



Gene by Gene serial:

Targeted sequencing of the GBE1 gene

Tested individual surname:	Sample: Buccal Swab	Date Received:
Tested individual name:	Customer serial:	Date Reported:
Tested individual I.D.:	Gene by Gene serial:	
Sex:	Date of Birth:	

Results

1. No variants were detected within the boundaries of the targeted region.

Targeted genomic regions

1. GBE1: Chr3: g.81691938T>C; c.986A>C. Results are reported with respect to NM_000158.3 and the NCBI annotation 105: GRCh37/hg19.

Discussion

1. Sequencing of the GBE1 gene yielded no variants in the targeted region.

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Gene by Gene is a College of American Pathologists (CAP) accredited and Clinical Laboratory Improvement Amendments (CLIA) certified clinical laboratory qualified to perform high-complexity testing. This test was developed and its performance characteristics determined by Gene by Gene. It has not been cleared or approved by the FDA. FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research. Only the genomic regions listed above were tested; there is a possibility that the tested individual is a carrier for additional, undetected mutations. Direct Sanger sequencing of a gene is unable to identify large deletions of a gene or part of it. Although molecular tests are highly accurate, rare diagnostic errors may occur that interfere with analysis. Sources of these errors include sample mix-up, trace contamination, and other technical errors. The presence of additional variants nearby may interfere with mutation detection. Genetic counseling is recommended to properly review and explain these results to the tested individual.