

Alzheimer's disease (AD)



AD is a progressive neurodegenerative disease responsible for 60-70% of dementia cases. Monogenic AD exist, yet over 95% of cases is caused by multiple genetic (and environmental) factors.

Polygenic risk scores (PRS)



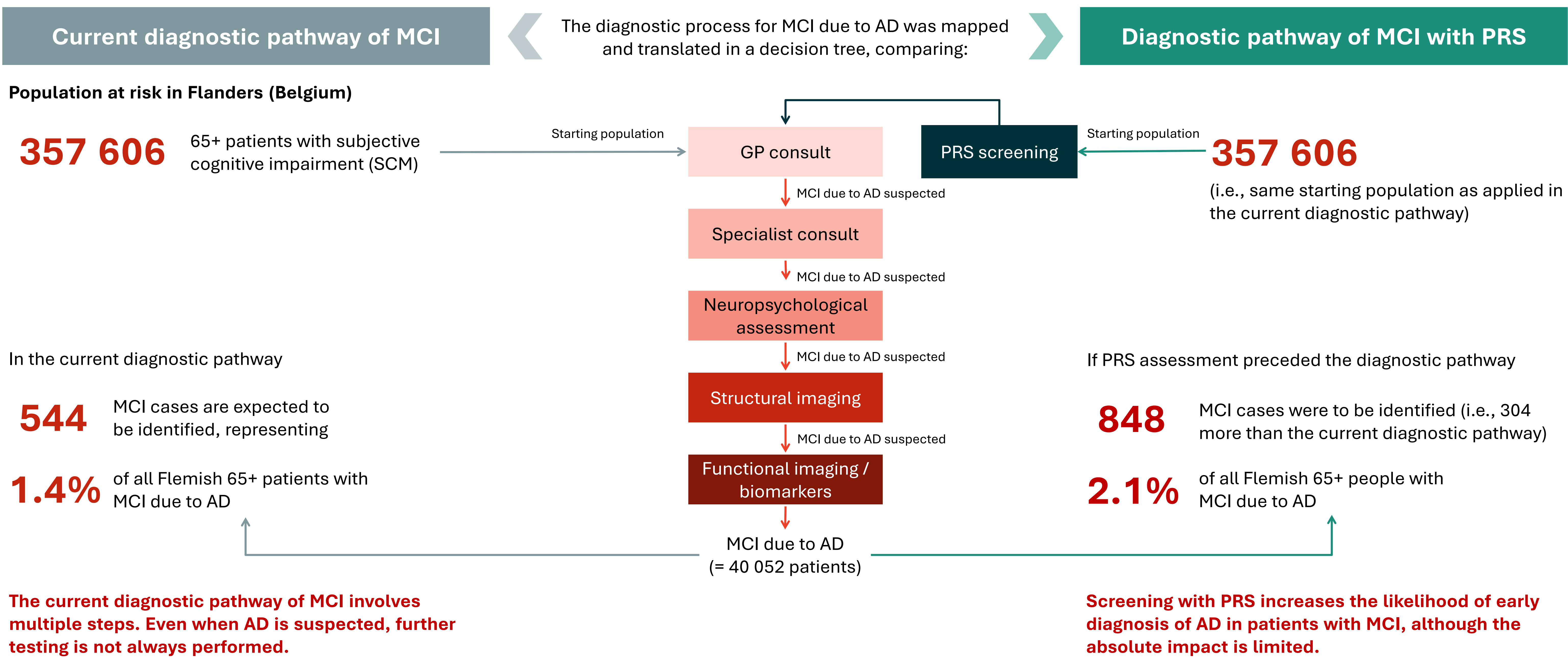
PRS integrate multiple genetic variants associated with a disease (here AD) in a single score, predicting an individual's likelihood of developing this disease.

Disease modifying treatments (DMT)



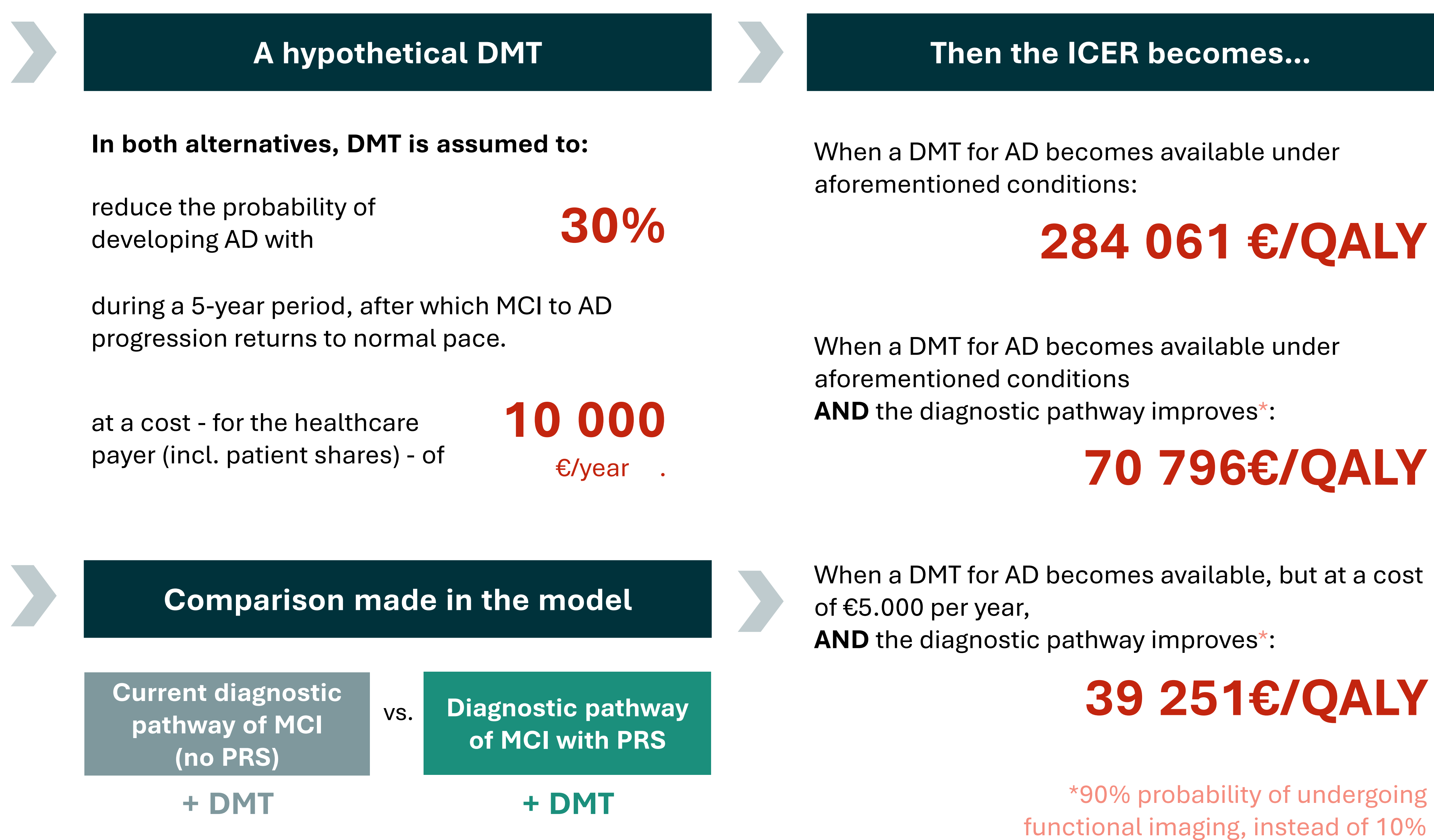
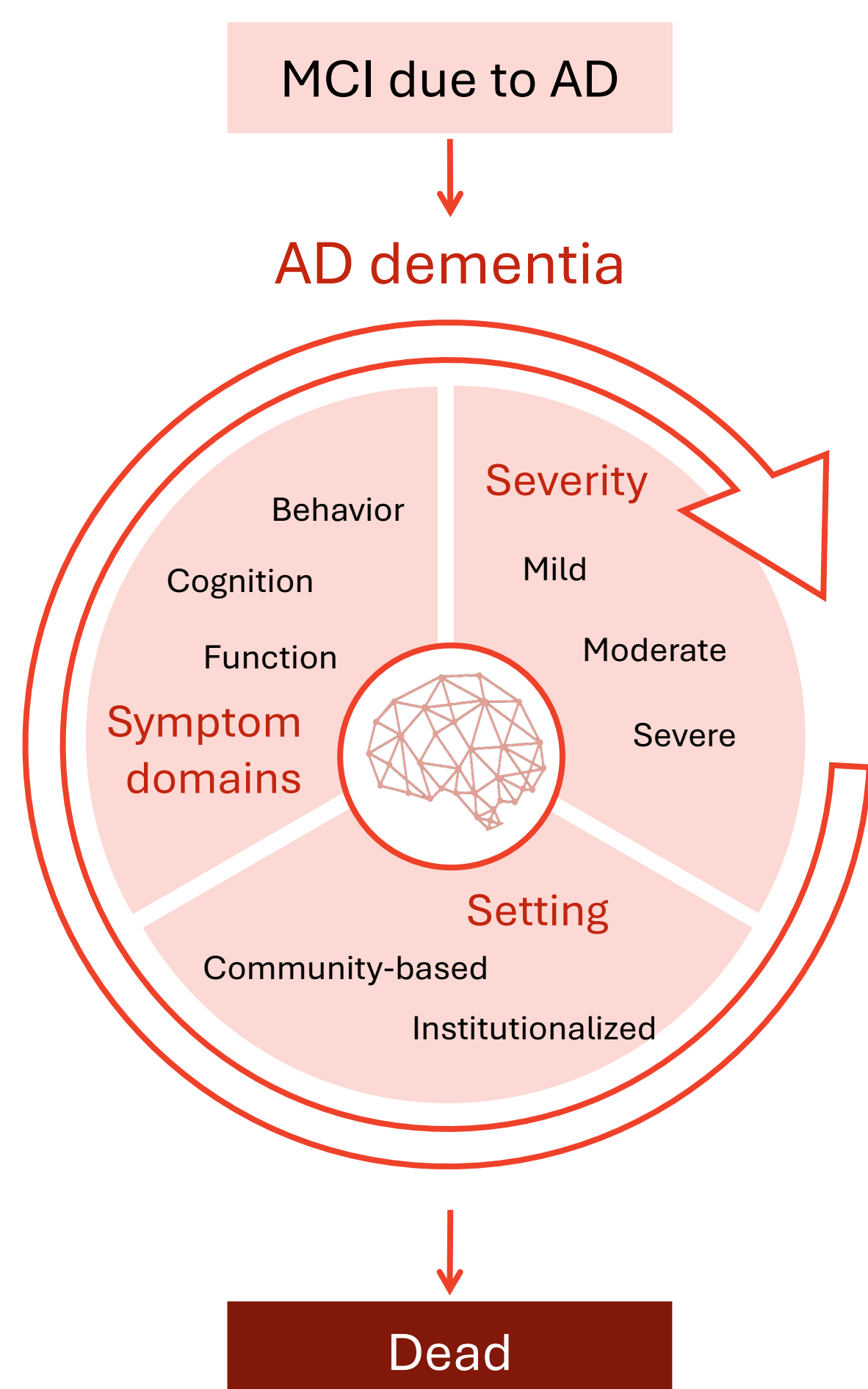
DMT are treatments that delay, slow down or reverse disease progression by targeting underlying causes or mechanisms. Up until now, no DMT for AD is available.

How can polygenic risk scores support early diagnosis of Alzheimer's disease?



Can PRS screening be cost-effective? Under which circumstances?

The end points of the decision tree were linked to a Markov model, leveraging the open-source model of Green et al. (2019):



Clinical implications

Combining PRS, a static measure, with more dynamic measures (e.g. biomarkers, transcriptomics, or epigenomics) might bring extra insights into the pathophysiological mechanisms of AD.

As long as no DMT for AD is available, PRS will have limited use in clinical practice.

When a DMT becomes available, PRS is only helpful if the diagnostic pathway of MCI becomes less costly and/or more efficient in diagnosing patients.

Health-economic implications

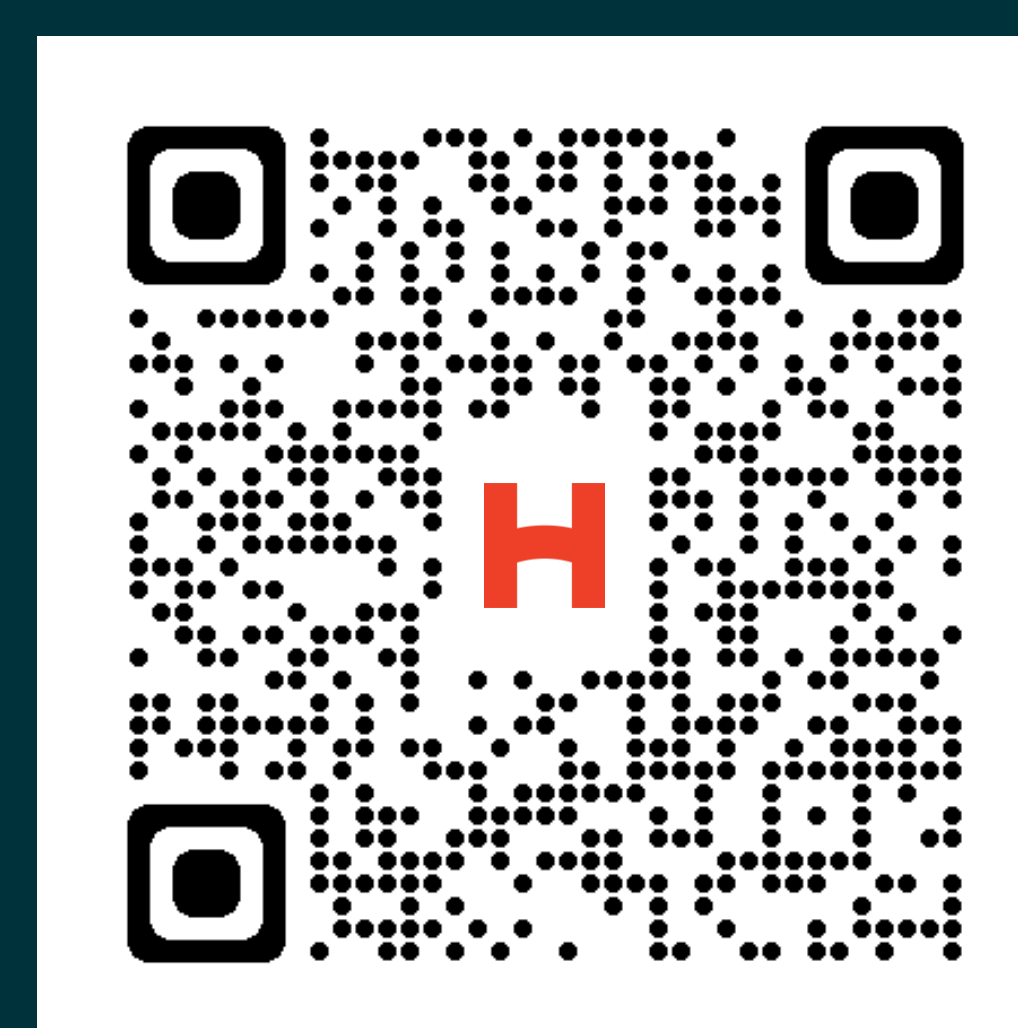
The cost-effectiveness of PRS-screening heavily depends on the overall MCI diagnostic pathway and DMT availability.

The actual merit of a value model lies in its adaptability, creating the possibility to evolve together with further research in PRS.

This study demonstrates the feasibility of an early health-economic evaluation of PRS as a screening tool for AD.

Abstract reference:

Werbruck A¹, Verdonck C¹, Abakkouy Y², Cleynen I², Vandenberghe R³, Vermeersch S¹, & Schoonaert L¹ (2024). Exploring the Implementation of Polygenic Risk Scoring in Alzheimer's Disease: An Early Value Model



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← Want to know more?