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Case Report Open Access

Pantothenate Kinase-Associated Neurodegeneration (PKAN) With a Typical "Eye of the Tiger": A Radiology Case Report

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Abstract

Pantothenate Kinase-Associated Neurodegeneration (PKAN), a subtype of neurodegeneration with brain iron accumulation (NBIA), presents with distinctive radiological features, notably the "eye of the tiger" sign. We report the case of a 7-year-old boy presenting with progressive developmental regression, dystonia, and dysarthria. Neuroimaging via MRI revealed bilateral and symmetric hyperintensity within the globus pallidus, surrounded by a hypointense area, indicative of the characteristic "eye of the tiger" sign, although not pathognomonic. PKAN, caused by mutations in the PANK2 gene, results in impaired coenzyme A synthesis, leading to neuronal destruction and iron accumulation in the basal nuclei. While the "eye of the tiger" sign is highly suggestive of PKAN, it can also be observed in other conditions, necessitating correlation with clinical findings and genetic testing for confirmation. This case underscores the importance of recognizing radiological patterns in guiding diagnosis and management of neurodegenerative disorders like PKAN.

Case Report

A 7-year-old boy was referred to the radiology department for the evaluation of progressive developmental regression, dystonia and dysarthria. The neurological examination revealed upper limb tremor, spastic paraparesis with rigidity, dystonic movements and dysarthria. His birth history was unremarkable, and developmental regression began at the age of 4. Laboratory tests and EEG results were normal.

The patient underwent scanning using a 1.5T MRI system, with the protocol involving 3D FLAIR images, a 3D T1-weighted images, susceptibility-weighted imaging (SWI), and diffusion-weighted imaging (DWI). Both T2 and SWI images revealed bilateral and symmetric hyperintensity within the globus pallidus, surrounded with a hypointensity area. No other lesions were observed, including other basal ganglia and substantia nigra.

This combination of finding is suggestive of a typical "eye of the tiger", characteristic of pantothenate kinase-associated neurodegeneration (PKAN), though not pathognomonic.

Discussion

PKAN, formerly called Hallervorden-Spatz syndrome, is the most usual type of neurodegeneration with brain iron accumulation (NBIA), accounting for half of the NBIA cases and has an estimated prevalence of 1-3/100000 [1].

It is an autosomal recessive disorder resulting from a mutation in the pantothenate kinase 2-gene (PANK2). PANK 2 is essential for the production of the "pantothenate kinase 2 enzyme", which regulates coenzyme A (CoA) synthesis. Insufficiency of this enzyme leads to the destruction of the phospholipid membrane, primarily in the basal nuclei and retina, resulting in secondary iron accumulation [1,2].

PKAN is subdivided into two main types, based on clinical presentation

1. **Classic PKAN:** (75%) characterized by an early onset (usually between 3 and 4) and a faster disease progression. Symptoms include dystonia, dysarthria, rigidity, choreoathetosis and retinopathy pigmentosa.

2. **Atypical PKAN:** (25%) characterized by a later onset (usually at the age of 14) and slower progression, often revealed by dysarthria, mild gait abnormalities, subtle dystonia and neuropsychiatric features.

Neuroimaging in PKAN reveals distinctive iron accumulation in the globus pallidus. As the disease progresses, it may also affect the substantia nigra. The typical MRI finding, known as "the eye of the tiger", is identified on T2 imaging as a round medial area of hyperintensity surrounded with a hypointense globus pallidus.

The hypointensity is best appreciated on T2*/SWI sequences due to susceptibility effects, correlating pathologically with areas of abnormal iron deposition, while the center area of hyperintensity "the eye" corresponds to neuronal loss with gliosis.

Over time, the hypointensity may dominate the radiological presentation, and the central hyperintensity can disappear [3-5].

This sign has been documented in various other conditions, including carbon monoxide poisoning, cortical basal ganglionic degeneration, multiple-system atrophy, and other forms of NBIA specially neuroferritinopathy [3-6].

Conclusion

While "the eye of the tiger" is a characteristic finding, it is not specific or sensitive for PKAN and should be correlated with clinical context, confirmed by genetic testing demonstrating PKAN2 gene mutation.

Ethics approval

Our institution does not require ethical approval for reporting individual cases or case series.

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Informed consent

Written informed consent was obtained from a legally authorized representatives for anonymized patient information to be published in this article.

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