

CASE REPORT

Extensive intracranial calcification of pseudo-TORCH syndrome with features of Dandy–Walker malformation

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ABSTRACT

Pseudo-TORCH syndrome or congenital infection-like syndrome is a group of conditions which resemble congenital infections such as those caused by toxoplasmosis, rubella, cytomegalovirus (CMV), herpes (TORCH) group of organisms, clinico-radiologically, but serological tests are negative for the organisms. One of the variety shows features such as microcephaly, extensive intracranial calcification showing gross resemblance to congenital CMV infection, making its other name as microcephaly intracranial calcification syndrome (MICS). Dandy–Walker malformation (DWM), in addition to posterior fossa large cyst, cerebellar vermis hypoplasia, and hydrocephalus is often associated with agenesis of the corpus callosum and callosal lipomas, dysplasia of the brainstem, and cerebellar hypoplasia or dysgenesis. But radiological features of DWM with microcephaly and intracranial calcification are very unusual and have been rarely reported in the literature.^[1] We report a case of infant showing clinical features suggestive of congenital CMV infection with negative serology and radiological imaging suggestive of DWM with extensive intracranial calcification. Pseudo-TORCH syndrome with radiological features of DWM is a congenital developmental abnormality. In spite of hydrocephalus, it does not require cerebrospinal fluid (CSF) diversionary procedure due to lack of increased intracranial pressure. Conservative management for seizure disorder is the optimal therapy.

Key words: Calcification, Dandy–Walker malformation, intracranial, pseudo-TORCH syndrome

Introduction

The etiology of extensive intracranial calcification in an infant mainly includes various congenital infections such as those caused by toxoplasmosis, rubella, cytomegalovirus (CMV), herpes (TORCH) group of organisms acquired during ante-natal period. Congenital CMV infection is the most common cause and results in microcephaly, mental retardation with radiological features of typical periventricular calcifications. Other rare causes include various genetic disorders such as Aicardi–Goutieres syndrome (AGS), Cockayne syndrome (CS), etc. Congenital CMV infection can be diagnosed serologically

by detecting CMV DNA using polymerase chain reaction (PCR) method. Pseudo-TORCH syndrome is a group of not so well-defined genetic diseases having autosomal recessive inheritance showing clinico-radiological features exactly similar to congenital CMV infection but serological tests are negative for the organisms. Dandy–Walker syndrome is a group of congenital hind brain anomaly with large posterior fossa cyst. Pseudo-TORCH syndrome with extensive calcium debris in cerebrospinal fluid (CSF) could lead to obstruction at fourth ventricle outlet leading to hydrocephalus and secondary enlargement of a posterior fossa cyst with cerebellar hypoplasia. Such a presentation of pseudo-TORCH syndrome could pose a difficulty in diagnosis as well as management planning, particularly for hydrocephalus.

Case Report

An 8-month-old male child presented with repeated episodes of generalized seizure and microcephaly since birth. Ante- and perinatal history were insignificant. Head circumference (42 cm) was grossly below the average value for that age. All the fontanelles were closed. Plain computerized tomography scan showed extensive intracranial calcification both periventricular and intraparenchymal calcifications. Calcifications were typically dense, thick, and chunky in nature and present all

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around the ventricles, within the cerebral hemispheres as well as in brain stem [Figure 1]. There was an associated moderate hydrocephalus with cerebral cortical atrophy. Interestingly, there was a large cyst in the posterior fossa with almost complete aplasia of cerebellar vermis [Figures 2 and 3] and hemispheres suggestive of Dandy–Walker malformation (DWM). The serological tests for CMV and other TORCH organisms were negative. Patients’ seizure was controlled with anti-epileptic drugs. Cysto- or ventriculoperitoneal shunt was not done as there were no features of raised intracranial pressure (ICP). After 6-month follow-up, patient is seizure free with anti-epileptics with almost normal development.

Discussion

Pseudo-TORCH syndrome has been a group of heterogeneous clinical disease entities having autosomal recessive pattern of inheritance with clinico-radiological features suggestive of congenital infections with TORCH group of organisms. They are characterized by the presence of microcephaly, seizures, neurological delay, hepatosplenomegaly, and thrombocytopenia associated with necrosis and calcification of the white matter quite similar to congenital CMV infection.^[2,3] Because of the high endemic rate of CMV exposure, serum antibody testing may not be used to accurately diagnose acute

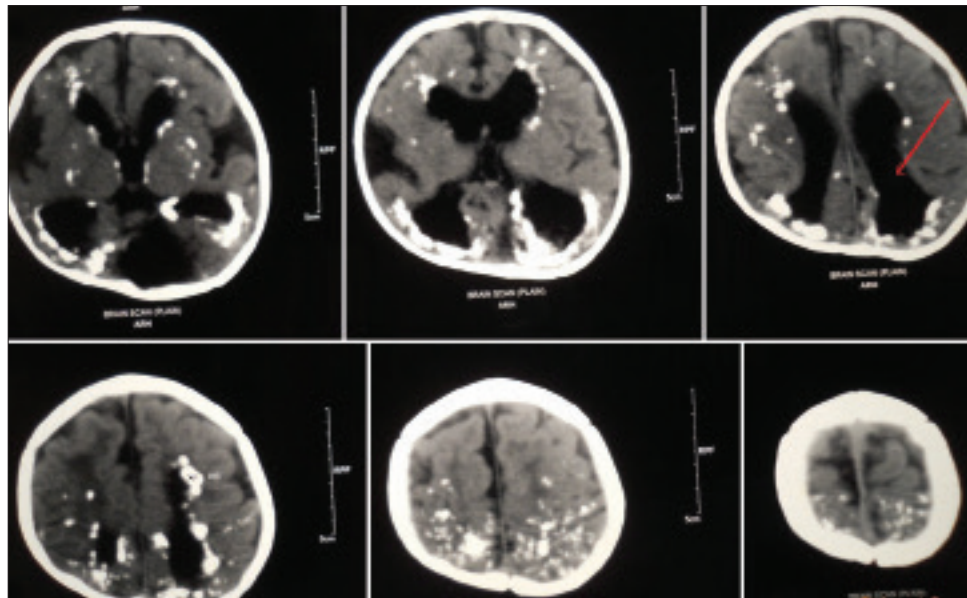


Figure 1: Extensive intracranial calcification, both periventricular and parenchymal, which are typically dense, chunky in nature with gross hydrocephalus (red arrow)

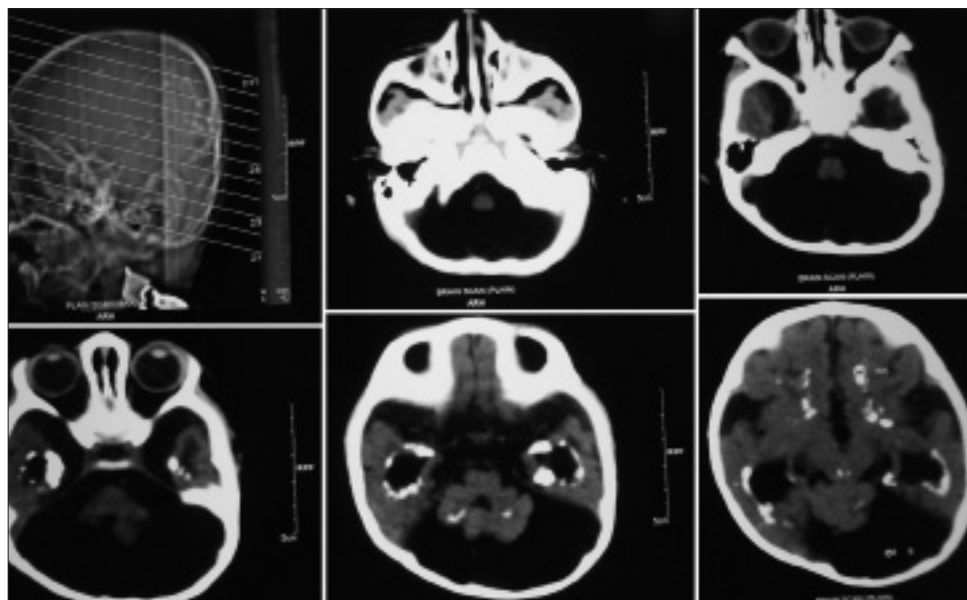


Figure 2: Large posterior fossa cyst with cerebellar hypoplasia, a feature suggestive of Dandy–Walker malformation (DWM)

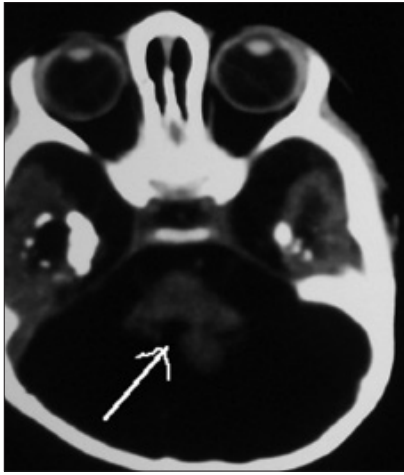


Figure 3: Vermianagenesis (white arrow) like that of Dandy–Walker malformation

CMV infection. Diagnosis of CMV-related infection relies on PCR analysis to detect CMV DNA within CSF; this method is highly sensitive and specific for detection of CMV infection. However, even after frantic search, serology remains negative for these organisms in pseudo-TORCH syndrome. Characteristically, in pseudo-TORCH syndrome, the calcifications are both intracranial and extracranial. The differential diagnosis of intracranial calcification in an infant includes congenital infections like TORCH group, various genetic conditions such as AGS, CS, fetal akinesia deformation sequence (FADS), and Fowler type. However, in all the above genetic conditions, calcifications were not typically periventricular rather they are diffuse cerebral in nature. In Aicardi syndrome, there is an associated raised interferon alpha in CSF and serum. This was negative in our case.

DWM, a congenital hind brain anomaly, is associated with a large posterior fossa and macrocephaly. It presents with features of delayed development with signs and symptoms of increased ICP. Various types of shunting procedures such as cystoperitoneal with or without ventriculoperitoneal shunts help in alleviating these symptoms. However, other than delayed development, all the other symptoms and signs were absent in our case. In the presence of extensive areas of necrosis and calcification, there may be calcified flecks and debris in CSF, which could have blocked the CSF drainage from

the ventricular system, leading to hydrocephalus and the cystic expansion of the fourth ventricle. This cystic expansion could have prevented the normal growth of cerebellum resulting in its hypoplasia.

The main difference between the pseudo-TORCH syndrome as in our case and DWM is the presence of symptoms and signs of raised ICP in the latter. Microcephaly is a regular feature of pseudo-TORCH syndrome, unlike the large head size in DWM. Thus, the latter requires cystoperitoneal with or without ventriculoperitoneal shunting for alleviating the symptoms. Such procedures are not required in pseudo-TORCH syndrome, as this is not associated with raised ICP. In pseudo-TORCH syndrome, there is a strong propensity for repeated episodes of seizures unlike DWM. However, the development of milestones remain almost normal in pseudo-TORCH syndrome, where as in DWM this is grossly affected by raised ICP and needs surgical intervention. Radiologically, there is also a large posterior fossa in DWM without calcifications.

Our case was significant in that it showed clinico-radiological features of congenital CMV infection as well as radiological findings of DWM. However, pseudo-TORCH syndrome with negative serology, secondary hydrocephalus with cerebellar hypoplasia and large cyst could strangely mimic other more common pathologies. Management of these lesions is purely conservative with no requirement for CSF diversionary procedures.

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