Genetic Risk for Alzheimer’s

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Our Work is About People and Science

The Alzheimer’s Association is a global organization working to advance care, support and research across the world.
Dementia is a Syndrome

- Dementia is a collection of symptoms related to cognitive decline
- Not everyone experiences memory loss
- Due to biological changes in the brain
- Alzheimer’s is most common cause
- Some causes of cognitive decline are reversible and not truly dementia
History of Alzheimer’s Disease

- Named after the late Dr. Alois Alzheimer, a German neuropathologist
- First described the disease in a landmark presentation about his patient, Auguste Deter, in 1906.
Hallmarks of Alzheimer’s Disease

Neuritic Plaques: Extracellular deposits of beta-amyloid

Neurofibrillary Tangles: Intracellular deposits of hyperphosphorylated tau

Neurodegeneration Cortical Atrophy

Healthy Brain

Advanced Alzheimer’s
Primary Prevention
Delay onset of AD pathology
• Decrease Aβ₄₂ production
• Prevent tangle formation

Secondary prevention
Delay onset of cognitive impairment in individuals with evidence of pathology
• Decrease accumulated Aβ burden
• Decrease neurodegeneration with anti-tau or neuroprotective agents

Tertiary prevention and treatment
Delay onset or progression of dementia
• Neuroprotection-prevent neuronal loss
• Enhance function of remaining neurons
• Neurotransmitter repletion

Clinical disease stage
Genetic Risk in Context

• Clinical diagnosis vs. biological Alzheimer’s disease
• Multiple risk factors
  – Age
  – Family history
  – Genetics (including Down syndrome)
• Modifiable risk factors
  – Cardiovascular
  – Education
  – Social and cognitive stimulation
  – Exercise
  – 35%
The Three “Buckets” of Genetic Risk

- Deterministic mutations
- APOE4
- Hundreds(?) of other genes
What do we know about APOE4 and risk?
What do we know about APOE4 and risk?
What about Genetic Testing

• In research
• Limitations
• In medical practice
• Direct to consumer
• Alzheimer’s Association position
THE END OF ALZHEIMER’S STARTS WITH YOU