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Fourth Ventricular Rosai–Dorfman Disease Mimicking Intraventricular Tumor in Young Adult: A Rare Case Report

Meghana V. Chougule[1] Shankaran Prabhakar[2] Nikhil Tadwalkar[3] Atul Ranade[1]

[1] Department of Neuropathology, Shanti Pathology Laboratory Cancer Diagnosis & Research Center, Kolhapur, Maharashtra, India
[2] Department of Neurosurgery, Gangamai Hospital, Solapur, Maharashtra, India
[3] Department of Neuropathology, Gangamai Hospital, Solapur, Maharashtra, India

Address for correspondence Meghana V. Chougule, MD (Pathology), Shanti Pathology Laboratory Cancer Diagnosis & Research Center, C.S. no 311, E ward, New Shahupuri, Kolhapur 416001, Maharashtra, India (e-mail: drmeghanachougule20@gmail.com).

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Abstract

Rosai–Dorfman disease (RDD) or sinus histiocytosis with massive lymphadenopathy can present with or without systemic disease. It is a benign histioproliferative disorder characterized by generalized lymphadenopathy, weakness, anemia, and rarely extranodal involvement. While RDD most commonly affects lymph nodes, extranodal involvement of multiple organs has been reported, including the central nervous system (CNS). However, CNS involvement in RDD is rare and is not well characterized. Isolated involvement of the fourth ventricle is even rarer. Such lesions may be mistaken for intraventricular tumors such as ependymoma or medulloblastoma. This report highlights the necessity to consider RDD as a differential diagnosis in case of intraventricular space-occupying lesion.

Keywords

- Rosai–Dorfman disease
- fourth intraventricular tumors
- extranodal
- lympho-histiocytic diseases
- emperipolesis

Introduction

Intracranial involvement of Rosai–Dorfman disease (RDD) is rare and occurs in older age group. Isolated intracranial involvement presents most commonly as dural lesions mimicking meningioma. Very few reports of fourth intraventricular location have been described in the literature. Histopathological hallmark of RDD is emperipolesis, which is the engulfment by macrophages of intact lymphocytes.[1] Only a single case of isolated intraventricular RDD has been described involving the fourth ventricle earlier by Morandi et al in 2000.[2] We present, perhaps, the second case of fourth intraventricular RDD with clinical, imaging, and histopathological findings with immunohistochemical (IHC) profile.

Case Report

A 20-year-old young man presented with altered sensorium. Patient had no history of fever, anemia, weight loss, and generalized lymphadenopathy or hepatosplenomegaly. On investigation, he was found to have a fourth ventricle tumor with obstructive hydrocephalus. He underwent ventriculoperitoneal shunt on emergency basis and was planned for definitive surgery once stabilized. Preoperative magnetic resonance imaging findings revealed a lobulated ovoid space-occupying lesion in the region of fourth ventricle measuring approximately 46 mm × 35 mm causing moderate obstructive hydrocephalus. No periventricular ooze of cerebrospinal fluid was noted. Moderate compression over

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pons and medulla with caudal protrusion of cerebellar tonsils by 10 mm secondary to mass effect in posterior fossa was noted. Suboccipital craniotomy with telovelar approach was done. The lesion was avascular, attached to choroid plexus. Complete excision was done and postoperative course was uneventful. The excised tissue was sent for histopathological examination.

Gross examination revealed multiple, firm, gray-white, nodular masses with yellowish areas aggregating to 6.5 cm $\times$ 5 cm $\times$ 2 cm. Cut-section revealed soft, yellow areas. Microscopic examination revealed a cellular lesion composed of polymorphous population of cells and numerous histiocytes of varying sizes with foamy or granular cytoplasm, exhibiting emperipolesis of lymphocytes along with marked infiltration by mature lymphocytes and plasma cells. Russell bodies were noted. Foci of fibrosis and focal clustering of blood vessels were seen. On IHC analysis, the histiocytes expressed strong positivity for S100 and CD68. The lymphocytic population was polymorphous and expressed CD3, CD20 positivity. *BRAFV600E* immunoreactivity was absent. On the basis of classical histomorphology and supportive IHC findings, a diagnosis of RDD of the fourth ventricle was confirmed.

The primary antibodies used for immunohistochemistry were S100 (clone beta-EP32; rabbit monoclonal antibody, Livermore, California, United States), CD68 (Clone-KP1, mouse monoclonal antibody, Pleasanton, California, United States), CD3 (rabbit polyclonal antibody, Livermore, California, United States), CD20 (clone L-26, mouse monoclonal antibody, Livermore, California, United States), Ki 67 (MIB 1 clone GM001 mouse monoclonal antibody, Pleasanton, California, United States), and *BRAFV600E* (clone RM8; rabbit monoclonal antibody, Livermore, California, United States).

Patient improved after surgery and was asymptomatic on follow-up. He underwent thorough clinical examination, which revealed no lymphadenopathy or hepatosplenomegaly and ultrasonography findings were normal to rule out systemic involvement. Postoperative computed tomography (CT) chest and abdomen was done to conclude this was an isolated intracranial disease.

Discussion

Rosai–Dorman disease (RDD) was originally described by French pathologist Destombes in 1965.[3] Subsequently, it was characterized as a distinct clinicopathological disorder in 1969 by Rosai and Dorfman.[4,5] It is a benign disease characterized by overproduction and infiltration of histiocytes within lymph nodes, most often those of the cervical region.[6] Other lymph node groups may also be involved.[7] Extranodal involvement is seen in 30% of the cases and may involve the skin, salivary gland, thyroid, and testis.[8] It usually presents in the second or the third decades of life as painless lymphadenopathy, fever, and weight loss. The cause of this condition remains unknown, although altered immune responses and infectious agents may play a role. RDD is a self-limited and seldom life-threatening disease that commonly does not require therapy.[7] It tends to involve the central nervous system (CNS) very rarely and in a slightly older age group.[9] Isolated intracranial involvement is even rare, with less than 150 cases described, and isolated intraventricular involvement is even rarer.[10] The dural-based masses mimic meningiomas. These lesions may be present in the suprasellar region, convexity, parasagittal region, cavernous sinus, petroclival region, and cerebellum. Two cases with locally aggressive features and dural sinus involvement have been reported by Toh et al.[11] Pachymeningitis may also be the mode of presentation.[12] Very rarely, parenchymal involvement mimicking lymphoma has been described.[13,14] Friconnet et al reported a case of RDD in a 30-year-old man presenting as an isolated mass arising from the right cerebellar peduncle and protruding into the fourth ventricle.[15] Morandi et al, in 2000, reported the first case of isolated fourth ventricular RDD in a 22-year-old woman.[2]

Patwardhan et al described the second case of isolated RDD in a 40-year-old man who presented as an enhancing lobulated mass expanding the atrium and the occipital horn of the lateral ventricle. Radiologically, the lesion resembled meningioma, as classically described, and the patient had no evidence of lymphadenopathy or hepatosplenomegaly. Thus, this was an isolated intraventricular presentation of RDD in the lateral ventricle.[16,17]

Our case is perhaps the second case in literature with sole involvement of fourth ventricular RDD in a 20-year-old man with altered sensorium. Imaging findings revealed a fourth ventricle space-occupying lesion mimicking tumor and causing obstructive hydrocephalus (►**Fig. 1**).

Radiological examination remains unable to establish the diagnosis by itself.[15]

The lesion was excised by telovelar approach exposing well-circumscribed, gray-white nodular lesion (►**Fig. 2**). Macroscopic examination revealed multiple fragmented firm, gray-white to yellow nodular masses aggregating to

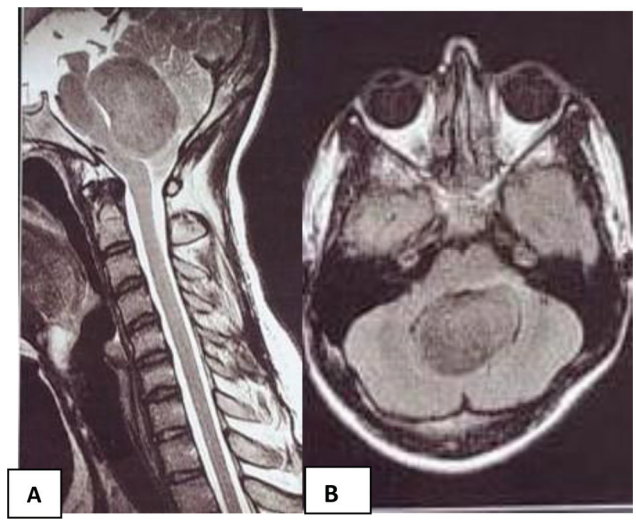

Fig. 1 (**A**, **B**) Magnetic resonance imaging revealed a lobulated ovoid space-occupying lesion in the region of fourth ventricle measuring approximately 4.6 cm $\times$ 3.5 cm causing moderate obstructive hydrocephalus. No periventricular ooze of cerebrospinal fluid was noted. Moderate compression over pons and medulla with caudal protrusion of cerebellar tonsils by 10 mm secondary to mass effect in posterior fossa was noted.

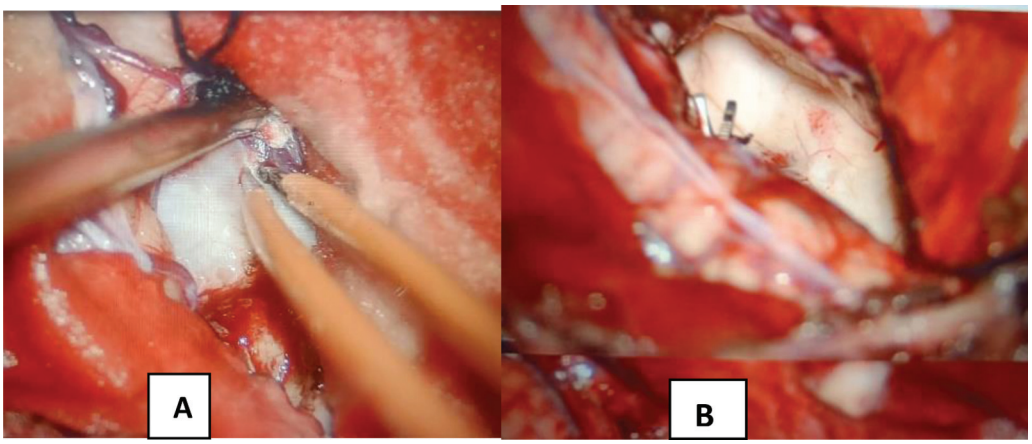

Fig. 2 Intraoperative images: (**A**) Telovelar approach exposing well-circumscribed, gray-white nodular lesion. Cerebellar tissue is seen at the 11 o'clock position. (**B**) Brain stem was seen at anterior aspect after total excision of the lesion.

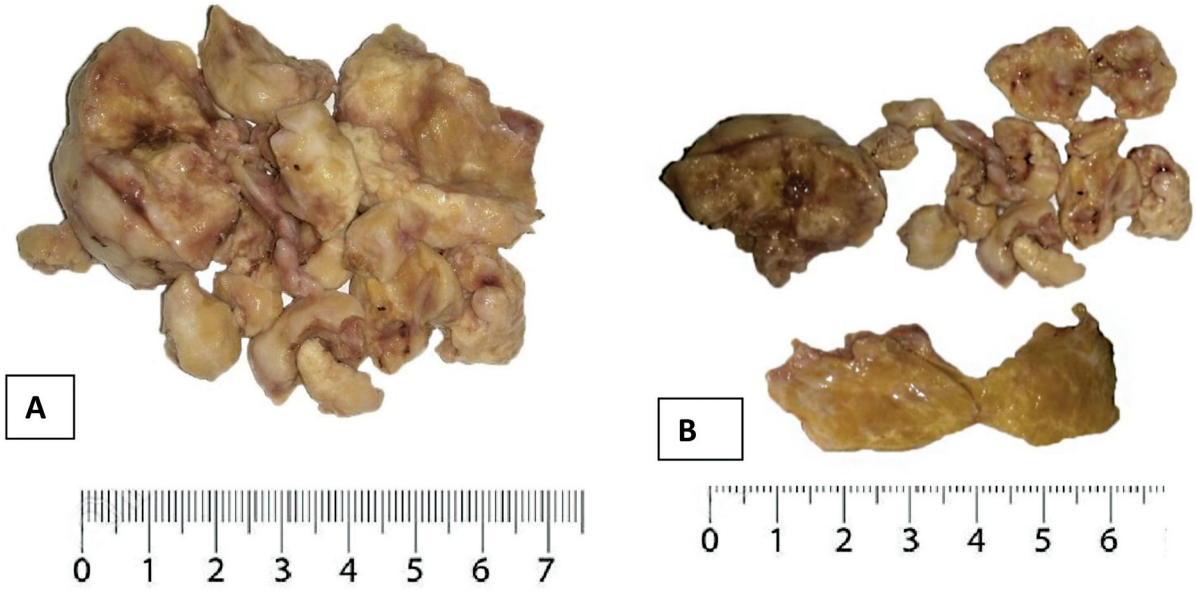

Fig. 3 (**A**) Gross examination: multiple fragmented firm; gray-white to yellow nodular masses aggregating to 6 cm × 5 cm × 2 cm. (**B**) Cut-section revealed soft, yellowish areas.

6 × 5 × 2 cm. Cut-section revealed soft, yellowish areas (►**Fig. 3**). Histology reveals a cellular lesion characterized by the accumulation of histiocytes with enlarged, round to oval hypochromatic nuclei and abundant eosinophilic cytoplasm, often containing engulfed intact inflammatory cells known as emperipolesis. Lesional histiocytes are usually associated with fibrosis and prominent inflammatory infiltrate comprised of plasma cells and lymphocytes. Occasional neutrophilic infiltrates and Russel bodies may be present (►**Fig. 4**).

Lymphohistiocytic diseases in the CNS are Langerhans' cell histiocytosis, Erdheim-Chester disease, RDD, Juvenile xanthogranuloma, and histiocytic sarcoma. The mean age of patient with RDD is 21 years. It forms solitary or multiple dural masses especially affecting the cerebral convexities, cranial base, cavernous sinuses, and parasagittal, suprasellar, and petroclival regions.[17]

In contrast to Langerhans' cell histiocytosis, eosinophils are rare. Emperipolesis is the hallmark of RDD.[15] Immunoprofile reveals immunoreactivity of histiocytes for S100 and CD68, and negativity for CD1a (►**Fig. 5**). *BRAFV600E* mutation is present in 50% of cases. Our case did not express immunoreactivity for *BRAFV600E*.

Retrospective history and thorough examination revealed no history of fever, anemia, weight loss, and generalized lymphadenopathy.

High-resolution CT and ultrasonography screening of thorax, neck, abdomen, and pelvis were normal. Thus, this lesion was concluded to be an isolated RDD of the fourth ventricle.

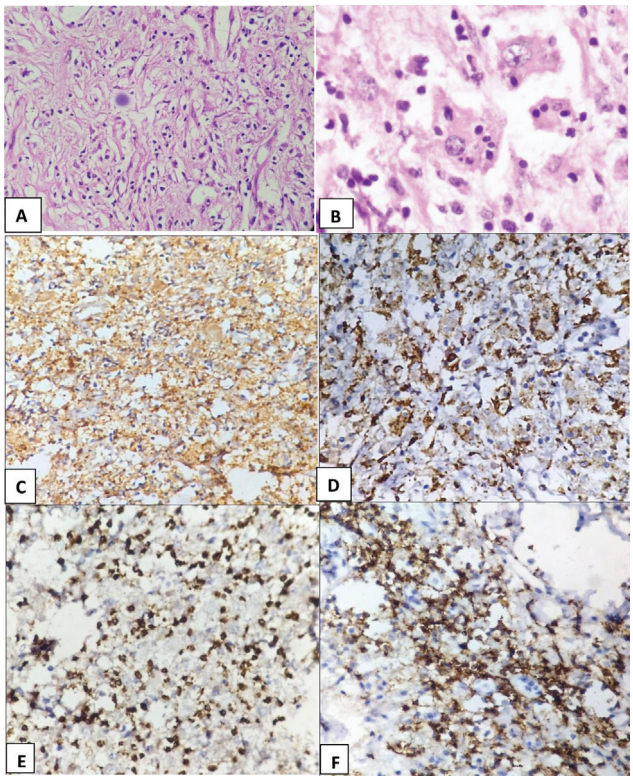

Fig. 4 Microscopy revealed (**A**) histiocytes with prominent emperipolesis admixed with lymphocytes, mature plasma cells, and Russell body (hematoxylin and eosin). (**B**) Histiocytes with emperipolesis. (**C**) Macrophages expressing S100. (**D**) Macrophages expressing CD68. (**E**, **F**) Lymphocytes are both CD3 and CD20 positive.

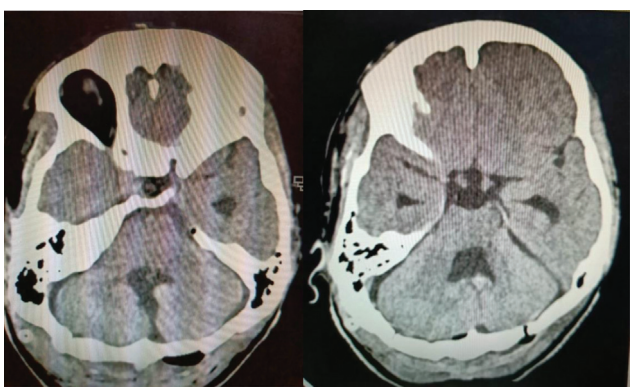

Fig. 5 Postoperative computed tomography scan revealed gross total excision of the lesion.

Conclusion

Fourth ventricular RDD is very rare and needs to be differentiated from commonly occurring fourth ventricular tumors such as ependymoma and medulloblastoma, or the rare glioneuronal tumors such as rosette forming glioneuronal tumor, in the young patients. Since diagnosis is based on histopathology, clinical and radiological suspicion does not alter the intraoperative aim of complete excision.

Patients' Consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patient understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflict of Interest

None declared.

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