CASE REPORT

Discussion and review of the literature following the case of a young man with Down’s syndrome and cerebral venous thrombosis


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Abstract
This review and discussion of the current literature is based on the case of a young man with Down’s syndrome (DS) and cerebral venous thrombosis (CVT).

Clinical case. Twenty-seven-year-old male who presented with headache, vomiting and left hemiparesis. After finding signs consistent with cerebral venous thrombosis on neuroimaging, anticoagulant treatment was started, and eventuated in a favorable clinical outcome.

Discussion. DS patients are predisposed to the occurrence of embolic stroke secondary to congenital heart disease. However, the causes of CVT in DS are uncertain, but probably have a multifactorial origin. There are to date two published cases of CVT in patients with DS.

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PALABRAS CLAVE
Trombosis venosa cerebral; Síndrome de Down; Tratamiento anticoagulante

Discusión y revisión de la bibliografía a partir de un caso de un varón joven con síndrome de Down y trombosis venosa cerebral

Resumen
Discusión y revisión de la bibliografía a partir de un caso de un varón joven con síndrome de Down (SD) y trombosis venosa cerebral (TVC).

Caso clínico. Varón de 27 años que comienza con cefalea, vómitos y hemiparesia izquierda. Tras encontrarse hallazgos en la neuroimagen compatibles con trombosis venosa cerebral, se inició tratamiento anticoagulante, con una evolución clínica favorable.

Discusión. Los pacientes con SD están predispuestos a la aparición de ictus embólicos secundarios a cardiopatías congénitas; sin embargo, las causas de TVC en el SD son inciertas, teniendo probablemente un origen multifactorial. Hasta la fecha hay dos casos publicados de TVC en pacientes con SD.

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Introduction

The most common heart diseases in Down’s syndrome (DS) are congenital (atrioventricular canal and interventricular septal defects). Recently published studies suggest that patients with DS may develop a Moyamoya type vasculopathy. There is a clear association between cerebral venous thrombosis (CVT) and a variety of pathologies. No clear association has been established between SD and CVT. We report the case of a young man with Down’s syndrome who presented clinical and radiological findings consistent with CVT.

Case report

A 27-year-old male with Down’s syndrome, who attended the hospital emergency department with headaches, vomiting and left hemiparesis. On physical examination he was found to be aware, oriented and collaborative, with a blood pressure of 150/75mmHg and heart rate of 68 bpm. The patient did not have a fever. Cardiopulmonary auscultation was normal. A neurological examination revealed esotropia in the basal position of the eye, left hemiparesis: ULL proximal 0/5, distal 4-/5, LLL proximal 3/5, distal 2/5, DTR present. The analysis performed in the emergency room showed no significant deviations in blood count or biochemical parameters. The coagulation study could not be performed due to an insufficient sample. An urgent cranial computed tomography (CT) was performed, which revealed indirect signs of CVT: hypersensitivity regions in the superior longitudinal sinus and left transverse sinus and stasis and hyperattenuation of some cortical veins (fig. 1). A cranial magnetic resonance image (MRI) scan confirmed the CT findings (fig. 2). The cause of this extensive thrombosis is still not clear. Anticoagulant therapy was initiated with heparin followed by Sintrom® for 1 month, which led to a favourable clinical course leaving him currently asymptomatic.

Discussion

Cerebral venous thrombosis was first described in 1825 by Ribes and enlarged upon in 1828 by Abercrombie. It is rare and difficult to diagnose, due to having polymorphic and relatively unspecific clinical features, so support from neuroimaging techniques is essential. The annual incidence of CVT is 3-4 cases per million inhabitants, mostly affecting young women. The most frequently affected area is the transverse sinus (86%), followed by the superior sagittal sinus (62%). Risk factors are grouped into genetic prothrombotic conditions (antithrombin deficiency, deficiency of proteins C and S, factor V Leiden mutation); acquired prothrombotic states (nephrotic syndrome, antiphospholipid antibodies); infections (otitis media, sinusitis, meningitis); inflammatory diseases (systemic lupus erythematosus, inflammatory bowel disease); haematologic conditions (leukaemia, thrombocytosis); drugs (oral contraceptives); mechanical causes, injuries and others.
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such as dehydration in children and neoplastic processes. The most common specific clinical manifestation is headache (>90% of cases), which may be accompanied by seizures (40%).

It is widely described in the literature that patients with DS are predisposed to embolic stroke secondary to congenital heart disease. Strokes in young patients with DS may also be secondary to Moyamoya type vasculopathy. The underlying mechanism of CVT in DS is currently uncertain. There are many diseases that could be responsible for CVT in this patient. Patients with DS have an increased risk of developing leukaemia. Hearing loss is another common problem in patients with DS, possibly related to the undiagnosed episodes of otitis media in this population. Patients with DS may therefore have an increased risk of CVT due to leukaemia or otitis media.¹

After reviewing the literature via the MEDLINE database using the PubMed search engine with keywords “Cerebral venous thrombosis and Down’s syndrome”, we found only two reported cases of DS and CVT.¹ ⁴

To understand the symptomatic variability of the process, it is necessary to distinguish between cerebral venous thrombosis, with local effects caused by venous obstruction (cerebral oedema, initially of ischaemic-cytotoxic origin and later vasogenic and reversible, and venous infarcts) and thrombosis of the large sinuses, which causes intracranial hypertension syndrome (the obstruction of the venous sinuses leads to an alteration in the reabsorption of cerebrospinal fluid, causing communicating hydrocephalus, which occasionally causes dilation of the ventricular system), although these processes occur simultaneously in most patients.³

Formerly, the “gold standard” for diagnosing CVT was arteriography. Currently, the most sensitive and least invasive diagnostic test is cranial MRI combined with MRI venography, which reveal the typical findings (hypointense signal on T1 and T2 in the thrombosed dural sinuses and absence of flow in the MRI venography).⁵ ⁶ Cranial CT is just as accurate as MRI for diagnosing this condition (100% sensitivity and specificity in multidetector CT with intravenous contrast), showing increased density in the dural sinuses (cord sign: 64.6% and 97.2% sensitivity and specificity, respectively, for the diagnosis of superficial venous thrombosis) and cortical veins (attenuated vein sign: 100% and 99.4% sensitivity and specificity, respectively, for the diagnosis of deep vein thrombosis) in the study without contrast; and the classic empty delta sign in the torcular Herophili in the study with contrast. Signs of venous infarction or haemorrhage can also be investigated, although these were not present in the case described.⁷ ⁸ These 2 imaging techniques are sufficiently sensitive and specific for a diagnosis, so it is not now normally necessary to perform a cerebral arteriogram. Treatment currently consists of anticoagulation with heparin, despite the risk of haemorrhagic transformation (40%), cranial hypertension with mannitol and endovascular thrombolysis (urokinase) in cases of very poor prognosis.⁹

Figure 2 Cranial MRI, TSE-T1 3D sequence with gadolinium. A: Sagittal section. Filling defects in the superior sagittal sinus (long arrows), torcular Herophili (double arrow) and straight sinus (short arrow) caused by thrombi. B: Axial section. Thrombus in the left transverse sinus (arrows). Permeable right transverse sinus.

Bibliography


