BJMHR
British Journal of Medical and Health Research
Journal home page: www.bjmhr.com

A Rare Case Report: Colpocephaly As A Cause Of Status Epilepticus

Makwana Darshana, Dhami Param*, Dipenkumar Patel, Kanzariya Umesh
Department of Medicine, B J Medical College and Civil Hospital, Ahmedabad

ABSTRACT

Colpocephaly is a rare congenital brain malformation resulting in disproportionately large occipital horns of lateral ventricles compared to frontal ones resulting from corpus callosal and other white matter developmental arrest. Colpocephaly can be associated with partial or complete agenesis of the corpus callosum, chiari malformations, lissencephaly, and microcephaly. Colpocephaly can present with headache, seizure disorder, visual impairment, movement abnormalities and muscle spasms as well as behavioral and cognitive dysfunction or can even be asymptomatic.[1] We report a case of a 21-year old male with colpocephaly presenting as a status epilepticus (SE) as an initial symptom along with incidental cardiac anomaly OS ASD (Ostium secundum atrial septal defect) found on routine screening.

Keywords: Colpocephaly, Status Epilepticus, Ostium Secondum Atrial Septal Defect, Ventriculomegaly, Corpus callosal agenesis

*Corresponding Author Email: dhamiparam77@gmail.com
Received 12 January 2023, Accepted 13 February 2023

Please cite this article as: Param D *et al*., A Rare Case Report: Colpocephaly As A Cause Of Status Epilepticus . British Journal of Medical and Health Research 2023.

INTRODUCTION

Colpocephaly is a rare form of congenital ventriculomegaly often associated with partial or complete agenesis of the corpus callosum.[2] Diagnosis is usually made during infancy due to associated neurodevelopmental disorders.[3] Initial discovery in adulthood is exceedingly rare.[4,5] Other associated organ system involvement is not well documented in previous literature which can affect overall prognosis of a patient.

CASE REPORT:

A 21-year old intellectually disabled male patient presented with new onset episodes of generalized convulsions lasting 3-5 min associated with frothing from mouth, up rolling of eyeballs, involuntary passage of urine and no consciousness recovery in between episodes. He had neither apparent past medical history nor any documents of the same. These is no significant family history. He had no recent history of fever, convulsions, visual difficulty, headache, breathlessness, palpitation or syncope.

On appearance to our ER, he was quickly assessed for vitals and he was tachypnic and tachycardic. SpO2 and sugar levels were within normal limits. General physical examination was unremarkable.

He was immediately managed to secure his airway and treated with IV lorazepam and loading dose of valproate. He, then, was urgently evaluated with NCCT brain which had obvious ventriculomegaly with possibility of corpus callosal agenesis (Figure 1). On further monitoring at 1hr post-presentation in ICU, he was still unconscious with minor tremulousness over both hands so was empirically put on midazolam drip and bedside EEG was done which showed generalized seizure discharges, so diagnosis of non-convulsive status epilepticus (NCSE) was kept.

His routine lab investigations (including ionized calcium and magnesium) were within normal limits. Chest X-ray and ECG were normal. Ultrasound of abdomen and pelvis showed prominent intrahepatic IVC (measures 20cm). CSF routine and culture reports were unremarkable. After stabilization, further special workup including MRI Brain (P+C) was suggestive of corpus callosal agenesis in both occipital horns suggestive of colpocephaly with "Racing car " appearance in horizontal cut and "Viking helmet" appearance in coronal section and absence of interventricular septum, likely lobar type holoprosencephaly with P/A ratio>3 (mentioned below in discussion) (Figure 2,3,4); 2D echo showed large OS ASD with L->R (left to right) shunt, EF=60%, Moderate TR (tricuspid regurgitation) with severe PH (pulmonary hypertension) and dilated RA (right atrium) and RV (right ventricle); CSF autoimmune panel was negative; COVID and H_1N_1 RTPCR were negative; thyroid profile was normal. Serum TORCH profile was also negative.

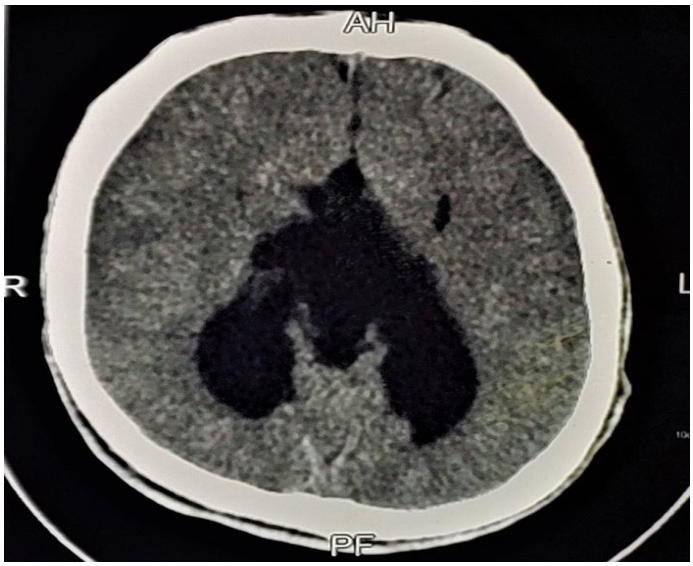

Figure 1: NCCT Brain showing dilated occipital horns of lateral ventricles

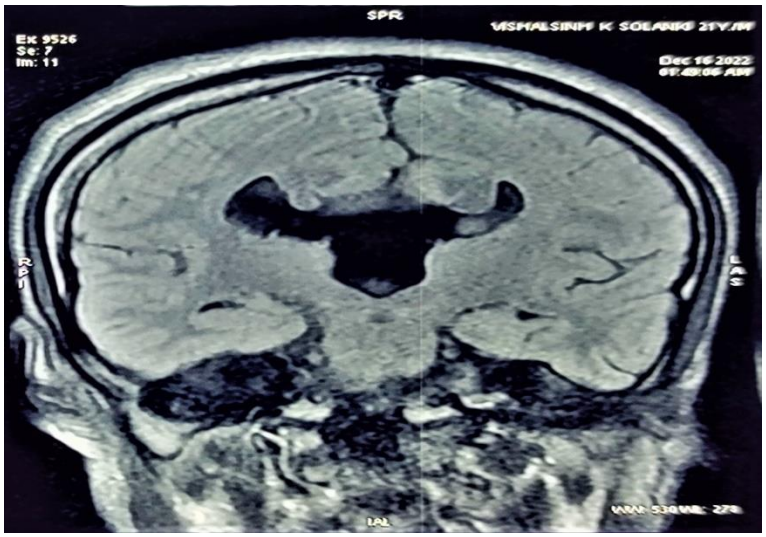

Figure 2: MRI Brain (FLAIR imaging) Coronal section showing absence of interventricular septum with "Viking helmet" appearance

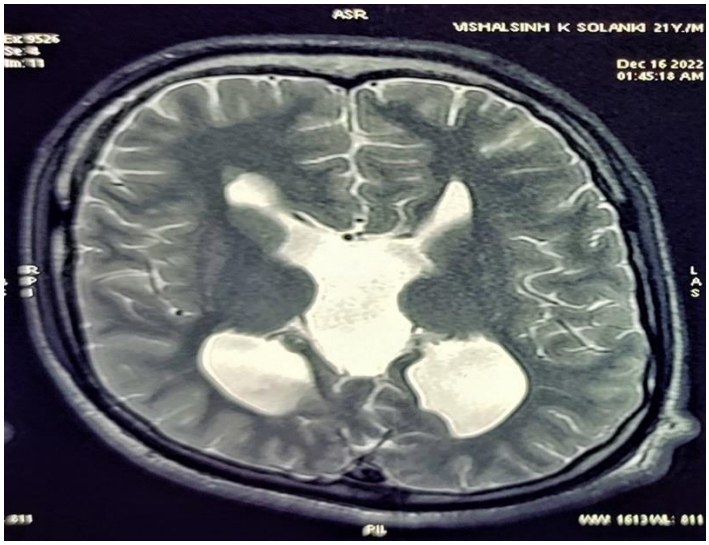

Figure 3: MRI Brain (T2W imaging) horizontal cut showing absence of interventricular septum and dilated occipital horns (P/A >3) with "Racing car " appearance

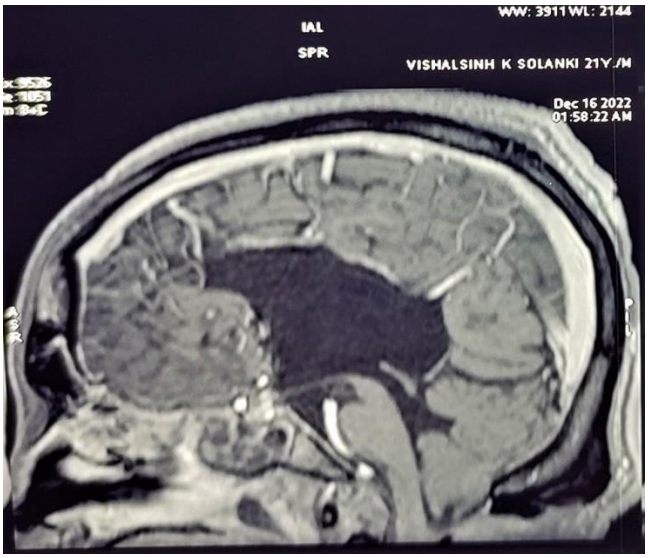

Figure 4: MRI Brain (T1W with contrast) sagittal section showing absent corpus callosum with high riding third ventricle

He had 2 more random episodes of focal convulsions after being off-midazolam drip each lasting ~2 min. After neurology consultancy, he was eventually put on four anti-epileptic drugs namely T. Valproate (500mg) TDS, T. Levetiracetam (500mg) TDS, T. Lacosamide (100mg) BD and T. Clobazam (10mg) OD.

Cardiologist opinion suggested to continue above same management with regular follow up in cardiology OPD. Patient was discharged after 2 days of convulsion-free monitoring.

DISCUSSION:

Colpocephaly is a term used to describe a congenital abnormal enlargement of the occipital horns of the lateral ventricles associated with normal frontal horns resulting from corpus callosal and other white matter suboptimal development. It can be associated with other neurological anomalies like partial or complete agenesis of the corpus callosum, chiari malformations, lissencephaly, micrognathia, microgyria, macrogyria, neurofibromatosis, myelomeningocele, cleft palate, microcephaly, cerebellar atrophy, optic nerve hypoplasia etc.[6,7] On review of literature, three adult colpocephaly cases have been described. Among them, two were previously normal subjects and one presented with psychiatric symptoms[8]. In general, some cases are thought to be X-linked or autosomal dominant inherited with incomplete penetrance.[9,10] Colpocephaly has been associated with trisomy 8 and trisomy 9,[11] Toxoplasma Gondii infection during pregnancy,[12] and maternal ingestion of ethanol, oral contraceptive medications, and other medications during pregnancy. Colpocephaly can present with headache, seizure, visual impairment, movement abnormalities, spasticity and moderate to severe intellectual disability or can even be asymptomatic up until found incidentally.[13] It is usually diagnosed by perinatal ultrasound in intrauterine life. After birth, radiologically, diagnosis of colpocephaly becomes more likely when the ratio of the posterior

horn to anterior horn of lateral ventricle width (P/A ratio) is ≥3. The P/A ratio along with non-obstructive ventriculomegaly and partial agenesis of corpus callosum helps to distinguish colpocephaly from normal pressure hydrocephalus, which is an important differential diagnosis.[14] Evans index and callosal angle can be used to differentiate between colpocephaly and normal pressure hydrocephalus. The Evans index shows the ratio of maximum width of the frontal horns of the lateral ventricles to the maximum internal diameter of the skull and a ratio of >3 indicates normal pressure hydrocephalus.[15,16] Colpocephaly is generally non-fatal though long term prognosis depends on other brain anomalies and other organ system involvement as it is present in this case which is an OS ASD(Ostium secundum atrial septal defect) with secondary PH and dilated RA and RV which ran asymptomatic all his life but later in life, prognosis depends on his underlying future cardiac status.

Colpocephaly is commonly diagnosed during infancy but can rarely be found in adults as a new onset convulsion. In addition to presenting feature, cardiac anomaly of OS ASD indicates towards its rare association. Colpocephaly usually remains asymptomatic in adult life but long term prognosis depends on other systemic involvement.

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