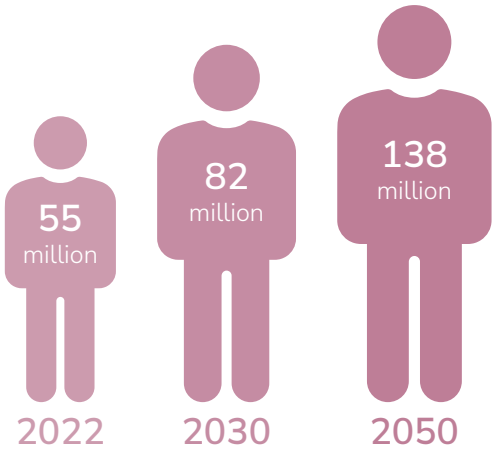
Alzheimer's Disease Risk Genetic Testing

Proactive Healthcare

Early Detection and Prevention with *APOE* Genotyping Testing

The impact of Alzheimer's disease

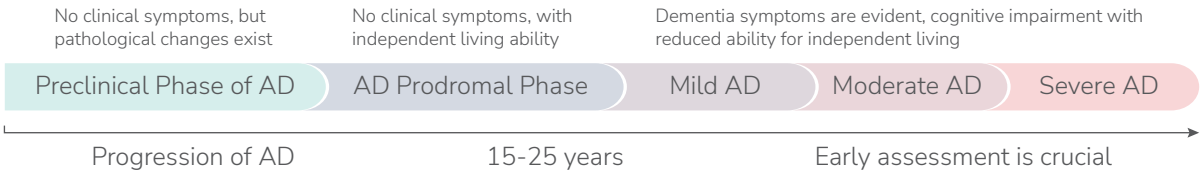
Alzheimer's disease (AD), the most common form of dementia (accounting for 60-80% of dementia cases), is a neurodegenerative disorder primarily affecting older adults, characterized by progressive cognitive decline and behavioral changes. According to the World Health Organization, AD and other forms of dementia rank seventh among the top ten causes of death worldwide. There is no cure for AD, and current treatments can only temporarily ease symptoms. Dementia, including AD, cost the global economy $1.3 trillion in 2019, with around 50% of costs related to care provided by informal caregivers. As the population ages, the prevalence of AD and related cognitive disorders is rising.



Note: the data from the "World Alzheimer Report 2022"[1].

Prevention of AD is key

AD's early symptoms mimic normal aging. The transition from normal cognition to mild cognitive impairment (MCI), and eventually to AD in elderly individuals, can take 15-25 years[2]. It is difficult to detect and identify in the early clinical stage. The Alzheimer's Association emphasizes that early diagnosis and intervention during the preclinical phase of AD is important to slow disease progression[3]. Alzheimer Europe also stresses the significance of early intervention in AD, advocating for the use of biomarkers to facilitate early detection and monitoring[4].



Genetics and AD Risk

The onset of AD is associated with various risk factors, including increasing age, infections, cardiovascular diseases, and unhealthy lifestyles[5]. **Among these, 60%-80% of the risk is attributed to genetic factors[6].**

APOE4 is a major genetic risk factor for AD, and its risk is gene dose-dependent. Homozygotes (carrying two *APOE4* alleles) have up to a 15-fold increased risk of developing the disease, whereas *APOE2* can reduce the risk of AD by nearly half and help extend lifespan[7].

Alzheimer's Disease Risk Genetic Testing (*APOE* Genotyping Testing)

The *APOE* Genotyping Testing uses MALDI-TOF MS to analyze two *APOE* gene loci, rs429358 and rs7412. These loci are linked to AD risk, aiding clinicians in evaluating the likelihood and progression of the disease.

APOE Allele Type	Genotype	rs429358	rs7412	Population Ratio	Risk Indication
E2	ε2/ε2	TT	TT	5-10%	Lower Risk
	ε2/ε3	TT	TC		
E3	ε3/ε3	TT	CC	70-80%	Normal
	ε2/ε4	TC	TC		
E4	ε3/ε4	TC	CC	10-15%	Higher Risk
	ε4/ε4	CC	CC		

Advantages

High Specificity

The MALDI-TOF MS technology detects low-abundance biomolecules through unique peaks and fingerprint features in mass spectrometry spectra.

Simple and Fast

Requires only a single reaction to produce easy and reliable results. Turnaround time is 10 working days.

Variety of Samples

Peripheral blood, oral swabs, genomic DNA, and dried blood spots can all be tested.

Applicable Population

- Individuals with cognitive impairment who need risk stratification.
- Individuals suspected of having AD and related cognitive disorders.
- High-risk individuals with a family history of AD.
- Individuals concerned about neurocognitive diseases and brain health.

Sample Requirements

- **Peripheral Blood:** ≥3 mL for adults, ≥2 mL for infants and children

- **Oral Swab:** Genotek oral swab preferred used according to manufacturer instructions

- **Genomic DNA:** more than 1 μg of purified DNA at a concentration of at least 5 ng/μL

- **Dried Blood Spot:** more than 3 spots

Service Workflow

1. Test Request Form and Informed Consent
2. Sample Collection
3. Sample Delivery
4. Sample Information Entry
5. Testing and Analysis
6. Report Issuance

Citations:
[1] Gauthier S, et al. World Alzheimer Report 2022
[2] Hodson R. Nature, 2018, 559(7715): S1
[3] Jack C R, Jr., et al. Alzheimer's & dementia: the journal of the Alzheimer's Association, 2024
[4] Alzheimer Europe. https://www.alzheimer-europe.org, 2023
[5] Breijyeh Z, et al. Molecules (Basel, Switzerland), 2020, 25(24)
[6] Gatz M, et al. Archives of general psychiatry, 2006, 63(2): 168-74
[7] Raulin A C, et al. Molecular neurodegeneration, 2022, 17(1): 72

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info@bgi.com