was referred to a cardiologist for further management for the atrial septal defect. He was started antiepileptics to control seizures and antioxidants. The patient regained complete acuity of vision at follow up after 2 months.

DISCUSSION

Cortical blindness results due to damage to bilateral occipital cortex simultaneously or sequentially with intact anterior visual pathway. When cortical blindness is associated with denial of blindness then it is called Anton's syndrome. Cortical blindness is one of the manifestations of cerebral forms of blindness that include homonymous hemianopia, blind sight, apperceptive or associative visual agnosia, visual neglect, prosopagnosia, simultagnosia, optic ataxia, palinopsia, etc [5].

Cortical blindness is usually diagnosed when there is loss of visual sensations in the absence of any ocular pathology, with preservation of the pupillary light reflexes, normal ocular movement, and normal fundus examination. The menace reflex although usually absent in cortical blindness, its presence does not rule it out. Visually evoked potentials are rarely normal in patients of cortical blindness and almost always associated with the absence of both posterior predominance and alpha reactivity in electroencephalogram [6,7]. The presence of associated neurological symptoms and signs helps in identifying the extent and cause of cortical blindness.

Ischemic stroke is the most common cause of cortical blindness. Tip of basilar artery strokes, sequential or simultaneous involvement of bilateral posterior cerebral artery, middle cerebral artery-posterior cerebral artery watershed infarcts can lead to these ischemic strokes. Apart from this cortical blindness can develop in the setting of hypoxic-ischemic encephalopathy, meningitis, systemic lupus erythematosus, posterior reversible leukoencephalopathy, polymorphic leukoencephalopathy, MELAS and dementing conditions like Heidenhain variant of Creutzfeldt-Jakob disease [8]. Cortical blindness can also occur as a transient phenomenon after traumatic brain injury, in migraine, as a complication of cerebral angiography or after epileptic seizures [9].

MRI of the brain can help in identifying the cause of cortical blindness and is the investigation of choice. The occipital cortex because of its relatively distal location from the central cerebral vasculature is sensitive to systemic hypoxia [9]. In patients with visual anosognosia or denial, lesion extends beyond striate cortex and into visual association areas [10]. The reason for denial can be due to false feedback to visual association area which is linked by a second visual system mediated by superior colliculus, pulvinar, and temporoparietal regions [10]. These areas receive inputs from the anterior visual pathway and hence, may be spared. In

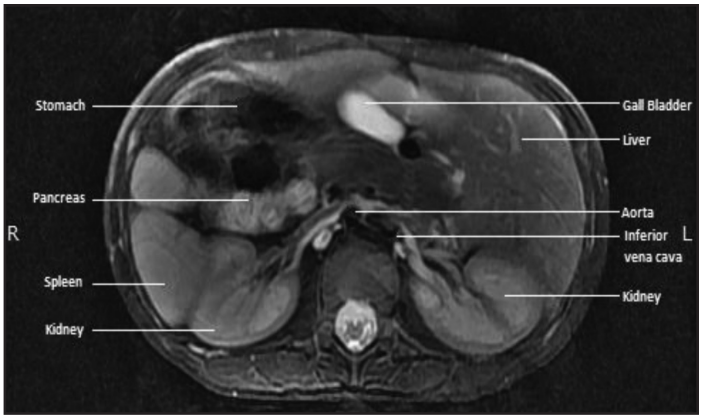

Figure 3: MRI abdomen showing evidence of situs inversus.

our patient, the cortical blindness was suspected based on clinical examination. Also, the presence of dextrocardia and wide fixed split first heart sound pointed toward the presence of some cardiac defect. 2D-ECHO, chest radiograph, ultrasound abdomen, and MRI abdomen revealed situs inversus with a large bidirectional atrial septal defect with severe valvular pulmonary stenosis.

The severe pulmonary stenosis leads to elevated risk for thrombosis. Multiple attacks of GE caused acidosis, depletion of volume, erythrocytosis, and hypoxia-induced activation of the prothrombotic system [11]. The secondary erythrocytosis caused by pulmonary stenosis may escalate blood viscosity; reduce cerebral blood flow, which makes the patient vulnerable to clot synthesis. Chronic hypoxemia also activates neutrophils and mononuclear cells that release vasoactive and chemotactic factors, resulting in endothelial injury [12]. This cardiac defect was largely asymptomatic but because of dehydration from loose stools, the defect was decompensated leading to increased shunt fraction and thereby hypoxia of cortical structures and thus leading to hypoxic-ischemic encephalopathy.

Platelets and endothelial cells interact and activated platelets increase thrombus formation by thrombin, which activates the coagulation cascade pathway. In addition, an impaired fibrinolytic system due to increased plasminogen activator-1 levels can contribute to thrombogenicity; thus, all these factors should also be taken under consideration for cerebrovascular accidents in such patients [13]. MRI brain showed gyral enhancement and signal intensity alteration in cortical and subcortical regions of parieto-oocipital and frontal regions as seen in hypoxic-ischemic encephalopathy. VEP and EEG were consistent with cortical blindness. The patient was referred to the cardiology department for management of cardiac defect. Also, parents were counseled to prevent any dehydration and to consult a doctor immediately in case of any acute illness to prevent further episodes of cortical ischemia. This is the first case of cortical blindness with Anton syndrome that occurred in a case of situs inversus. Our patient completely regained vision after two months.

CONCLUSION

Cortical blindness should be considered in a patient with a normal anterior visual pathway. Among various causes of cortical blindness, hypoxic-ischemic brain damage secondary to cyanotic heart disease can be a rare presentation. Detailed complete clinical evaluation is important to identify the possible etiology in the case of cortical blindness. The clinicians are advised to consider structural heart disease in childhood stroke. It will help in early diagnosis and management resulting in good clinical outcome.

REFERENCES

1. Roach ES, Golomb MR, Adams R, Biller J, Daniels S, Deveber G, *et al*. Management of stroke in infants and children: a scientific statement from a Special Writing Group of the American Heart Association Stroke Council and the Council on Cardiovascular Disease in the Young. Stroke. 2008;39:2644-91.
2. Odegard KC, Zurakowski D, DiNardo JA, Castro RA, McGowan FX Jr, Neufeld EJ, *et al*. Prospective longitudinal study of coagulation profiles in children with hypoplastic left heart syndrome from stage I through Fontan completion. J Thorac Cardiovasc Surg. 2009;137:934-41.
3. Reitsma DC, Mathis J, Ulmer JL, Mueller W, Maciejewski MJ, DeYoe EA. Atypical retinotopic organization of visual cortex in patients with central brain damage: congenital and adult onset. J Neurosci. 2013;33:13010-24.
4. Neurologic disorders caused by the lesions in specific parts of the cerebrum. In: Ropper AH, Samuels MA, Klein JP. Adams and Victor's Principles of Neurology. 10th ed. China: McGraw-Hill Education; c2014. p. 455-85.
5. Papageorgiou E, Hardiess G, Schaeffel F, Wiethoelter H, Karnath HO, Mallot H, *et al*. Assessment of vision-related quality of life in patients with homonymous visual field defects. Graefes Arch Clin Exp Ophthalmol. 2007;245:1749-58
6. Kwong Yew K, Abdul Halim S, Liza-Sharmini AT, Tharakan J. Recurrent bilateral occipital infarct with cortical blindness and anton syndrome. Case Rep Ophthalmol Med. 2014;795837.
7. Bharati S, Sharma MK, Chattopadhay A, Das D. Transient cortical blindness following intracardiac repair of congenital heart disease in an 11-year-old boy. Ann Card Anaesth. 2017;20:256-8.
8. Parmar HA, Trobe JD. Hypoxic-ischemic encephalopathy with clinical and imaging abnormalities limited to occipital lobe. J Neuro Ophthalmol. 2016;36:264-9.
9. Zwicker JC, Sila CA. MRI findings in a case of transient cortical blindness after cardiac catheterization. Catheter Cardiovasc Interv. 2002;57:47-9
10. Kondziella D, Frahm-Falkenberg S. Anton's syndrome and eugenics. J Clin Neurol. 2011;7:96-8.
11. Odegard KC, Zurakowski D, Hornykewycz S, DiNardo JA, Castro RA, Neufeld EJ, *et al*. Evaluation of the coagulation system in children with two-ventricle congenital heart disease. Ann Thorac Surg. 2007;83: 1797-803.
12. Tefferi A, Barbui T. Polycythemiavera and essential thrombocythemia: 2017 update on diagnosis, risk-stratification, and management. Am J Hematol. 2017;92:94-108.
13. Patnaik MM, Tefferi A. The complete evaluation of erythrocytosis: congenital and acquired. *Leukemia.* 2009;23:834-44.

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