Basal ganglia T1* hyperintensity and chronic liver failure in a patient with Budd Chiari syndrome and MTHFR mutation in homozygous state

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C.T., female,78-year-old
diabetes mellitus, obesity, heart failure (NYHA II), Barrett's esophagus and chronic lymphedema with episodes of lymphangitis

In 90's, a diagnosis of hepatic veins thrombosis (**Budd Chiari syndrome**) associated with early stage liver cirrhosis was made

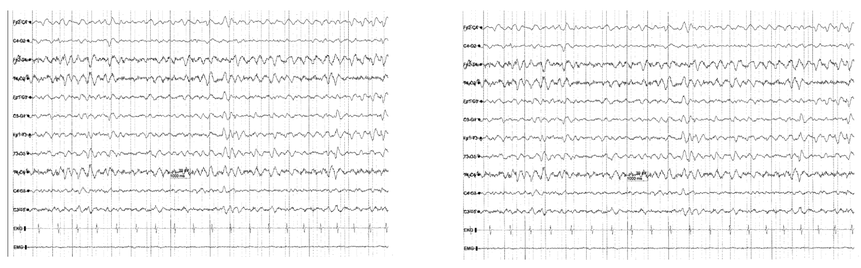
- ✓ the patient **never** took oral contraceptives and blood tests for hepatitis and autoimmunity were negative
- ✓ **no abdominal masses** were detected.
- ✓ on laboratory evaluation for inherited thrombophilia, the **c677t MTHFR mutation** in homozygous state was found, along with plasmatic hyperhomocysteinemia

Last year the findings of anemia and high levels of indirect bilirubin required an Internal Medicine ward

- ✓ abdominal TC scan **confirmed the diagnosis of Budd Chiari syndrome** and showed severe cirrhosis with portosystemic shunt.
- ✓ laboratory tests **excluded hemolytic anemia** and **myeloproliferative syndrome**, as **JAK-2** evaluation was **negative**.

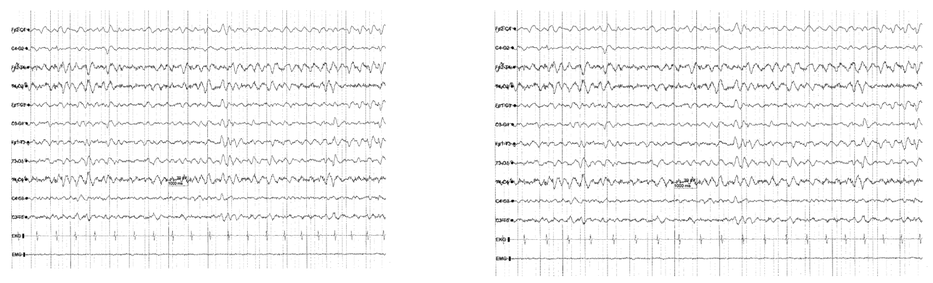
In March 2015, due to onset of a **severe acute impairment of consciousness without focal signs**, she was admitted to our neurological department

- ✓ **Neurological Examination:** mild kinetic and postural tremor, more evident in the right side, was the only clinical feature of extrapyramidal system involvement; this signs was clearly different from "asterixis" typical of hepatic encephalopathy
- ✓ **EEG**

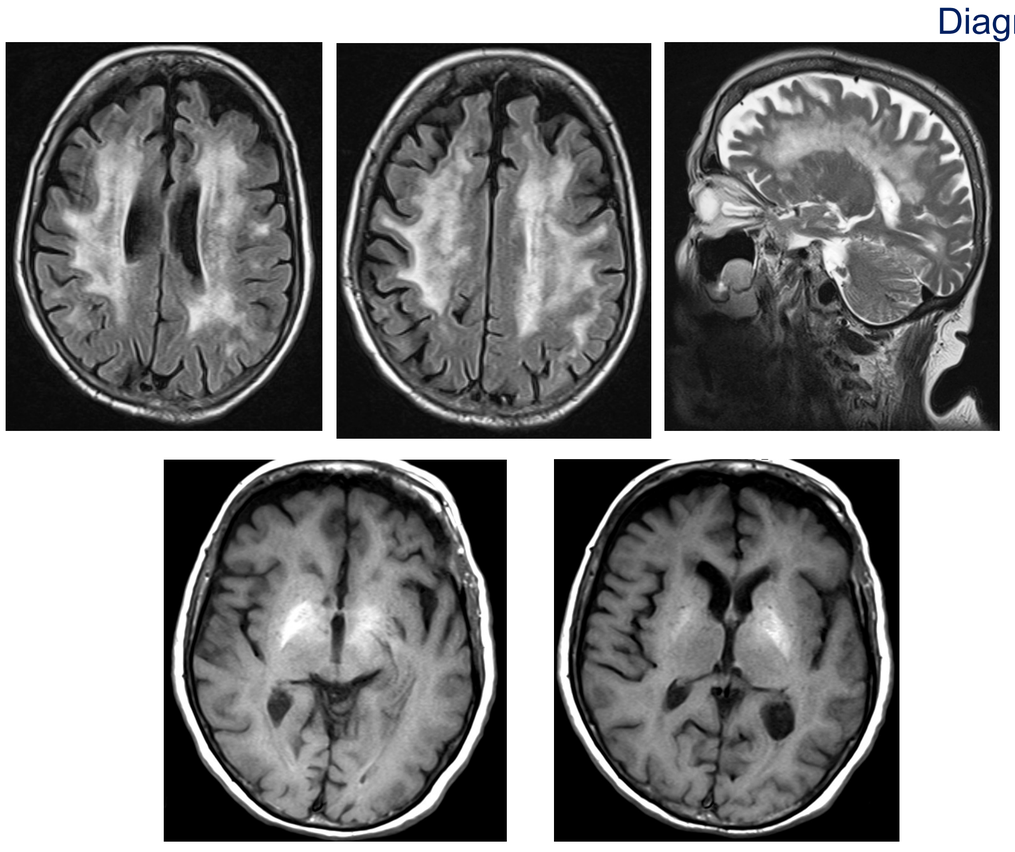


- ✓ **Blood tests:** plasmatic hyperammonemia

After the specific therapy,
the state of consciousness progressively improved,
along with the almost complete normalization of EEG



Diagnosis: **acute hepatic encephalopathy**



- ➢ **widespread** and **marked bilateral** and **symmetrical leukoencephalopathy**
- ➢ **bilateral T1*hyperintensity of pallidal, red and dentate nuclei**, compatible with deposits of paramagnetic material, in the first instance **manganese**

We have considered the aforementioned tremor as a manifestation of **Acquired Hepatocerebral Degeneration (AHCD)**, in which manganese has a crucial role.

We hypothesize that **homozygotic MTHFR mutation** was the thrombofilic substrate in the development both of **brain leukoencephalopaty** and **Budd Chiari syndrome.**

Recent literature identifies MTHR mutation and hyperhomocysteinemia as **indipendent risk factors for Budd Chiari syndrome**, which can lead to cirrhosis.

cirrhosis-related portosystemic shunt may led to manganese systemic and brain spreading , expecially in basal ganglia

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XLV CONGRESSO NAZIONALE 10-13 OTTOBRE 2015 – GENOVA

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