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PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION MOLECULAR DIAGNOSTIC CENTER

Pantothenate kinase-associated neurodegeneration (PKAN) (previously known as Hallervorden-Spatz syndrome) is a progressive neurodegenerative disorder with brain iron accumulation. Symptoms include progressive dystonia, dysarthria, rigidity, and retinitis pigmentosa. Classical PKAN has its onset in the first decade of life; atypical PKAN presents with later onset and slower disease progression. The disease is autosomal recessive and is caused by mutations in the pantothenate kinase 2 (*PANK2*) gene.

REASONS FOR REFERRAL

- Confirmation of clinical diagnosis in patients with classical or atypical PKAN
- Carrier testing of family members of PKAN patients
- Prenatal diagnosis

TESTING METHODOLOGY

Mutation scanning: Our laboratory uses PCR followed by denaturing high performance liquid chromatography (dHPLC) to screen the entire *PANK2* gene for heteroduplex DNA, indicating a DNA sequence change. Samples containing a change are directly sequenced to determine the presence of a disease-causing mutation or a single nucleotide polymorphism.

Mutation Analysis

>98% detection rate in patients with eye-of-the-tiger sign on MRI

Test reporting follows the ACMG guidelines.

SPECIMEN REQUIREMENTS

Blood: ACD (Solution A or B):

Adult: 5 mL
Child: 5 mL
Infant: 2-3 mL

Requisition form must accompany specimen including ethnicity, clinical and family history information. Prior to any genetic testing we recommend genetic counseling. To receive forms and information about prenatal diagnostic testing, please contact our laboratory.

TURNAROUND TIME: 3-4 weeks

CPT CODES: 83891, 83898x7, 83909x7, 83912

PKAN info 0205