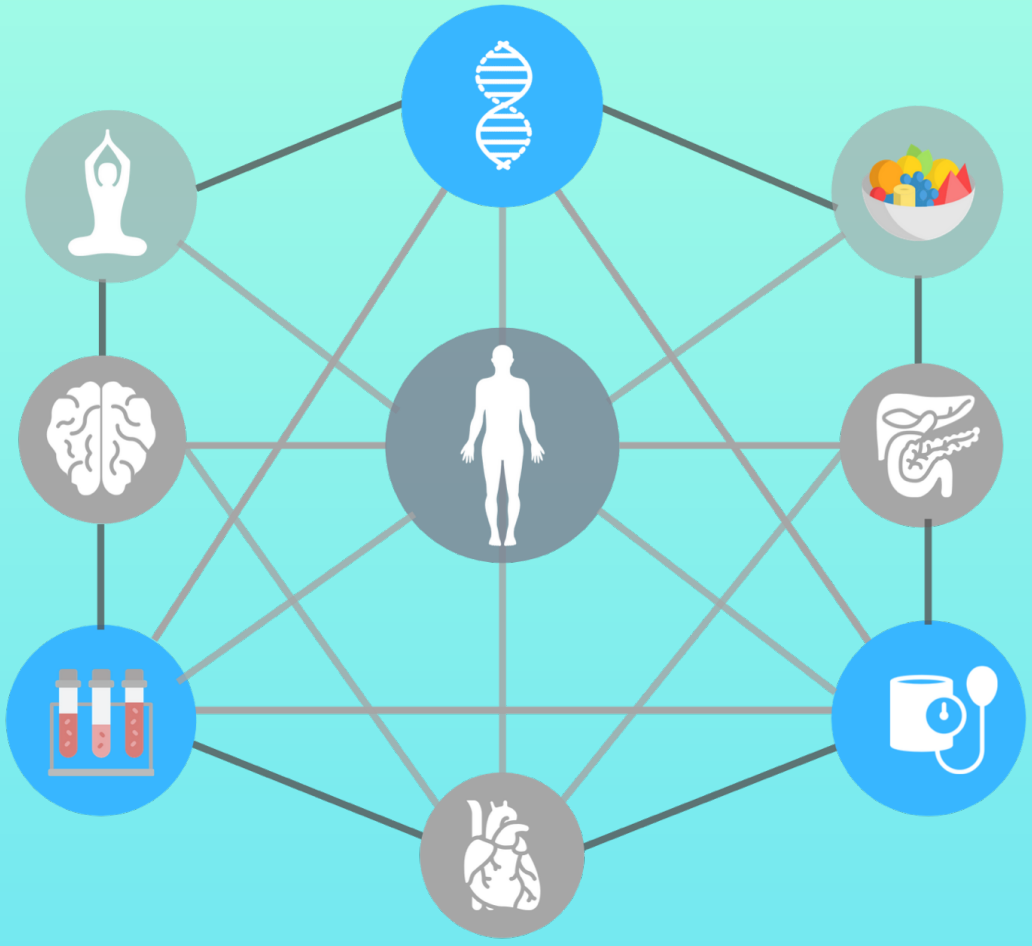

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For better clinical outcomes

Genetic Test Report

DG08081904

Report on 08 Aug 19

Name: Ajay
Age: 19
Sex: Male
Phone: +919215400152
Patient unique ID: DG08081904

Sample type: Blood (DNA)
Sample Collected on: 08 Aug 19
Referred by: Dr. Sandhya Koche
Clinical Diagnosis: Peripheral Neuropathy
Test Requested: Genotyping array

Clinical Information:

The patient is presented with weakness in lower limbs, tingling and burning sensations.

Disease tested for:

Peripheral Nervous System Disease and Chronic Inflammatory Demyelinating Polyneuropathy (CIDP)

Test result :

Negative for CIDP, 6 likely pathogenic heterozygous variations found in TTN gene (indicates slighly elevated risk of Peripheral Neuropathy.

Variant Type	Variant ID	Chr:Position	Genotype	Gene affected	Consequences	Implicatedin
Single nucleotide variation(SNV)	rs3829747	2:178532834	TC	TTN(Titin)	Missense Variant	Peripheral Neuropathy

The patient was screened for 65 genes and 5192 SNVs for Peripheral Neuropathy.

Gene description:

aaaaaaaaaaaaa

Additional findings*:

Disease Name	No. of genes considered	No. of SNPs considered	Average population risk	Your risk	Remarks
Abnormality of the Achiles tendon	30	1368	0.66	1.777786	2.6 x Higher Risk

Interpretation:

Altered TTN function could pose a high risk of developing Tibial Muscular Dystrophy, along with higher risk of abnormalities of the Achilles tendon could affect the muscles in the front of lower leg and result in steppage gait. Additionally, the abnormalities of the Achilles tendon can lead to pain and swelling in the heel and foot, as Achilles tendon connects the bones of your heel to your calf muscles and allows you to flex your foot and point your toes.

Recommendations:

Limitation of test:

Test results are interpreted in the context of clinical findings, family history and other laboratory data. Only variations in genes potentially related to the proband's medical condition are reported. Rare polymorphisms may lead to false negative or positive results. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. If results obtained do not match the clinical findings, additional testing should be considered.Specific genetic events like copy number variants, translocations and repeat expansions may not be reliably detected with targeted Genotyping microarray. In addition, due to limitations in technology, certain regions may either not be covered or may be poorly covered, where variants cannot be confidently detected. Also, genotyping array only identifies known polymorphism. Clinical exome is recommended for finding novel variations. Further sanger sequencing might be need to validation of certain mutations.

Disclaimer:

Any preparation and processing of a sample from patient material provided to WELLOWISE by a physician, clinical institute or a laboratory (by a "Partner") and the requested genetic and/or biochemical testing itself is based on the highest and most current scientific and analytical standards. However, in very few cases genetic or biochemical tests may not show the correct result, e.g. because of the quality of the material provided by a Partner to WELLOWISE or in cases where any test provided by WELLOWISE fails for unforeseeable or unknown reasons that cannot be influenced by WELLOWISE in advance. In such cases, WELLOWISE shall not be responsible and/or liable for the incomplete, potentially misleading or even wrong result of any testing if such issue could not be recognized by WELLOWISE in advance.

Reported verified by:

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Chief Scientist

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