arcensus

Your genetic report

John Doe

Subject ID XXXXXXXX

your guide

welcome

Dear John

Welcome at Arcensus!

About 30 years ago, I diagnosed my first patient with a complicated genetic disease using an innovative molecular test.

Back then, we all had a vision of how molecular knowledge could help detect diseases quickly and early and how we could use genetic knowledge to preventively offer the best medicine to our patients.

Here, the central mission is to make the benefits of good, transparent, and always relevant genetic analysis available to as many people as possible worldwide.

We want to close with a commitment to you: We will not be perfect, but we will listen to you; we will ensure that we treat our partners and our colleagues with respect, passion, humility, and integrity.

Yours
Dr. Arndt Rolfs, MD
Professor for Neurology

about you

arcensus-diagnostics.com

John Doe

Male- 167 cm; 70 kg
DOB 12.01.2003- 18 years old
Dummy street 10; dummy country
Dummy @email.com
Sample collection date: 12.08.2021

Subject ID: xxxxxxxxx
Family ID: yyyyyyyyy
Product: **myLifeHeart™**
Sample type: Buccal swab
Sample received on: 18.09.2021

Your symptoms

This information helps us to determine which genetic variants are most relevant to you

- Fatigue and weakness
- Shorten of breath

Your family history

Your father suffers from heart problems since he turned 40 years

Additional medical documents

No documents attached

your results

myLifeHeart™ Genetic Test

Date of report: **18.09.2021**

Findings related to your symptoms (**primary**)

SCN5A gene	**What does this mean?**
One pathogenic variation detected in this gene	This test found that you have a change to one of the two copies of the SCN5A gene. This result means that the genetic diagnosis of Brugada syndrome type 1 is confirmed. Brugada Syndrome is a rare and potentially life-threatening heart rhythm disorder that is sometime inherited. People with Brugada syndrome have an increased risk of having irregular heart rhythms beginning in ventricles, which are the lower chambers of the heart. This disease is subject to preventive measures and treatment in case it is necessary. Based on this result, there is a high likelihood that other close relatives might be at risk of developing this disease.

Findings unrelated to your symptoms (**incidental**)

	What does this mean?
No pathogenic variations detected	According to the ACMG* guidelines, the genetic test did not reveal any variant in the genes unrelated to your family history or clinical manifestations. Your genetic data is continuously checked with the latest clinical research and stays up to date in a rapidly evolving medical field.

**ACMG= American College of Medical Genetics and Genomics (https://www.acmg.net/)*

Prof. Arndt Rolfs, MD
Medical Director

Héctor Rodrigo Mendez, MD
Human Geneticist

next steps

Our recommendations

	Please, consider genetic testing to clarify the inheritance of this variant (mother or father) and objectively inform recurrence risk and risk for other close relatives
	Contact your attending physician to discuss options concerning preventive, monitoring, and potential treatments.
	Genetic counselling is recommended

Learn more

To understand more about **dilated cardiomyopathy disease**, please scan the QR Code or go to the website: **https://medlineplus.gov/genetics/condition/brugada-syndrome/**

Talk to us

A free 15- minute counseling session with Prof. Arndt Rolfs, MD is included in your purchase of this test. Please schedule your appointment with us here: https://calendly.com/arcensus/15min

Or email us at counseling@arcensus-diagnostics.com

additional information

SCN5A genetic variant details

Gene name	SCN5A
Transcript	NM_001099404.2
cDNA change	c.5587G>T
Protein change	p.(Glu1864*)
Zygosity	Heterozygous
Clinical significance	Pathogenic

Use of anonymized data

Your personalized data will be stored and re-analysed quarterly for at least 12 months, based on your consent. Thus, during this period you will be informed about latest scientific findings, benefit from continuously improved diagnosis, and if the case, for analysis of further family members. After that 12-month period you have the possibility to prolong the data storage and quarterly updated reporting by signing a further annual subscription period.

All data (personal, genomic) are stored and handled under the EU General Data Protection Regulation (GDPR) and Health Insurance Portability and Accountability ACT (HIPAA) regulations.

By agreeing with the separate consent for research use, you provide consent for the pseudonymized long-term storage of your DNA sample and genomic data set, and their use in research activities. The research activities are essential to the continuous improvement of the early diagnosis, treatment, and prevention of any disease; the insights we may gain from your data could make an important contribution to these efforts.

Did you know?

Genetics is rapidly evolving with the growing potential to interpret more and more mutations in the future. This increase in better scientific interpretation of mutations will make it possible to continuously improve the world's knowledge in describing the clinical consequences of given genetic mutations.

Arcensus myLifeHeart™ genomic testing for cardiovascular diseases offers the optimal package with the best conditions to diagnose cardiovascular diseases or to investigate the risk of their development – based on the complete analysis of more than 450 genes

Glossary

ACMG	The American College of Medical Genetics and Genomics (ACMG) is an organization composed of biochemical, clinical, cytogenetic, medical, and molecular geneticists, genetic counselors and other health care professionals committed to the practice of medical genetics.
cDNA change	In genetics, complementary DNA (cDNA) is DNA synthesized from a single-stranded RNA. A cDNA change indicates a DNA mutation or variation in a particular gene.
Incidental findings	Incidental findings are observations, results, or other findings that may occur during analysis but are unrelated to the goals of the analysis. In genetics, the incidental findings are reported based on ACMG guidelines or internal evidence and have medical value for patient care.
Pathogenic variation	A genetic variation that increases an individual's susceptibility or predisposition to a certain disease or disorder. Broadly, a pathogenic variation is known as mutation.
Primary findings	Primary findings describe pathogenic variations in a gene or genes that are relevant to the diagnostic indication for which the sequencing was ordered.
Protein change	In genetics, a protein change reflects the impact of the mutation identified at DNA level on the protein. The mutations can affect the sequence, structure, stability, interactions, activity, abundance and other properties of the protein.
SCN5A gene	The ***sodium voltage-gated channel alpha subunit 5*** (SCN5A) gene belongs to a family of genes that provide instructions for making sodium channels. The sodium channels containing proteins produced from the SCN5A gene are abundant in heart (cardiac) muscle cells and play key roles in these cells' ability to generate and transmit electrical signals. These channels play a major role in signaling the start of each heartbeat, coordinating the contractions of the upper and lower chambers of the heart, and maintaining a normal heart rhythm.

Transcript	The transcript represents a copy of the genetic information stored into a complementary strand of RNA (messenger RNA or mRNA).
Zygosity	Each individual receives one chromosome set from the mother, and one from the father. Zygosity is the degree to which both copies (from mother and from the father) of a chromosome have the same genetic sequence. Heterozygous means that only one chromosome has the genetic variation (or mutation); the other chromosome pair is normal (or wild type) Homozygous means that both chromosomes have the same genetic variation (or mutation). Compound heterozygous means that both chromosomes have genetic variations, but not the same. One mutation is inherited from the mother, one mutation from the father.