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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: Female

Specimen Collected: 12-Sep-22 13:15

Dysautonomia, Familial (ELP1) 2 Variants **|Received: 12-Sep-22 13:16** **Report/Verified: 12-Sep-22 13:54**

Procedure	Result	Units	Reference Interval
Fam.Dysautonomia (ELP1),Specimen	Whole Blood		
Fam.Dysautonomia (ELP1),Allele 1	**c.2204+6T>C** *		
Fam.Dysautonomia (ELP1),Allele 2	Negative		
Fam.Dysautonomia (ELP1),Interp	See Note f1 i1		

Result Footnote

f1: Fam. Dysautonomia (ELP1), Interp

Indication for testing: Carrier screening or diagnostic testing for familial dysautonomia.

Positive: One pathogenic variant, c.2204+6T>C, was detected in the ELP1 gene; therefore, this individual is at least a carrier of familial dysautonomia. Genetic counseling is recommended. This individual's reproductive partner should be offered screening for the disorder. At-risk family members should be offered testing to determine carrier status for the identified variant.

This result has been reviewed and approved by Yuan Ji, Ph.D.

Test Information

i1: Fam. Dysautonomia (ELP1), Interp

BACKGROUND INFORMATION: Dysautonomia, Familial (ELP1), 2 Variants
CHARACTERISTICS: Familial dysautonomia is a debilitating disease caused by abnormal development and survival of sensory, sympathetic, and parasympathetic neurons. Symptoms include gastrointestinal dysfunction, vomiting and autonomic crises, recurrent pneumonia, altered sensitivity to pain and temperature, scoliosis, and cardiovascular instability. Other characteristics include infantile hypotonia, deteriorating wide-based ataxic gait, and decreased life expectancy.
INCIDENCE: 1 in 3,600 Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: ELP1 pathogenic variants.
VARIANTS TESTED: p.R696P (c.2087G>C) and c.2204+6T>C.
CLINICAL SENSITIVITY: 99 percent in Ashkenazi Jewish individuals, unknown in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Variants other than p.R696P (c.2087G>C) and c.2204+6T>C will not be detected. Diagnostic errors can occur due to rare sequence variations.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Test Information

i1: Fam. Dysautonomia (ELP1), Interp
Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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