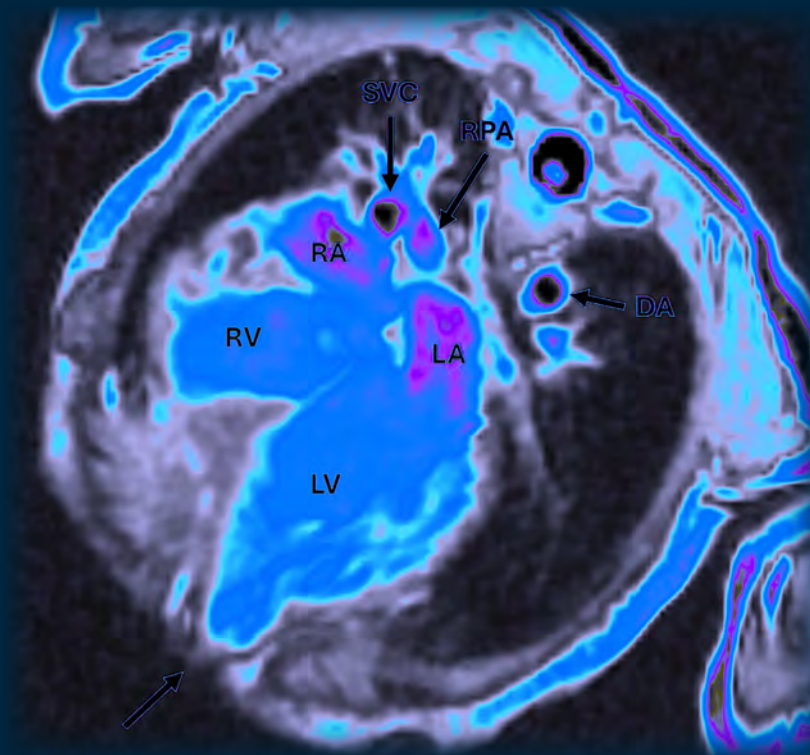


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Cerebral Vein
Thrombosis

Testicular Torsion

Trichobezoar

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Sphenopalatine Block for
Migraine Headaches in
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Cerebral Vein Thrombosis

Zackary C. Kon, BS; Richard B. Towbin, MD; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

Cerebral venous thrombosis is a rare neonatal condition with an incidence of up to 6.4 per 100,000 infants and a mortality rate nearly double that of older children. It encompasses thrombosis of the dural venous sinuses as well as the cerebral veins, often with multivessel involvement. Neonates are particularly susceptible due to an immature hemostatic system, with major risk factors, including congenital heart disease, complicated delivery, hypoxia, and sepsis. Clinical presentation is often nonspecific; however, seizures are the most common manifestation. Cranial ultrasonography with Doppler is the preferred initial imaging modality, while MRI with MR venography is the diagnostic standard for defining thrombus extent and associated parenchymal injury.

Keywords: central nervous system, brain, hematologic

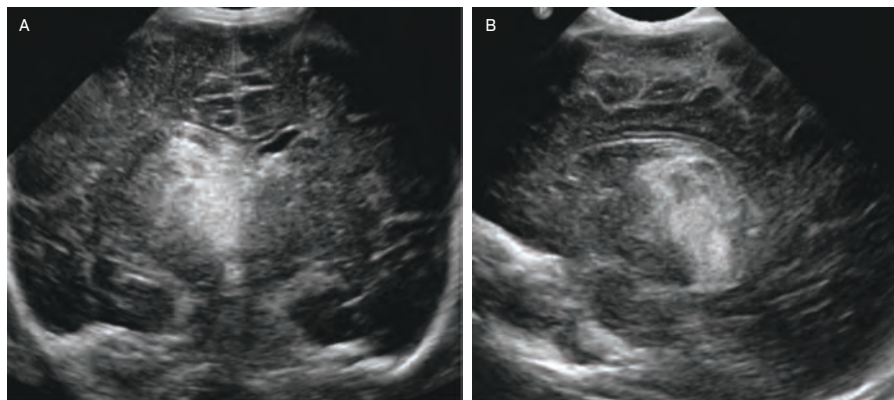
Case Summary

A term neonate was born to a 30-year-old mother with fever (100.7°C), gestational hypertension, and vaginal culture positive for *Mycoplasma* and *Ureaplasma*, confirming chorioamnionitis. At delivery, the APGAR scores were 2 and 9 at 1 and 5 minutes, respectively, and the birth weight was 4.85 pounds. The newborn was afebrile, had grunting respiration with intercostal retraction, tachycardia, and had a seizure at 12 hours of age. Abnormal laboratory values included 9 nucleated red blood cells, an elevated C reactive protein (7.16), and increased bands and neutrophils. This patient was diagnosed with fetal inflammatory response syndrome (FIRS).

Imaging Findings

A head US (HUS) (Figure 1) performed at 22 hours of age showed intracranial hemorrhage involving the area of the

Figure 1. (Head US Coronal (A) and Sagittal (B) performed at 22 hours of age showing intracranial hemorrhage involving the area of the right caudate nucleus and thalamus (arrows) and intra third ventricular blood (not shown). There was associated local mass effect and left to right midline shift.



right caudate nucleus, thalamus, and intra third ventricular blood. There was associated local mass effect and left-to-right midline shift.

MRI (Figure 2) performed on day of life 2 showed intraventricular hemorrhage, involving the right lateral ventricle greater than left, and third ventricle,

intracerebral hemorrhage involving the right basal ganglia and thalamus and diffusion restriction in the same region.

Diagnosis

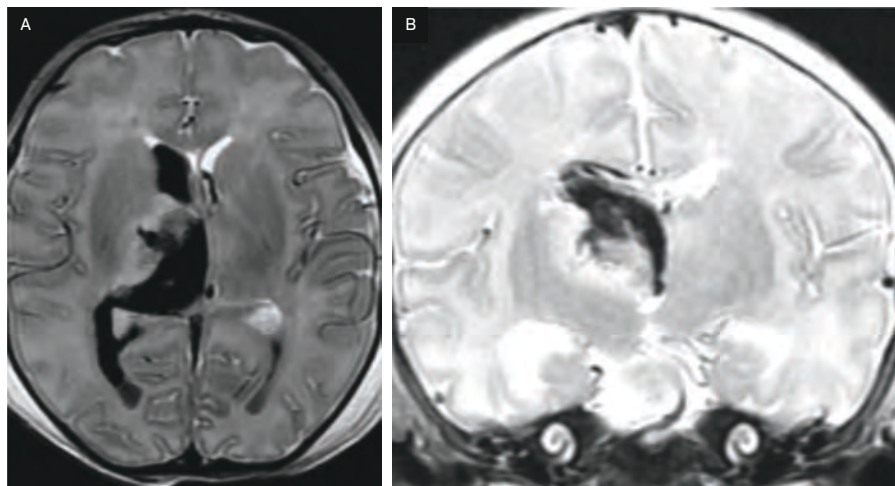
Cerebral venous thrombosis (CVT) involving the internal cerebral vein (ICV).

The differential diagnoses related to the imaging findings include cerebral infarction, grade 4 germinal matrix hemorrhage, coagulopathy, birth trauma, hypoxic-ischemic injury with secondary hemorrhage, FIRS, and vascular malformations.

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Figure 2. MRI performed on day 2 of life. (A, B) Axial and coronal T2-weighted images showing a large hemorrhagic mass bulging into the right frontal horn (arrowhead), bilateral intraventricular hemorrhage (arrow), involving the right lateral ventricle greater than left, and third ventricle, intracerebral hemorrhage involving the right basal ganglia and thalamus and diffusion restriction in the same region.



Discussion

CVT is a rare pediatric condition with an annual incidence of 1.1 per 100,000, increasing to 6.4 per 100,000 in infants and approximately 2.6 to 12/100,000 live births per year.¹ Neonates account for most cases and experience nearly 3 times the mortality rate of older children (9.9% vs 3.5%).^{2,3} CVT encompasses thrombosis of the dural venous sinuses (CVST) and cerebral veins (CVT). Venous sinus thrombosis affects large dural conduits, including the transverse (85%) and superior sagittal sinuses (42%). CVST is most common in the first 28 days and represents 30-50% of pediatric cases. CVT affects smaller vessels, including cortical veins (12%), deep medullary veins (4%), and deep cerebral veins such as the ICV and great cerebral vein (vein of Galen) (each 8%). Multivessel involvement is frequent (65%).⁴ There is a male predominance accounting for about 60% of cases.

Unlike arterial strokes, which result from abrupt arterial occlusion and produce infarction confined to well-defined arterial territories,¹ venous strokes arise from impaired cerebral venous

outflow. The resulting elevation in venous pressure reduces the arteriovenous perfusion gradient, disrupts the blood-brain barriers, and promotes vasogenic edema with frequent hemorrhagic transformation. Consequently, venous infarctions do not conform to arterial boundaries and are more commonly hemorrhagic, reflecting venous hypertension and capillary rupture rather than primary arterial ischemia. Given the extensive collateralization of the cerebral venous system, venous strokes present with an indolent and nonspecific clinical presentation compared with abrupt focal neurological deficits precipitated by arterial occlusion.^{5,6}

While CVST is more readily identified on imaging, the deep cerebral venous system is often underrecognized despite its strong association with hemorrhagic injury and poor outcomes. The ICV serves as the principal drainage pathway for the deep white matter, basal ganglia, thalami, and choroid plexus. Numerous deep medullary veins converge into the subependymal veins, including the thalamostriate, septal, and longitudinal caudate veins, which unite near the foramen of Monroe to form the paired

ICVs. The ICVs course posteriorly within the tela choroidea that forms the roof of the third ventricle, collecting atrial and choroidal tributaries before draining into the vein of Galen and subsequently the straight sinus. Obstruction along this pathway may result in venous congestion, hemorrhagic infarction, and intraventricular hemorrhage.¹

Neonates are particularly vulnerable to CVT and CVST due to an immature hemostatic system with reduced procoagulant and anticoagulant factors, creating a fragile equilibrium predisposing to thrombosis.⁷ Risk factors include congenital heart anomalies (48%), complicated delivery (38%), hypoxia (31%), and inflammation or sepsis (21%).⁸

Clinical presentation is often nonspecific. Up to 27% of preterm infants may be asymptomatic.⁴ Seizures are the most common clinical manifestation (52%), followed by lethargy (36%) and irritability (12%).⁸ Diagnosis is typically made postnatally, with a median age at diagnosis of 10 days of life.¹ In utero CVT is rare and primarily reported as isolated case reports.

Cranial ultrasonography with Doppler is the preferred initial imaging modality in neonates due to its safety profile, lack of ionizing radiation, and ability to demonstrate abnormal increase in parenchymal echogenicity and mass effect and venous distention with intraluminal hyperechoic thrombus.^{9,10} MRI with MR venography (MRV) is the modality of choice for defining thrombus extent and detecting associated parenchymal injury and bleeding. CT and CT venography are acceptable alternatives, though with slightly lower sensitivity (0.79 vs 0.82) and specificity (0.90 vs 0.92).⁶ MRI findings include both direct and indirect signs. Direct visualization of thrombus and intracranial hemorrhage varies by clot age. Acute thrombi (1-4 days) appear iso- to hypointense on T1-weighted images and hypointense on T2-weighted images, potentially mimicking normal flow voids. Subacute thrombi (5-16 days)

demonstrate T1 and T2 hyperintensity due to methemoglobin, while chronic thrombi (>16 days) show variable signal characteristics.¹⁰ Contrast-enhanced or time-of-flight MRV may confirm absent flow in large dural sinuses, though visualization of deep medullary veins remains challenging.¹⁰

Indirect imaging findings reflect venous congestion and parenchymal injury. Superior sagittal sinus thrombosis often produces parasagittal hemorrhage, whereas transverse sinus thrombosis may result in basal temporal, cerebellar, or tentorial hemorrhage. Cortical vein thrombosis may present as a superficial crescentic hematoma with underlying cortical ischemia.¹⁰

Deep venous thrombosis produces characteristic imaging patterns that reflect specific venous drainage territories. Thrombosis of the deep medullary vein results in the “iris pattern,” characterized by radially oriented hypointense linear structures along the lateral ventricles on T2-weighted or susceptibility-weighted imaging representing venous congestion or thrombosis as these veins converge toward the subependymal veins. Thrombosis of the basal vein of Rosenthal produces the “striato-hippocampal pattern,” involving the insula, hippocampus, ventral basal ganglia, ventral thalamus, and choroid plexus of the temporal horn. Thrombosis of the ICVs or straight sinus results in “thalamocaudate pattern,” characterized by infarction or hemorrhage of the ipsilateral thalamus, caudate nucleus, and choroid plexus. Bilateral pattern strongly suggests straight sinus thrombosis. In term neonates, the presence of intraventricular hemorrhage with associated thalamic hemorrhage should prompt evaluation of the ICVs as impaired venous drainage of the choroid plexus via the superior choroidal vein into the ICV is a common mechanism underlying intraventricular hemorrhage in this population.¹⁰

Management is primarily supportive, including seizure control and correction of dehydration, anemia, and infections. Anticoagulation is not routine due to limited evidence; however, current data suggest reduced severity of CVT by nearly 66% (RR, 0.33; 95% CI, 0.18-0.58) and reduction in long-term neurological deficits (odds ratio 0.46, 95% CI 0.23-0.94).¹¹

Neonatal CVT carries higher mortality than in older children (9.9% vs 3.5%), and approximately 48% of survivors experience long-term neurological deficits.³ Poor outcomes are associated with prematurity, traumatic delivery, deep venous involvement, and hemorrhagic infarction, whereas isolated superficial sinus thrombosis and normal neurological examination at presentation predict more favorable outcomes.¹²

Conclusion

CVT is a rare neonatal condition with an incidence of up to 6.4 per 100,000 infants and a mortality rate nearly double that of older children. It encompasses thrombosis of the dural venous sinuses as well as the cerebral veins, often with multivessel involvement. Neonates are particularly susceptible due to an immature hemostatic system, with major risk factors, including congenital heart disease, complicated delivery, hypoxia, and sepsis. Clinical presentation is often nonspecific; however, seizures are the most common manifestation. Cranial ultrasonography with Doppler is the preferred initial imaging modality, while MRI with MRV is the diagnostic standard for defining thrombus extent and associated parenchymal injury.

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Testicular Torsion

Tamer I. Elashyi, MS; Richard B. Towbin, MD; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

Testicular torsion is a cause of acute scrotal pain and represents a true surgical emergency. Torsion of the spermatic cord compromises blood flow to the testis and, if not promptly addressed, can lead to irreversible ischemic damage. Timely surgical exploration and orchiopexy are essential for testicular salvage. Color Doppler US serves as a valuable diagnostic tool; particularly, clinical findings are equivocal.

Keywords: genitourinary, scrotum, malposition

Case Summary

An adolescent male presented with 4 days of left testicular pain and swelling.

Imaging Findings

US of the scrotum (Figure 1) showed the left testis to be enlarged with heterogeneous echotexture and no color Doppler flow.

Diagnosis

Testicular torsion.

The differential diagnosis for acute scrotal pain in children and adolescents includes testicular torsion, torsion of the testicular or epididymal appendage, epididymo-orchitis, and trauma.

Discussion

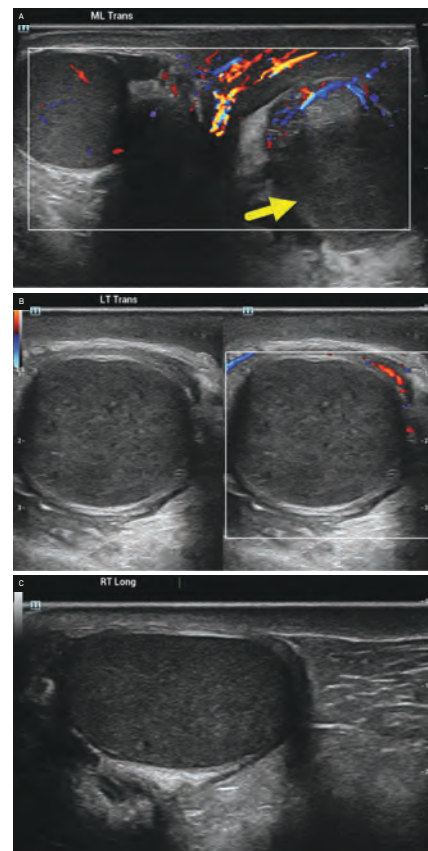
Testicular torsion is a relatively rare but urgent condition, affecting approximately 5.9 per 100,000 males under the age of 18 each year.¹ It occurs when the testis and spermatic cord twist, leading to reduced

arterial perfusion and obstructed venous drainage. This vascular compromise can quickly progress to testicular ischemia if not promptly addressed. Torsion most often occurs in males with a bell-clapper deformity, a congenital abnormality in which the testis lacks normal posterior anchoring and is free to rotate within the tunica vaginalis.²

Testicular torsion most commonly affects prepubertal and adolescent males. It is more common after 10 years of age as rapid pubertal growth is considered a contributing factor.² Additional risk factors include an undescended testis, prior episodes of intermittent torsion, scrotal trauma, and the presence of a bell-clapper deformity. Patients typically present with sudden onset of severe unilateral testicular pain, often accompanied by nausea, vomiting, and sometimes lower abdominal or scrotal pain.^{3,4}

To aid in clinical decision-making, the Testicular Workup for Ischemia and Suspected Torsion (TWIST) score⁵ can be used to stratify the risk of torsion based on physical exam findings. The 7-point scoring system includes testicular

Figure 1. (A) Transverse color Doppler US of the scrotum showing the left testis (arrow) to have absent internal blood flow. There is relative surrounding hyperemia. (B) Transverse image of the left testis with and without color Doppler showing the testis to be enlarged, have a mildly heterogeneous echotexture, and mild surrounding hyperemia. (C) Longitudinal US image of the normal right testis for comparison.



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swelling (2 points), hard testis (2 points), absent cremasteric reflex (1 point), nausea or vomiting (1 point), and high-riding testis (1 point).⁵ Scores of 0-2 indicate low risk, 3-4 intermediate risk, and 5-7 high risk for the presence of torsion. US is typically reserved for patients in the intermediate-risk group, while high-risk patients may proceed directly to surgical exploration.⁶ The TWIST score demonstrates high diagnostic performance, with a sensitivity of 95.5%, specificity of 97.2%, positive predictive value of 93%, and negative predictive value of 97%.^{4,6}

US is the primary imaging modality used to diagnose testicular torsion. Grayscale findings in acute torsion may include testicular enlargement, abnormal orientation of the affected testis within the scrotum, and a spermatic cord knot representing the site of twisting. In the early phase of torsion, the testis may appear homogeneously echogenic. As ischemia progresses, the echotexture becomes increasingly heterogeneous.

Color flow Doppler US is more sensitive for definitive diagnosis. The hallmark finding is absent or significantly decreased intratesticular blood flow. Another key diagnostic feature is the whirlpool sign, which represents blood flow within a twisted spermatic cord.⁷ A positive finding warrants immediate urologic consultation to prevent irreversible ischemic damage.

Prompt restoration of blood flow to the ischemic testis is critical, with irreversible damage typically occurring after 4-8

hours of torsion.⁸ Manual detorsion may be attempted when immediate surgical intervention is unavailable, but its success rate is variable, ranging from 26% to 80%. Despite this, surgical intervention remains the gold standard and should always be prioritized over manual detorsion.

Manual detorsion is an emergency bridge to surgery procedure that is indicated to urgently restore blood flow to a torsed testicle. It usually involve laterally rotating the testes since most torsions are medially rotated. Therefore, the left testes is rotated in a clockwise manner and the right counterclockwise. The end point is to decrease pain and make the spermatic cord feel normal. This usually occurs after 180-720 degrees of rotation.⁸ Surgical management includes detorsion and orchiopexy, in which the testis is sutured to the scrotal wall to prevent recurrence. If the testis is nonviable, orchiectomy is performed. In cases where a bell-clapper deformity is identified, bilateral orchiopexy is recommended, as the anomaly is bilateral in up to 80% of patients.⁹

Conclusion

Testicular torsion is a cause of acute scrotal pain and represents a true surgical emergency. Torsion of the spermatic cord compromises blood flow to the testis and, if not promptly addressed, can lead to irreversible ischemic damage. Timely surgical exploration and orchiopexy

are essential for testicular salvage. Color Doppler US serves as a valuable diagnostic tool; particularly clinical findings are equivocal.

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Trichobezoar

Nikolas Tsiouplis, BS; Richard B. Towbin, MD; Douglas C. Rivard, DO; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

Trichobezoar is a collection of hair and undigestible matter found in the stomach or small intestine. Patients are often asymptomatic for years before the condition is diagnosed, either through incidental imaging or an acute complication. Common symptoms are abdominal pain and vomiting, with serious complications, including intestinal obstruction, ulceration, peritonitis, and visceral perforation. Surgery is the key treatment modality, with laparoscopic techniques becoming the preferred method. Radiograph and US are helpful for diagnosis, but CT is the most useful imaging test. Psychiatric management is critical in these patients to prevent recurrence.

Keywords: gastrointestinal, intestine, foreign body

Case Summary

A teenage girl presented with abdominal pain and loss of appetite. On physical exam, the abdomen was soft with a palpable mass in the epigastric region.

Imaging Findings

Figure 1: Scout image from CT scan showing distended stomach (arrows) with mixed density intragastric contents conforming to the gastric lumen shape (star). Figure 2: Axial non enhanced CT of the abdomen showing intragastric mass with mixed density and attenuation diagnostic of bezoar

Diagnosis

Trichobezoar.

In children, the imaging differential diagnosis includes gastric masses, including lymphoma and neuroblastoma, and other types of bezoars, such as

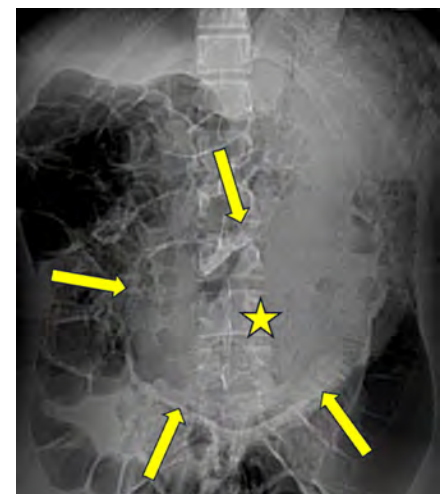
phytobezoar or pharmacobezoar and foreign bodies.

Discussion

Bezoars are compactions of foreign materials within the gastrointestinal (GI) tract.¹ A trichobezoar is characterized by a conglomeration of hair.¹ Trichobezoars can be classified based on their location—gastric, small bowel, or Rapunzel syndrome,² with gastric trichobezoar being the most common.² Rapunzel syndrome is a rare but serious complication of trichobezoar in which a long tail extends from the stomach into the small bowel.³

Trichobezoar is a rare condition, comprising only 6% of all bezoars,⁴ but it most commonly presents in young females. A case series of 17 patients reported median age of presentation at 7 years and a 1:4.7 M:F ratio.⁴ Trichophagia, a psychiatric condition characterized by the consumption of hair,⁴ Trichophagia

Figure 1. Scout image from abdominal CT scan showing distended stomach (arrows) with mixed density intragastric contents conforming to the gastric lumen shape (star).



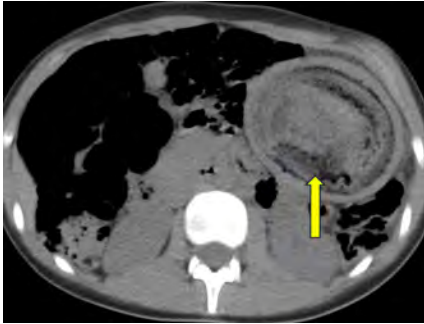
is commonly associated with trichotillomania, an impulse-control disorder in which patients compulsively pull out their own hair.⁵

Patients are usually asymptomatic for several years before the onset of GI symptoms.⁶ Common presenting symptoms include abdominal pain, vomiting, abdominal distension, hair in the feces, and a palpable epigastric mass.⁴ Serious complications include intestinal obstruction, gastric ulceration, acute peritonitis, malnutrition, and bowel perforation.^{2,4} Bowel perforation is likely

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Figure 2. Axial nonenhanced abdominal CT showing an intragastric mass with mixed density and an inner rim of gas bubbles (speckled appearance) with the attenuation diagnostic of bezoar.



a result of pressure necrosis of the gastric or small intestinal wall leading to ulceration and eventual perforation, most often involving the lesser curvature. Consequently, surgery is the recommended treatment for this condition. In the past, open surgery with gastrotomy or enterotomy was the most common technique for trichobezoar removal.⁴ Currently, laparoscopic or endoscopic methods have been described and increasingly utilized because of their minimally invasive nature.^{7,8} In severe cases with jejunal perforation, surgical options include intestinal repair or resection with anastomosis.⁹

Multiple imaging modalities can be used to confirm the diagnosis of trichobezoar when paired with clinical findings. A gastric-shaped, partial opacification will be observed on plain radiograph.⁴ An upper GI series shows one or more, mobile gastric filling defects that mold to the gastric luminal contour and can be associated with a high-grade obstruction.⁴ Abdominal US may show a hyperechoic mass in the stomach or small bowel with posterior shadowing as a result of the air within the mass.⁹ CT is a superior diagnostic exam that can be utilized to characterize the size and location of the mass and determine if

obstruction or other complications are present.³ On CT, trichobezoar exhibits low attenuation with a “mottled gas trapping giving it a speckled” appearance similar to particulate matter.^{10,11} The mass is generally heterogeneous and contains interspersed gas.¹ In an examination with contrast, the intraluminal mass will be surrounded by the contrast.¹¹ Endoscopy is diagnostic in cases of gastric trichobezoar.⁸ In cases where the type of bezoar is unclear prior to surgery, the diagnosis can be made by gross pathology after specimen extraction.⁴

Rapunzel syndrome can be a serious complication of trichobezoar, with postoperative mortality of 8%, which is attributed to surgical complications and sepsis.^{4,12} To prevent recurrence, treatment of the underlying psychiatric disorder is advised. Patients with trichotillomania should receive behavioral therapy and pharmacologic treatment with selective serotonin reuptake inhibitors⁵ (fluoxetine, escitalopram).⁵ Unfortunately, there is weak evidence of direct benefit. It appears that behavior therapy is more effective.

Conclusion

Trichobezoar is a collection of hair and undigestible matter found in the stomach or small intestine. Patients are often asymptomatic for years before the condition is diagnosed, either through incidental imaging or an acute complication. Common symptoms are abdominal pain and vomiting, with serious complications including intestinal obstruction, ulceration, peritonitis, and visceral perforation. Surgery is the key treatment modality, with laparoscopic techniques becoming the preferred method. Radiograph and US are helpful for diagnosis, but CT is the most useful

imaging test. Psychiatric management is critical in these patients to prevent recurrence.

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Ectopia Cordis

Stephen Yao, BS; Richard B. Towbin, MD; Ranjit Philip, MD; Jason Johnson, MD; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

This case illustrates the critical importance of early prenatal diagnosis, multidisciplinary coordination, and multimodality imaging in the management of ectopia cordis. Advanced imaging not only facilitates accurate diagnosis but also plays a central role in guiding resuscitative strategies and surgical planning. Cardiac MRI provides complementary information to the CT scan with advantages of excellent intracardiac detail (especially for complex anatomy such as double outlet right ventricle, which is commonly seen in ectopia cordis) and functional/flow information.

Keywords: cardiothoracic, cardiac, congenital

Case Summary

A Gravida 1 woman was referred to the maternal-fetal medicine clinic following abnormal findings on routine prenatal US. Sonographic evaluation revealed a structurally abnormal anterior chest wall defect with a beating heart visualized partially outside the thoracic cavity, consistent with ectopia cordis. Further evaluation revealed an omphalocele and a suspected diaphragmatic defect. Subsequent detailed fetal echocardiography demonstrated associated complex intracardiac anomalies, including atrioventricular canal defect with double-outlet right ventricle and pulmonary atresia. A core multidisciplinary team convened to plan the delivery of this high-risk infant, led by maternal-fetal medicine and involving fetal cardiology, neonatology, pediatric cardiothoracic surgery, and pediatric surgery.

She was born at term with expected SaO₂ of 75-80%, and prostaglandins was

initiated for ductal dependent pulmonary blood flow. The prenatal diagnosis was confirmed by transthoracic echocardiography (TTE), and she was palliated with a Blalock-Taussig-Thomas shunt for pulmonary blood flow. During her initial hospitalization, she also underwent omphalocele repair. Other comorbidities included bilateral hydronephrosis of the kidneys.

Planning for further surgical palliation involved multimodality imaging, including cardiac MRI, cardiac catheterization, and CTA, which confirmed a large anterior chest wall defect with cardiac herniation and associated intracardiac defects.

Imaging Findings

The initial postnatal TTE at bedside confirmed the prenatal diagnosis of double outlet right ventricle, with the aorta arising from the right ventricle and a large ventricular septal defect with

Figure 1. Postnatal transthoracic echocardiogram 2-D 4-chamber view in systole showing the relative size of ventricles and the large ventricular septal defect (*). The right ventricle gives rise to the aorta. Ao, aorta; LA, left atrium; LV, left ventricle; RV, right ventricle.



pulmonary atresia (Figure 1). Cardiac catheterization was done for hemodynamic assessment, which also revealed the dagger-like extension of the left ventricle beyond the sternum (Figure 2) that was also demonstrated with CTA (Figure 3). Cardiac MRI confirmed the moderately dilated left ventricle with its apex below the skin extending beyond the sternum and the right ventricle giving rise to the great arteries (Figures 4, 5).

Diagnosis

Ectopia cordis.

Ectopia cordis may occur as an isolated anomaly or as part of a broader syndromic spectrum, most notably the pentalogy of Cantrell or limb-body-wall complex. In

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Figure 2. Cardiac catheterization angiogram: left ventricle (10 mL at 15 mL/second via power injection in straight anteroposterior projection). A catheter courses retrograde through the aortic arch into the right ventricle across a ventricular septal defect into the left ventricle. The contrast flows from the left ventricle through a large conoventricular (outlet) ventricular septal defect into the right ventricle and out the aorta. The apex of the left ventricle extends inferiorly coming to a sharp point below the sternum (arrow). The apex is also positioned very anteriorly in the chest wall. Ao, aorta; LV, left ventricle; RV, right ventricle.

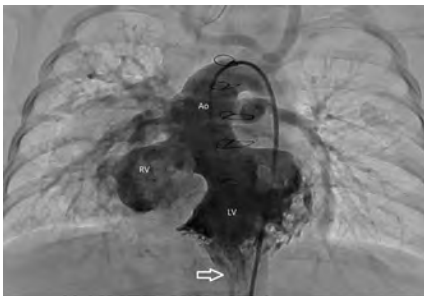


Figure 3. Sagittal contrast-enhanced CT angiogram of the chest demonstrating the left ventricular herniation through the anterior chest wall defect (arrow). LV, left ventricle.



this case, the involvement of the heart, pericardium, diaphragm, sternum, and abdominal wall makes the diagnosis of pentology of Cantrell.

Van Allen et al, Russo et al, and many others have outlined various criteria for the diagnosis of limb-body-wall complex, also known as cylosomas. Limb-body-wall complex is an anomaly consisting of 2 of the following 3 fetal anomalies: (1) thoraco-abdominoschisis or abdominoschisis, (2) limb defects, and (3)

Figure 4. 4-chamber SSFP at end-diastole. The left ventricle apex is just below the skin through a defect in the sternum and ribs (arrow). The right ventricle is normal in size with the left ventricle moderately dilated. The left and right atrium are normal in size. DA, descending aorta; LA, left atrium; LV, left ventricle; RA, right atrium; RPA, right pulmonary artery; RV, right ventricle; SVC, superior vena cava.

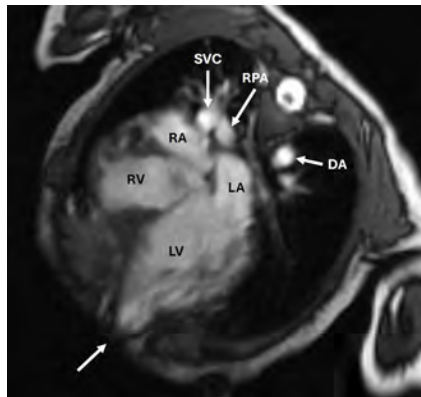
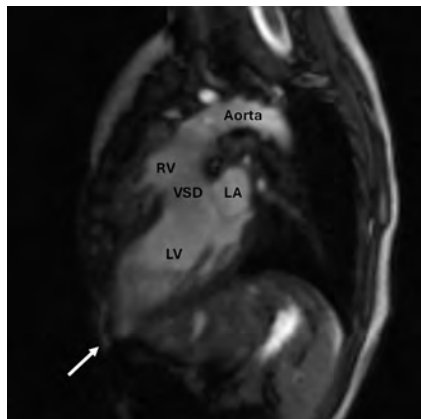


Figure 5. Left ventricular outflow tract SSFP at end-diastole. The left ventricle apex is just below the skin through a defect in the sternum and ribs (arrow). The left ventricle is moderately dilated, and the right ventricle is normal in size. The aorta arises from the right ventricle. There is a large ventricular septal defect. LA, left atrium; LV, left ventricle; RV, right ventricle; VSD, ventricular septal defect.



cranio-facial abnormalities, such as cleft lip/palate, encephalocele, or exencephaly.

Discussion

Ectopia cordis is a rare and severe congenital malformation characterized

by partial or complete extrusion of the heart outside the thoracic cavity, resulting from failure of midline mesodermal fusion during early embryogenesis.¹⁻⁴ It is frequently associated with intracardiac defects and extracardiac anomalies such as omphalocele and diaphragmatic hernia, contributing to its historically poor prognosis.

In this case, the pulmonary atresia necessitated early palliative cardiac surgery to provide pulmonary blood flow. The prenatal diagnosis was crucial for preparing for immediate postnatal resuscitation and management, including surgical planning. However, until 1981, ectopia cordis could only be diagnosed upon delivery, and prior to 1988, there were no reported cases of prenatal diagnosis of this malformation before the 3rd trimester of gestation.⁵

In the modern day, ectopia cordis can be diagnosed as early as 10 weeks of gestation, but it is typically diagnosed at the beginning of the 2nd trimester via a fetal echocardiogram, which remains the primary diagnostic tool.^{5,6} The combination of fetal echocardiography and MRI enables precise prenatal characterization, facilitating delivery planning, resuscitation preparedness, and multidisciplinary coordination. While fetal echocardiography remains the cornerstone of diagnosis, fetal MRI provides superior delineation of extracardiac anatomy and spatial relationships but may not be routinely available at all centers.

Postnatally, TTE plays a central role in delineating intracardiac anatomy and assessing hemodynamics, including ventricular function and associated structural defects such as ventricular septal defects. Cardiac MRI provides enhanced spatial resolution and tissue characterization, allowing for improved visualization of extracardiac relationships and more comprehensive functional assessment. CTA is particularly valuable in preoperative planning, as it offers high-resolution delineation of both

cardiovascular and extracardiac anatomy. In addition, CTA enables accurate assessment of thoracic cavity dimensions and lung volumes, which are critical in determining the feasibility, timing, and surgical approach for repair in patients with ectopia cordis.⁷ Despite advances in prenatal diagnosis and perioperative care, outcomes remain guarded and are highly dependent on the extent of cardiac exposure, associated anomalies, and feasibility of surgical reconstruction. Overall, the survival rate is approximately 10% depending on gestational age and associated cardiac and extracardiac anomalies. This case highlights the evolving role of multimodality imaging in optimizing early management strategies for this rare but life-threatening condition.

Conclusion

This case illustrates the critical importance of early prenatal diagnosis,

multidisciplinary coordination, and multimodality imaging in the management of ectopia cordis. Advanced imaging not only facilitates accurate diagnosis but also plays a central role in guiding resuscitative strategies and surgical planning. Cardiac MRI provides complementary information to the CT scan with advantages of excellent intracardiac detail (especially for complex anatomy such as double outlet right ventricle, which is commonly seen in ectopia cordis) and functional/flow information.

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Osteogenesis Imperfecta

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Abstract

Osteogenesis imperfecta is caused by mutations in the COL1A1 or COL1A2 genes, resulting in defects in collagen structure or quantity. The clinical spectrum ranges from mild bone fragility to perinatal lethality. Clinical presentation is variable and may include recurrent fractures, osteopenia, hearing loss, vascular fragility, blue-gray sclera, and skeletal deformities. Prenatal diagnosis and monitoring typically involve US and chorionic biopsy with genetic testing, while postnatal evaluation includes radiography, dual-energy x-ray absorptiometry, and confirmatory genetic testing. Management focuses on reducing fracture risk and improving function through bisphosphonates, physical therapy, and surgical stabilization.

Keywords: musculoskeletal, bones, congenital, syndrome

Case Summary

A 5-week-old neonate presented to the emergency department with decreased movement of the right lower extremity.

Imaging Findings

Initial radiograph of the right femur (Figure 1) revealed a transverse, angulated fracture. A subsequent skeletal survey (Figure 2) demonstrated multiple rib fractures, a left clavicle fracture, a fracture through the C2 vertebral body, and numerous Wormian bones within the skull. Head CT (Figure 3) confirmed the C2 vertebral body fracture and multiple Wormian bones and additionally showed absence of the lamina dura surrounding unerupted teeth.

Diagnosis

Osteogenic imperfecta.

The differential diagnosis in a child with frequent fractures includes nonaccidental trauma, idiopathic juvenile osteoporosis, hypophosphatemia rickets, osteopetrosis with renal tubular acidosis, as well as various skeletal disorders such as Cole-Carpenter syndrome, Bruck syndrome, severe fibrous dysplasia of bone, and idiopathic autosomal recessive hypophosphatasia. In infants, additional differential diagnoses can include achondrogenesis type 1 and thanatophoric dysplasia.

Discussion

Osteogenesis imperfecta, also known as brittle bone disease, is most commonly an autosomal-dominant disorder, occurring in approximately 1 in 15,000-20,000 live births.¹ The condition encompasses multiple subtypes, with severity ranging from mild bone fragility to perinatal lethality in type 2 disease.² An overview of the 4 most common subtypes is included in Table 1.

Figure 1. Radiograph of the right femur showing a transverse fracture of the mid diaphysis.



Most cases are caused by mutations in the COL1A1 and COL1A2 genes, which encode type 1 collagen, leading to either reduced collagen production or structural abnormalities.² In osteogenesis imperfecta type 1, frameshift mutations with premature stop codons result in decreased collagen synthesis, a thinner bone matrix, and impaired skeletal mineralization.² In contrast,

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Figure 2. (A) Frontal image of the chest, (B) oblique view of the chest, (C) and lateral view of the skull and cervical spine obtained as part of a skeletal survey show multiple healing rib fractures (arrowheads), including multiple fractures within the same rib, a left clavicle fracture (arrow), a fracture through the pedicles of C2 (white arrow), and multiple Wormian bones (dashed arrow).

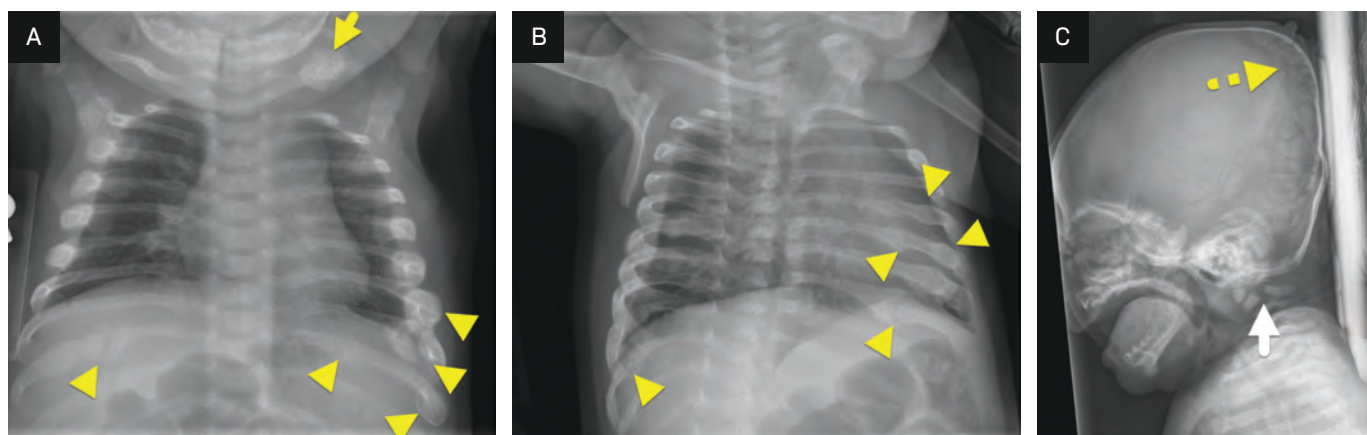
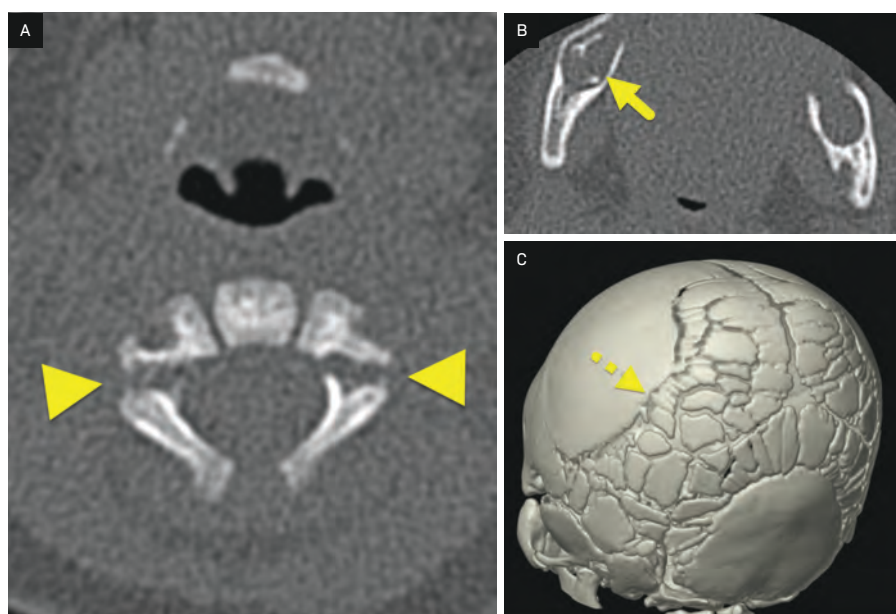


Figure 3. (A) Axial CT image through the C2 vertebral body showing a fracture of both pedicles (arrowheads). (B) Axial CT image through the mandible showing the absence of the normal lamina dura surrounding the teeth (arrow). (C) 3-dimensional reconstruction of the skull showing multiple Wormian bones (dashed arrow).



types 2-4 are typically associated with substitution or deletion mutations that produce structurally defective collagen, causing bone fragility and abnormal bone matrix formation.²

Clinically, osteogenesis imperfecta is characterized by recurrent fractures, often beginning in early childhood with the onset of ambulation.¹ Additional skeletal findings include osteopenia, short stature, scoliosis, pectus excavatum, and hearing

loss.³ Beyond the skeleton, the connective tissue disorder affects multiple organ systems, leading to features such as blue or gray sclera, dentinogenesis imperfecta with translucent teeth, joint hyperlaxity, restrictive pulmonary disease, sensory and motor disturbances, vascular fragility, and nephrolithiasis.^{2,3}

Diagnosis of osteogenesis imperfecta involves a combination of clinical evaluation, imaging, and genetic testing.

In patients with a known family history, prenatal evaluation typically begins with chorionic villus sampling and DNA sequencing.⁴ Alternatively, incidental findings on routine prenatal US, such as bone shortening, decreased bone echogenicity, fractures, and rib beading, may raise suspicion for osteogenesis imperfecta and prompt further genetic testing.⁵ US can also be used to monitor for complications in fetuses known to have the condition.⁴ Transvaginal US is preferred for its ability to detect skeletal abnormalities as early as 14 weeks of gestation, with 3-dimensional imaging offering superior diagnostic detail when combined with 2-dimensional images.^{4,6} Fetal MRI can provide additional information, particularly for evaluating extra-skeletal complications such as restricted lung volume in osteogenesis imperfecta type 2.⁴

When diagnostic evaluation begins postnatally, radiography is typically the initial imaging modality of choice. Skeletal surveys are particularly useful for monitoring patients with osteogenesis imperfecta, especially during periods of rapid growth. Radiographic findings may include fractures and diaphyseal bone deformities, flattening of the skull base, basilar invagination, multiple Wormian bones, rib beading, and generalized osteopenia.^{4,6} In osteogenesis imperfecta type 3, characteristic features

Table 1. Overview of the 4 Most Common Subtypes of Osteogenesis Imperfecta, Including Inheritance Pattern, Pathophysiology, Unique Clinical Features, and Typical Severity of Presentation

Subtype	Inheritance	Pathophysiology	Unique Clinical Features	Severity
Type 1	Autosomal dominant	Reduced amount of collagen with normal structure	Minimal deformities, blue sclera, conductive hearing loss, mildly impaired growth, dentinogenesis imperfecta in subtypes 1B/1C	Mild
Type 2	Autosomal dominant OR recessive	Severe disruption of collagen function	Extreme bone fragility, beaded ribs, crumpled femora on radiograph	Severe to lethal
Type 3	Autosomal dominant OR recessive	Reduced amount and/or structural dysfunction of collagen	Blue sclera in infancy, severe bone fragility with progressive deformities, basilar invagination, brittle teeth	Mild to severe
Type 4	Autosomal dominant OR recessive	Reduced amount and/or structural dysfunction of collagen	Normal sclera, moderate to severe bone deformity, impaired growth, dentinogenesis imperfecta in subtype 4B	Moderate to severe
There are at least 17 additional subtypes that have been identified. ¹⁻⁵				

include a “bamboo cane” appearance of abnormally remodeled long bones and “popcorn” calcifications around the knees.⁴ Definitive genetic diagnosis of osteogenesis imperfecta is confirmed via skin biopsy.⁴

Because radiography lacks sensitivity for assessing bone density, dual-energy x-ray absorptiometry (DXA) is often used to supplement evaluation and monitoring of osteopenia in patients with osteogenesis imperfecta.⁵ CT and MRI are not routinely used but may be helpful for better visualization of cranial structures or soft tissues in select cases.^{4,5}

As there is no known cure for osteogenesis imperfecta, management relies on a multidisciplinary approach aimed at reducing fractures and maximizing mobility. Bisphosphonates, such as cyclic intravenous pamidronate and oral risedronate, are commonly used to increase bone density and

reduce fracture risk.¹ Surgical stabilization with intramedullary rods and physical therapy are also frequently employed.¹ Emerging treatments under investigation include mesenchymal stem cell therapy, setrusumab (a sclerostin-neutralizing antibody), and bone marrow transplantation.^{1,7,8}

Conclusion

Osteogenesis imperfecta is caused by mutations in the COL1A1 or COL1A2 genes, resulting in defects in collagen structure or quantity. The clinical spectrum ranges from mild bone fragility to perinatal lethality. Clinical presentation is variable and may include recurrent fractures, osteopenia, hearing loss, vascular fragility, blue-gray sclera, and skeletal deformities. Prenatal diagnosis and monitoring

typically involve US and chorionic biopsy with genetic testing, while postnatal evaluation includes radiography, DXA, and confirmatory genetic testing. Management focuses on reducing fracture risk and improving function through bisphosphonates, physical therapy, and surgical stabilization.

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Sphenopalatine Block for Migraine Headaches in Children

Noah A. Ivak, BA; Richard B. Towbin, MD; Carrie M. Schaefer, MD; Alexander J. Towbin, MD

Abstract

Migraine headaches can significantly impact both the physical and psychological well-being of children and adolescents, and have been shown to have a considerable prevalence within this population. It is therefore reasonable to further explore treatment modalities that can alleviate or resolve migraine symptoms. Sphenopalatine ganglion block represents one of these modalities and has demonstrated promising and successful outcomes when treating migraines in the pediatric population.

Keywords: head and neck, face, headaches, interventional radiology

Case Summary

A teenage girl presented with a history of chronic migraine headaches that interfere with her daily activities.

Imaging Findings

A sphenopalatine ganglion (SPG) block was performed under intermittent lateral fluoroscopic guidance with the patient in a supine position and neck extended. The nasal mucosa was locally anesthetized bilaterally using 4% lidocaine spray, followed by 2% viscous lidocaine 5 minutes later. The SphenoCath (Figure 1) was positioned over the middle turbinate and contrast was injected to confirm satisfactory catheter tip position. Then, 2 ccs of 4% lidocaine was slowly injected, and the patient was asked not to swallow to retain local anesthetic in the pterygopalatine fossa (Figure 2).

Figure 1. SphenoCath (Dolor Technologies, Clarefield, Utah).



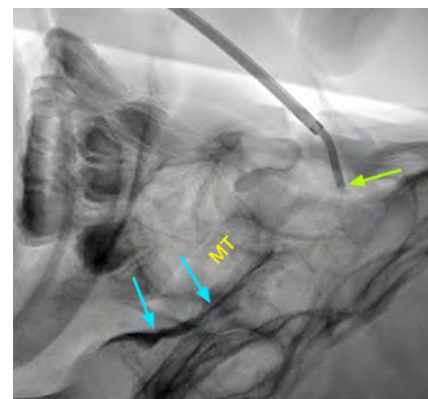
Diagnosis

Migraine Headache.

Discussion

The SPG is the largest of the 4 parasympathetic ganglia of the head and neck. Other common names synonymous for SPG include pterygopalatine ganglion, Meckel ganglion, or nasal ganglion. The SPG resides in the pterygopalatine fossa that is located at or above the level of the posterior to the lateral insertion of the middle nasal turbinate and maxillary sinus. The SPG is approximately 1.0-1.5 mm deep to the posterior nasal mucosa. The SPG receives 3 main inputs: sensory, parasympathetic, and

Figure 2. (A) Coned lateral fluoroscopic spot image demonstrating that the catheter tip (green arrow) is in a satisfactory position above the middle turbinate. Contrast outlining the nasopharynx (teal arrows) with confirming satisfactory position prior to injection of 2% lidocaine. MT, middle turbinate.



sympathetic.¹ The sensory input originates from the maxillary nerve, which gives off 2 ganglionic branches to the SPG, largely without synapsing, and leading into the greater and lesser palatine nerves that are responsible for sensation in the palate, uvula, tonsils, soft palate, and oral mucosa.^{1,2} The sympathetic input arises from the superior cervical ganglion, through the deep petrosal nerve, which, in turn, joins the greater petrosal nerve to form the Vidian nerve. The sympathetic fibers do not synapse in the SPG. The

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last input is the parasympathetic fibers that originate in the superior salivatory nucleus of the brainstem and eventually lead into the greater petrosal nerve and then the Vidian nerve.^{1,2} These parasympathetic fibers do synapse in the SPG and give rise to post-ganglionic parasympathetic fibers that innervate the lacrimal gland, nasal mucosa, and meningeal vessels.^{2,3} Parasympathetic innervation of the meningeal vessels can lead to vasodilation and neurogenic inflammation, leading to activation of trigeminal nociceptors, which, in turn, can cause headache pain.³ Therefore, using a block to inhibit the SPG may attenuate this mechanism of meningeal vasodilation and neurogenic inflammation, providing a basis for headache relief. Given this anatomical framework, it is reasonable to hypothesize that targeting the SPG can have an influence on chronic headache pain. Accordingly, several studies have investigated whether modulation of the SPG can alleviate or resolve headache symptoms.⁴

The literature shows improvement of symptoms of migraines using SPG-targeted treatment modalities. Targeting the SPG results in pain reduction for cluster headaches (CH) and migraines in adults, either via anesthetic block (Ansarinia et al) or electrical stimulation (Tepper et al).^{5,6} Tepper et al reported stimulation of the SPG resulted in 61% of CH completely resolving within minutes, and 77% of the patients with CH experienced a >50% reduction in headache intensity.⁵ Ansarinia et al reported 10 patients with migraines who had the SPG blocks and 2 patients were pain-free following 3 minutes of stimulation, and 3 of the patients had pain reduction.⁶ It is shown that targeting the SPG can both affect CHs and migraines. While these findings are compelling in the adult population, evidence in pediatric populations is less available.

When compared with adults, SPG treatment modalities for migraines in the pediatric population are sparse. Migraines

in the pediatric population can have a significant toll on the well-being of the patient. In a meta-analysis, it was shown that the prevalence of migraine in children and adolescents was 11%, showing a significant portion of this population being affected, warranting further research into treatment modalities.⁷ SPG block is a relatively safe and minimally invasive procedure that can be used to treat migraine headaches. A retrospective study found that out of a total of 489 SPG blocks that were performed on patients (ages 6-26 years) who had migraine or status migrainosus, 100% of them achieved pain reduction with a mean preprocedural pain score of 5.7 and a post-procedure pain score of 2.4 assessed using the visual analog scale.⁸ Lastly, in a prospective study of 17 adolescents (mean age of 14 years, 82% female) who completed the SPG block series for chronic headaches, 59% reported meaningful improvement, with 53% of the population maintaining this improvement 3 months after.⁹

SPG block in pediatric patients can be initiated using a phased local anesthesia approach using aerosolized 4% lidocaine spray and 2% lidocaine gel at different time intervals and using different methods of delivery (eg, cotton swab).⁸ This approach was successful in mitigating the specific anxiety and nervousness of children and adolescents, to allow for insertion of the SphenoCath (Dolor Technologies, Clarefield, Utah) into the nares. The SphenoCath is advanced utilizing intermittent fluoroscopic guidance and brought to the anterosuperior nasal cavity where the curved inner catheter is deployed superior to the middle nasal turbinate. 1 mL of isovue-300 is injected to confirm the transit of fluid towards the pterygopalatine fossa, and once confirmed, 2 mL of 4% lidocaine is slowly injected.⁸ Following removal of the catheter, the patient is maintained supine with neck extension for 10 minutes to allow for proper absorption and full contact of the administered lidocaine through the nasal mucosa.⁸

In Kouri et al, a non-image-guided approach was used. 0.3 mL of 0.5% bupivacaine was administered in each naris while the patient was in an upright seated position using a Tx360 nasal applicator device (Tian Medical, Libertyville, Illinois). The tip of the catheter is intended to be positioned posterior to the inferior nasal turbinate, although, without imaging confirmation, the catheter tip location is uncertain. The Tx360 device sprays anteriorly, superiorly, and laterally in order to fully cover the SPG.⁹ It is important to specifically note that the 2 studies use different anatomical landmarks: the image-guided approach positions the catheter tip adjacent to the expected location of the SPG with the patient in supine position, while the other delivers anesthetic while the patient is seated upright with uncertain catheter position and a wide spray zone is created to locally anesthetize the SPG.^{8,9} These differences are a reflection of the variation in delivery method and guidance used. Although typically well-tolerated, SPG block can result in some minor adverse effects.

Adverse effects resulting from SPG block are typically local and transient. Reported side effects include nasal or palatal numbness, ipsilateral tearing, and an unpleasant taste. Epistaxis, bleeding, and infection have also been described, particularly with injection-based approaches.⁴ Overall, SPG block appears to be a safe and well-tolerated procedure for migraine treatment in pediatric patients.

Conclusion

Migraine headaches can significantly impact both the physical and psychological well-being of children and adolescents and has been shown to have a considerable prevalence within this population. It is therefore reasonable to further explore treatment modalities that can alleviate or resolve migraine symptoms. SPG block represents one of these modalities and has demonstrated promising and successful

outcomes when treating migraines in the pediatric population.

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