**Classical vs. Atypical**

Classical PKAN onsets during the first decade of life and has a quick rate of progression. Atypical PKAN may onset anywhere between the first and third decade of life and has a slower rate of progression. Symptoms overlap between the two forms, but some symptoms are more commonly found in one form than the other. Dystonia, spasticity, choreoathetosis, and gait abnormalities are typical of both forms.

<table>
<thead>
<tr>
<th></th>
<th>Classical</th>
<th>Atypical</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dysarthria</td>
<td></td>
<td>+</td>
</tr>
<tr>
<td>Retinopathy</td>
<td>+</td>
<td></td>
</tr>
<tr>
<td>Developmental Disabilities</td>
<td>+</td>
<td></td>
</tr>
<tr>
<td>Psychiatric symptoms</td>
<td></td>
<td>+</td>
</tr>
</tbody>
</table>

*An affected individual is not guaranteed to develop these symptoms; rather, these symptoms are found most commonly in individuals with this form of PKAN.*

*Psychiatric symptoms can manifest as changes in personality, Obsessive Compulsive Disorder, and Attention Deficit Hyperactivity Disorder.*

Other than time of onset there is no way to test for which type of PKAN an individual will develop. However, if the affected individual has an affected family member or members, they can expect their prognosis to closely follow the rest of the family, but this is not a guarantee.

**Words to Know**

Following diagnosis, it is common for affected individuals and their families to research PKAN. This list is meant to familiarize you with medical terms you will likely come across in your research.

- **Choreoathetosis** – irregular migrating muscle contractions distinguished by twisting and writhing
- **Dysarthria** – speech abnormalities including stuttering, slurring, or soft or raspy speech
- **Dystonia** – the involuntary muscle contractions and spasms of the head, neck, arms, and legs; can also affect mouth and throat
- **Eye of the Tiger** – a characteristic MRI finding where the formation of light regions, representing swelling and fluid buildup, surrounded by dark regions, iron accumulation, resembles the eyes of a tiger. This finding will normally confirm PKAN.
- **PANK2** – the gene responsible for production of enzymes that prevent iron accumulation in the brain
- **Retinopathy** – deterioration of the retina in the eye
What is PKAN?
Pantothenate Kinase-Associated Neurodegeneration (PKAN) is one of ten Neurodegeneration with Brain Iron Accumulation (NBIA) disorders. Specifically, PKAN is the result of a mutation in the PANK2 gene and is usually characterized by an “Eye of the Tiger” finding on an MRI scan. Genetically, PKAN is characterized as an autosomal recessive disorder. When both parents are carriers for the disorder, they have a 25% chance of having a child with PKAN. There are two forms of PKAN: classical and atypical.

PKAN is a progressive disorder consisting of periods of deterioration and stability in regard to their ability to walk, speak, and swallow. During periods of stability individuals won’t have any significant changes in their abilities. Periods of stability can last from a couple weeks to years. During periods of deterioration the individual experiences a decrease in capabilities. Periods of deterioration can last as long as a couple days to weeks.

Management
Currently there is no cure for PKAN, but there are therapies available to manage symptoms and increase the quality of life for affected individuals. What therapies an affected individual receives is dependent on the symptoms exhibited not on the form of PKAN. Though these aren’t the only therapies available to individuals with PKAN, these are usually the first therapies recommended to them.

Drug Therapy:
- Botulinum toxin or Botox (dystonia and spasticity)
- Trihexphenidyl (dystonia)
- Baclophen (dystonia and spasticity)
  - Oral or pump
- Clonazepam (dystonia and spasticity)
- Deferiprone (dystonia)

Non-Drug Therapy:
- Occupational Therapy
- Physical Therapy
- Speech Therapy
- Deep Brain Stimulation (dystonia)

What’s Next?
Following diagnosis, assessments are necessary to determine severity, identify needs, and address concerns of the affected individual. Necessary assessments include:

- Neurological exam
- Genetic counseling

Based on symptoms the following evaluations may also be necessary:

- Eye exam
- Speech evaluation
- Walking evaluation
- Nutritional and swallowing evaluation

Because PKAN is a progressive disorder these evaluations will become routine to ensure that the affected individual’s changing needs are being met.

Long-Term Care Options:

- Adaptive Aids (walkers and wheelchairs)
- Gastronomy tube (also called a G-tube)
- Caregivers
- Hospice

Resources:

- NBIA Disorders Association
- NBIAcure
- NBIA Alliance

*Though PKAN is housed under the umbrella term NBIA, it is important to note that every NBIA has its own unique set of symptoms and experiences.

Clinical trials offer opportunities to test medications that directly target the disorder and not just the symptoms. For more information about PKAN clinical trials visit ClinicalTrials.gov and search PKAN.