Cerebral sinus venous thrombosis in a patient with complete gonadal dysgenesis

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Abstract

Cerebral sinus venous thrombosis (CSVT) is a rare form of venous thrombosis (VTE), but nevertheless one important cause of stroke in young women. Complete gonadal dysgenesis (CGD) is a rare congenital disorder of sex development characterized by normal female genitalia and fibrotic non-productive gonads in which hormone replacement therapy (HRT) is crucial to induce secondary sex characteristics. We describe a case of a patient with CGD in which CVST was the first manifestation of antiphospholipid syndrome (APS). A 49 years old 46, XY patient with CGD undergoing HRT was admitted to the emergency ward complaining of right sided, pulsating headache, nausea, vomiting and decreased visual acuity for the previous five days. Physical exam was unremarkable, without neurologic deficits. Blurred optic disc edges on ophthalmologic evaluation suggested increased intracranial pressure. Brain CT venography revealed thrombosis of the right lateral and left transverse sinus and filling defects of right internal jugular vein (IJV). Brain MRI confirmed right lateral sinus thrombosis, extending to right IJV. Treatment with low molecular weight heparin and acetazolamide was started with progressive symptomatic improvement. The patient was discharged after bridging to warfarin. An extensive hypercoagulable workup at presentation and 12 weeks later revealed a lupus anticoagulant (LAC) screen positive with DRVVT confirmation. No malignancy was identified in a whole-body CT-scan. HRT was stopped. CSVT is a multifactorial condition with gender-related specific causes. Despite the potential increased risk of venous thrombosis related with HRT and gonadal tumours in patients with CGD, we could not find in the literature any prior report of CSVT associated with this condition.