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Rare disease detected by head ultrasound in an infant hospitalized for vomiting and irritability

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A 6 months old boy was referred to IIIrd Pediatric Unit Cluj-Napoca with pyloric stenosis suspicion. The symptoms started about 2 months before admittance with projectile vomiting, irritability, and failure to thrive. Initially physical exam was normal. Abdominal ultrasound did not revealed abnormal finding and gastro-duodenal passage through the pylorus was normal. The patient received symptomatic treatment without clinical improvement. During the admittance period he presented persisting irritability and started to have intermittent abdominal colic.

A head ultrasound was performed, shown above (see details in fig 1). To confirm the diagnosis a cerebral magnetic resonance examination was performed (fig 2). Also, a cardiac ultrasonography was performed.

Questions:

1. What is your diagnosis?
2. Which clinical sign in this case is specific for the patient's disease?
3. What do you expect to find at the echocardiography?

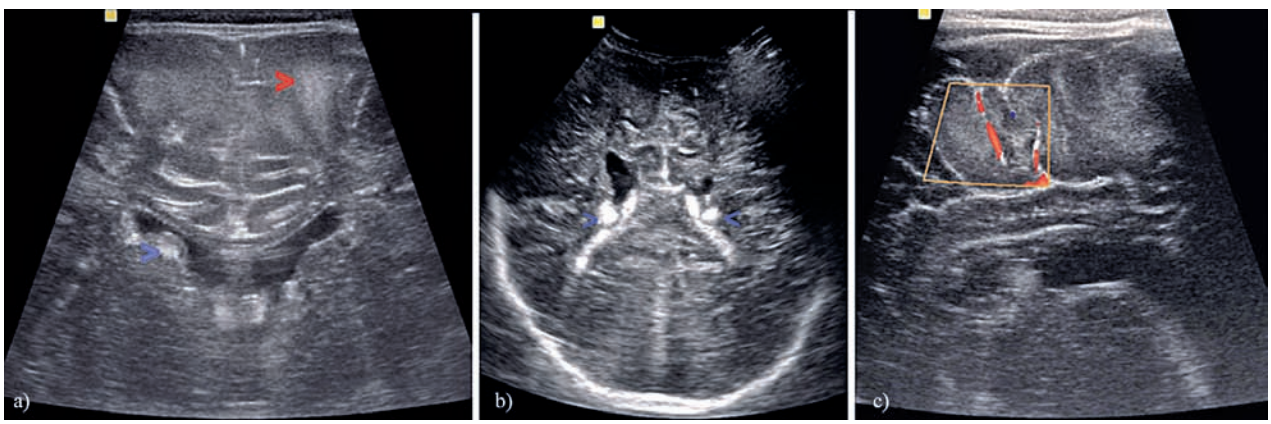


Fig 1. Head ultrasound: a) Coronal view at foramen Monro level; b) Coronal view at lateral ventricules level; c) Color Doppler interrogation on a sagital section. Ventricular system is symmetric and without enlargement. There are bilateral, multiple subependimal hyperechoic nodules (blue arrowheads). The nodules measure less than 10 mm in diameter, are homogeneous, well limited and without acoustic shadow. Cerebral parenchyma presents abnormal hyperechoic areas of the the white matter obvious at the subcortical level and there is abnormal poor differentiation between cortex and subcortical withe matter echogenicity (red arrowhead); these areas present a normal blood flow at the color Doppler interrogation.

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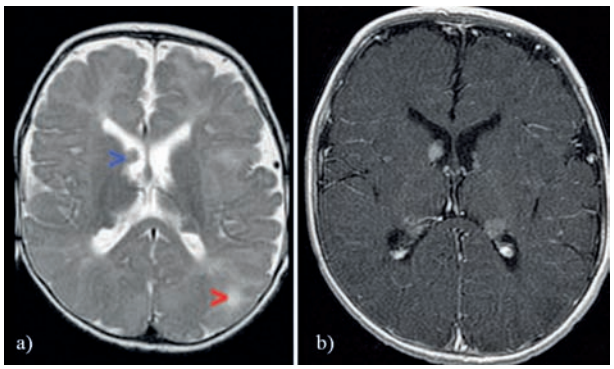


Fig 2. Cerebral magnetic resonance: a) T2 sequence – axial view; b) postcontrast study – axial view. Cerebral magnetic resonance confirms the ultrasound findings. There are multiple subependimal nodules (blue arrowheads) and withe matter hyperintense signal areas visible on T2 sequence located bilateral at the level of the migration lines (red arrowhead). Subependimal nodules presents contrast enhancement (From "Scandia Imagistica" Cluj-Napoca Collection, with permission).

Answer QUIZ vol 13 no. 2

Large cervical mass in a patient with a history of breast cancer

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Answer to questions

1. The differential diagnosis should include cervical adenopathy and a primary tumor mass of the cervical region. Adenopathy could be related to breast, lung, esophagus, respiratory tract or gastric cancer. The origin of a primary mass could be the upper gastro-intestinal and respiratory tract and the thyroid.

2. The right internal jugular vein is partially filled with a material of mixed echogenicity, which is in continuity with the cervical mass. Color Doppler ultrasound reveals that the thrombotic material in the internal jugular vein is vascularized and there is continuity between its vessels and those of the tumor. The presence of vessels in the thrombus is diagnostic of a tumor thrombus, by invasion of the internal jugular vein.

3. In this case, given the extensive tumoral thrombosis, adenopathy is improbable and the most likely diagnosis is a primary malignant mass. The position, lack of visualization of the right thyroid lobe and structure of the mass (calcifications) suggest a malignant thyroid tumor with direct invasion of the internal jugular vein.

The pathology report for this patient described a malignant epithelial proliferation, most likely of thyroid origin. This diagnosis was supported by the high thyroglobulin blood level (7684 ng/ml, normal values < 78 ng/ml) and the history of radiation therapy.

Overall, the main causes of internal jugular vein thrombosis are catheterization, surgical neck interventions, trauma, coagulation disorders and tumors of the head, neck and mediastinum [1]. Microinvasion of cervical veins is not uncommon in thyroid cancers and can only be diagnosed by the pathologist [2]. However, macroinvasion is uncommon and has most often been described in follicular and anaplastic thyroid tumors [3]. It may also occur, but is even more unusual, in well-differentiated thyroid tumors, such as papillary carcinoma [3]. Several case reports and small case series have been published where the tumor thrombus affected not only the jugular veins, but also the mediastinal veins, extending into the right atrium [1]. In the case of our patient, contrast-enhanced CT excluded invasion of the tumor in the mediastinum; it showed that the tumor thrombus did not extend below the junction of the right subclavian and internal jugular veins.

Bilateral venous thrombosis is also possible [2], but it was excluded by US and CT in our case.

For this patient, ultrasound was very useful to identify the presence and cervical extent of the mass, as well as the nature of the venous thrombus, which was an important clue to the diagnosis. However, invasion of the veins of the superior mediastinum and right atrium could only be excluded by CT.

The presence of venous thrombosis is considered a sign of aggressive behavior and the surgical treatment is more difficult in these cases. In spite of this, most authors recommend surgical resection, with ligation of the thrombosed internal jugular vein, accompanied by thrombectomy in cases where the thrombi extend in the mediastinal veins [4].

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