# SUREGX a division of Suretox laboratory

# PARKINSON'S-ALZHEIMER'S-DEMENTIA

# SAMPLE REPORT

Client: <<docnametitle>>

Ordered By:

Physician: <<refphys>> Test: Complete Cancer Panel Patient Name: <<patname>>

DOB: <<dob>>
Gender: <<sex>>

Specimen Type: Oral Swab/Saliva

Accession #: <<accession>>

Collected: <<collectdtm>>
Accessioned: <<entrydtm>>
Reported: <<reportdtm>>

# Discover<sup>™</sup> Parkinson-Alzheimer-Dementia Risk Assessment Report

Gene	Classification	Zygosity	Variant Transcript
Negative	Negative	Negative	Negative

## **Negative Result: No Clinically Significant Alterations Detected**

**Additional Results: Variants of Uncertain Significance Identified** 

Location:

Consequence/Type: ClinVar/dbSNP:

**Sample Level Metrics** 

Amplicon Mean Coverage: Uniformity of Coverage: Low Coverage Regions:

Patient genomic DNA (gDNA) was isolated from patient sample(s) (buccal swab or spittle sample) using bead-based extraction chemistry. gDNA is quantified and quality checked following extraction. Targeted coding exons are isolated through the library preparation process utilizing the panel created by Paragon Genomics for identification of variants within genomic regions with known hereditary neurodegenerative disease relationships. PCR amplification of regions of interest is followed by sequencing on the Illumina Next Generation Sequencing (NGS) platform. The bioinformatics pipeline utilized prioritizes variants based upon their consequence to protein function and ultimately their clinical significance. Variants were interpreted manually using locus specific databases, literature searches, and other molecular biological principles. As a result of updates to knowledge in the field of genomics, variant classification and/or interpretation may change over time as more information becomes available. Only variations of clinical significance (primary findings) are included in this report, secondary/incidental sequence variant(s) are not.

#### Interpretation

#### Recommendations

- Always speak with a medical professional regarding your results
- Genetic counseling is recommended to discuss the implications of these test results
  - If you would like to discuss these results in further detail, please call our office directly at (201) 791-7293 to schedule an appointment with one of our genetic counselors.

### Parkinson-Alzheimer-Dementia Panel:

Parkinson's disease Discover, Alzheimer's disease Discover and Dementia disease Discover

Targeted Regions for "Parkinson-Alzheimer-Dementia Panel", includes the whole regions of the genes indicated below:

APOE, APP, ATP13A2, CHCHD10, CSF1R, DCTN1, DNAJC6, DNMT1, FBXO7, FUS, GBA, GCH1, GRN, HNRNPA2B1, HTRA2, LRRK2, MAPT, NOTCH3, PARK7, PINK1, POLG, PSEN1, PSEN2, SNCA, TAF1, TARDBP, TBK1, TH, TREM2, TYROBP, UBQLN2, VCP, VPS35

Ph: (201) 791-7293 Fx: (866) 425-4630 www.suretoxlab.com 241Molnar Drive Elmwood Park, NJ 07407 Director: Robert Rush, Ph.D. Assistant Director: Seshamma Reddy, M.D., Ph.D. CLIA#: 31D2063148

Page **1** of **2** 



# PARKINSON'S-ALZHEIMER'S-DEMENTIA

SAMPLE REPORT

Patient Name: <<pathame>> Accession #: <<accession>>

#### **Test Indication:**

\*All genes in panel are run for each patient, only clinically significant variants are reported. While not all genes will be found to be wildtype in their genetic code, any variations seen and not reported were not clinically significant and therefore not known to cause disease according to current research.

#### **Test Limitations**

Variants may exist in genes other than the ones listed in this panel or in novel ways that have yet to be clinically characterized. As such, this test does not represent all of the possible variations in the entire exome or genome of the individual. Further testing should be discussed with a medical professional or genetic counselor based upon the coverage regions of this test and potential ability of other testing to elucidate information that may be clinically significant. Only the protein coding regions of these genes are included in the analysis and reported herein. Variations outside the sequenced regions of these genes are not included in the genetic risk analysis. Certain types of mutations other than single nucleotide changes or smaller deletions/duplications may not be identified based on the current Next Generation Sequencing analysis technology used for this test. Examples of mutations that are not covered under this test: mosaicism, chromosomal aberrations, insertions or deletions larger than 50bp, or copy number variants. False negatives can occur if DNA quality is suboptimal (but passes minimum thresholds for quality control), if patient has received a recent bone marrow transplant. Based on study design and technology differences between laboratories, reported variants may not be the same from the same subject. The sensitivity and specificity of these tests varies between facilities due to differences in chemistry and bioinformatics analysis.

#### Disclaimer

This report was generated using the materials and methods described above, which required the use of various reagents, protocols, instruments, software, databases, and other items, some of which were provided or made accessible by third parties. A defect or malfunction in any such reagents, protocols, instruments, software, databases, and/or other items may compromise the quality or accuracy of the report. The report has been generated based on, and incorporates references to various scientific manuscripts, references, and other sources of information that were prepared by third parties that describe correlations between certain genetic mutations and particular diseases (and/or certain therapeutics that may be useful in ameliorating the effects of such diseases). Such information and correlations are subject to change over time in response to future scientific and medical findings. SureTox makes no representation or warranty of any kind, expressed or implied, regarding the accuracy of the information provided by or contained in such manuscripts, references, and other sources of information. If any of the information provided by or contained in such manuscripts, references, and other sources is later determined to be inaccurate, the accuracy and quality of the report may be adversely impacted. SureTox is not obligated to notify you of any impact that future scientific or medical research findings may have on the report. The report must always be interpreted and considered within the clinical context, and a physician should always consider the report along with all other pertinent information and data that a physician would prudently consider prior to providing a diagnosis to a patient or developing and implementing a plan of care for a patient. The report should never be considered or replied upon alone in making any diagnosis or prognosis. The manifestations of many diseases are caused by more than one gene variant, a single gene variant may be relevant to more than one disease, and certain relevant gene variants may not have been considered in the report. In addition, many diseases are caused or influenced by modifier genes, epigenetic factors, environment factors, and other variables that are not addressed by the report (or that are otherwise unknown). As such, the relevance of the report should be interpreted in the context of a patient's clinical manifestations. Medical knowledge annotation is constantly updated and reflects the current knowledge at the time. The test performance characteristics were determined by SureTox. The report was generated by SureTox as required by the CLIA 1988 regulations. The report, and the tests used to generate the report, have not been cleared or approved by the U.S. Food and Drug Administration (FDA), since FDA has determined that such clearance or approval is not necessary. This laboratory is CLIA certified to perform high complexity testing.

Ph: (201) 791-7293 Fx: (866) 425-4630 www.suretoxlab.com 241Molnar Drive Elmwood Park, NJ 07407

Director: Robert Rush, Ph.D. Assistant Director: Seshamma Reddy, M.D., Ph.D. CLIA#: 31D2063148