dante labs

DANTE LABS™ TEST RESULTS

KIT ID: SAMPLE

Sensory Test

Introduction

The Sensory Test is based on Whole Genome Sequencing Test. As such, it analyzes all Common and Rare Variants Associated with Sensation Disorders, including Hearing Disorders, Olfaction Disorders, Somatosensory Disorders, Taste Disorders, Vision Disorders. Along with environmental factors, Genetics plays a key role in the regulation of Sensation Disorders.

In our analysis, we did not find any pathogenic variants.

Genes/Locations included in report:

- (0)	AHR (0)	NSF (0)	CPA6 (0)	ERC2 (0)	MGAM (0)	PRH1 (0)
SIK3 (0)	ACOD1 (0)	BTBD3 (0)	DHX15 (0)	DIRC3 (0)	HHLA1 (0)	NCAM1 (0)
OR2M7 (0)	OR4D6 (0)	OR5A1 (0)	PCDH9 (0)	PPM1H (0)	SPON1 (0)	STAB2 (0)
STAT6 (0)	ZPLD1 (0)	ADRA1D (0)	ARL17B (0)	PLPPR1 (0)	PTCHD4 (0)	STK32B (0)
THEMIS (0)	BTF3P11 (0)	CLEC12B (0)	NDUFA10 (1)	OR14C36 (0)	TAS2R14 (0)	TAS2R19 (0)
TAS2R38 (2)	KIAA1147 (0)	PPARGC1A (0)	AC0186302 (0)	LINC01768 (0)	AC000372.1 (0)	AC003975.1 (0)
AC011481.1 (0)	AC018630.2 (0)	AC024224.2 (0)	AC025265.1 (0)	AC073332.1 (0)	AC108748.1 (0)	AC112198.2 (0)
AL109838.1 (0)	AL139350.1 (0)	AL355297.1 (0)	AL512658.1 (0)			

Variants Found:

Gene/Loc	Chr: Pos	RSID	Phenotype Name	Zygosity	Variant	Allele Frequency	Significance	Review Status
TAS2R38	chr7:141672604	rs10246939	Phenylthiocarbamide tasting	HET	T>C	0.52057	drug response	
TAS2R38	chr7:141673345	rs713598	Phenylthiocarbamide tasting	HET	C>G	0.50479	drug response	
NDUFA10	chr2:240897186	rs77216981	Leigh syndrome	HET	C>T	0.04054	uncertain significance	★

Individual Variant Interpretations:

rs10246939 - NM_176817.5(TAS2R38):c.886A>G (p.Ile296Val)

Kim et al. (2003) identified an 886A-G transition in the PTC gene, resulting in an ile296-to-val (I296V) substitution (rs10246939). This polymorphism, in conjunction with other SNPs in the gene, give rise to the ability to taste or not taste phenylthiocarbamide (see 171200).

PubMed PMID: 12595690

rs713598 - NM_176817.5(TAS2R38):c.145G>C (p.Ala49Pro)

Within the PTC gene, Kim et al. (2003) found 3 common polymorphisms that influence the ability to taste phenylthiocarbamide (see 171200). One was a 145G-C transversion, resulting in an ala49-to-pro (A49P) substitution (rs713598).

PubMed PMID: 12595690

rs916977 - NM_004667.5(HERC2):c.1598+247A>G

In 3 independent genomewide association studies of a total of 1,406 persons and a genomewide linkage study of 1,292 relatives, all from the Netherlands, Kayser et al. (2008) found that the HERC2 variant rs916977 showed a gradient-wise (clinal) allele distribution across 23 European populations that was significantly correlated to iris color variation (227220), with the C allele, associated with blue eyes, being more common in northern Europe and the T allele, associated with brown eyes, more common in southern Europe. Analysis of rs916977 together with the 3 SNPs in intron 1 of the OCA2 gene identified by Duffy et al. (2007) (611409.0013) revealed significant genomewide association for only the HERC2 SNP (P = 3.53 x 10(-18)).

PubMed PMID: 17236130
PubMed PMID: 18252221

In our analysis, we did not find any related conditions

Methods

Extraction

Before sequencing, DNA extraction and library preparation processes were carried-out by automated liquid handling robots. Sequencing was completed using the NovaSeq 6000 instrument (Illumina).

The Nextera DNA Flex (Illumina) library was used during sequencing.

Analysis

Primary and secondary analysis was performed on the Illumina DRAGEN platform. Our secondary analysis extends the GATK 'best practices' pipeline. This includes Variant Quality Score Recalibration

It is important to note that applying a filter will not remove any data from the VCF file; it will just annotate the "FILTER" column. Variants with the "PASS" annotation are considered high quality and may, therefore, be used for advanced downstream analysis.

Sequence data is primarily aligned to the GATK GRCh37 reference genome and mitochondria is aligned to the Revised Cambridge Reference Sequence (NC_012920.1). Additional references may have been requested though tertiary analysis is not conducted on variant calls using references other than GRCh37.

Limitations

Test results are not interpretations. All variants reported in the genes included in the panel are reported.

Rare polymorphisms may lead to false-negative or false-positive results.

Due to limited read length and other contributing technical limitations, repeat expansions (e.g. in the Huntington gene, the SCA-genes, the myotonic dystrophy repeat region, and other similar regions) cannot be assessed with the applied method

Disclaimer

Any preparation and processing of a sample from saliva collection kit to Dante Labs by a customer is assumed to belong to the email used by the customer at the moment of kit registration on the Dante Labs Genome Manager platform before the shipment of the specimen to the laboratory.

The analysis and reporting conducted by Dante Labs are based on information from one or more published third-party scientific and medical studies.

Because of scientific and medical information changes over time, your risk assessment for one or more of the conditions contained within this report may also change over time. For example, opinions differ on the importance and relative weights given to genetic factors. Also, epidemiological data isn't available for some conditions, and this report may not be able to provide definitive information about the severity of a particular condition. We recommend asking your healthcare provider to correctly interpret them. Therefore, this report may not be 100% accurate (e.g., new research could mean different results) and may not predict actual results or outcomes.

This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained.

Contact

Please contact contact@dantelabs.com for more information on the contents of this report, our analysis methodology, and the limitations of this process.