INVITAE

INVITAE DIAGNOSTIC TESTING RESULTS

Patient name:	KENNETH MCKENZIE	**Sample type:**	Blood	**Report date:**	18-MAY-2022
DOB:	22-FEB-1947	**Sample collection date:**	11-MAY-2021	**Invitae #:**	RQ2054253-1
Sex assigned at birth:	Male	**Sample accession date:**	13-MAY-2021	**Clinical team:**	Shayna Richards Jenna Kay
Gender:		**Patient ID (MRN):**	20000161842		

Reason for testing

Diagnostic test for a personal history of disease

Test performed

Sequence analysis and deletion/duplication testing of the 168 genes listed in the Genes Analyzed section.

- Invitae Arrhythmia and Cardiomyopathy Comprehensive Panel
- Add-on Preliminary-evidence Genes for Arrhythmia and Cardiomyopathy
- Add-on Sudden Unexpected Death in Epilepsy (SUDEP) Genes

ADDENDED REPORT

This report supersedes RQ2054253 (26-MAY-2021) and includes new information.

- Re-review of the available evidence indicates that the ALPK3 variant is associated with autosomal dominant ALPK3-related conditions.

RESULT: POSITIVE

One Pathogenic variant identified in ALPK3. ALPK3 is associated with autosomal recessive and autosomal dominant cardiomyopathies.

One Pathogenic variant identified in ACADVL. ACADVL is associated with autosomal recessive very long chain acyl-CoA dehydrogenase deficiency.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
ALPK3	c.4004_4005dup (p.Ala1336*)	heterozygous	PATHOGENIC
ACADVL	c.848T>C (p.Val283Ala)	heterozygous	PATHOGENIC
AKAP9	c.3744A>C (p.Glu1248Asp)	heterozygous	Uncertain Significance
SCN3B	c.584+2dup (Intronic)	heterozygous	Uncertain Significance
GAA	c.271G>A (p.Asp91Asn)	heterozygous	Benign (Pseudodeficiency allele)

About this test

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This diagnostic test evaluates 168 gene(s) for variants (genetic changes) that are associated with genetic disorders. Diagnostic genetic testing, when combined with family history and other medical results, may provide information to clarify individual risk, support a clinical diagnosis, and assist with the development of a personalized treatment and management strategy.

Next steps

- This is a medically important result that should be discussed with a healthcare provider, such as a genetic counselor, to learn more about this result and the appropriate next steps for further evaluation, treatment and/or management. This result should be interpreted within the context of additional laboratory results, family history and clinical findings.
- Consider sharing this result with relatives as they may also be at risk. Details on our Family Variant Testing program can be found at www.invitae.com/family.
- One or more variants were identified that are not known to cause disease. See the GAA variant(s) in the Variant Details section for more information.
- Register your test at www.invitae.com/patients to download a digital copy of your results. You can also access educational resources about how your results can help inform your health.