Management Interventions for Hereditary Hemorrhagic Telangiectasia with Complications of Pulmonary Arteriovenous Malformations and Brain Arteriovenous Malformations: Case Report

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ABSTRACT

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant disorder characterized by the presence of vascular abnormalities that impact many organ systems. We present a case of a 6-year-old child diagnosed with HHT, who has experienced repeated epistaxis and had a family history of the condition. The patient complained of hypoxemia and seizures, Chest CT and cranial MRI confirmed the presence of both pulmonary and cerebral arteriovenous fistulas. A genetic test indicated an ENG gene mutation, which led to the diagnosis of HHT type I. Both the pulmonary and intracranial arteriovenous fistulas were embolized through intervention. The pulmonary arteriovenous fistula was treated by embolizing it with a spring coil, which effectively blocked the venous sac and the feeding arteries. As a result of the embolization, the child's hypoxemia totally resolved and returned to normal. The intracranial arteriovenous fistula was treated by embolization using a double microcatheter technique with spring coil along with Onyx. There were no complications such as cerebral hemorrhage or cerebral infarction following the embolization procedure, and there was no recurrence of seizures. Our case demonstrates that interventional embolization is an effective treatment for pulmonary and cerebral vascular abnormalities in symptomatic HHT.

INTRODUCTION

Hereditary hemorrhagic telangiectasia (HHT) is a multifaceted genetic condition resulting from abnormalities in multiple genes. It is often characterized by abnormal connections between arteries and veins, as well as widening of small blood vessels, in various organs including the skin, nasopharynx, gastrointestinal tract, liver, lungs, and brain [1]. Most HHT patients have mutations in either the ENG gene (HHT1) or the ACVRL1 gene (HHT2). HHT type 1 is characterized by a higher occurrence of pulmonary arteriovenous fistulas, intracranial arteriovenous fistulas, and gastrointestinal bleeding [2]. According to the HHT guidelines, intervention can be performed to treat symptomatic vascular malformations using interventional procedures [3]. Both pulmonary arteriovenous malformation (PAVM) and Brain arteriovenous malformation (BAVM) have been documented, but the occurrence of both conditions together is rare. We present a case report of a 6-year-old male patient who was identified with HHT type I after appropriate medical examinations. The patient exhibited both PAVM and BAVM. The child's hypoxemia and seizure symptoms were managed with interventional embolization.

PRESENTATION OF CASE

The child is a 6 year old boy who presented with convulsions and MRI suggestive of cerebral hemorrhage on the 6th day of life and was given conservative treatment. The child presented with epilepsy 2 times 3 years ago. Unconsciousness, lip bruising, left limb twitching, lasting 3-5 minutes, EEG suggested epilepsy, no cerebral vascular malformation and other abnormal manifestations, then oral oxcarbazepine control is good. Because of the lack of regular medication, seizures occurred again, lasting for 1 hour, cranial MRI suggests cerebrovascular malformations, right middle cerebral artery M2 branch-right transverse sinus, sigmoid sinus arteriovenous fistula, left posterior cerebral artery developmental variations. 1 year ago, parents unintentionally found that both hands, fingers, feet, lips bruising, and then gradually appeared after the activity of chest tightness and wheezing. Chest enhancement CT showed a pulmonary arteriovenous fistula in the lower lobe of the right

lung, and the right lower pulmonary artery was dilated and connected to the dorsal segment of the lower lobe of the right lung, which was significantly strengthened by a vascular mass and returned to the right pulmonary vein. The child had a family history of intermittent nosebleeds. Genetic testing revealed an ENG mutation, which led to the diagnosis of HHT type I.

Endovascular embolization

A surgical procedure was carried out under the administration of general anesthesia using tracheal intubation. The Seldinger technique was used for puncturing the right femoral vein and a 5F vascular sheath (Terumo) was inserted. Heparin was then injected at a dosage of 100 U/Kg. A 0.035-inch guidewire (Apert) was used to guide the selection of a 5F pigtail catheter (Cordis) for pulmonary arteriography. The results revealed a pulmonary arteriovenous fistula in the right lower lobe of the lungs, along with a dilated right lower pulmonary artery connecting to the dorsal segment of the right lower lobe. Additionally, there was thickening observed in the drainage vein. These findings were confirmed after further examination of the lesion. Once the lesion was clarified, the 7F StearEase long sheath (Xianjian□China) was exchanged for a 5F Cobra2 catheter (Terumo) and inserted into the right pulmonary artery. The artery that supplies the pulmonary arteriovenous fistula was observed to be divided from the main trunk, and the angiogram (Figure 4) showed the thickening of the draining vein. The road map procedure involved inserting a small guide wire into the thin tube, identified as a microcatheter. The microcatheter was then carefully positioned inside the venous sac space of the arteriovenous fistula. Next, the coil springs were released one by one, following the path of the sac and the arteries that supply blood to it. The specific combinations of coil springs are as follows: Boston Scientific Interlock 16-40, 16-40, 18-40, 18-40, and 2-40, 22-60, 22-60, and 14-30. There are 4 instances of the Cook MWCE-18-14-10 NESTER. Additionally, there is 1 instance of the Boston Scientific Complex Helical-18 with a diameter ranging from 10mm to 14mm, as seen in Figure 5. The final screening revealed the absence of an arteriovenous fistula or draining vein (Figure 6).

Embolization of the intracranial arteriovenous fistula was carried out after a month. Following the administration of general anesthesia, a puncture was made in the right femoral artery and a 6F vascular sheath (Terumo) was inserted to replace it. The patient was then given a systemic dose of heparin at a rate of 100 U/kg. A 0.035 Terumo guidewire with hydrophilic coating was inserted, A 6F Envoy Guiding MPD catheter□Cordis□ was then introduced. Selective cannulation was performed to access the right common carotid artery. In this position, an arteriovenous fistula was observed in the right middle cerebral artery M2 branch, specifically in the right transverse sinus and sigmoid sinus. The size of the fistula measured approximately 15.35*16.69 mm. The drainage vein appeared thickened, with a diameter of 2.97 mm. Subsequently, the decision was made to carry out arteriovenous fistula embolization. Two SL-10 microcatheters (1.7F,Stryker) were inserted. The tip of the microcatheter was positioned both near and far from the center of the arteriovenous fistula, using a microguidewire (Microvention Traxcess-14) as a guide. The fistula and the draining vein were observed multiple times. The Stryker Target spring coils of varying sizes (5-20, 4-15, 3-8, and 4-10) were put in a certain order. Once released, the blood flow in the fistula decreased and the flow rate of the draining vein also dropped. Additionally, the Onyx-18 liquid embolic agent (EV3) was injected through the microcatheter. The arteriovenous fistula didn't exist in the final image, and there was no leakage of the contrast agent. Additionally, there were no blockages or bleeding in the arteries within the head

DISCUSSION

HHT, also known as Rendu–Osler–Weber disease□is a genetic disorder caused by mutations in signaling pathway genes that control TGF-B1 in endothelial cells. These mutations affect the regulation of angiogenesis and vascular integrity, resulting in the dilatation of capillaries and the creation of arteriovenous malformations [5]. Curaçao's 4 criteria for diagnosing HHT include: (1) frequent and recurring nosebleeds; (2) expansion of small blood vessels in the mucous membranes of the lips, mouth, face, and fingers; (3) presence of abnormal blood vessel formations in organs such as the brain, liver, and lungs; and (4) having a close family member who has been diagnosed with HHT using the same criteria. Patients who fulfill at least three out of the four criteria are classified as having definitive HHT, while meeting two out of the four criteria is indicative of probable or suspected HHT [6]. As patients get older, certain symptoms of HHT may become evident. In one case, the youngster exhibited hypoxemia and seizures, as well as a pulmonary arteriovenous fistula observed on a chest CT scan and an intracranial arteriovenous fistula detected on a cranial MRI. The diagnosis of HHT was made based on the presence of nasal hemorrhage, a family history of the disease, and the fulfillment of three clinical signs. Subsequent analysis of genetic screening indicated a mutation in the ENG gene, namely HHT type I. HHT patients can be classified into five distinct categories, with HHT I and HHT II accounting for around 90% of all HHT cases [7]. ENG codes for endothelin, a receptor of transforming growth factor (TGF)-β type III. Research has demonstrated that deficiency of endothelin causes delayed remodeling of the developing vascular capillary plexus, enhanced proliferation of endothelial cells, and localized enlargement of veins, resulting in the formation of arteriovenous shunts [8]. HHT I is characterized by a higher occurrence of PAVMs, BAVMs, and gastrointestinal bleeding, while HHT II is associated with a higher occurrence of hepatic vascular malformations and high-output heart failure [9].

PAVM is responsible for approximately 50% of all patients diagnosed with HHT [10]. Pulmonary arteriovenous fistula is an anomalous connection between the pulmonary artery and pulmonary vein, causing blood to bypass the capillaries and flow from the right side of the heart to the left side. This disrupts gas exchange and hinders the filtration of venous thromboembolism, resulting in potential complications such as transient ischemic attacks (TIAs), stroke, and the development of brain abscesses

[11]. The youngster had cyanosis in the nail beds of both hands and had hypoxemia, indicated by an oxygen saturation of 80%. As a result, treatment was necessary. Currently, it is suggested to perform endovascular embolization on PAVM that have supplying arteries larger than 2-3 mm in order to decrease the possible risk of embolic stroke [12].

The embolization of pulmonary arteriovenous fistulae remains a topic of debate, with previous suggestions to only embolize the supplying artery [13]. However, a recent study from Japan discovered that combined embolization of the supplying artery and the venous sac resulted in a significant reduction of fistula during long-term follow-up. This finding was based on a retrospective analysis comparing embolization of the supplying artery alone with simultaneous combined embolization of the supplying artery and the venous sac [14,15]. Dustin discovered that the simultaneous embolization of both the cystic lumen and the supplying artery resulted in a greater occlusion rate compared to embolization of the artery alone over long-term follow-up [16]. We employed controlled coils to occlude both the venous sac and the supplying artery. The initial coil used was a 20-60 mm controlled coil, which was 30% larger than the supplying artery. This larger size ensured better contact with the vessel wall, preventing displacement and migration into the body circulation. Additionally, it allowed for achieving the highest possible packing density [11,12]. Consistent with the present findings, our use of coils embolization to treat a pulmonary arteriovenous fistula led to a notable enhancement in the patient's oxygen levels. Specifically, there was an increase in oxygenation from 80% prior to the procedure to 96%. Additionally, there was a decrease in hemoglobin levels and a reddish discoloration of the nail bed observed in the child. Currently, coils are the predominant choice for embolizing pulmonary arteriovenous fistulas. Additionally, Amplatzer vascular plugs (AVPs), which are dense and expandable nitinol mesh devices used to block blood vessels, are also employed [17]. AVPs, in comparison to coils, present a reduced likelihood of device movement and fewer metal artifacts. This is advantageous for postprocedural imaging follow-up [18].

Approximately 10% of individuals diagnosed with HHT type I exhibit concurrent cerebrovascular abnormalities [19]. In the reported population, the risk of cerebral hemorrhage in patients with HHT is 1%, Morgan noted that infants and children in particular are at high risk for sudden intracranial hemorrhage [20], which is consistent with the history of cerebral hemorrhage in our child's young children. Seizures are another common presentation in patients with HHT, seen in 10% of cases [21]. Josephson showed that patients with hemorrhagic bavms are at a higher risk of subsequent seizures [22]. The exact cause of the seizures is not well understood and may be related to changes in vascular dynamics. Systematic review and meta-analysis suggest that approximately 20% of patients with HHT cerebral arteriovenous malformations develop hemorrhages associated with arteriovenous malformations, and approximately 50% of patients with HHT cerebral arteriovenous malformations develop symptoms associated with cerebral arteriovenous malformations, including headaches, seizures and/or focal neurologic deficits [19]. In a recently published study from the Cerebrovascular Malformation Consortium's HHT Investigative Group, 153 patients with HHT cerebral AVMs were followed for an average of 3 years. The authors found an overall hemorrhage rate of 1% per year in unruptured AVMs and a rupture rate of 0.4% per year [23].

Currently, HHT combined with cerebrovascular malformations are categorized on imaging features into brain arteriovenous fistulae, nidus-type arteriovenous malformations, and capillary vascular malformations, of which approximately 10% are Direct high-flow fistulous arteriovenous shunts [24]. Krings provides the most extensive and detailed account of cerebral arteriovenous malformation (AVM) characteristics in individuals with HHT. The predominant observation is that cerebral vascular malformations associated with HHT are typically seen in the upper region of supratentorial and superficial compartment [21]. Simple arteriovenous fistulas warrant a more intensive therapeutic approach. The child had an arteriovenous fistula in the M2 segment of the middle cerebral artery, which extended to the transverse sinus and sigmoid sinus. We performed a transfemoral intervention using a double microcatheter. Initially, we used a spring coil to embolize the venous sac lumen and reduce the flow rate. This technique minimizes the backflow of the non-adherent liquid embolization material, Onyx gel, towards the artery end. It also reduces the injection time and enhances the dispersion of the gel, hence improving the effectiveness of the embolization process. The spring coils create a solid barrier when combined with the Onyx adhesive, resulting in a decrease in the deformed vascular mass and venous pressure. Mont 'Alverne successfully treated a big arteriovenous fistula by using spring coils, resulting in total relief of symptoms [25]. In a study conducted by Gross, 213 patients who were treated for arteriovenous fistulae were divided into groups. The surgical and endovascular groups showed comparable rates of obliteration and symptom resolution rates [26].

CONCLUSION

HHT is a complex genetic disorder involving multiple systems, Genetic screening can be utilized to detect possible further organ abnormalities, while radiology plays a crucial role in both diagnosing and treating the condition. Endovascular embolization is a reliable and secure method for treating pulmonary arteriovenous fistulas and cerebral arteriovenous malformations.

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FIGURES

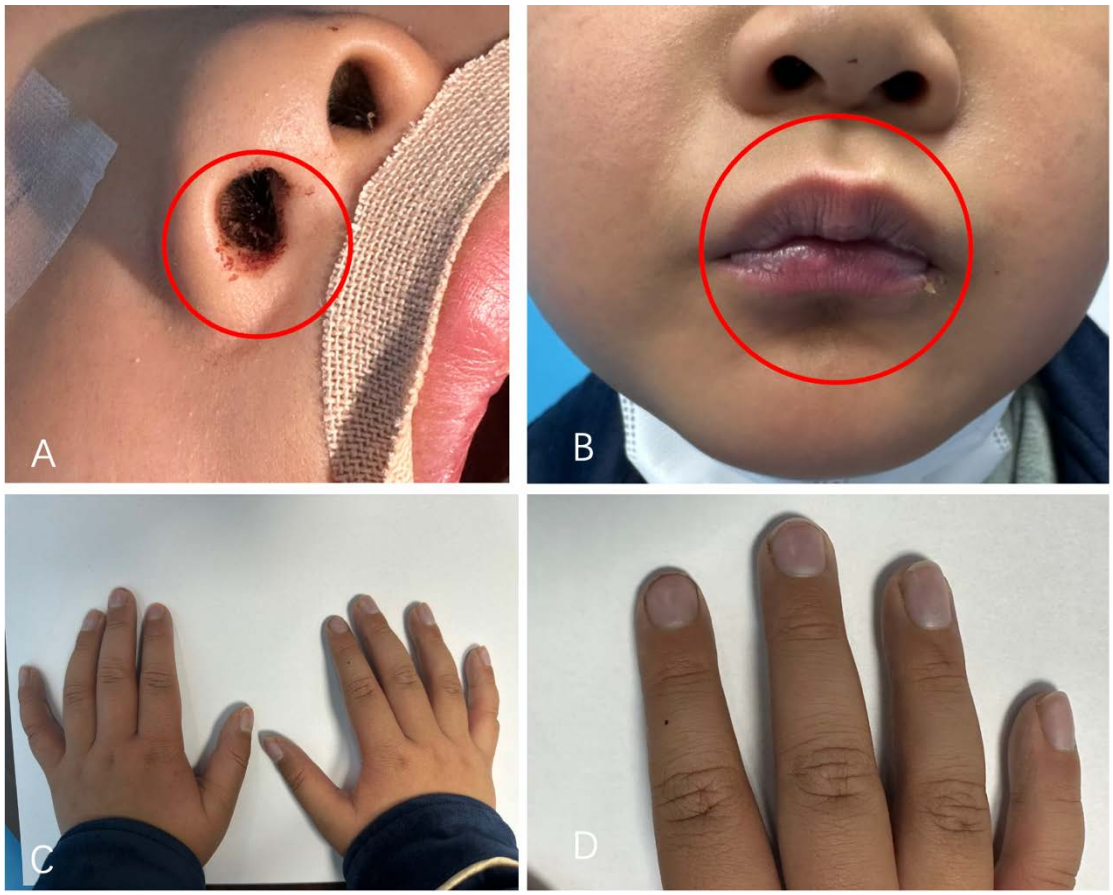

Figure 1: Physical examination. A: Recurrent epistaxis on the right side; B to D□Cyanosis of the lips and nail beds of both hands.

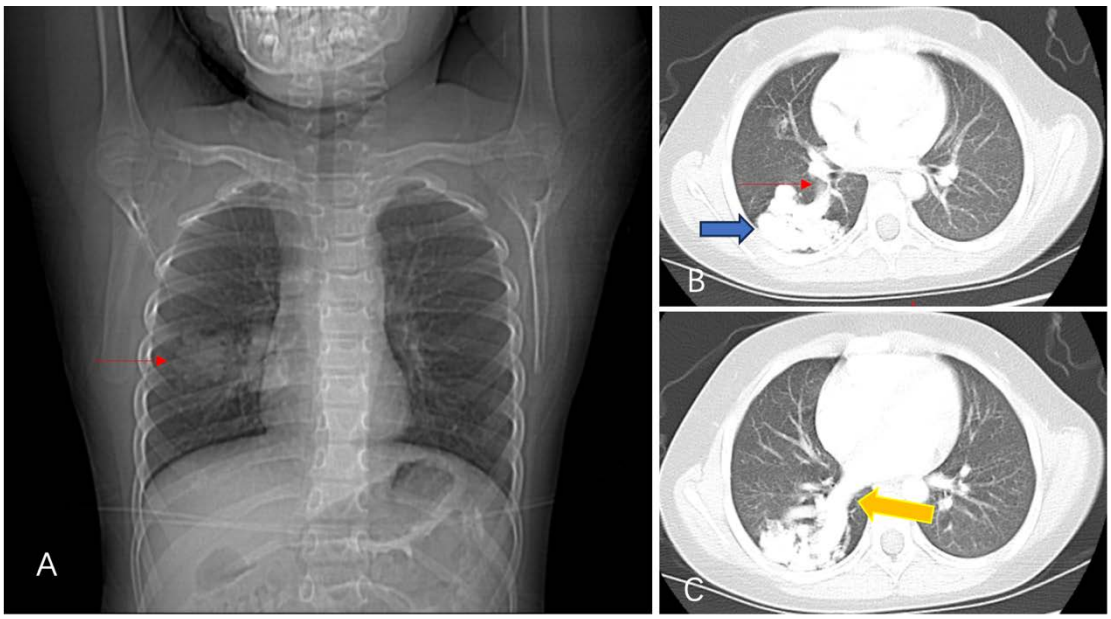

Figure 2: Chest Contrast-enhanced CT: A. substantial increase in mass was observed in the dorsal region of the lower lobe of the right lung, with the lesion measuring around 47*30*36 mm. B. The feeding artery originated as a branch of the pulmonary artery, and separation was observed within the cystic cavity. C. The right lower pulmonary vein exhibits thickening of the draining vein.

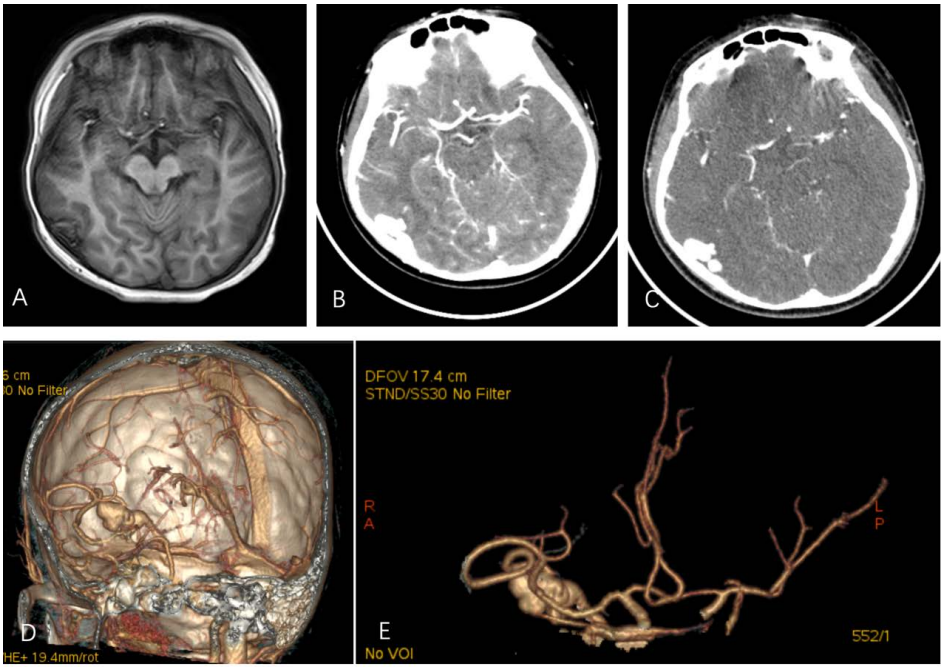

Figure 3: Cranial Contrast-enhanced CT and MRI: cerebrovascular malformation, right middle cerebral artery M2 segment branch-right transverse sinus, sigmoid sinus arteriovenous fistula.

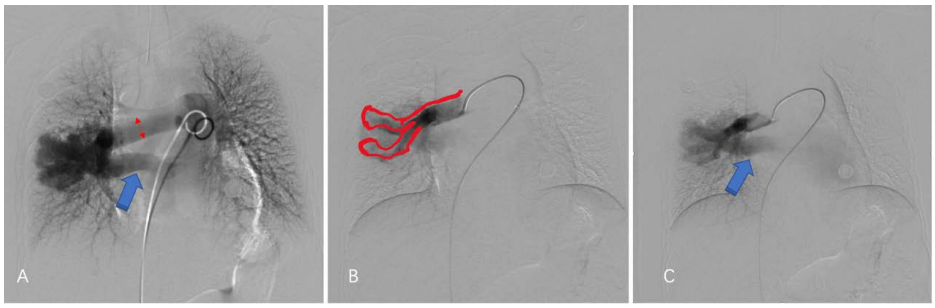

Figure 4: A. 5F pigtail catheter (Cordis) was implemented for pulmonary arteriography, which revealed the presence of a pulmonary arteriovenous fistula in the lower lobe of the right lung. The fistula is characterized by a dilated right lower pulmonary artery that connects to the dorsal segment of the right lower lobe of the lung. Additionally, there is thickening observed in the draining vein. B. 5F Cobra2 catheter (Terumo) was inserted into the right pulmonary artery, and contrast was used to visualize the feeding artery of the pulmonary arteriovenous fistula, which was separated from the main trunk. C. The visualization reveals an increase in the diameter of the draining vein.

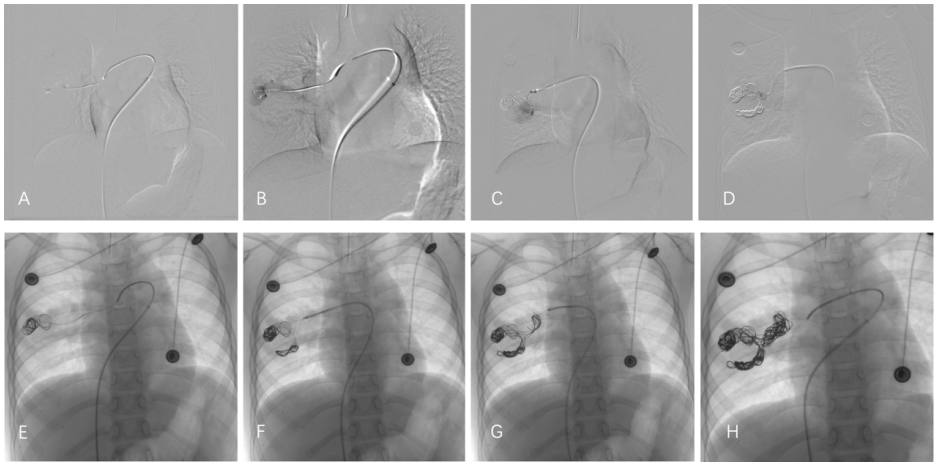

Figure 5: A-D. The microguide wire is utilized to guide the microcatheter into the lumen of the arteriovenous fistula capsule and the feeding arteries. E-H demonstrate the sequential process of coil springs embolization within the lumen of the venous sac and the feeding artery.

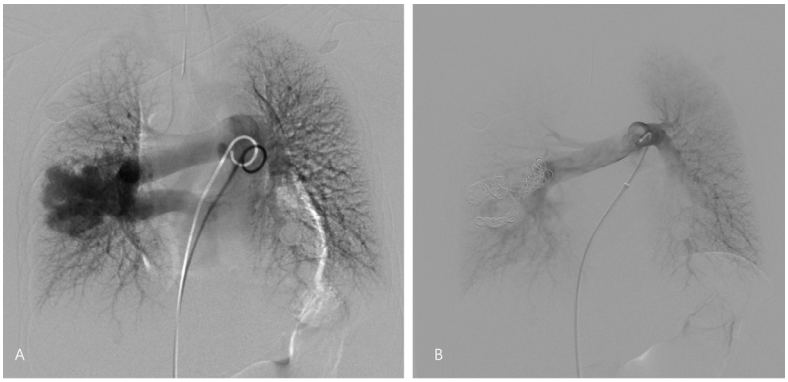

Figure 6: A Preoperative Angiography B. Postoperative Angiography. Arteriovenous fistula and draining vein not visualized.

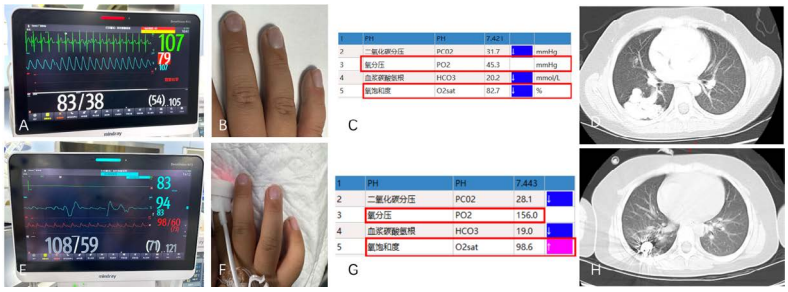

Figure 7: Comparisons between the preoperative and postoperative conditions. Figures A-D display preoperative vital signs, nail bed color, blood gas analysis, and CT results. Figures E-H depict the results of the surgery after it was performed. The oxygen saturation levels were increased, the nail bed turned crimson, and the pulmonary arteriovenous fistula resolved.

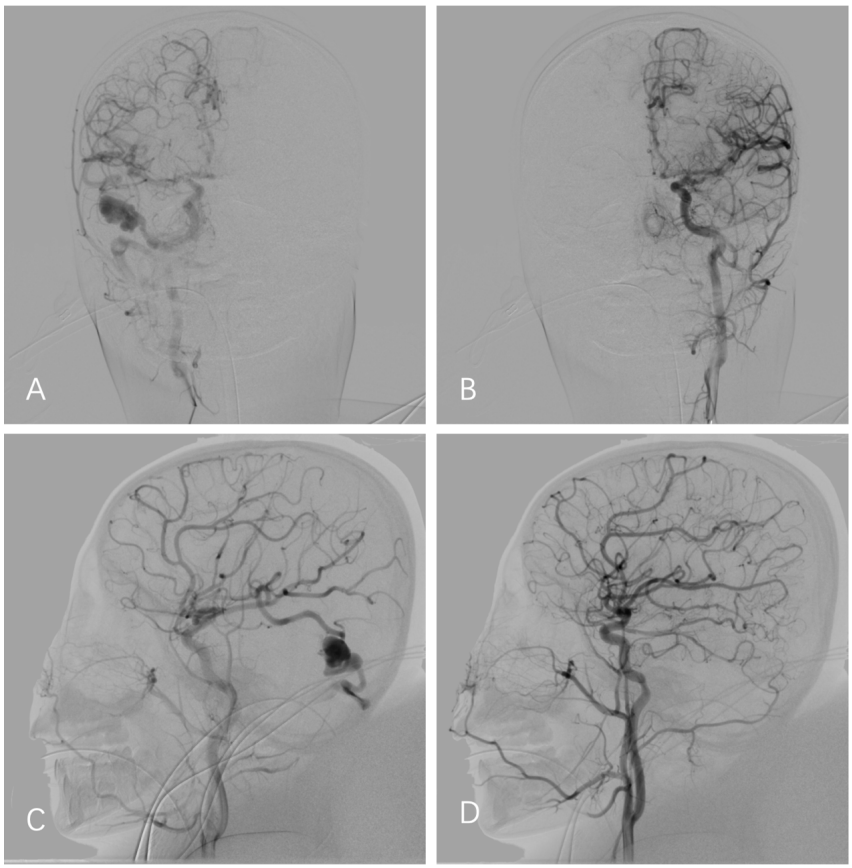

Figure 8: Figures A and C depict the right orthopantomogram and lateral contrast images, illustrating the presence of an arteriovenous fistula between the middle cerebral artery branch-transverse sinus and sigmoid sinus. Figures B and D display the left orthopantomogram and lateral angiography, respectively, revealing no abnormalities.

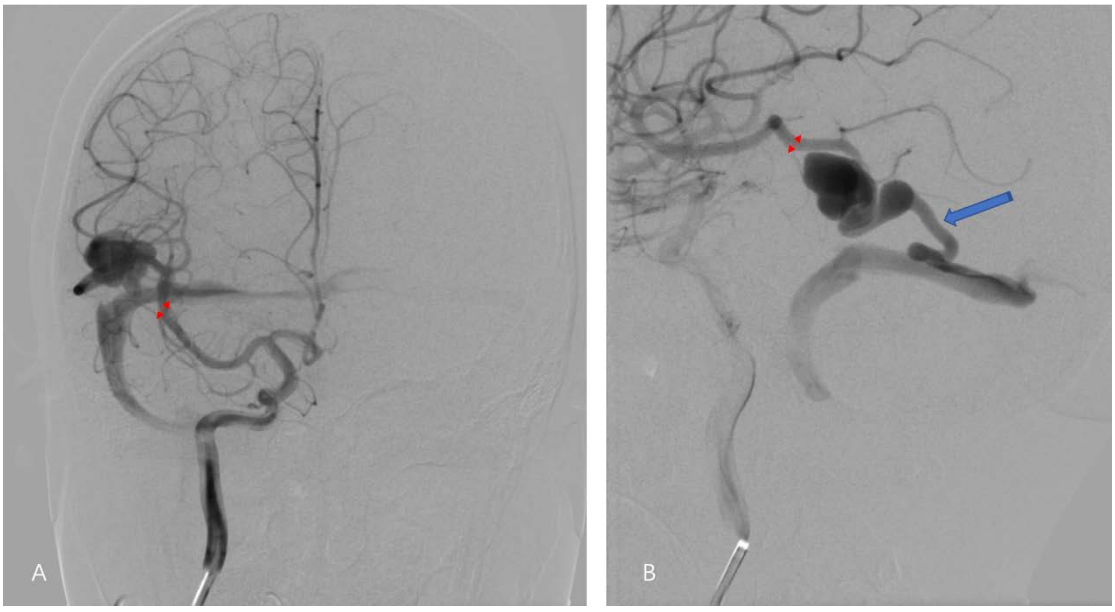

Figure 9: Frontal and lateral view, red arrows are blood-supplying arteries, blue arrows are draining veins.

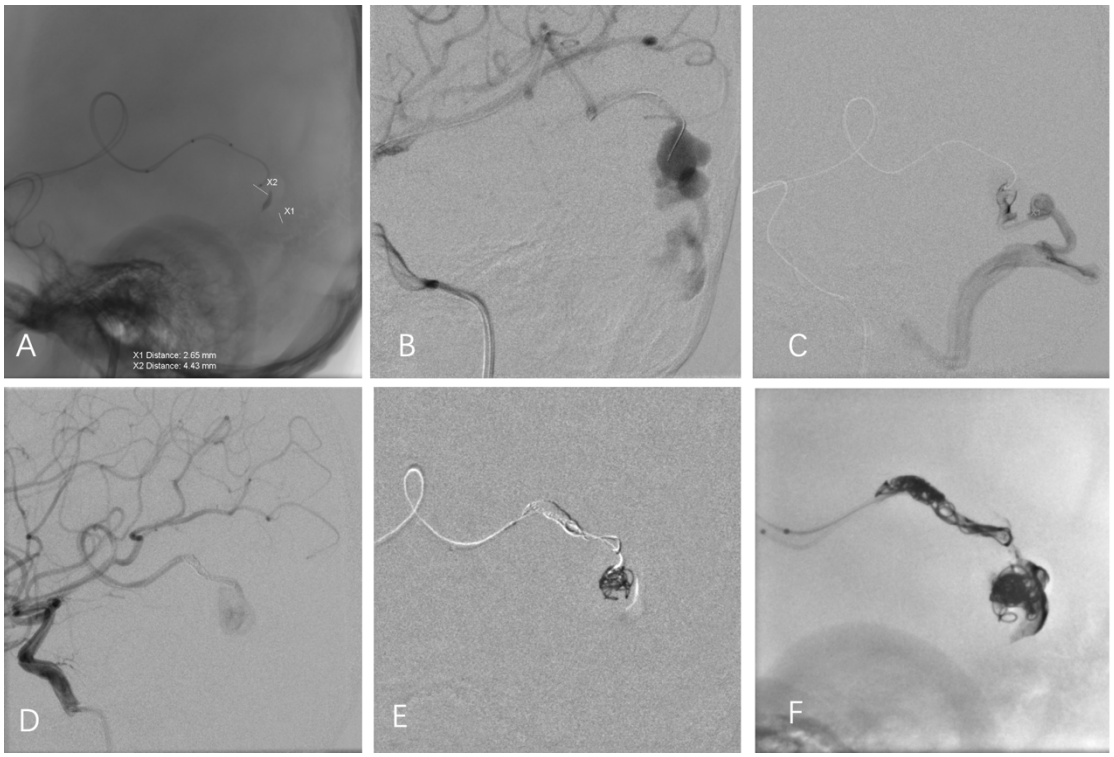

Figure 10: A-C. Micro guide wire (Traxcess-14 guidewire) together with the microcatheter (SL-10 (1.7F) × 2 (Stryker)) were selected to be inserted into the cystic lumen of the arteriovenous fistula and the blood-supplying artery, respectively. The angiography procedure was used to determine the exact location of the arteriovenous fistula. Figure D demonstrates the use of coil springs embolization to reduce the flow rate in the blood-supplying artery. Figure E shows coil springs embolization being performed in the cystic cavity. Figure F illustrates the process of Onyx glue embolization.

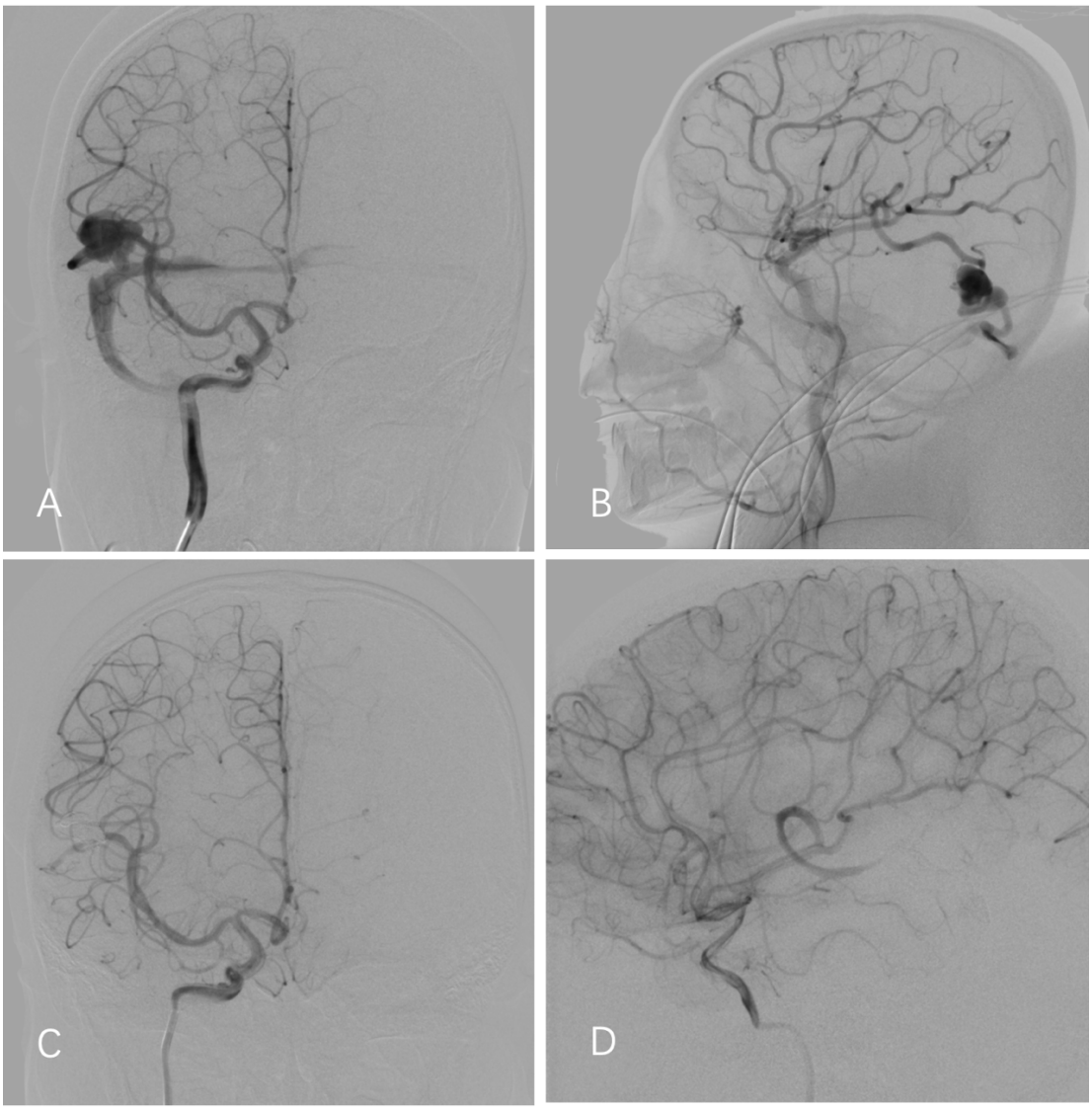

Figure 11: A and B display preoperative orthostatic and lateral photographs, C and D exhibit postoperative orthostatic and lateral imaging, revealing the complete resolution of the arteriovenous fistula.

KEYWORDS

Hereditary hemorrhagic telangiectasia; Pulmonary arteriovenous malformations; Brain arteriovenous malformation; ENG gene; Embolization

ABBREVIATIONS

HHT = Hereditary Hemorrhagic Telangiectasia
PAVM = Pulmonary Arteriovenous Malformations
BAVM = Brain Arteriovenous Malformations