



Patient Name Jane Alver

Date of Birth 06/21/1985

Race/Ethnicity Hispanic

Gender Female

Ordering Physician Dr. Alpha

Date of Report 05/01/2022

Test Type AOA

ACC # A001234-2-02

Result Description

Your patient had a positive result in the SCN5A gene [c.204T>A; p.Tyr68Ter] and is associated with Brugada syndrome 1; Dilated cardiomyopathy, 1E; Long QT syndrome type 3. The specific genetic variant that was found was g.38633104A>T and is classified as Pathogenic.

Your patient had a positive result in the SCN5A gene [c.2440C>T; p.Arg814 Trp] associated with Dilated cardiomyopathy, 1E. The specific genetic variant that was found was g.38686038G>A and is classified as Pathogenic.

Interpretation Limitations

The interpretations set forth in this report are based only upon current scientific knowledge and technology. Each variant is interpreted independently of all other variants. This test attempts to use current scientific knowledge to identify possible genetic variants; however, current scientific knowledge about the function of variants, genes, and other portions of the genome, and the ways in which genetic disorders are inherited, is incomplete. For example, variants in different genes may sometimes interact to cause disease, and variants may modify the phenotype of a monogenic disease. It cannot be excluded that pathogenic variants were missed due to limitations inherent in the analysis method used in this test.

These results should be interpreted in the context of an individual's medical evaluation, family history, and racial/ethnic background. The data, interpretations and results of this test are not intended to recommend or discourage any specific treatment plan, product or course of action in an individual's medical care.

The identification of some variants, including those associated with disease, is limited by the current state of knowledge in the genomics field and the annotations of variants in currently available public and private databases. Variants found in the individual that are benign, likely benign, or of uncertain significance, as identified in the medical literature based on ACMG criteria, are not reported. Please note that variant classification and/or interpretation may change over time if and when more information becomes available.

General Limitations: This test evaluates only the genes and variants listed; other variants and/or genes may exist that are not detected by this test which may or may not be clinically relevant. This test does not account for all variations in the individual tested. Absence of genes and/or variants that are not detected by this test does not rule out the possibility that an individual has different predicted phenotypes due to the presence of other variants and/or genes that are not detected by this test or due to other factors such as drug-drug interactions, comorbidities and lifestyle habits. The phenotype of the reported variant may be substrate-dependent. Clinical interpretation of these results must be made in the context of other factors such as gender, age, weight, ethnicity, disease state, diet, organ function, concomitant therapy, and drug-drug interactions. Clinical correlation is advised. Only a physician, pharmacist or other healthcare professional should advise on the use of the medications prescribed.