**Down Syndrome**

**Program Description and Objective**

Life expectancy in the population of persons withDown syndrome (DS) has increased dramatically in recent years and is now over 60 years in developed countries. Accordingly, it has also increased the interest in those complications that occur in adulthood and senescence. Amongst the later, Alzheimer’s disease (AD) is especially important. By the age of 40, all subjects present AD pathological changes in the brain. Although not all individuals with DS will develop dementia. The prevalence of AD increases with age, from 20-50% when aged 54 to 59 years, to 60-75% when older than 60 years. The main reason for this high risk is related to an increased accumulation of the amyloid beta (Aβ) peptide in the brain as Trisomy 21 in DS involves, among others, the over-expression of the amyloid precursor protein (APP). Indeed, the chromosome 21 carries the APP gene (Wiseman et al., 2015). This ultra-high risk and universal pathological changes in adults with DS has led the scientific community to conceptualise DS as a form of genetically determined AD.However, there is a gap in knowledge about the factors influencing the emergence of AD in DS given the variability in the age of presentation. Other factors (genetic or environmental) may play an important role in the development of AD. Detecting early changes related to Alzheimer’s disease in adults with Down syndrome can be difficult due to the presence of intellectual disability and other health issues. The diagnosis of Alzheimer’s disease in persons with Down syndrome requires appropriate evaluation tools and a specialized team with significant experience. The goal of this project (Horizon 21 DS consortium (H21 consortium)) is the harmonization of diagnostic protocols so that a single assessment scale for clinical investigation (in clinical investigations, neuropsychology, biochemistry, blood and CSF, imaging) and monitoring of patients during treatment can be obtained data can be compared and combined.

**Methods**

**Participants:** Greek patients with trisomy 21, aged 35 years old or over and without diagnosed Alzheimer’s disease, to be recruited and monitored. Participants will undergo two assessments within 6 months (baseline and one or more follow-ups). Exclusion criteria are: severe ID, significant sensory impairments, or other acute illness that preclude cognitive testing.

**Clinical evaluation**: physical exam, complete neurological examination (including SPES/SCOPA scale and Tinetti Balance and Gait Scale), structured medical history, CAMDEX-DS interview, Neuropsychiatric inventory, Dementia questionnaire for people with learning disabilities, functional scale.

**Cognitive evaluation**: A series of cognitive tests will be administered: the Kaufman brief intelligence test (K-BIT), the CAMCOG-DS test, the Cued Recall test, the Picture Cancellation Task, the Cats and Dogs test (inhibition), the Verbal Fluency test, the Barcelona test, the Digit Span (forward/ backward test).

**For more information**

**https://horizon-21.org/alzheimers-disease-and-t21/**