Alzheimer Risk Assessment Report

&

APOE GENOTYPING

阿茲海默風險評估

(高風險報告範本)

Patient:	Hospital ID:
Age: 73 歲	Physician:
Gender: Female	Specimen ID: AZ#
Education:	Sampling Date: 2022/ /

Lab Requisition #:
Specimen Type: Touch Exosome DNA
Date Reported:
Clinical Indication: 1. Episodes of depression (憂鬱症史): 2. Head traumas (頭部挫傷): 3. Concomitant cardiovascular disorder (伴發性心血管疾病): 4. Diabetes (糖尿病): 5. Thyroid disease (甲狀腺疾病): 6. Others (其他疾病):

RESULTS:

ALZHEIMER RISK ASSESSMENT(阿茲海默症即時性風險評估)

Alzheimer's disease risk genes	**APOE**	**SORTILIN 1**	**RISK LEVEL**
Deterioration 退化程度	+++	++	*HIGH RISK* 高風險
Functional Implication 功能指標	Cholesterol carrier in the brain 腦部膽固醇載體	beta-amyloid (Aβ) peptide deposition β澱粉樣蛋白胜肽堆積	
TEST RESULT: Moderate risk of cognitive impairment 中度認知能力受損			

INTERPRETATION

WARNING 警示

Mild Dementia 輕度失智

當特定欄位為" -" 時，該基因目前"無退化風險".
當特定欄位為" +" 時，該基因目前已出現"低度退化風險".
當特定欄位為" ++" 時，該基因目前已出現"中度退化風險".
當特定欄位為" +++" 時，該基因目前已出現"高度退化風險".

APOE ε4 ALLELE BURDEN(APOE 先天遺傳基因分型)

	APOE LOCUS (APOE 基因位區)					
SNP (rs429358)	(C; C)	(C; T)	(C; T)	(T; T)	(T; T)	(T; T)
SNP (rs7412)	(C; C)	(C; C)	(C; T)	(C; C)	(C; T)	(T; T)
Genotype 基因型	**APOE ε4/ε4**	**APOE ε3/ε4**	**APOE ε2/ε4**	**APOE ε3/ε3**	**APOE ε2/ε3**	**APOE ε2/ε2**
ε4 Allele Burden ε4 等位基因序列負荷	2	1	1/2	0	-1	-2
Population Frequency	2.5%	13.5%	1.2%	74.8%	7.3%	0.7%
Risk 測得之風險	~10x	~5x	~2x	1x	~1/2x	~1/4x

The ε4 allele of the apolipoprotein E (APOE) gene is the strongest genetic risk factor and is associated with an increased risk for both sporadic and familial forms of Alzheimer disease.

載脂蛋白基因 ε 4等位序列（APOE ε 4）是最強大的阿茲海默風險遺傳因數，與偶發性阿茲海默症（Sporadic AD）和家族性阿茲海默症(Familial AD)風險的增加相關。

INTERPRETATION

APOE STATUS: ε2/ε3 HOMOZYGOTE GENOTYPE

阿茲海默失智中度風險

RECOMMENDATION

Regular Alzheimer exam every SIX (6) months.

(建議每 6 個月作一次阿茲海默失智風險檢查)

INTENDED USE

The AlzheiNostics Test is a composite genetic test, including polymorphism genotyping of apolipoprotein E gene (*APOE*) and **DNA** differential energetic mapping in the APOE locus. The test is intended to assess both the real-time as well as the predictive risk to the development of Alzheimer's disease.

AlzheiNostics 檢測是一種複合型基因檢測，包含載脂蛋白（APOE）基因的多態分型和 APOE 位區 DNA 等差能階的檢索。該檢測對阿茲海默症當下發生的即時性風險及未來發生的預測性風險同時評估。

PRINCIPLE

The genetic contribution to late-onset Alzheimer's disease (LOAD) is now well established, with heritability estimates ranging from 58 to 79%. Located on Chromosome 19, the APOE gene represents the largest genetic risk factor for LOAD to date, with genetic variation producing three isoforms: ε2 (protective), ε3 (neutral and most common form) and ε4, which is associated with increased risk for LOAD. In addition to this predictive AD risk factors, irregular gene activities such as under-expression of APOE gene correlate directly to the real-time risk of Alzheimer's disease.

目前已完全確立晚發性阿茲海默症（LOAD）乃溯源於遺傳，估計有 58%至 79%的元素源自遺傳。APOE 基因位於 19 號染色體上，是晚發性阿茲海默症(LOAD)迄今為止最大的遺傳危險因數，其基因變異可產生三個等形蛋白： ε 2（保護性）、 ε 3（中性， 為最常見的形式）及與 LOAD 風險增加有關的等位序列 ε 4。除了以上這三種預測性的風險因數外，非正常的基因活動，譬如 APOE 基因的低下表達，也與阿茲海默症的即時性風險有著直接關聯。

The AlzheiNostics Test analyzes the genomic DNA to differentiate the energetic level of the APOE gene and directly correlates the irregular APOE gene activities in the context of DNA aging to the real time risk of Alzheimer disease development. The test results can be cross-examined and further verified by the **ε**4 allele burden, which in turn serves as a predictive risk factor for LOAD.

AlzheiNostics 檢測分析全基因組 DNA 來區分 APOE 基因的能量位階，並將 APOE 基因的非正規活動以 DNA 老化為內涵與阿茲海默症的即時性風險直接串聯。此檢測結果並可進一步對 ε 4 等位序列負荷進行交叉查驗 LOAD 的風險。

CAUTION

The AlzheiNostics Test is specific to Alzheimer dementia and is not intended for non-AD neurodegenerative disorders. The test results should NOT be used as a stand-alone guide for any medical decision in the absence of ancillary diagnostic procedure. Deviations from the "Sample Collection Procedure" recommended for the AlzheiNostics Test may compromise the performance of the test.

AlzheiNostics 檢測是專門為區分阿茲海默症而設，不適用於非 AD 神經退行性疾病。在沒有輔助診斷程序的情況下，不應將檢測結果用作任何醫療決策的獨立指南。沒有按照 AlzheiNostics 檢測之「樣本收集步驟」進行採樣可能會損害檢測結果的準確度。

Sign: *BING LING, MD* Date: 2022 / /