

Non-migratory cerebrospinal fluid hydrothorax following ventriculoperitoneal shunting in KIF7-Related Joubert syndrome with acrocallosal features: A case report

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ABSTRACT

Joubert syndrome is a rare neurodevelopmental disorder characterized by cerebellar vermis hypoplasia and a broad clinical and genetic spectrum. Some KIF7-related Joubert syndrome cases show overlapping acrocallosal features, including additional central nervous system malformations and hydrocephalus requiring neurosurgical intervention. Although ventriculoperitoneal shunting is commonly used in pediatric hydrocephalus, intrathoracic complications are rare. We report a term-born male patient with KIF7-related Joubert syndrome with acrocallosal features who developed pleural effusion 20 months after ventriculoperitoneal shunt placement. Thoracic ultrasonography demonstrated bilateral pleural effusion. β 2-transferrin analysis confirmed the presence of cerebrospinal fluid (CSF) in the pleural fluid. Imaging studies revealed an intact shunt system without intrathoracic migration or mechanical failure. Exploratory laparoscopy identified no macroscopic diaphragmatic defect, and the overall clinical course supported presumed impairment of peritoneal CSF absorption with secondary pleural accumulation. CSF-related pleural effusion should be considered in VP-shunted children presenting with respiratory symptoms even in the absence of catheter migration. After alternative causes have been excluded, impaired peritoneal absorption with transdiaphragmatic CSF passage may be considered a plausible but unproven mechanism.

Keywords: Cerebrospinal fluid, Joubert syndrome, hydrothorax, KIF7, pleural effusion, ventriculoperitoneal shunt

Introduction

Joubert syndrome is a rare neurodevelopmental disorder, most commonly inherited in an autosomal recessive manner, characterized by hypoplasia or agenesis of the cerebellar vermis (1,2). The clinical spectrum includes hypotonia, global developmental delay, episodic hyperpnea/apnea, and abnormal oculomotor findings (3). The characteristic “molar tooth sign” on magnetic resonance imaging (MRI), resulting from vermian hypoplasia, thickened superior cerebellar peduncles, and fourth ventricle configuration, is a defining radiological feature (2,3).

Joubert syndrome exhibits marked clinical and genetic heterogeneity, with several subtypes described (1,2). Some patients may show overlapping acrocallosal features, including corpus callosum agenesis, polydactyly, and hydrocephalus (1,3). KIF7 mutations have been linked to acrocallosal features and more severe central nervous system malformations (4). Hydrocephalus is reported more frequently in patients with acrocallosal features, particularly in KIF7-related cases, increasing the likelihood of neurosurgical intervention (1,4).

Ventriculoperitoneal (VP) shunt placement is a commonly used treatment for pediatric hydrocephalus. While infection and mechanical dysfunction are the most frequent complications, intrathoracic complications are rare but

potentially life-threatening (5,6). These include catheter migration and cerebrospinal fluid (CSF)-related pleural effusion without catheter displacement (6). In pediatric patients, limited peritoneal absorptive capacity may facilitate transdiaphragmatic CSF passage into the pleural cavity, even in the absence of shunt migration or a diaphragmatic defect (5,7,8).

Here, we present a pediatric patient with KIF7-related Joubert syndrome with acrocallosal features who developed non-migratory CSF hydrothorax following VP shunt placement, and we discuss the diagnostic and therapeutic implications of this rare shunt-related complication.

Case Report

A term-born male patient with a prenatal diagnosis of hydrocephalus was evaluated postnatally after polydactyly and dysmorphic facial features were noted. Genetic analysis identified a homozygous KIF7 variant consistent with the clinical and radiological diagnosis of KIF7-related Joubert syndrome.

The patient was referred to the neurosurgery outpatient clinic because of rapidly increasing head circumference and vomiting. Examination revealed hypotonia, global



Figure 1: Axial T2-weighted brain magnetic resonance image demonstrating the characteristic molar tooth sign of Joubert syndrome, including hypoplasia of the cerebellar vermis and thickened superior cerebellar peduncles (highlighted by a red circle).

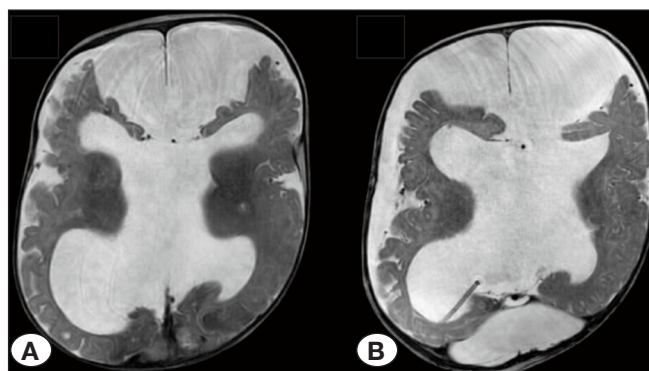


Figure 2: Pre- and post-shunt axial T2-weighted brain magnetic resonance images. **A)** Preoperative image demonstrating ventriculomegaly consistent with hydrocephalus. **B)** Postoperative image obtained after ventriculoperitoneal shunt placement showing a reduction in ventricular size.

developmental delay, and episodic hyperpnea/apnea. Cranial MRI showed findings consistent with Joubert syndrome, including the characteristic molar tooth sign (Figure 1). Based on hydrocephalus, corpus callosum agenesis, polydactyly, and the homozygous KIF7 variant, he was considered to have KIF7-related Joubert syndrome with acrocallosal features. At approximately 5 months of age, a ventriculoperitoneal (VP) shunt with a medium-pressure, non-programmable pediatric valve was placed for progressive hydrocephalus, with postoperative imaging showing a reduction in ventricular size (Figure 2).

Twenty months after VP shunt placement, he presented with fever and respiratory distress. Thoracic ultrasonography revealed bilateral pleural effusion, measuring 54 mm on the right and 20 mm on the left. Chest imaging demonstrated bilateral pleural effusion, more pronounced on the right side

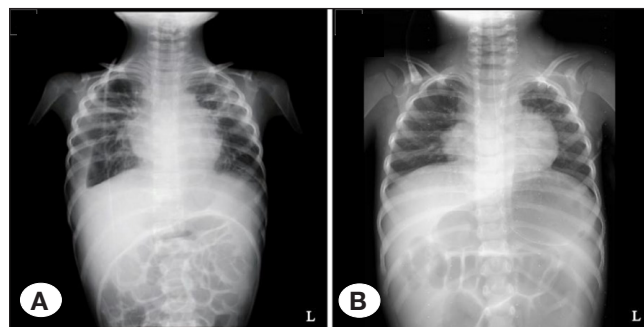


Figure 3: Chest imaging before and after pleural drainage and temporary external ventricular drainage. **A)** Anteroposterior chest radiograph demonstrating bilateral pleural effusion, more pronounced on the right side, consistent with the patient's clinical presentation of respiratory distress. **B)** Follow-up chest radiograph after ventriculoperitoneal shunt removal, temporary external ventricular drainage, and chest tube removal demonstrating resolution of the pleural effusion.

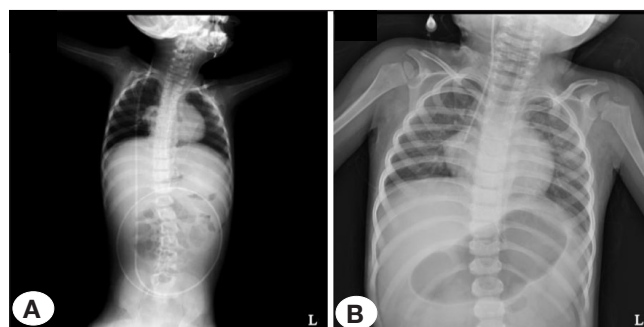


Figure 4: Shunt configuration before and after definitive revision. **A)** Anteroposterior chest and abdominal radiograph demonstrating an intact ventriculoperitoneal shunt, with the distal catheter tip located within the abdominal cavity and no evidence of intrathoracic migration. **B)** Follow-up radiograph after ventriculoatrial shunt placement demonstrating the final ventriculoatrial shunt configuration.

(Figure 3A). Thoracentesis and right-sided chest tube placement were performed. Evaluation for infectious, malignant, and inflammatory causes was negative. Persistent chest tube drainage raised suspicion of CSF-related pleural effusion, and $\beta 2$ -transferrin analysis confirmed the presence of CSF in the pleural fluid.

Cranial imaging, shuntography, and thoracic computed tomography demonstrated an intact VP shunt system, with the distal catheter located within the abdominal cavity and no evidence of intrathoracic migration or mechanical failure (Figure 4A). Pleural fluid analysis was consistent with a transudate according to Light's criteria. Exploratory laparoscopy revealed no macroscopic diaphragmatic defect. In the absence of catheter migration, ascites, or a macroscopic diaphragmatic defect, the findings supported presumed impairment of peritoneal CSF absorption with possible secondary transdiaphragmatic passage.

Because of persistent CSF drainage from the pleural space, the VP shunt was removed and temporary external ventricular drainage (EVD) was established to divert CSF away from the peritoneal cavity and allow stabilization before definitive shunt reconstruction. The pleural effusion regressed rapidly after EVD placement. After temporary external ventricular

drainage, clinical stabilization, and exclusion of CSF infection, fenestration of the posterior fossa cystic lesion communicating with the fourth ventricle was performed to improve intracranial CSF circulation. Definitive CSF diversion was then achieved by ventriculoatrial shunt placement. This route was preferred because persistent CSF-related pleural effusion during VP shunting supported presumed failure of the peritoneal absorptive pathway, whereas ventriculopleural shunting was avoided because of the recent CSF-related pleural effusion. Follow-up chest imaging after VP shunt removal, EVD placement, and chest tube removal demonstrated resolution of the pleural effusion (Figure 3B). The final ventriculoatrial shunt configuration was subsequently confirmed radiologically (Figure 4B). No recurrence was observed during 12-month follow-up, and the patient had no clinical or radiological evidence of recurrent pleural effusion or shunt-related cardiopulmonary complications at the last visit.

Discussion

Intrathoracic complications of ventriculoperitoneal (VP) shunts are rare but potentially life-threatening (5, 6). They may result from intraoperative thoracic injury, distal catheter migration, or, more rarely, CSF-related pleural effusion without catheter migration. The latter is thought to result from impaired peritoneal CSF absorption, allowing fluid to pass into the pleural space through transdiaphragmatic pathways or microscopic pleuroperitoneal communications (6).

Most CSF-related hydrothorax cases are associated with catheter migration, whereas non-migratory cases are less common. In non-migratory cases, CSF ascites is frequently observed; therefore, pleural effusion without catheter migration or ascites, as in our case, represents an exceptionally rare presentation (6,9). A recent literature review identified 19 reported cases of CSF-related pleural effusion without VP shunt migration, 14 of which occurred in pediatric patients (10). Selected representative pediatric cases are summarized in Table I (7,8,10-14).

Another clinically relevant feature is the delayed onset of pleural effusion, which developed approximately 20 months after VP shunt placement. This suggests that non-migratory CSF hydrothorax may present as a late complication, as also observed in previous cases (8,10). Delayed presentation may reflect gradual saturation of peritoneal CSF absorptive capacity, with subsequent passage of CSF into the pleural cavity through microscopic pleuroperitoneal communications (6, 8). Age-related changes in CSF production and growth-related anatomical factors may also contribute, although these explanations remain speculative in the present case.

The bilateral nature of the effusion also deserves comment. Although bilateral, it was markedly right-predominant, consistent with the proposed mechanism of transdiaphragmatic CSF passage through the right hemidiaphragm (6,8). Bilateral involvement is less typical, as most reported non-migratory CSF hydrothorax cases are unilateral (10). In our case, bilateral effusion may have resulted from bilateral microscopic pleuroperitoneal communications, mediastinal redistribution, or transudative

redistribution due to sustained CSF volume load. These mechanisms remain speculative because no macroscopic diaphragmatic defect was identified.

Several findings supported a shunt-related CSF pleural effusion without catheter migration. β 2-transferrin confirmed CSF in the pleural fluid. Imaging showed an intact shunt system with the distal catheter located within the abdominal cavity, and laparoscopy showed no macroscopic diaphragmatic defect. Rapid regression after VP shunt removal and diversion away from the peritoneal cavity further supported the peritoneal cavity as the presumed source of CSF overload. Together, these findings support presumed impairment of peritoneal CSF absorption as the most plausible mechanism, although this was inferred indirectly rather than definitively demonstrated.

Shunt overdrainage was also considered. Excessive CSF diversion may theoretically exacerbate peritoneal absorptive overload, a mechanism consistent with previous explanations for non-migratory CSF hydrothorax (6,8). However, the patient had a medium-pressure, non-programmable pediatric valve, overdrainage was not suspected clinically, and follow-up cranial imaging did not show slit ventricles or subdural collections. Therefore, overdrainage was considered less likely, although it cannot be completely excluded as a minor contributor.

The underlying KIF7-related Joubert syndrome with acrocallosal features may have provided additional disease-specific context. KIF7 mutations are associated with complex central nervous system malformations, which may theoretically alter CSF dynamics (4,15). However, increased CSF production or altered CSF dynamics remains speculative in this case because CSF production rate and opening pressure were not measured despite temporary external ventricular drainage.

Management was guided by presumed failure of the peritoneal cavity as an absorptive site. Temporary EVD was used after VP shunt removal to stop further CSF delivery to the peritoneum, allow effusion resolution, and provide a controlled interval before definitive diversion. Fenestration of the posterior fossa cystic lesion communicating with the fourth ventricle was performed to improve intracranial CSF circulation and reduce possible cyst-related compartmentalization. VP shunt revision alone was not favored because the distal catheter was correctly positioned and there was no mechanical failure. Ventriculopleural shunting was avoided because of the recent CSF-related pleural effusion. VA shunting was preferred to bypass the presumed impaired peritoneal absorptive pathway, while choroid plexus coagulation was considered less suitable because definitive CSF diversion was required in the setting of progressive hydrocephalus and complex intracranial abnormalities (8, 10). Although VA shunting prevented recurrence during 12-month follow-up, longer-term surveillance remains necessary because of the known risks of VA shunts.

To the best of our knowledge, this is the first reported case of non-migratory CSF hydrothorax in a patient with KIF7-related Joubert syndrome with acrocallosal features. Nevertheless, the principal clinical novelty of this case is

Table I: Selected pediatric cases of non-migratory CSF hydrothorax after VP shunting

Author (year)	Age	Interval from VP shunt to effusion	Effusion / VP shunt side	Ascites	Diaphragmatic defect	Diagnostic evidence	Treatment strategy	Recurrence / follow-up
Hadzikaric (2002) ¹²	16 months	16 months	Left / right	No	NR	Imaging + clinical correlation	VA shunt	No early recurrence; discharged 5 days after VA shunt
Adeolu (2006) ⁷	8 years	6 weeks	Right / right	NR	NR	Imaging + clinical correlation	Shunt revision	NR
Smith and Cohen (2009) ¹⁴	14 months	2.5 months	Right / right	Yes	No gross defect	β 2-transferrin assay	Externalization+VA shunt	NR
Patel (2011) ¹³	4 months	~ 3.5 months	Right / right	No	No	β 2-transferrin assay	Shunt revision + VA shunt	Recurred after repositioning; none after VA shunt; duration NR
Chuen-im (2012) ¹¹	5 years	Several years	Right / right	NR	No	β 2-transferrin assay	VATS / pleurodesis, CPC	NR
Kim (2015) ⁸	3 years	3 years	Right / right	Yes	NR	β 2-transferrin assay	Externalization+distal revision	No recurrence at 6 months
Schmid (2023) ¹⁰	4 years	2 years	Right / right	NR	Yes / diaphragmatic CSF fistula	Imaging + clinical correlation	Externalization+CPC	No recurrence; shunt-free at 2.5-year follow-up
Present case	25 months	20 months	Bilateral, R > L / right	No	No macroscopic defect	β 2-transferrin assay	VP shunt removal + EVD, cyst fenestration, VA shunt	No recurrence during 12-month follow-up

NR: not reported, **VP:** ventriculoperitoneal, **VA:** ventriculoatrial, **EVD:** external ventricular drainage, **CPC:** choroid plexus coagulation, **VATS:** video-assisted thoracoscopic surgery. Data were recorded as NR when not reported in the original article or available literature review. Superscript numbers correspond to the references listed in the manuscript.

the occurrence of non-migratory CSF hydrothorax without ascites or a macroscopic diaphragmatic defect. The KIF7-related Joubert phenotype provides additional context, but the main diagnostic message is that shunt-related hydrothorax may occur despite an intra-abdominal distal catheter. From a practical standpoint, CSF-related pleural effusion should be suspected in VP-shunted children with unexplained respiratory distress, persistent or recurrent pleural effusion, transudative pleural fluid, or continuous high-volume pleural drainage, even when the distal catheter remains intra-abdominal (6,8,10). Diagnosis should include chest imaging, shunt series or CT, pleural fluid analysis, and CSF-specific testing such as β 2-transferrin or β -trace protein when available (10, 14). Treatment should be individualized and may include pleural drainage, distal catheter revision, temporary shunt externalization or EVD, conversion to VA shunting, or CSF production-reducing procedures such as choroid plexus coagulation in selected cases (8,10).

Conclusion

This case highlights that shunt-related CSF pleural effusion may occur even in the absence of catheter migration or a macroscopic diaphragmatic defect. After exclusion of alternative causes, impaired peritoneal CSF absorption should be considered a presumed mechanism. CSF-related pleural effusion should therefore be suspected in VP-shunted children presenting with unexplained respiratory symptoms or persistent pleural effusion, allowing timely diagnosis and appropriate management.

Contribution of the authors

Study conception and design: TA, MAE, ŞSB; data collection: TA, MAE; analysis and interpretation of results: TA, MAE, ŞSB; draft manuscript preparation: TA, MAE. Critical revision of the manuscript and supervision: MAE, ŞSB. All authors reviewed the results and approved the final version of the article.

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Conflict of interest

The authors declare that there is no conflict of interest.

Patient confidentiality: Written informed consent was obtained from the patient's parent/legal guardian for participation in the study and publication of the patient's clinical information.

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