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Case Report

A large growing occipital meningocele with Dandy–Walker syndrome: A case report and review of the literature

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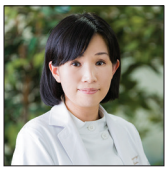

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ABSTRACT

Background: Dandy–Walker syndrome (DWS) is a well-known developmental anomaly. An occipital meningocele (OMC) is recognized as a malformation that is relatively often associated with DWS, but the association of DWS with OMC has been reported in approximately 40 cases. We present herein a rare clinical course of DWS with OMC, in which the sac was small at birth and became progressively larger.

Case Description: A 5-day-old baby boy was referred to our hospital due to OMC. He was born at 33 gestational weeks due to premature rupture of the membranes. He was diagnosed as having DWS associated with OMC. The OMC was covered with skin and its maximum diameter at birth was 3 cm. Magnetic resonance imaging showed an occipital bone defect and continuity of the fourth ventricle, posterior fossa cyst, and OMC sac. The aqueduct was patent, and no hydrocephalus was found. The OMC sac increased progressively with moderate hydrocephalus and reached 7 cm at the age of 54 days when his weight was 2508 g. A cystoperitoneal shunt and repair were performed after sinus venography by contrast computed tomography (CT). At the age of 1 year and 8 months, he had moderate developmental disabilities.

Conclusion: In most cases reported, the OMC was relatively small, and large and giant sizes were reported in only six cases. Almost all cases remained the same size as at birth and underwent surgical intervention as early as possible. It was possible to understand the relationship between the occipital bone defect and abnormal running of sinuses such as the superior sagittal sinus, torcular Herophili, and transverse sinus preoperatively from the CT venography (CTV) image. CTV may be an effective and important method for safely performing repair and shunt.

Keywords: Cyst-peritoneal shunt, Dandy–Walker syndrome, Occipital meningocele

INTRODUCTION

Dandy–Walker syndrome (DWS) is a well-recognized developmental anomaly of the brain consisting of cerebellar vermis hypoplasia or agenesis, cystic dilatation of the fourth ventricle, enlarged posterior fossa, and a high tentorium. Many congenital malformations of the central nervous system such as hydrocephalus, agenesis of the corpus callosum, cerebral grey

abnormalities, heterotopias, syringomyelia, aqueductal stenosis, malformation of the inferior olivary nuclei and dendritic nuclei, and hypoplasia of the pyramidal tracts are associated with DWS.[6] In addition, systemic anomalies include cleft palate, limb anomalies, cardiac malformation, urogenital abnormalities, skin capillary angioma, and so on, like PHACES syndrome.[6] The prognosis of DWS is often determined by the severity and diversity of these associated anomalies.[3,6] Occipital meningocele (OMC) is relatively rare among the malformations combined with DWS, reported to occur in 16% of cases with DWS.[3] The incidence of OMC was reported to be 0% in 72 cases of DWS.[10] We present herein a case of DWS associated with OMC and a rare clinical course of the OMC sac which was small at birth and became progressively larger.

CASE PRESENTATION

A boy was suspected of having DWS at 31 gestational weeks on fetal ultrasonography based on the findings of a cyst in the posterior fossa. He was delivered vaginally at 33 gestational weeks due to premature rupture of the membranes. His general condition was good (Apgar score 8/9), with a birth weight of 1969 g and a head circumference of 31 cm. However, he was found to have a ventricular septal defect and pulmonary hypertension which resulted in polypnea and retracted breathing. Therefore, he was transferred to our institution 5 days after birth for further evaluation and treatment. There was no family history of any congenital malformation or neural tube defect. An occipital bone defect with a skin-covered mass, 3 cm * 3 cm * 1 cm in size, was observed. A hairless part covered with fibrous tissues was found above the mass [Figures 1a and b]. In addition, cleft lip and palate and constrictive bands of the limbs suggestive of amniotic band syndrome were observed, and bilateral 2nd, 3rd, and 4th fingers were already partially absent at birth. He underwent pulmonary artery banding for pulmonary hypertension and the ventricular septal defect at the age of 11 days. Three-dimensional computed tomography (CT) at the age of 5 days showed sagittal suture dilatation and an occipital bone defect with swelling in the occipital region [Figure 1c]. Magnetic resonance imaging (MRI) demonstrated a cystic cerebrospinal fluid (CSF) intensity small swelling in the occipital region communicating with a posterior fossa cyst through a defect in the occipital bone suggestive of OMC [Figures 1d and e]. A large posterior fossa, vermis hypoplasia, and a posterior fossa cyst communicating with the fourth ventricle were observed, suggestive of DWS. The aqueduct was patent, and expansion of the third and lateral ventricles was not seen. These findings were consistent with the diagnosis of DWS with an OMC, and he was treated conservatively due to his low weight and no findings of hydrocephalus. In the genetic testing, there were no abnormalities in chromosome, G band, and subtelomere. Then, the occipital cystic swelling by meningocele progressively increased. The size of the subcutaneous mass in the occipital region was 7 cm * 7 cm * 2.5 cm at the age of 50 days [Figures 2a and b]. MRI at the age of 50 days showed a markedly increased OMC communicating with a posterior fossa cyst and fourth ventricle, as well as mild ventricular enlargement indicating hydrocephalus [Figures 2c and d]. CT showed that both the occipital bone defect and sagittal suture dilatation increased. Furthermore, contrast CT venography (CTV) demonstrated the run of the sinuses and veins in a defective part of the bone [Figure 2e]. The left transverse sinus ran along the left edge of an occipital bone defect, but there was neither sinus nor veins in its right ma

rgin. Therefore, the meningocele repair followed by cystic peritoneal shunt (CP-S) surgery was scheduled at the age of 54 days, with a patient's weight of 2508 g and head circumference of 34 cm. The surgery was performed under general anesthesia. First, the constriction bands of the right ankle and the right first digit were debanded by plastic surgeons in a supine position. Next, a CP-S with pressure set to 5 cm H2O valve was placed (Codman Hakim Programmable Valve, Johnson and Johnson Medical Ltd, New Brunswick, NJ, USA), placing the cyst tube from the right edge of the bone defect. The body was changed to the prone position. The meningocele sac was decompressed by draining due to the CP-S surgery. A skin incision was created around the hairless part above the meningocele sac, the redundant skin was excised with fibrous tissues, and a dural margin was defined all around. When the dura was incised, the left transverse sinus ran along the left edge of the meningocele sac [Figure 3a]. No neural tissues were seen inside the sac, suggestive of a meningocele. The redundant dura was excised, and the dural margin was sutured to achieve a watertight closure. The occipital bone defect was not repaired considering further bone growth, and the skin was closed esthetically in layers. No surgical complications were encountered, and an MRI 2 months after surgery showed that the ventricles and the cyst had shrunk postoperatively [Figures 3b and c]. The shunt pressure setting increased gradually and reached 14 cm H_2O 5 months after the operation, confirming the size of the ventricles and the meningocele sac. The circumference of the head was almost equivalent to that appropriate for age. Cheiloplasty and debanding of the constriction band of the left wrist were performed at the age of 6 months, and the patient was discharged home at the age of 7 months. At the age of 1 year and 8 months, the cyst side tube of the CP-S was replaced with a new one due to its occlusion and he had moderate developmental disabilities.

DISCUSSION

A cranial meningocele consists of the herniation of only dura mater through a skull defect, whereas an

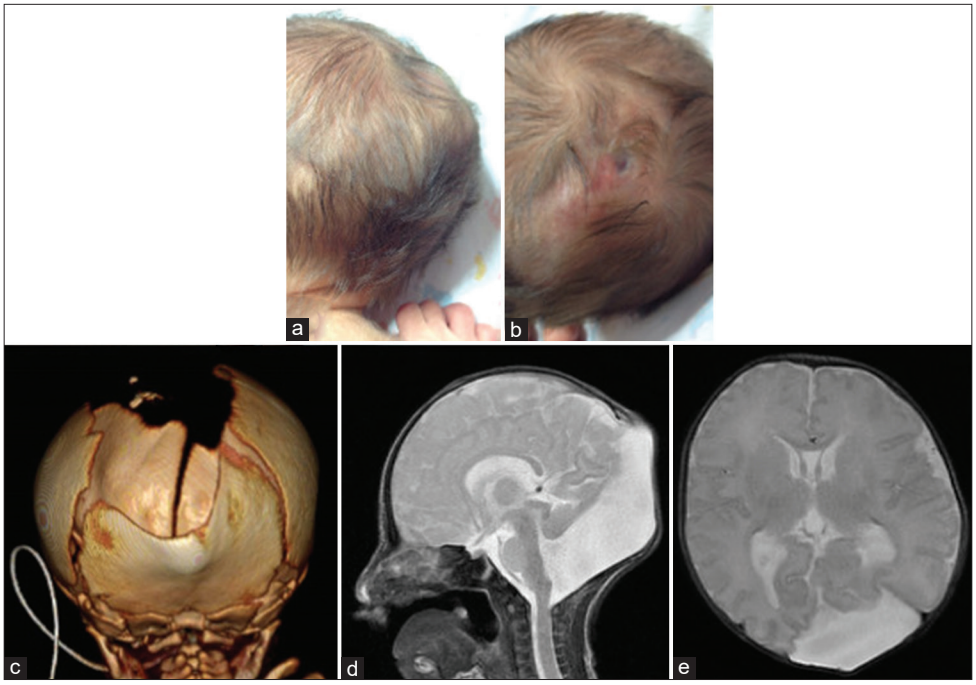

Figure 1: Images of the patient at the age of 5 days. (a and b) Clinical photographs show the skin-covered mass of size 3 cm*3 cm*1 cm with a hairless part covered with fibrous tissues. (c) Three-dimensional computed tomography shows sagittal suture dilatation and an occipital bone defect. (d and e) On sagittal and axial magnetic resonance imaging, a large posterior fossa, vermis hypoplasia, and a posterior fossa cyst communicating with the fourth ventricle can be seen, confirming the Dandy–Walker syndrome (DWS). The cystic cerebrospinal fluid-intensity small swelling in the occipital region is a large cyst communicating with a posterior fossa cyst through a defect in the occipital bone, suggesting an occipital meningocele associated with DWS. Hydrocephalus and occlusion of the aqueduct are not found.

encephalocele includes the herniation of dura mater with neural tissue.[3,16] Cephalocele is the term that includes both cranial meningocele and encephalocele. The skull defect is more often in the occipital bone, and the overlying head skin is usually intact. The association between cephalocele and DWS was first reported in 1887.[7] There have been only approximately 40 cases of OMC associated with DWS reported, and Table 1 summarizes 32 cases for which details could be found, along with the present case.[3,4,7-9,11,13,14,16,17,20]

DWS with OMC

Table 1 shows that almost all cases of OMC with DWS were diagnosed at birth and underwent neurosurgical intervention in infancy. OMC can be of the atretic type, in small, large, or giant sizes.[8] Talamonti *et al.* classified OMC sacs into three groups by diameter, small, <5 cm; large, 5–9 cm; and giant, >9 cm.[16] In most cases, the OMCs were relatively small, and large and giant sizes were reported in only 12 cases. The size of an OMC usually increases progressively as raised intracranial pressure leads to compensatory CSF escape into the sac as children grow, but no case of progressive meningocele sac enlargement could be found, except for one case[16] and the present case. Almost all cases remained the same size from birth, even if they were large or giant. Because surgery is promptly performed in early infancy when DWS with OMC is diagnosed, early operation may prevent the progression of small OMCs. Given the information in Table 1, it appears that the prognosis of the DWS with OMC is not necessarily good. In particular, the large and giant cases tend to have much higher mortality than the small cases. However, surgical treatment of DWS with OMC remains controversial. The treatment strategy that should be selected from among shunting alone, repair of OMC alone, or both has also not been determined.

OMC repair and shunting

In Table 1, only six patients having DWS associated with OMC underwent only OMC repair as the first surgery, two patients underwent shunt surgery as the second treatment, and the other four patients died. The outcome of only repair was poor. The effects of hydrocephalus on intracranial pressure are mitigated by a skull-deficient meningocele sac and an open fontanelle.[3,9] Therefore, it is thought that OMCs compensate for the increased intracranial pressure. Thus, the repair of OMC

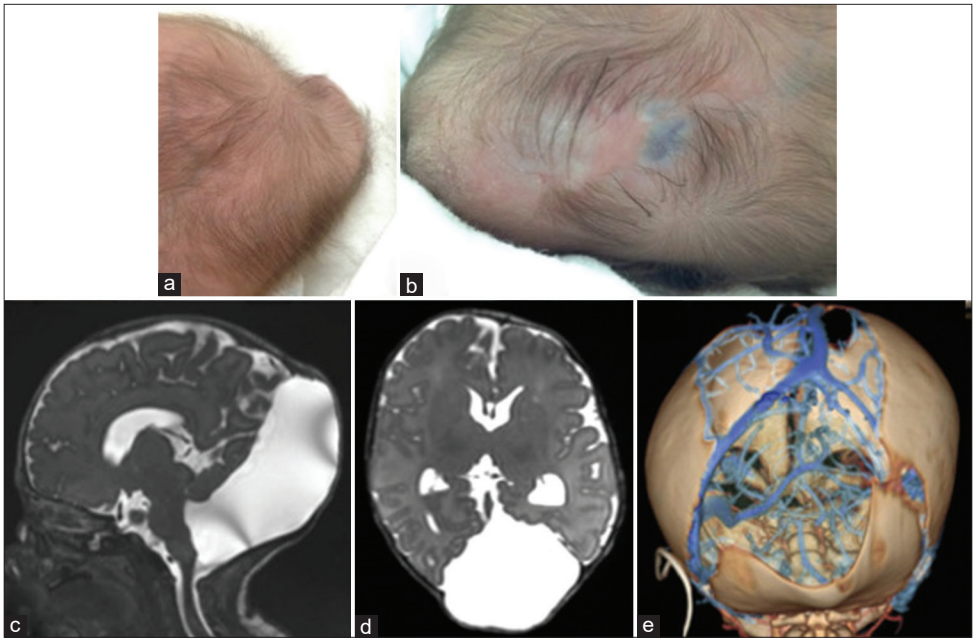

Figure 2: Images of the patient at the age of 50 days. (a and b) Clinical photographs show that the skin-covered subcutaneous mass in the occipital region has enlarged to 7 cm*7 cm*2.5 cm. The hairless part covered with fibrous tissues is further thinned, and the inside of the dura is seen through it. (c and d) On sagittal and axial magnetic resonance imaging, an occipital meningocele communicating with a posterior fossa cyst and fourth ventricle, and mild ventricular enlargement indicating hydrocephalus are seen. (e) Three-dimensional computed tomography venography demonstrates the run of the sinuses and veins in a defective part of the bone. The left transverse sinus runs along the left edge of an occipital bone defect, but there is neither sinus nor veins in its right margin. Both the occipital bone defect and sagittal suture dilatation are enlarged.

changes CSF flow dynamics and also further diminishes surface area for CSF resorption. The pathology of DWS has not yet been clarified, and the association of DWS with OMC is also more unclear. However, we consider that curative treatment of DWS with OMC cannot be achieved by surgical meningocele repair alone, and it likely requires shunt procedures.

Bindal *et al*. recommended only shunt placement as the initial surgical treatment of DWS with OMC and that surgical closure of the OMC may not be necessary.[3] In Table 1, nine patients underwent cystoperitoneal shunt (CP-S) or a ventriculoperitoneal shunt (VP-S) alone as the first surgery, and their OMC size was small in six cases, large in two cases, and giant in one case. In the six small OMC cases with only shunt placement, all six patients survived. Therefore, in the case of a small OMC, it may be possible to perform only shunt placement as the first surgery and then choose whether to proceed to repair depending on the clinical course.

On the other hand, of the three patients with a large or giant OMC who underwent only shunt placement two patients died [Table 1]. All four patients, including the present case, who were treated by both shunt surgery and repair for a large or giant OMC simultaneously as the first surgery, survived. It has been reported that shunting increases the likelihood of developing decubitus ulcers due to the edges of the occipital bone protruding, in addition to the high risk of shunt infection and frequent shunt malfunction.[3,17] OMC repair in addition to shunting may provide a better outcome than shunt placement alone in DWS with large or giant OMC. The present case was diagnosed as having DWS associated with a small OMC at birth. Considering the previous case reports, if shunting could have been performed relatively early after birth, OMC repair might have been skipped. However, the patient's low weight due to premature birth and surgery for a congenital heart malformation made early shunt placement impossible. Therefore, hydrocephalus and OMC were observed carefully, and repair and shunting were performed when his weight exceeded 2500 g to prevent infection.

CP-S or VP-S

There is no clear guideline for deciding between CP-S and VP-S for patients having DWS with OMC. As the initial shunt placement in the cases of DWS with OMC that we found, including the present case, 15 patients underwent CP-S, six patients underwent VP-S placement, and four patients underwent combined shunt surgery (CP-S plus VP-S) [Table 1]. CP-S was selected more often than VP-S as the first surgery for DWS with OMC. Patency of the aqueduct is the

Table 1: The characteristics of cases of DWS associated with OMC.

Case	Size	Age at first operation	First treatment	Additional treatment	after initial surgery	Hydrocephalus	Aqueduct patency	Outcome	Author
1	Small	1 month	CP-S/repair	VP-S	1 month	-	NR	good	Kojima. 1982[7]
2	Small	at birth	VP-S	CP-S/repair	1 week	+	NR	good	
3	Small	at birth	CP-S			+	NR	normal-intelligece	Bindal. 1991[3]
4	Small	5 months	repair	CP-S	1 month	+	NR	normal-intelligece	
5	Small	1 week	CP-S			+	NR	severe development delay	
6	Small	at birth	CP-S/repair	VP-S	NR	+	NR	borderline-intelligence	
7	Small	at birth	CP-S/repair			+	NR	severe development delay	
8	Small	4 months	CP-S	repair	6 months	+	NR	moderately retarded intelligence	
9	Small	1 week	CP-S/repair	VP-S	1 week	+	NR	survival	
10	Small	1 week	VP-S	repair	4 week	+	NR	normal-intelligece	
11	Small	1 week	CP-S			-	+	normal-development	Shuto. 1999[14]
12	Small	1 day	CP-S/VP-S/repair			+	-	survival	Yuceer. 2007[20]
13	Small	7 weeks	CP-S/repair			+	+	survival	
14	Small	2 weeks	CP-S/repair			+	+	survival	
15	Small	1 day	CP-S/repair			+	+	survival	
16	Small	4 months	CP-S/VP-S/repair			+	-	survival	
17	Small	2 weeks	CP-S/repair			+	+	survival	
18	Small	1 day	CP-S/VP-S/repair			+	-	survival	
19	Small	1 day	repair			-	NR	death	Kotil. 2008[8]
20	Large	1 month	VP-S			+	NR	good	
21	Large	1 day	VP-S			+	NR	death	
22	Large	2 months	repair			-	NR	death	
23	Large	1 month	CP-S/repair			+	NR	good	
24	Large	3 months	repair			-	NR	death	
25	Large	1 day	CP-S/VP-S/repair			-	NR	good	
26	Large	2months	CP-S/repair			-	+	moderately retarded intelligence	Present Case
27	Giant	1 week	CP-S	repair repair	2 months 3 months	+	+	death	Todo. 1993[17]
28	Giant	10 months	repair			-	NR	death	Muzumadar. 2004[11]
29	Giant	6 days	NR			+	NR	NR	Cakmak. 2008[4]
30	Giant	8 years	repair	CP-S repair	3 weeks 3 months	-	+	good	Talamonti. 2011[16]
31	Giant	1.5 years	VP-S/repair			+	+	survival	Mankotia. 2016[9]
32	Giant	at birth	conservative treatment			+	NR	death	Rana. 2021[13]

Size: size of occpital meningocele (OMC), Small: OMC<5cm, Large: OMC=5 to 9cm, Giant: OMC>9cm, CP-S: cystoperitoneal shunt, VP-S: ventriculoperitoneal shunt, NR: not recorded, DWS: Dandy-Walker syndrome, +: having

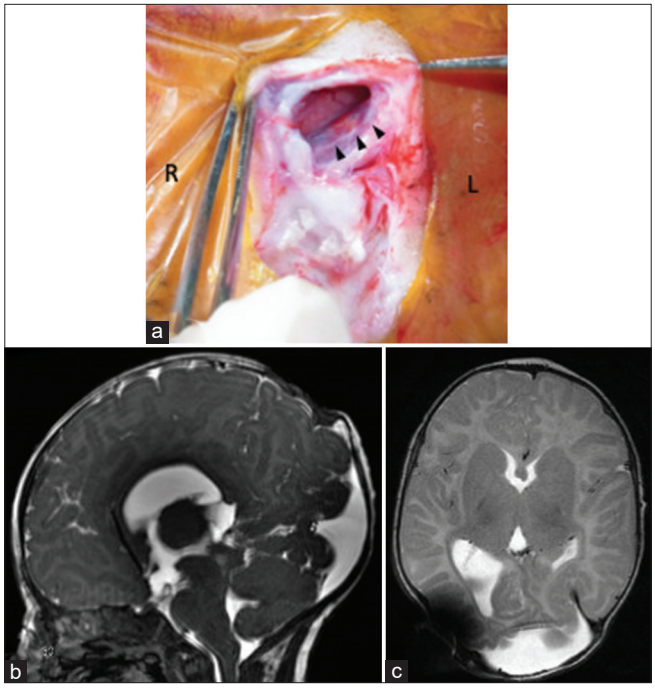

Figure 3: (a) Intraoperative photograph shows the left transverse sinus (black arrow) running along the left edge of the meningocele sac after the dura incision. (b and c) On sagittal and axial magnetic resonance imaging 2 months after surgery, the contracted occipital meningocele and ventricles are seen.

prerequisite for a single shunt.[15] Some authors recommended VP-S for DWS as the first surgical treatment due to its simplicity, low risk of complications, and achievement of early decompression of the supratentorial compartment.[3,12] However, some authors pointed out that VP-S placement can lead to the development of acquired aqueductal stenosis and isolated fourth ventricles, and they recommended CP-S.[1,2,5] The peritoneal shunt from the posterior fossa cyst appears to be the more logical placement because it maintains downstream flow in the aqueduct. Catheter placement on the cyst side in the edge of the skull defect may be easier because OMC is continuous with the posterior fossa cyst through an occipital bone defect. It is necessary to consider carefully which shunt should be selected taking into account the clinical condition of the dilated cyst and ventricles. In the present case, CP-S was selected because the enlargement of the posterior fossa cyst and the OMC sac was more marked than that of the ventricles. Since the ventricles were also reduced by CP-S placement, it was thought that VP-S was not necessary. The pressure setting of the shunt system was increased incrementally in small steps after the operation to maintain the patency of the aqueduct.

Usefulness of CTV

To safely perform shunting and repair for patients having DWS with OMS, it is necessary to understand the anatomy of the cyst. In particular, the relationship between the OMC sac and the vein or sinus is important. Verma *et al*. reported that magnetic resonance venography (MRV) may show a dilated venous sinus and torcula, and it aids in proper surgical planning in cases of occipital encephalocele.[18] In the present case, CTV demonstrated more clearly the running of the sinus in the OMC sac, whereas MRV showed defects in most of the sinus. MRV has the disadvantage of the lack of flow-related enhancement due to artifacts and the necessity of anesthesia. However, if we can get a detailed image of the sinus by MRV, CTV is unnecessary considering the risk of additional radiation and iodinated contrast medium. Yamashita *et al*. reported that three-dimensional CTV (3DCTV) showed the superior sagittal sinus (SSS) and skull defect, and it was used as a guide for the cranioplasty in a patient with a parietal encephalocele.[19] The relationships between the occipital bone defect and the abnormal running of the sinuses, such as the SSS, torcular herophili, and transverse sinus, were observed preoperatively, and it was possible to safely perform repair and CP-S using the 3DCTV image as a guide. We think that it is important to obtain information about the sinuses and veins, whether from CTV or MRV, considering the anatomical variability of pediatric veins.

CONCLUSION

Although almost all cases remained the same size as at birth and underwent surgical intervention as early as possible, the present case of OMC that developed progressively and reaching a large size was rare. Similar to most cases, both shunt placement and repair were performed at the same time as the first treatment. Confirming significant enlargement of the OMC sac despite mild hydrocephalus, CP-S was selected, and the outcome was good. It was possible to understand the relationship between the occipital bone defect and abnormal running of the sinuses preoperatively from the CTV image. CTV may be an effective and important method for safely performing repair and shunting.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Nil.

Conflicts of interest

There are no conflicts of interest

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The author(s) confirms that they have used artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript or image creations.

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