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2450 Holcombe Blvd, Houston, TX 77021

Patient Name:	[redacted]			Shirley Lasch,	
Date of Birth:	[redacted] 1961	Lab Number:	708032	Institute for Neurodegenerative Disorders	
Gender:	M	Family #:	555661	Tel No. :	203-401-4323
Hospital/MR #:		Date Collected:	December 11, 2016	Fax No:	203-508-1515
Accession #:		Date Received:	December 21, 2016	CC: Cheryl Halter, M.S. Fax # : 317-278-4507	
Sample Type:	SALIVA	Date Reported:	January 26, 2017	CC: Laura Heathers, Fax # : 317-278-1100	
Test Code:	60130				
Indication:	No Indication Provided				

GBA Expanded Mutation Panel

708032-1833326

RESULTS: **No GBA variants detected**

INTERPRETATION

The sample was tested as part of the Parkinson's Progression Markers Initiative (PPMI) research study. Parkinson's disease is a chronic and progressive movement disorder that affects approximately 1-2% of the population. It is a complex disorder, with both environmental and genetic risk factors contributing to development of the condition. Specific changes (i.e., mutations) in the GBA gene has been associated with Parkinson disease risk. A person who carries at least 1 copy of the mutation(s) being tested (i.e., a heterozygous mutation) has an increased chance of developing Parkinson's disease, compared to a non-carrier. The performed targeted mutation analysis will only identify the mutations listed in the methodology section in the GBA gene. Testing will not identify any other variants or mutations that may be present within the GBA gene, or any other genes. Genetic counseling is recommended and is available through the PPMI research study.

METHODOLOGY:

This analysis was performed using PCR and pyrosequencing. The mutations tested are: c.84dupG (historically known as 84GG), c.115+1G>A (historic nomenclature: IVS2+1G>A), c.1226A>G (p.N409S historically known as p.N370S) and c.1448T>C (p.L483P historically known as p.L444P). Individuals being studied should understand that rare diagnostic errors may occur. Possible sources of diagnostic errors include sample mix-ups, erroneous paternity identification, and genotyping errors. Genotyping errors can result from trace contamination PCR, from maternal contamination of fetal samples, from rare genetic variants which interfere with analysis, from mosaicism at levels below standard detection and other sources.

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Christine M. Eng, M.D.
Medical Director

Linyan Meng, Ph.D.
Assistant Laboratory Director

This test was developed and its performance characteristics determined by Baylor Miraca Genetics Laboratories DBA Baylor Genetics (CAP# 2109314/ CLIA# 45D0660090). It has not been cleared or approved by the FDA. The laboratory is regulated under CLIA as qualified to perform high-complexity testing. This test is used for clinical purposes. It should not be regarded as investigational or for research.

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