Spinocerebellar Ataxia Type 1: \textit{ATXN1} CAG Repeat Analysis

\textbf{Test Code: MATXN}
\textbf{Turnaround time: 3 weeks}
\textbf{CPT Codes: 81401 x1}

\section*{Condition Description}

Spinocerebellar Ataxia Type 1 (SCA1) is characterized by progressive cerebellar ataxia, dysarthria and eventual bulbar dysfunction. SCA1 is an autosomal dominant ataxia caused by expansion of the CAG repeat tracts within the \textit{ATXN1} gene. The overall prevalence of autosomal dominant ataxia is estimated at 1-3/100,000.\textsuperscript{1,2} Various studies have shown that SCA1 accounts for approximately 6\% of the autosomal dominant cerebellar ataxias.\textsuperscript{3-5}

References:


\section*{Genes}

\textit{ATXN1}

\section*{Indications}

This test is indicated:

- Confirmation of a clinical suspicion of spinocerebellar ataxia type 1.

\section*{Methodology}

CAG repeat number is determined by PCR amplification and fragment size analysis. Additional evaluation for the presence of CAT trinucleotides that interrupt the CAG repeat tract is done by a \textit{SfaNI} restriction enzyme digest.

\section*{Detection}

Nearly 100\% of CAG expansions in the \textit{ATXN1} gene will be detected by this assay.

\section*{Reference Range}

Normal: 6-36 CAG repeats.
Mutatable normal: 36-38 CAG repeats.
Full Penetrance: 39+ CAG repeats.

\section*{Specimen Requirements}

* Preferred specimen type: Whole Blood

\section*{Type: Whole Blood}

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.