McARDLE DISEASE AND CAVERNOUS ANGIOMA: A CASE REPORT

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CASE REPORT

The patient was a 41 years old female, without previous history of neurological disease or familiar history pf neurological disease, admitted to San Martino Hospital in July 2014 for intense headache, with scotoma and phosphenes in right eye. The urgent brain CT **(FIGURE 1)** scan showed hyper density at optic chiasm and left optic tract, with extension to left temporo-mesial areas. The neurological examination (strength, sensibility, reflexes) was negative. The cerebral MRI with Gadolinium and Angio Sequences revealed absence of enhancement of the lesion, compatible with spontaneous or secondary hemorrhage **(FIGURE 2)** . Other exams were performed: Visive fields examination (relative defects in right infero-lateral fields), cerebral angiography (negative for aneurysm of other vascular malformations- **FIGURE 3).**

The follow up MRI (performed after about 4 months after the onset of symptoms) showed progressive reabsorption of hemorrhagic components, without enhancing of the lesion, suggesting the hypothesis of **cavernous angioma of left optic tract (FIGURE 4)** ; the patient was treated with symptomatic drugs for headache with good response. The neurological examination was always normal, except for intermittent scotoma and phosphenes in right eye.

Blood test performed during the hospitalization revealed repeated high levels of CPK till 3000 U/L, with gradual reduction without normalization (but always > 400 U/L). We performed Electromiography (EMG) at four limbs (negative), DBS for Pompe (negative) and muscular biopsy showing a suffering muscle for accumulation pathology, with marked reduction of coloration for myophosforilase, compatible with **glycogenosis type 5 (McArdle disease). The diagnosis was confirmed by genetic test.**

The patient complained of fatigue and muscle weakness, expecially after physical exercise since few years; she had normal mental and physical development and she did not report any familiar history of neuromuscular disease.

DISCUSSION

McArdle disease (glycogenosis type V or myophosphorylase deficiency) is an inborn disorder of skeletal muscle carbohydrate metabolism characterized by failure of muscle glycogen breakdown, with autosomic recessive inheritance. [1].

Glycogenosis, expecially Pompe Disease (glycogenosis type 2) can involve other tissue than skeletal muscle, in which is present glycogen accumulation, as **liver, hearth, smooth muscle, brain, liver, spleen ,salivary glands, kidney and blood vessels**. [2]. Some cases of vascular malformation are reported in Pompe disease; brain vascular abnormalities were reported in late onset included basilar artery dolichoectasia, internal carotid dilatative artheriopathy and aneurysm of basilar, internal carotid or cerebralis media arteries and can be a serious complication of late onset cases provoking bleeding leading to a severe neurological deficits or death. [3]

This is a rare case described of brain vascular abnormalities founded in a patient with McArdle disease; the causal association of cavernous angioma and the muscular disorder can be conceivable on the basis of literature data regarding glycogen accumulation in other tissue than skeletal muscle, in this case the endothelium of brain vessels.

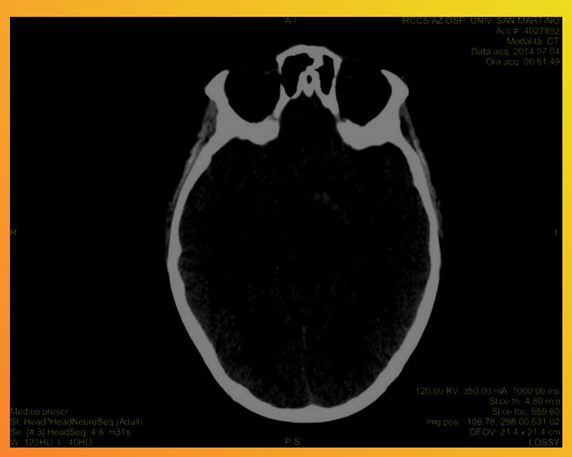

Figure 1: Cerebral CT scan

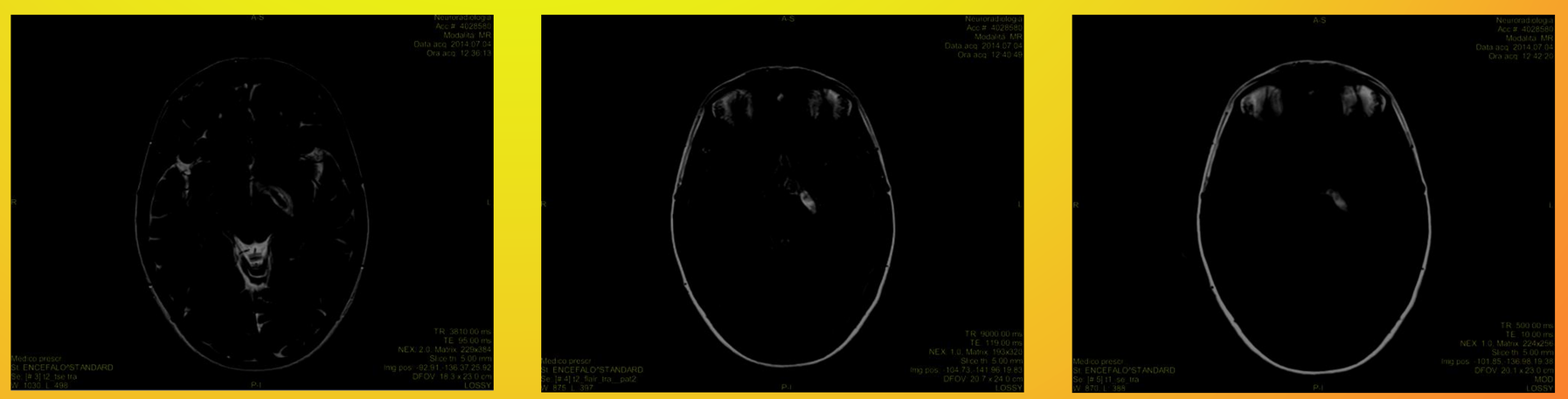

Figure 2: Cerebral MRI (T2-FLAIR and T1 sequences at admission)

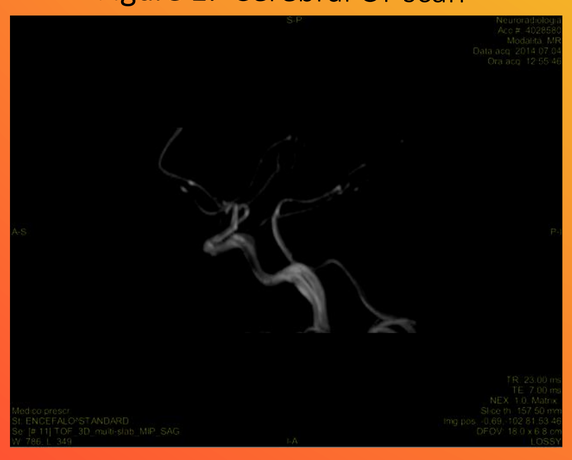

Figure 3: Cerebral angiography

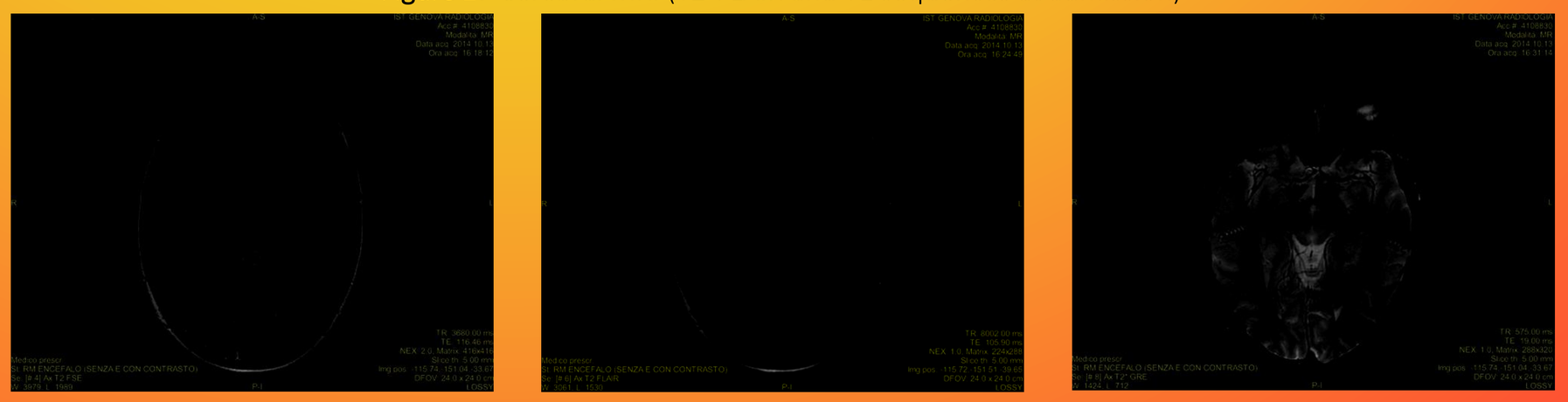

Figure 4 Cerebral MRI (T2-T1 and T2 * GE sequences)performed after 4 moths.

References:
1-Filosto M, Todeschini A. et al. Non muscle involment in late-onset glycogenosis II. Acta Myol. 2013 Oct; 32(2); 91-4.
2-Kohn TA, Noakes et al. McArdle disease does not affect skeletal muscle fibre type profiles in humans. Biol Open. 2014 Nov 28; 3 (12): 1224-7.
3-Sacconi S, Bocquet JD et al. Abnormalities of cerebral arteries are frequent in patients with late-onset Pompe disease. J Neurol 2010; 257:1730-1733.