Biotin thiamine responsive basal ganglia disease
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Imaging sequences
Imaging Technique: MR
Special Focus: Metabolic disorders Case Type: Clinical Cases
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Patient: 6 years, female

Clinical History:

6-year-old girl with a history of epileptic seizures, dystonia and mental retardation was admitted for a control brain MR. Seizures began at the age of 1 month. EEG at that time was abnormal over the central region. Leigh's disease was suspected initially. However, genetic testing showed a SLC19A3 gene deletion.

Imaging Findings:

Extensive areas of volume loss and signal change involving cortex and subcortical white matter of parietal and occipital lobes, the dorsal part of superior frontal gyri and the parasagittal precentral region is seen on T1 and T2-weighted images (Fig. 1, 2, 3).
Bilateral symmetric necrosis of putamina can be seen on both T1 and T2-weighted images on the level of basal ganglia. Similar changes can be appreciated in dorsolateral thalami (Fig. 4, 5). Subtle T2 hyperintensities are present in both caudate heads (Fig. 5).
Findings are consistent with severe chronic sequelae of biotin thiamine responsive basal ganglia disease.

Discussion:

Background:
Biotin thiamine responsive basal ganglia disease (BTBGD) is an autosomal recessive neurometabolic disease [1]. Mutation in SLC19A3 gene which encodes membrane thiamine transporter protein is a hallmark of BTBGD. It usually presents from ages 3-10 but can manifest in various periods of life, from the newborn to adulthood [3, 4].

Clinical Perspective:
It is characterised by 3 stages. Stage 1: fever, followed by subacute encephalopathy, vomiting and confusion. Stage 2: acute encephalopathy with seizures, quadripareis or quadriplegia, loss of developmental milestones, dysphagia and dysarthria. Stage 3: chronic encephalopathy with complete loss of comprehension and, if not treated, death [3].

Imaging Perspective:
Characteristic MR findings include bilateral necrosis in the basal ganglia, especially in the heads of caudate nuclei and putamina but with no involvement of globi pallidi.
Severe oedema and cortical hyperintensity is observed during the acute and subacute crisis. Treatment with biotin
and thiamine may cause resolution of the cortical abnormalities but the basal ganglia abnormalities typically persist. MR spectroscopy in the acute phase may show increased lactate and decreased NAA [2]. Symmetrical basal ganglia lesions are caused by various systemic or metabolic conditions, namely Wernicke’s encephalopathy (WE), mitochondrial diseases (Leigh's disease, MERRF) and CO poisoning. The main MR difference between WE and BTBGD is that mamillary bodies are not involved in the latter. Main difference between MERRF, CO poisoning and BTBGD is that BTBGD does not involve globus pallidus while the difference with Leigh's disease is that the latter less frequently involves the cerebral cortex [2].

**Outcome:**
BTBGD should be considered in every paediatric patient with acute onset extrapyramidal symptoms and bilateral symmetrical abnormal MR signals of caudate nuclei and putamina. Biotin and thiamine treatment is recommended until BTBGD is excluded.

Early administration of biotin and thiamine is crucial as it results in improvement within days while lack of treatment may result in death or permanent neurologic sequelae such as quadriplegics [1, 2].

**Differential Diagnosis List:** Biotin thiamine responsive basal ganglia disease, Wernicke encephalopathy, Leigh syndrome, MERRF, Toxic encephalopathy (CO poisoning), Wilson disease, Juvenile Huntington disease

**Final Diagnosis:** Biotin thiamine responsive basal ganglia disease

**References:**
Description: T1 sagittal images show extensive areas of volume loss involving predominantly subcortical white matter of parietal and occipital lobes as well as the dorsal aspects of superior frontal gyri. Origin: Department of Radiology, University Medical Centre Ljubljana, Slovenia
Description: Sagittal T2 images show extensive areas of volume loss and signal change involving cortex as well as subcortical white matter of frontoparietal and occipital region. Origin: Department of Radiology, University Medical Centre Ljubljana, Slovenia
Description: Low intensity signal and volume loss is clearly present in both putamina. Subtle changes are also present in both thalami. Findings are consistent with atrophy. Origin: Department of Radiology, University Medical Centre Ljubljana, Slovenia
Description: Axial T2 images show extensive symmetric bilateral atrophy cortically and subcortically in frontoparietal regions. Origin: Department of Radiology, University Medical Centre Ljubljana, Slovenia
Description: Hyperintensity with volume loss is visible in putamina and in lateral aspects of both thalami. Subtle hyperintensities can be appreciated in caudate heads. Findings are consistent with chronic sequelae of BTBGD. Origin: Department of Radiology, University Medical Centre Ljubljana, Slovenia