**MOTHER AND DAUGHTER WITH HEMORRHAGIC STROKE: MERE COINCIDENCE?**

Bárbara Cristina Vieira de Aquino1, Stone Sam Nogueira do Nascimento2, Diogo Barbalho Chaves2, Rodrigo Alencar e Silva3, Clécio de Oliveira Godeiro Júnior3

1. Department of Neurology, Hospital das Clínicas da Faculdade de Medicina de Ribeirão Preto - USP, Ribeirão Preto, Brazil
2. Hospital Promater, Natal, Brazil
3. Department of Neurology, Hospital Universitário Onofre Lopes, Natal, Brasil

**Corresponding author:**

Bárbara C. V. de Aquino

Email: [barbaracvdeaquino@gmail.com](mailto:barbaracvdeaquino@gmail.com)

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A 31-year-old woman underwent laparoscopic right oophoroplasty, due to ovary cyst and endometriosis suspect, after several episodes of abdominal pain, worsened by menstrual period. There was no history of other comorbidities. Her mother died by hemorrhagic stroke, with no confirmed etiology, after hysterectomy surgery.

In 2º post-operative day, she presented in emergency room with refractory nausea and vomits. Upper digestive endoscopy showed mild edematous esophagitis with superficial tear, bulboduodenitis, pangastritis and a peptic ulcer (Sakita stage H2). She was medicated with symptomatic drugs and discharged from hospital 9 days after.

The next day (12º post-operative day), she returned to hospital due to acute abdominal pain, constipation, dysarthria and lower limb weakness. There were no history of fever, vomit or headache. Blood pressure was mildly high (154x93 mmHg) and other vital signs were normal. Laboratory tests showed severe hyponatremia (Na 103 mEq/L) and abdominal computed tomography (CT) showed bowel distension, with no obstructive image. Brain CT was unremarkable. She was hospitalized and hypertonic saline was administered.

The next day, she presented sudden headache, numbness (Glasgow Come Scale 7) and flaccid right hemiparesis. She also had hematic appearance of the urine by the urinary catheter. Head CT reveal left frontal intraparenchymal hematoma (25ml), with mass effect and hemoventriculus - ICH score = 2 (FIGURE 1).

She was intubated and underwent surgical drainage of the hematoma and monitoring intracranial pressure. In 5º postoperative day of hematoma drainage, patient presented seizures and increased intracranial pressure, treated with phenytoin. Control brain CT showed no relevant changes. In 9º postoperative day, she presented areflexic tetraparesis, initially attributed to pontine myelinolysis (after correction of severe hyponatremia).

There are several underlying pathological conditions associated with intracerebral hemorrhage (ICH): hypertension, amyloid angiopathy and ruptured vascular malformation are most common. Other less common underlying etiologies of nontraumatic ICH include: arteriovenous and other vascular malformations, cerebral venous thrombosis, hemorrhagic infarction, primary or metastatic tumor, central nervous system infection, mycotic intracranial aneurysm, Moyamoya disease, cerebral vasculitis, cerebral hyperperfusion syndrome, reversible cerebral vasoconstriction syndrome (RCVS), sickle cell disease and bleeding disorders.1-2

Patient brain MRI did not show any evidence of tumor, amyloid angiopathy or infection. Angiographic study was unremarkable (no evidence of arteriovenous malformation, venous thrombosis, Moyamoya disease or aneurism). Blood tests did not reveal coagulopathy, infection, sickle cell disease, renal impairment or autoimmune disease.

As a known metabolic cause of RCVS, porphyria is a genetic and hereditary condition3, which may cause ICH. This condition may also cause flaccid and areflexic tetraparesis due to polyneuropathy, severe hyponatremia due to syndrome of inappropriate antidiuretic hormone secretion, abdominal pain and red/brown color of urine.4

Urine porphobilinogen level of patient was high and genetic panel for porphyria was requested, which was positive for variegate porphyria (pathogenic variant in heterozygous in PPOX gene – FIGURE 2). Patient was treated with hematin and high glucose supply, presenting slow progressive clinical improvement.

**Legend:**

FIGURE 1: left image shows hematic appearance of the urine by the urinary catheter. Right image exhibit patient head CT revealing left frontal intraparenchymal hematoma (25ml), with mass effect and hemoventriculus.

FIGURE 2: genetic panel for porphyria was positive for variegate porphyria (pathogenic variant in heterozygous in PPOX gene).

**References:**

1. Cordonnier C, Demchuk A, Ziai W, Anderson CS. Intracerebral haemorrhage: current approaches to acute management. Lancet. 2018;392(10154):1257.
2. Gross BA, Jankowitz BT, Friedlander RM. Cerebral Intraparenchymal Hemorrhage: A Review. JAMA. 2019;321(13):1295.
3. Webb AJ, Ingale H, Irani SR, Hu MT. Acute variegate porphyria presenting with reversible cerebral vasoconstriction. Clin Neurol Neurosurg. 2016;146:102.
4. Ramanujam VM, Anderson KE. Porphyria diagnostics - Part 1: A brief overview of the porphyrias.Curr Protoc Hum Genet. 2015;86:17.20.1. Epub 2015 Jul 1.