Case Report

Choroid Plexus Papilloma with Dandy Walker Variant: Co-existence or Association- Case Report

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Abstract

Choroid plexus tumors are rare intracranial tumors which account for 0.4-0.6% of all brain tumors. Choroid plexus tumors represent a spectrum of neoplasms derived from papillary epithelium of normal choroid plexus, including well-differentiated papilloma (WHO grade I), intermediate form as atypical Choroid Plexus Papilloma (WHO grade II) and highly aggressive choroid plexus carcinomas (WHO grade III). Though rare, it is responsible for the communicating hydrocephalus in children due to overproduction of cerebrospinal fluid. Due to advances in molecular biology and better understanding of the tumorigenesis of choroid plexus papilloma, now it is established that several genetic syndromes and central nervous sytem abnormalities are associated with this tumor. Here, we reported a case of a 10 months old child who presented with sudden deterioration of consciousness level and after thorough evaluation, diagnosed as a case of Choroid Plexus Papilloma with Dandy Walker Variant. Till date, this is the first reported case of the association/ co-existence of such two conditions which needs further evaluation.

Key words: Choroid plexus papilloma, lateral ventricle, trigone.

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INTRODUCTION

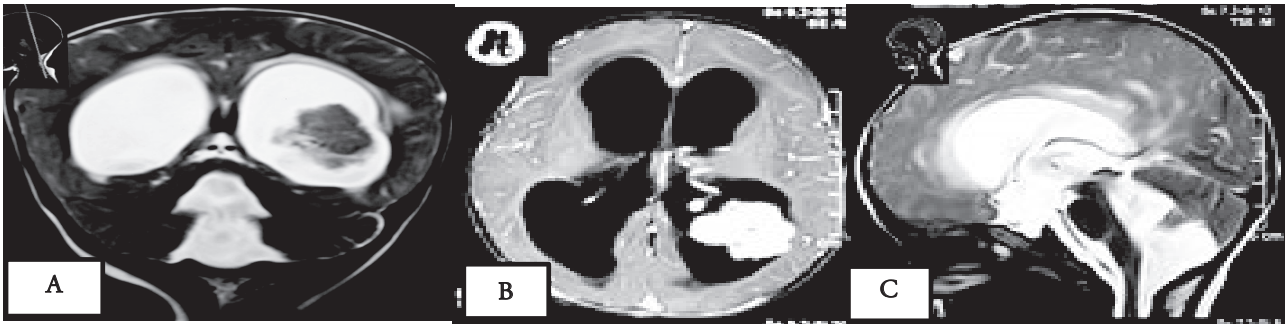
Choroid plexus papillomas (CPP) are rare, benign tumors representing 0.3%–0.6% of all intracranial tumors, with a male to female ratio of 1.2:11. Although generally found within the ventricular system, they can arise ectopically in the brain parenchyma or disseminate throughout the neuraxis. While generally considered benign, these tumors possess a complex biology, which is especially true for choroid plexus carcinomas (CPC). Although the majority of choroid plexus tumors are sporadic, there has been significant work to examine associations with specific genetic mutations[2-5]. In addition to Li-Fraumeni syndrome, other genetic disorders may play a role in the development ofchoroid plexus tumors, like Aicardi syndrome, Rhabdoid predisposition syndrome etc.[6] Although there was a reported case showing concomitant presence of choroid plexus hyperplasia and Dandy Walker Variant (DWV) along with other congenital malformations[7], till now our case is the first reported case showing choroid plexus papilloma with Dandy Walker Variant.

CASE REPORT

A 10 months old female child was brought to us with history of multiple episodes of vomiting followed by deterioration of the consciousness level within 3 days. She was immediately hospitalized and treated conservatively. Despite treatment, there was progressive deterioration of the consciousness level. There was no history of fever, convulsion and contact with TB patient.

On examination of the central nervous system (CNS), baby was drowsy and poorly responsive. Glasgow coma scale (GCS) was E2V2M4 (8 out of 15), pupils were bilaterally equal and sluggishly reacting to light. Plantar response was bilaterally extensor. Occipito-frontal circumference (OFC) was 50cm which was more than the size of an infant. There were no other congenital abnormalities on clinical examination. Our provisional diagnosis was hydrocephalus. The magnetic resonance imaging (MRI) of the brain revealed an intraventricular mass in the trigone of the left lateral ventricle attached by a pedicle, with features of communicating hydrocephalus. There was vermian hypoplasia and large posterior fossa cyst communicating with fourth ventricle [Figure 1].

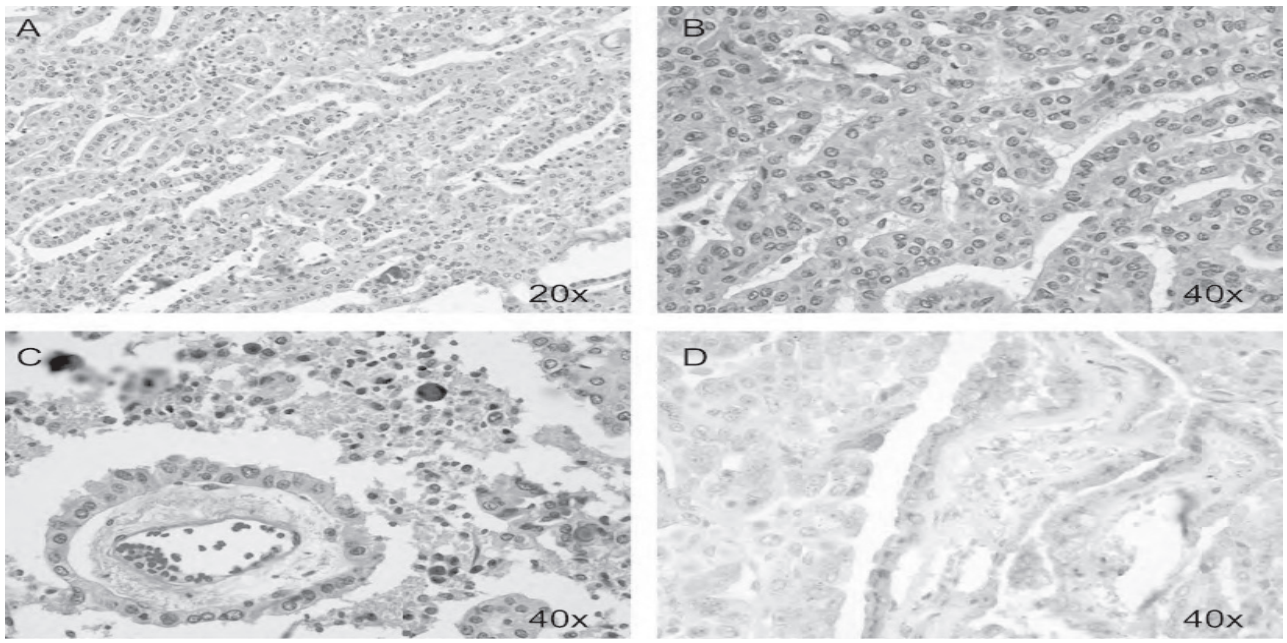
Figure 1 : MRI of brain with contrast – T2 weighted image coronal section showing irregular iso- to hypo-intense intraventricular lesion with vermian hypoplasia (A), communicating hydrocephalus (B), homogenously contrast enhancing lesion attached with a vascular pedicle (C).



Considering patient's condition, we did ventricular tapping through right Kocher's point immediately after admission. On the same day, she underwent right sided ventriculoperitoneal (VP) shunt surgery. At 1st post-operative day (POD), patient's GCS became E4V5M6 (15 out of 15). Her general condition was improved and vomiting was subsided. After 2 weeks, she was operated through left parieto-occipital craniotomy and removal of tumor through transcortical trans-ventricular approach. Corticotomywas done through posterior part of left superior parietal lobule. The highly vascular, lobulated, friable tumor was exposed; its pedicle was clamped with temporary aneurysm clip and cauterized. After that, the tumor was removedin piecemeal fashion. External ventricular drain (EVD) was placed and removed at 5th POD. Her post-operative period was uneventful.

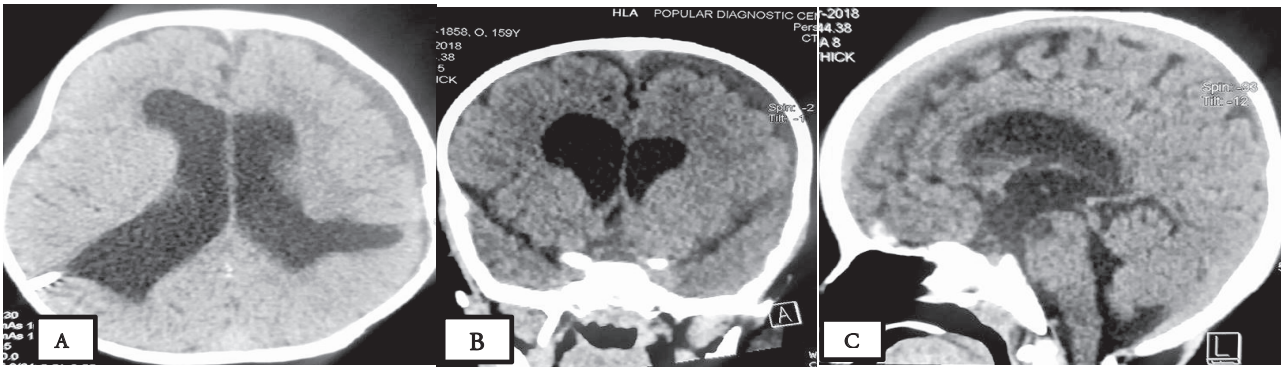
Specimen consists of multiple friable pieces of tissue measuring in between 0.5 cm and 0.3 cm. Histopathologic examination showed a benign papillary tumor composed of delicate fibrovascular connective tissue fronds, covered with single layer of cuboidal to columnar epithelium. No malignancy was seen. A diagnosis of choroid plexus papilloma was made. Immunohistochemistry was not performed in our case. Representative histopathologial photomicrographs were shown in Figure 2.

Figure 2: Representative hematoxylin/eosin and immunehistochemical staining of a CPP. These tumors feature a single layer of cuboidal or columnar epithelium in a papillary configuration (A and B) covering a fibrovascular core (C). The presence of transthyretin on immunohistochemical staining (D) can aid in confirming the diagnosis.



The patient was followed-up with a computed tomography (CT) scan 6 months after surgery which showed a decrease in the size of the ventricles, with normal appearing choroid plexus. There was no tumor residual [Figure 3]. All the signs and symptoms in the child were resolved in the 6 months follow-up.

Figure 3: Postoperative follow up CT scan of brain after 6 months of surgery showing no recurrence of tumor with diminished size of the ventricles (A, B, C).



DISCUSSION

CPPs are rare intracranial neoplasms with an estimated occurrence of 0.3%–0.6% of all intracranial tumors, with a male to female ratio of 1.2:11. According to the classification of WHO, these neoplasms ranges from well-differentiated papillomas (WHO grade I) to highly aggressive choroid plexus carcinomas (WHO grade III), with rare intermediate forms referred to as "atypical CPP". More than 80% of supratentorial CPPs present under20 years of age, while infratentorial CPPs are almost evenly distributed across all ages .The median ages at diagnosis for tumors in the lateral ventricle, third ventricle, fourth ventricle, and cerebellopontine angle are 1.5 years, 1.5 years, 22.5 years, and 35.5 years, respectively1. CPPs have also been detected in utero, suggesting a congenital origin for a subset of these tumors[8]. Our patient was diagnosed at the age of 10 months.

Symptoms are due to overproduction of cerebrospinal fluid (CSF) or obstruction of CSF outflow, depending on tumor location and size. Regardless of age, patients typically present with signs and symptoms of increased intracranial pressure including headache, visual disturbance, nausea, vomiting, seizures, hydrocephalus, papilloedema, cranial nerve deficits and gait impairment[9].

Choroid plexus tumors have a well-recognized grading system but their histologic appearances do not predict their behavior. Sometimes, CPPs can possess aggressive features simulating high-grade lesions. On the other hand, CPPs with evidence of parenchymal invasion or loss of normal architecture have been found to exhibit clinically benign behavior, with good long-term outcomes after surgical resection[10]. Cases of CSF dissemination along the neuraxis and malignant transformation are also reported[11].

Although the majority of choroid plexus tumors are sporadic, there has been a lot of works in an attempt to find associations with specific genetic mutations[2-5]. Li-Fraumeni syndrome, characterized by germline mutations of the p53 tumor suppressor, is a known predisposing factor for the development of choroid plexus tumors, particularly choroid plexus carcinomas[12-14]. Other genetic disorders may play a role in the development of choroid plexus tumors, like-Aicardi syndrome which is an X-linked disorder characterized by the triad of total or partial agenesis of the corpus callosum, chorioretinal "lacunae," and infantile spasms[15]. Though Wieselthaler et al. in 2002 reported a case showing concomitant presence of choroid plexus hyperplasia and Dandy Walker Variant along with other congenital malformations[7], our reported case showed choroid plexus papilloma in a patient with Dandy Walker Variant with features of communicating hydrocephalus.

Regarding molecular pathways associated with CPPs, several genes in the TRAIL pathway, along with E-cadherin, are known to be methylated in CPPs, resulting in decreased tumor necrosis factor– induced apoptosis and increased cell migration, respectively. Aberrant signaling through PDGFR and Notch3 are associated with tumor formation and growth. Mutations of TP53 and hSNF5/INI1 (SMARCB1), which plays an integral role in chromatin remodeling, have been shown to promote CPP formation, while methylation of TWIST1, an inhibitor of p53, and stratifin, a regulator of the G2 checkpoint, are also associated with CPPs[6].

The choice of treatment for CPPs is maximum safe surgical resection. Previous report proposed preoperative embolization as an adjunct to surgical resection but it is rarely successful due to the small caliber of feeding vessels[6]. Reports of chemotherapy use in patients with CPP are limited. The CPT-SIOP-2000 study by the International Society of Pediatric Oncology reported on the use of etoposide, vincristine, and either carboplatin or cyclophosphamide for the treatment of various choroid plexus tumors[7]. Regarding radiotherapy, most authors

agreed that radiation should be reserved for recurrent or malignant lesions, not residual tumor after resection, particularly given the indolent nature of most CPPs[18-22]. In our case, we did gross total resection of the tumor without any obvious per-operative complication and 6 months follow-up image showed no tumor recurrence.

CONCLUSION

Till date, this is the first reported case of the association/co-existence of choroid plexus papilloma with Dandy Walker variants which needs further evaluation.

ABBREVIATION:

CNS	: Central Nervous System
CPC	: Choroid Plexus Carcinoma
CPP	: Choroid Plexus Papilloma
CSF	: Cerebrospinal Fluid
CT	: Computed Tomography
DWV	: Dandy Walker Variant
EVD	: External Ventricular Drain
GCS	: Glasgow Coma Scale
MRI	: Magnetic Resonance Imaging
OFC	: Occipito-frontal circumference
POD	: Post-operative Day
VP	: Ventriculoperitoneal
WHO	: World Health Organization

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