Neuropathy ataxia retinitis pigmentosa syndrome

NARP, NARP syndrome

Neuropathy ataxia retinitis pigmentosa syndrome, also known as NARP, is a rare mitochondrial condition where the mitochondria are not able to produce ATP molecules properly. ATP molecules are the cell's main energy source. Mitochondria are parts of a cell that help turn the energy we get from food into energy that the body can use. They are also important in the communication between body parts and in creating other materials the body needs. Mitochondrial conditions can cause a variety of signs and symptoms in many parts of the body, particularly those that use a lot of energy like muscles and the brain.

Genetics

NARP is an inherited genetic condition, meaning it is passed down in a family. NARP is caused by changes in the mitochondrial genome (mtDNA) or set of DNA contained in the mitochondria of a cell. NARP is typically caused by changes, called mutations or variants, in the gene *MT-ATP6*. Mitochondria are inherited from the mother through her eggs. People typically do not inherit any mitochondria from sperm, making it very unlikely for mitochondrial conditions to be passed down from a father. Both males and females can have NARP.

Frequency

The exact frequency of NARP is not known. It is thought to be less common than a similar condition called Leigh syndrome, which is thought to affect approximately 1 in 40,000 people.

Signs and Symptoms

NARP can cause a range of signs and symptoms typically beginning in childhood or early adulthood. These symptoms can vary widely between people who have the condition, even among members of the same family. Always check with your provider if new symptoms appear or you are concerned. Signs and symptoms may include:

- Numbness, tingling, or pain in the arms and legs (sensory neuropathy)
- Muscle weakness and problems with balance and coordination (ataxia)
- Changes in the light-sensitive tissue that lines the back of the eye (the retina). In some cases, these changes cause vision loss. (retinitis pigmentosa).
- Learning differences and developmental delays
- Loss of intellectual function (dementia)
- Seizures (epilepsy)
- Hearing loss
- Changes in the electrical signals of the heart (cardiac conduction defects)

People with NARP can be stable for years but have episodes of increased symptoms, often happening when someone is sick with a virus.

Diagnosis

NARP can be diagnosed by:

- Measuring biochemical markers in blood and cerebrospinal fluid (the fluid surrounding the brain and spinal cord)
- Brain studies and imaging looking for changes in the structure of the brain or seizure activity (MRI,

MRS, EEG)
- Heart studies looking for rhythm changes (EKG)
- Nerve studies (nerve conduction velocity study/NCV) to look for nerve function
- Performing a genetic test to look for changes in genes known to cause NARP

NARP is not included on newborn screening panels. If there is a known family history of NARP, prenatal testing can be performed on amniotic fluid (the fluid surrounding a baby) or chorionic villi (a specific part of the placenta). Results interpretation of this testing for mitochondrial conditions is complicated and genetic counseling is recommended.

Treatment and Management

Before beginning any treatment or therapy, please consult with your physician.

As of 2022, there is no FDA-approved therapy for NARP. Treatment and management of NARP are symptomatic and supportive. This may include:

- Traditional medications for seizures called antiepileptic drugs
- Physical therapy and aerobic exercise to support muscle function
- Traditional treatment of heart conditions
- Hearing aids or cochlear implants for hearing loss
- Avoidance of mitochondrial toxins like certain drugs, tobacco, and alcohol
- Mitochondrial supplements

People with NARP typically work with several healthcare providers regularly, including:

- Neurology for the muscles and brain
- Ophthalmology for the eyes
- Cardiology for the heart
- Audiology for hearing

All those living with or caring for someone with NARP must have an emergency protocol letter. These letters, which are written and signed by a doctor, share details about prescribed treatment during crises and in emergency room settings.

Clinical Trials

For specific details on clinical trials visit www.mitoaction.org/clinicaltrials or www.clinicaltrials.gov.

Resources

- Neuropathy Ataxia Retinitis Pigmentosa Syndrome - The National Institutes of Health
- Maternally Inherited Leigh Syndrome and NARP Syndrome - National Organization for Rare Disorders

Connecting with others who are impacted by a rare disease allows for important information to be shared about day-to-day life, prevents isolation, and gives hope. Please contact MitoAction for peer support opportunities at 888-MITO-411 or email mito411@mitoaction.org.

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