

References

- Gutiérrez Gutiérrez G, Díaz-Manera J, Almendrote M, Azriel S, Eulalio Bárcena J, Cabeza García P, et al. Clinical guide for the diagnosis and follow-up of myotonic dystrophy type 1 MD1 or Steinert's disease. *Neurología*. 2020;35:185–206.
- Johns MW. A new method for measuring daytime sleepiness: the Epworth sleepiness scale. *Sleep*. 1991;14:540–5.
- American Academy of Sleep Medicine. In: Darien IL, editor. The International Classification of Sleep Disorders — Third Edition (ICSD-3). 3rd ed. American Academy of Sleep Medicine; 2014.
- Mills RJ, Young CA. A medical definition of fatigue in multiple sclerosis. *QJM*. 2008;101:49–60.
- Maestri M, Romigi A, Schirru A, Fabbrini M, Gori S, Bonuccelli U, et al. Excessive daytime sleepiness and fatigue in neurological disorders. *Sleep Breath*. 2019.
- Romigi A, Franco V, Placidi F, Liguori C, Rastelli E, Vitran G, et al. Comparative sleep disturbances in myotonic dystrophy types 1 and 2. *Curr Neurol Neurosci Rep*. 2018;18:102.
- Romigi A, Albanese M, Liguori C, Placidi F, Marciani MG, Massa R. Sleep-wake cycle and daytime sleepiness in the myotonic dystrophies. *J Neurodegener Dis*. 2013;2013:692026.
- Romigi A, Izzi F, Pisani V, Placidi F, Pisani LR, Marciani MG, et al. Sleep disorders in adult-onset myotonic dystrophy type 1: a controlled polysomnographic study. *Eur J Neurol*. 2011;18:1139–45.
- Laberge L, Bégin P, Montplaisir J, Mathieu J. Sleep complaints in patients with myotonic dystrophy. *J Sleep Res*. 2004;13:95–100.
- Laberge L, Gagnon C, Jean S, Mathieu J. Fatigue and daytime sleepiness rating scales in myotonic dystrophy: a study of reliability. *J Neurol Neurosurg Psychiatry*. 2005;76:1403–5.
- Laberge L, Gallais B, Auclair J, Dauvilliers Y, Mathieu J, Gagnon C. Predicting daytime sleepiness and fatigue: a 9-year prospective study in myotonic dystrophy type 1. *J Neurol*. 2020;267:461–8.
- Hermans MCE, Merkies ISJ, Laberge L, Blom EW, Tennant A, Faber CG. Fatigue and daytime sleepiness scale in myotonic dystrophy type 1. *Muscle Nerve*. 2013;47:89–95.
- Gallais B, Gagnon C, Forgues G, Côté I, Laberge L. Further evidence for the reliability and validity of the Fatigue and Daytime Sleepiness Scale. *J Neurol Sci*. 2017;375:23–6.
- Gagnon C, Heatwole C, Hébert LJ, Hogrel J-Y, Laberge L, Leone M, et al. Report of the third outcome measures in myotonic dystrophy type 1 (OMMYD-3) international workshop Paris, France June 8, 2015. *J Neuromuscul Dis*. 2018;5:523–37.

A. Romigi

IRCCS Neuromed Istituto Neurologico Mediterraneo, Sleep Medicine Center, Pozzilli (IS), Italy

E-mail address: andrea.romigi@gmail.com.

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Ischaemic stroke secondary to paradoxical embolism as a consequence of superior vena cava syndrome by a displacement of a ventriculoatrial shunt*



Infarto cerebral por embolismo paradójico secundario a síndrome de vena cava superior por malposición de un catéter de derivación ventriculoauricular

Dear Editor:

Superior vena cava syndrome (SVCS) is an infrequent condition characterised by a partial or total obstruction of blood flow through the superior vena cava due to extrinsic compression, infiltration, or thrombosis. Progression is variable and sometimes slow, and the condition can even be life-threatening; therefore, it requires a precise diagnosis and early treatment.^{1,2}

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We present the case of an 82-year-old woman, who was partially dependent and was using a colostomy bag due to perforated diverticulitis. In 2013, she underwent radiosurgery for a petroclival meningioma measuring 4 cm. She later developed non-communicating hydrocephalus due to external compression of the meningioma. As ventriculo-peritoneal shunt was contraindicated due to the history of colostomy, she underwent ventriculoatrial shunt (VAS) implantation in December 2014 (Fig. 1).

In May 2019, the patient consulted our department due to global aphasia and right hemiparesis (muscle strength of 2/5), with onset upon waking. Given suspicion of stroke, we performed a multimodal CT scan, which revealed distal occlusion of the M1 segment of the left middle cerebral artery and favourable mismatch. The patient underwent mechanical thrombectomy, with angiography showing complete reperfusion. Despite this, neurological symptoms improved only slightly. A follow-up CT scan performed 24 hours after the procedure revealed an ischaemic lesion involving the left lentiform nucleus and insula.

During her stay at the stroke unit, the patient presented oedema in the right arm, hindering the insertion of peripheral venous catheters. On the fifth day after admission, we also observed oedema in the face and contralateral arm; examination of the upper limbs yielded normal results. Given the suspicion of SVCS, we performed a Doppler ultrasound of the supra-aortic trunks, which revealed thrombosis of the internal jugular veins. We also requested a non-contrast chest, abdomen, and pelvis CT scan, which initially ruled out extrinsic venous compression or tumour and

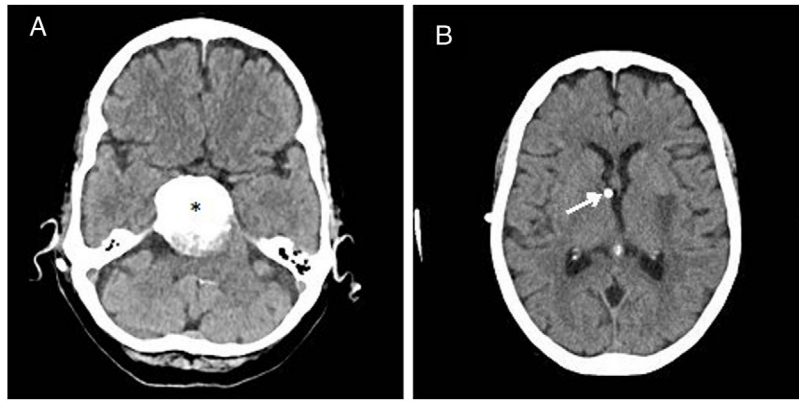


Figure 1 (A) Cranial CT scan showing the petroclival meningioma (asterisk) that caused compressive hydrocephalus, leading to the placement of a ventricular shunt. (B) CT scan showing the proximal end of the VAS at the right lateral ventricle.

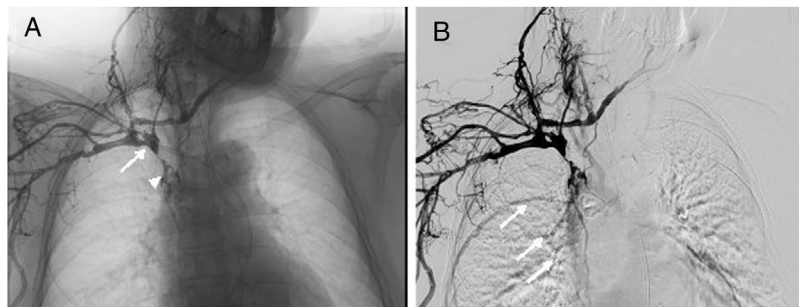


Figure 2 Venography showing contrast administration after catheterisation of the right cephalic vein. (A) Obstruction of the superior vena cava (arrow), with filiform passage of contrast to the right atrium (arrowhead); (B) collateral filling of thoracic wall veins towards the azygos vein (arrows).

detected a displacement of the distal end of the VAS in the superior vena cava. A venography of the right arm confirmed thrombosis of the right subclavian vein, the brachiocephalic trunk, and the superior vena cava, with the latter showing contrast passage to the right atrium (Fig. 2). The venous system of the contralateral arm could not be studied due to the inability to insert a peripheral catheter.

After reassessing the patient's clinical situation, and having verified the correct functioning of the VAS, we opted for conservative treatment with enoxaparin at a therapeutic dose of 1 mg/kg every 12 hours. In the following weeks, we observed complete resolution of the facial oedema and partial resolution of the oedema in the arms.

Aetiological study of stroke included a transthoracic echocardiography without echo-enhancing agents, which yielded normal results, and a transcranial Doppler ultrasound right-to-left shunt test, which showed positive results, suggesting a paradoxical embolism from the superior vena cava thrombosis.

Global aphasia and right hemiparesis persisted. After clinical stability was achieved, the patient was discharged with prescription of anticoagulation therapy with warfarin at 32 mg weekly, for an indefinite period of time.

SVCS is a complex clinical syndrome whose aetiology has changed over time. The most frequent cause is currently malignant mediastinal neoplasm, especially small-cell lung cancer and non-Hodgkin lymphoma.³ However, the increasing use of semi-permanent intravascular (venous catheters,

haemodialysis) and cardiac devices (pacemakers, defibrillators) has significantly contributed to the appearance of new cases, representing the first non-neoplastic cause of SVCS.^{4,5}

Ventricular shunt is one of the most widely used neurosurgical procedures in the treatment of hydrocephalus. Over the past 20 years, ventriculoperitoneal shunts have preferentially been used due to the technical challenge and the cardiopulmonary and renal complications observed with VAS.^{6–8} However, no published study has reported cases of SVCS, as observed in our patient. We can only refer to the literature published in the 1960s on children undergoing VAS placement due to hydrocephalus of non-neoplastic origin.^{9–11} The postulated mechanism states that atrial contraction during the cardiac cycle favours retrograde transmission of the movement to the rest of the catheter, whereas proximal displacement promotes thrombosis due to reduced mobility of the distal end in the superior vena cava.¹²

Cardioembolic aetiology accounts for 25% of ischaemic strokes. Less frequent cardioembolic causes include patent foramen ovale, which is present in 25% of the general population and is diagnosed in up to 40% of younger patients with otherwise cryptogenic stroke.¹³

From a therapeutic point of view, endovascular procedures (local fibrinolysis, percutaneous angioplasty) constitute the first line of treatment for SVCS secondary to intravascular devices.¹⁴ The functional prognosis of our patient, the risk associated with invasive techniques, and

the proper functioning of the VAS led us to opt for conservative treatment, achieving partial symptom resolution.

The interest of this case resides in its unusual form of presentation: ischaemic stroke secondary to paradoxical embolism, which has not previously been reported. In fact, a retrospective series of 70 patients with ischaemic stroke of infrequent aetiology reported no cases of this clinical manifestation.¹⁵

In conclusion, considering the possible development of SVCS, it is essential to continuously monitor patients with semi-permanent intravascular devices, as the complications may be catastrophic.

References

1. Pench-Alonso B, Arredondo-Ruiz P, González-Galván LM, Fermín-Hernández C. Síndrome de vena cava superior: diagnóstico y tratamiento. *Med Int Mex.* 2018;34:403–11.
2. Zimmerman S, Davis M. Rapid fire: superior vena cava syndrome. *Emerg Med Clin North Am.* 2018;36:577–84.
3. Cheng S. Superior vena cava syndrome: a contemporary review of a historic disease. *Cardiol Rev.* 2009;17:16–23.
4. Kalinin RE, Suchkov IA, Shitov II, Mzhavanadze ND, Povarov VO. Venous thromboembolic complications in patients with cardiovascular implantable electronic devices. *Angiol Sosud Khir.* 2017;23:69–74.
5. Labriola L, Seront B, Crott R, Borceux P, Hammer F, Jadoul M. Superior vena cava stenosis in haemodialysis patients with a tunnelled cuffed catheter: prevalence and risk factors. *Nephrol Dial Transplant.* 2018;33:2227–33.
6. Niu H, Wu H, Luo W, Wang K, Zhao L, Wang Y. Ventriculoatrial shunt as a feasible regimen for certain patients of hydrocephalus: clinical features and surgical management. *Acta Neurol Belg.* 2019;10:1007.
7. Hung AL, Vivas-Buitrago T, Adam A, Lu J, Robison J, Elder BD, et al. Ventriculoatrial versus ventriculoperitoneal shunt complications in idiopathic normal pressure hydrocephalus. *Clin Neurol Neurosurg.* 2017;157:1–6.
8. McGovern RA, Kelly KM, Chan AK, Morrissey NJ, McKhann GM 2nd. Should ventriculoatrial shunting be the procedure of choice for normal-pressure hydrocephalus? *J Neurosurg.* 2014;120:1458–64.
9. Guidetti B, Occhipinti E, Riccio A. Ventriculo-atrial shunt in 200 cases of non tumoural hydrocephalus in children: remarks on the diagnostic criteria, postoperative complications and long-term results. *Acta Neurochir (Wien).* 1969;21:295–308.
10. Nugent GR, Lucas R, Judy M, Bloor BM, Warden H. Thrombo-embolic complications of ventriculo-atrial shunts. Angiocardiac and pathologic correlations. *J Neurosurg.* 1966;24:34–42.
11. Piatt JH Jr, Hoffman HJ. Cor pulmonale: a lethal complication of ventriculoatrial CSF diversion. *Childs Nerv Syst.* 1989;5:29–31.
12. Overton MC 3rd, Derrick J, Snodgrass SR. Surgical management of superior vena cava obstruction complicating ventriculoatrial shunts. *J Neurosurg.* 1966;25:164–71.
13. Arboix A, Alió J. Acute cardioembolic stroke: an update. *Expert Rev Cardiovasc Ther.* 2011;9:367–79.
14. Sfyrouras GS, Antonopoulos CN, Mantas G, Moulakakis KG, Kakisis JD, Brountzos E, et al. A review of open and endovascular treatment of superior vena cava syndrome of benign aetiology. *Eur J Vasc Endovasc Surg.* 2017;53:238–54.
15. Arboix A, Bechich S, Oliveres M, García-Eroles L, Massons J, Targa C. Ischemic stroke of unusual cause: clinical features, etiology and outcome. *Eur J Neurol.* 2001;8:133–9.

J. Molina-Gil*, S. Calleja-Puerta, M. Rico

Servicio de Neurología, Hospital Universitario Central de Asturias, Oviedo, Asturias, Spain

* Corresponding author.

E-mail address: javimol1993@hotmail.com (J. Molina-Gil).

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Periventricular heterotopia: broadening of the clinical spectrum of the clathrin 1 gene (*CLTC*) pathogenic variants*



Heterotopías periventriculares: ampliación del espectro clínico de las variantes patogénicas del gen de la clatrina 1 (*CLTC*)

Dear Editor:

The *CLTC* gene encodes the clathrin heavy chain 1 (CHC1) protein.^{1,2} This structure enables the formation of lattices

in clathrin-coated vesicles by facilitating the intracellular membrane traffic of receptors, endocytosis of certain macromolecules, and stability of the mitotic spindle during the metaphase.³ This protein is expressed in greater abundance in the developing brain.⁴ Loss-of-function (LoF) mutations of the *CLTC* gene are associated with autosomal dominant mental retardation-56 (MIM#617854), although they have also been reported in patients with epilepsy and other neurodevelopmental disorders.^{3–5}

We present the case of a girl with a previously unreported de novo mutation of the gene and periventricular heterotopia detected with a brain magnetic resonance imaging (MRI) study.

Our patient is a girl with no relevant family history, and personal history of patent ductus arteriosus and bone alterations (spina bifida occulta and mild rib hypoplasia). Head circumference has consistently been in the 10th percentile. Fig. 1 shows the phenotype. At the age of 5, she began to present epileptic seizures (typical absence and generalised tonic-clonic seizures), which were controlled with ethosuximide and valproic acid; seizures returned when the

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