High signal intensity in basal ganglia and talamus in pediatrics diseases

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Learning objectives

- To review the most common pediatric diseases with hyperintense basal ganglia and thalamus injuries in the sequences of magnetic resonance imaging (MRI) T2-weighted and FLAIR.

- To establish a differential diagnosis between them according with the rest of radiology findings for a diagnostic approach.
Background

Basal ganglia are subcortical neuronal structures that form a circuit of interconnected nuclei. They are responsible for the initiation and integration of the movement.

They receive information from the cerebral cortex and the brain stem, process it and project back to the cortex, brain stem, and spinal cord to contribute to the coordination of movement.

The basal ganglia are composed of the following nuclei: 1) caudate, 2) lenticular (formed by the putamen and the external and internal globus pallidum), 3) and the amygdaloid complex.

The thalamus is composed of a series of ovoid nuclear complexes. It originates in the diencephalon being the most voluminous structure of this zone. It extends from the Monro’s foramen to the quadrigeminal plate.

Bilateral hyperintense injuries in basal ganglia and thalamus in the sequences of magnetic resonance (MRI) with TR prolongation may be present in numerous pediatric diseases, so it is necessary to establish a good differential diagnosis.
Findings and procedure details

1. MITOCHONDRIAL DISEASES

Mitochondrial diseases are a heterogeneous group of disorders caused by defects in the production of intracellular energy. Symptoms related to involvement of all organs have been described but the tissues that require more energy production are the most vulnerable, such as the brain and muscle.

1.1 Leigh disease (subacute necrotizing encephalomyelopathy)

It is mitochondrial disease that results from a disorder in the respiratory chain production of adenosine triphosphate.

Clinical manifestations are a consequence of a progressive dysfunction of the basal ganglia and white substance. The main finding are central hypotonia, developmental regression or arrest, ophtalmoplegia, ataxia and even respiratory dysfunction in advanced stages of the disease.

MR finding includes (T2W and FLAIR): bilaterally and symmetrically high signal intensity in long TR sequences (T2W and Flair in basal ganglia and thalamus.

Putamen is the most affected nucleus of the basal ganglia. The dorsomedial nucleus of the thalamus is also frequently affected.

**Figure 1 and figure 2.**

Axial diffusion-weighted images, during the acute phase, may show restriction but other times we observed a high signal intensity in axial diffusion-weighted images (figure 3) without evident restriction of diffusivity in the apparent diffusion coefficient map. **Figure 4.**

One way to establish the differential diagnosis with other entities that attend with high signal intensity in basal ganglia is by MRI spectroscopy in which we will observe an increase of the lactate peak in basal ganglia. **Figure 5.**

1.2. Glutaric aciduria type I
It is a rare disorder of organic acid metabolism caused by deficiency of glutaryl-CoA dehydrogenase. Improper degeneration of amino acids results in increased levels of glutaric acid. Accumulation of glutaric acid causes neurotoxicity in the basal ganglia and frontotemporal cortex which can lead to progressive dystonia, hypotonia, permanently impaired speech and seizures.

The most important MRI (T2W and Flair) findings are the high signal intensity in basal ganglia and thalamus.

In acute phases, like Leigh's disease, these lesions may show diffusion restriction.

The severity of the symptoms will depend on the involvement of the basal ganglia and this is the most important, but not the most frequent, neuroradiological manifestation of Glutaric aciduria type I.

Other associated findings would be: high signal intensity of subcortical white matter, markedly widened Sylvian fissures with wide open opercula "bat wings" (considered the most frequent radiological findings) and frontotemporal atrophy or hypoplasia.

Because dietary and drug therapy may alter the natural history of the disease (diet low in proteins and carnitine supplements), early diagnosis of such patients is critical.

1.3 Mytochondrial myopathy, Encephalopathy, Lactic Acidosis and Stroke (MELAS)

It is a mitochondrial disease with a very poor prognosis. Clinical manifestations: encephalomyopathy, ictal episodes and lactic acidosis. Like most mitochondrial diseases, it is characterized by failure to produce TPA in the affected cells.

Patients with MELAS syndrome usually appear healthy at birth with normal early development, then exhibit delayed growth, episodic vomiting, seizures, and recurrent cerebral injuries resembling stroke.

MR imaging demonstrates multiple cortical and subcortical infarct-like high signal intensity lesions that cross vascular boundaries, along with varying degrees of generalized cerebral and cerebellar atrophy. The parietal and occipital lobes and the basal ganglia are frequently involved. Figure 6.

1.4 Kearns-Sayre syndrome (KSS)
It is a rare sporadic multisystem mitochondrial disorder affecting muscle, the central nervous system and endocrine organs. It usually presents before the age of 20 with external ophthalmoplegia, atypical pigmentary retinopathy, heart block and cardiomyopathy, increase of proteins in CSF and cerebellar syndrome.

Histopathology it shows spongiform degeneration and vacuolization of nerve tissue. There is white and grey matter injury. Basal ganglia and thalamus can be affected by spongy degeneration, neuronal loss and gliosis. The cerebellum and the cervical spinal cord may be affected by spongy degeneration. Calcium deposit in basal ganglia can often be seen.

As in the rest of mitochondrial diseases, the neuroradiological findings are not specific to any of the mitochondrial disease types.

Cerebral and cerebellar atrophy are the most common findings observed in MRI in Kearns-Sayre syndrome, although alterations are also seen in the white matter and nuclei of the base. Figure 7 and Figure 8.

2. HYPOXIC-ISCHAEMIC BRAIN INJURY

Hypoxia-ischemia lesions have different forms of presentation; whether the patient is a full-term (RNAT) or preterm (RNPT) newborn.

In both full term and preterm, severe hypoperfusion will affect metabolically active regions such as the basal ganglia, even the brainstem of the brain and cerebellum.

These lesions will be visualized as high signal intensity T1W and variable signal in T2W (low signal intensity in the first two weeks, chronic phase high signal intensity due to glyosis).

Axial diffusion-weighted images show marked restriction with low signal in the ADC map.

3 NEUROCUTANEOUS SYNDROMES.

3.1 Neurofibromatosis type 1
Neurofibromatosis type I (NF-1), formerly known as von Recklinghausen disease, is an autosomal dominant genetic disorder classified as a neurocutaneous syndrome or phakomatosis. Diagnosis is usually established in childhood based on a series of well known major and minor criteria.

MR images (T2W) show multiple focal hyperintense lesions of the basal ganglia, cerebellar white matter, brainstem and bilateral thalami. No mass effect or contrast enhancement is observed. The exact nature of these lesions is unknown but are thought to be areas of myelin vacuolization vs. small hamartomas. It is important to recognize these lesions as they occur in 70-90% of NFI patients.

Figura 9 y 10.

Patients with NF-1 also have a higher risk of developing astrocyt tumors they are usually of low grade and have a better prognosis than the general population. The most frequent localization is in the brain stem and cerebellum. Figura 11.

4. SUBSTANCE ACCUMULATION DISEASES


It usually begins in adolescence with the presence of hepatic (hepatitis and cirrhosis) and neurological disorders (dysarthria, dystonia, ataxia, parkinsonisms and psychiatric problems)

The disease causes progressive degeneration of the neurons of grey nucleus and of the brainstem. The lesions that occur in the nervous system result from an intoxication of these neurons by the anomalous deposits of copper. They accumulate in the putamen, globus pallidum, caudate, thalamus and brainstem.

MRI findings show high signal intensity and atrophy in putamen nuclei.

Other areas of the brain and brain stem may be affected, such as the substance nigra, periaqueductal gray substance and tegmentum.

5. THALAMIC TUMORS.

Thalamic tumors are very rare.
They are usually unilateral and the most frequent histology is astrocytomas. Bilateral thalamic glioma is a low grade astrocytoma; however, due to its deep location, they have a very poor prognosis.

The most frequent clinical manifestations: headache, vomiting, apathy, even dementia.

MRI findings:; the tumor showed high signal intensity (T2W sequences) and isointense (T1W sequences). After administration of contrast, these tumors do not characteristically enhance.

An 11-year-old girl with oppressive headache that wakes her up at night and does not give up with painkillers. Some isolated vomit. Two weeks of evolution.

Intravenous noncontrast cranial CT showed bilateral supratentorial midline lesion producing lateral ventricle and third ventricle hydrocephalus. Figure 12

MRI shows high signal intensity (T2W) in both thalamus, with mass effect on the III ventricle and secondary obstructive hydrocephalus. Figure 13.

6. INFECTIOUS PATHOLOGY.

Both the basal ganglia and thalamus can be affected in the course of infectious diseases either by viruses, opportunistic fungal infections or even by the presence of abscesses.

Acute disseminated encephalomyelitis (ADEM) is the most common form of post-infectious encephalitis. It usually appears after a viral infection or more infrequently, postvacunal.

The initial clinical course is variable, may present with nonspecific infectious symptoms, acute or subacute neurological dysfunction. The initial evolution was variable but with a tendency towards total clinical resolution with steroid treatment.

The most frequent location of the lesions is the subcortical white matter, frontal or parietal, the involvement of the basal ganglia has been described in up to half of the children with acute disseminated encephalomyelitis.
Fig. 1: MRI (FLAIR): Bilateral high signal intensity in basal ganglia (white arrows) and thalamus (red arrow)

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Fig. 2: MRI (Coronal T2W): Bilateral high signal intensity injuries in putamen nuclei (whites arrows)

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Fig. 3: Axial diffusion-weighted images. High signal intensity in basal ganglia

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Fig. 4: Apparent diffusion coefficient map.

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Fig. 5: MRI spectroscopy. Lactate peak

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Fig. 6: MRI (T2W) Thickening of the cerebral cortex. Bilateral high signal intensity in thalamus.

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Fig. 7: Axial CT: low density un basal ganglia.

Fig. 8: MRI (T2W) Bilateral high signal intensity in basal ganglia and thalamus.

**Fig. 9:** MRI (FLAIR)

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Fig. 10: MRI (T2W)

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Fig. 11

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Fig. 12: Axial CT

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Fig. 13: Coronal T2W

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Conclusion

There are a lot of pediatrics diseases with high signal intensity injuries in basal ganglia and thalamus.

It is important to know the diseases associated with these findings and also, the rest of the associated radiology findings. In this way, we can establish an early diagnosis and appropriate treatment to improve prognosis.
References