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The University of Dublin

Understanding dementia in people with an intellectual disability: A model to diagnose dementia

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Ospidéal
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Daughters of Charity
Disability Support Services



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gcuntas diomhaoin
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OUTLINE

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- Ageing and Intellectual Disability
- The genetic link between Down syndrome and Alzheimer's disease
- Prevalence of dementia in people with Down syndrome
- Careful attention to differential diagnosis
- Brain Health
- The newly established National Intellectual Disability Memory Service
- Future directions



Ageing with Down syndrome

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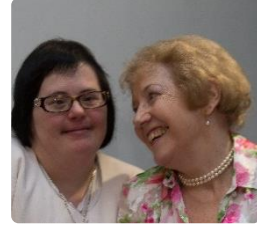


THIS IS A SUCCESS STORY

Opportunities to:

- Promote lifelong health and wellbeing
- Maintain independence
- Reorient services

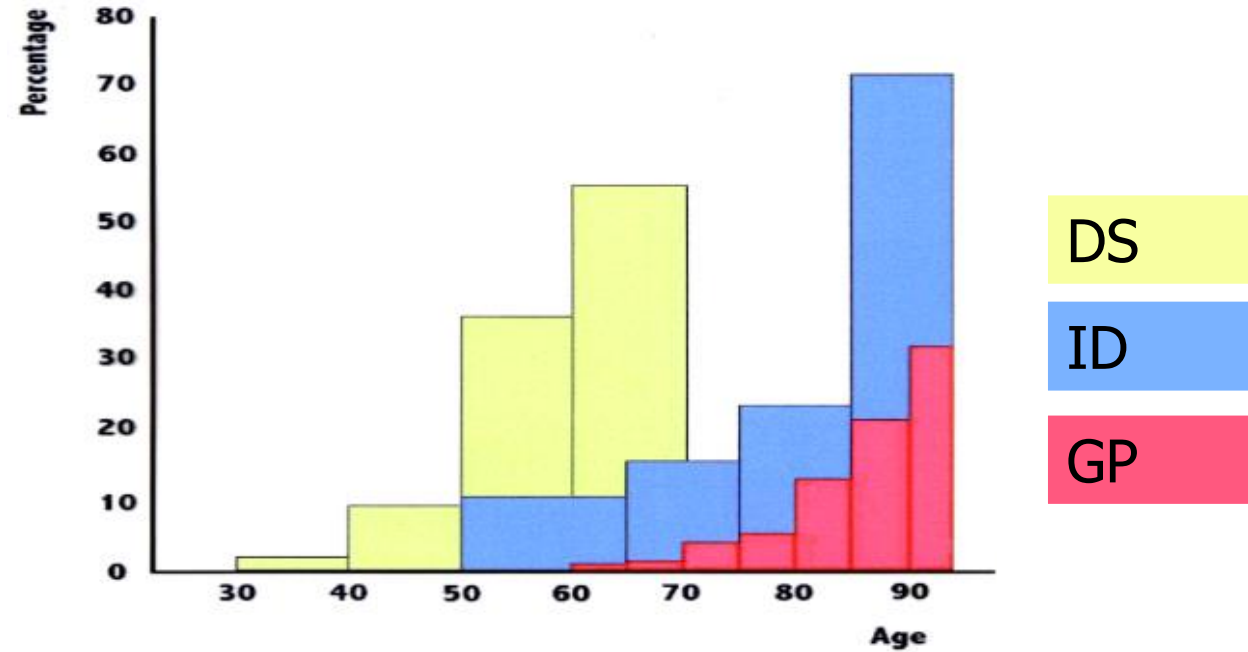
High rates of morbidity and mortality and
we need to address health inequities





Cognitive Ageing

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- The incidence of dementia could be up to **five times higher** than in people without ID (Strydom *et al*, 2013)
- **Much higher rates** in people with Down syndrome (McCarron *et al*, 2014, 2016; Strydom *et al*, 2010)



Dementia in people with Down syndrome **under** the age of 40 years is very uncommon and rarely seen

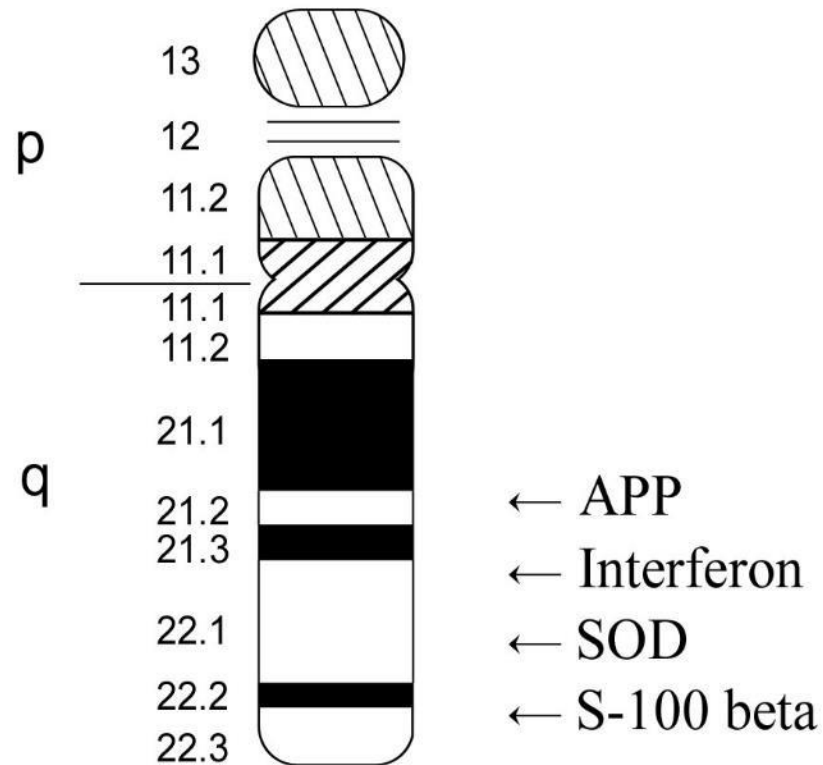


Why is there an association between Down syndrome and Alzheimer's?



Neuropathology of Dementia in People with Down syndrome

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- The gene coding for Amyloid Precursor Protein (APP) is on chromosome 21
- Individuals with DS have 3 copies of this gene and produce excess APP
- Over many years, this excess APP leads to β - amyloid plaque formation and eventually AD



Diagnosing Dementia in People with Down syndrome is **Highly Complex**

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- Assessment tools for dementia in the general population **are not appropriate** for people with learning disabilities.
- For example, widely-used tools such as the Mini-Mental State Examination (MMSE; Folstein & Folstein, 2001) assume the pre-morbid level of functioning to have been within the average range.
- There is **no single battery of assessments** universally used by Intellectual Disability teams, but some useful and validated tools are available.
- Critical need for the standardization of validated assessment tools to diagnose and stage dementia in people with DS, considering pre existing level of ID
- Alzheimer's Biomarker Consortium Down syndrome (ABC-DS) and the European Horizon 21 Consortium are validating various batteries of screening and cognitive tests for use in clinic settings



Diagnosing Dementia in People with Down syndrome is **Highly Complex**

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- The clinical presentation of dementia in persons with ID can differ.
- Personality and behavioural changes seem to occur earlier.
- Standardized tests often prove difficult and inaccessible.
- There may be communication difficulties for all involved.
- Improvised care environments undermine patient-centred planning.
- There may be a lack of base line data (personally and historically).
- High staff turnover limits symptom recognition.



Importance of a Brain Health Check

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People with Down syndrome >40 years

Number of tools available

- National Task Group-Early Detection Screen for Dementia (NTG-EDSD) (Moran JA, et al 2013)

Not a diagnostic instrument –early AD type changes and /or potentially reversible conditions

- Cognition, memory, and executive functioning, communication, behavior and personality etc.

- Dementia Screening Questionnaire for Individuals with Intellectual Disabilities (Deb S,et al 2007)



Careful attention to differential diagnosis

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Consider other physical or mental health problems and ensure the proper management of existing health problems such as:

- Depression or other mental illness
- Sensory impairment (vision and hearing)
- Thyroid impairment
- Sleep apnoea
- B12 & folate deficiency
- Medical problems (drug interaction, infection, pain, epilepsy)
- Major life events (separation, bereavement, moving)
- Catatonic Regression

Understanding the Process of Change: Early Stage

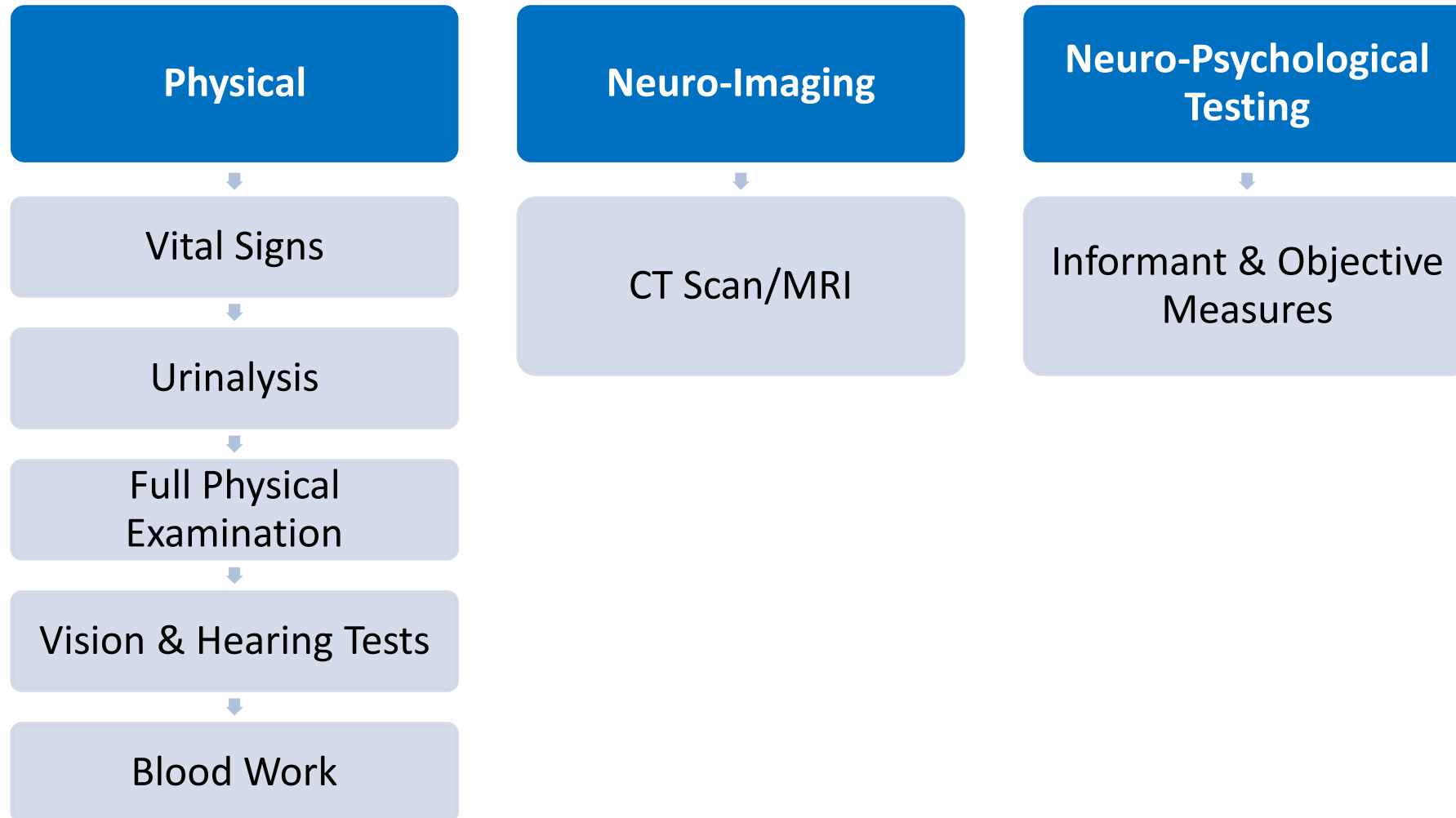
At this stage, the person is showing signs of decline from their usual level of functioning in the following areas:

- Subtle changes in behaviour and mood.
- Performance at day placements deteriorate.
- Memory problems, particularly for recent events.
- Ability to learn new information is affected
- Language and word finding problems.
- Decline in social, community and daily living skills.
- Disorientation.
- Difficulties with steps, stairs and kerbs due to depth perception problems.



Diagnostic Work-up: Some components

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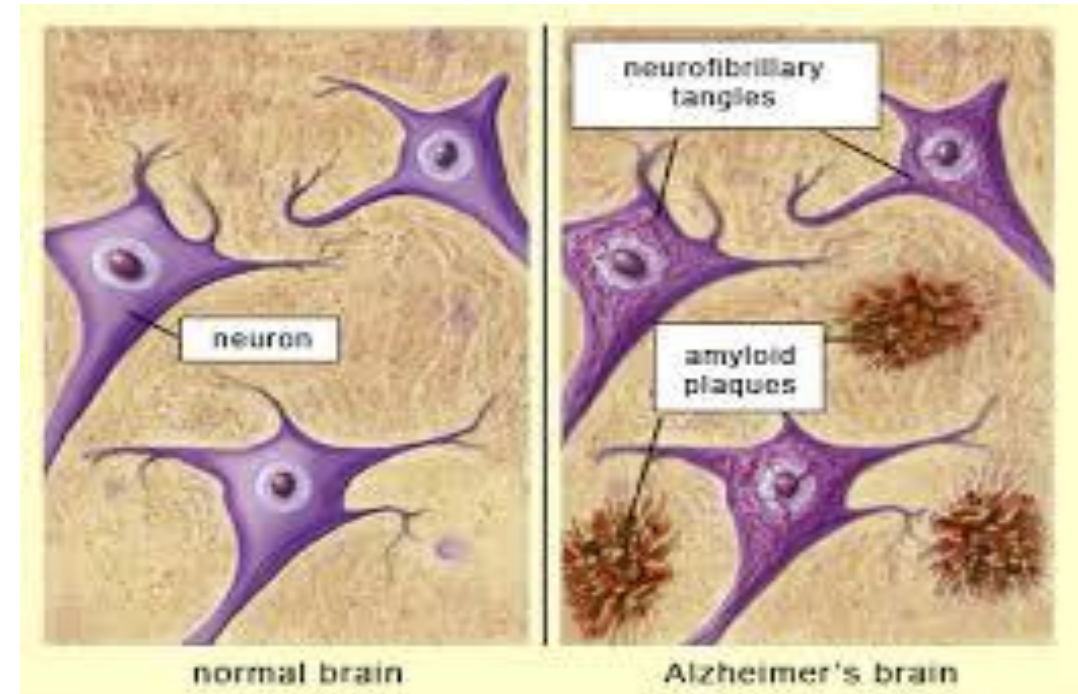




Correlating cognitive decline with new and innovative biomarkers

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To understand the role of existing biomarkers and validate new biomarkers to further assist with differential diagnosis in the clinic setting



- Promising work on Amyloid PET and tau PET biomarkers in adults with Down syndrome and their relationship to clinical diagnosis (Handen BL. 2020; Strydom A, et al 2018)
- Blood based bio markers –neurofilament light (NF-L)-excellent measure of neurodegeneration and a strong correlation with brain atrophy and clinical status (Rafii MS, et al 2019)



A Prospective 20 Year Longitudinal follow-up of Dementia in Persons with Down syndrome

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A prospective 20-year longitudinal follow-up of dementia in persons with Down syndrome

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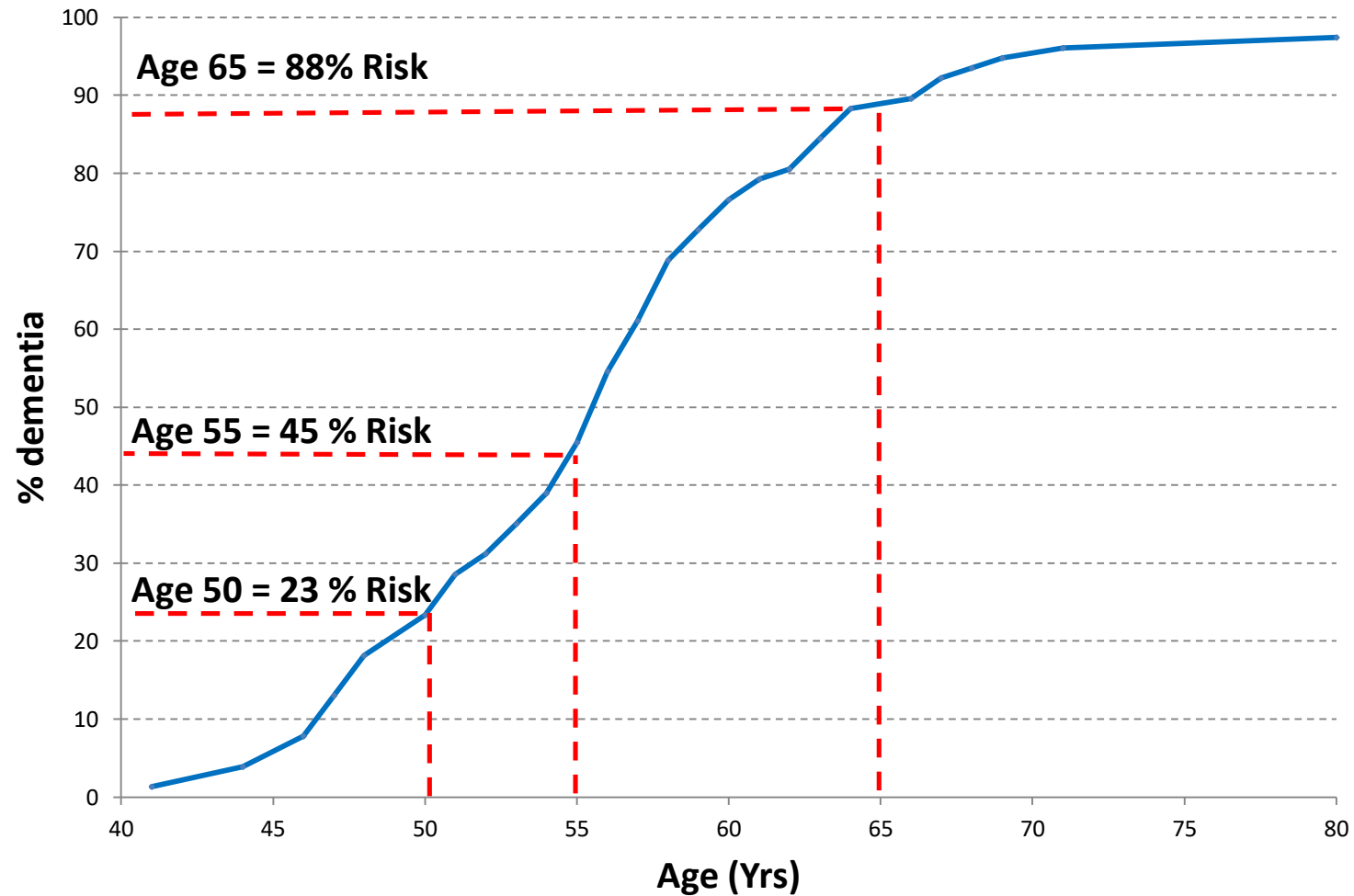
³ Daughters of Charity Disability Support Service, Dublin, Ireland



Risk Trajectory By Age

Dementia in Persons with Down syndrome

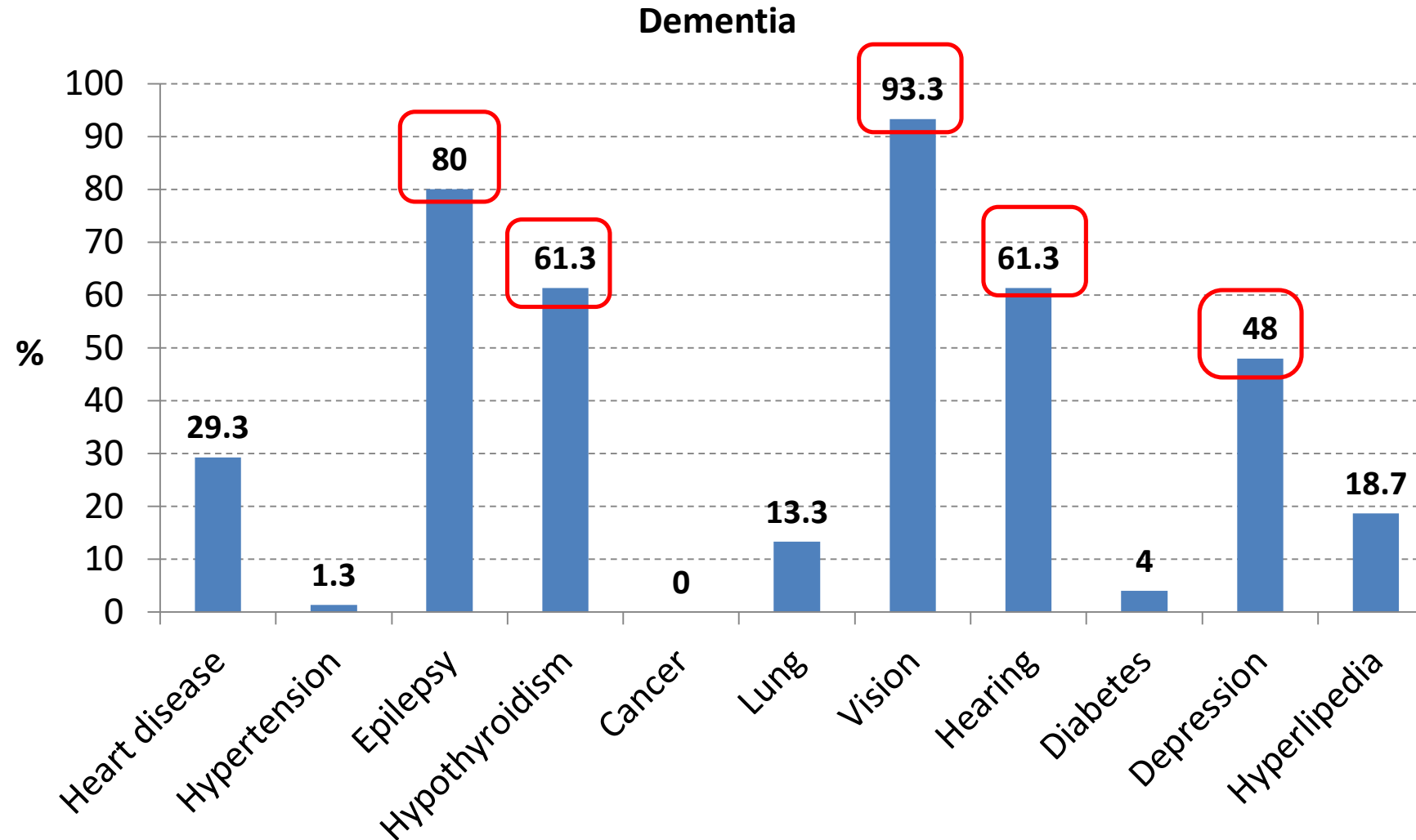
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Dementia Co-Morbidities

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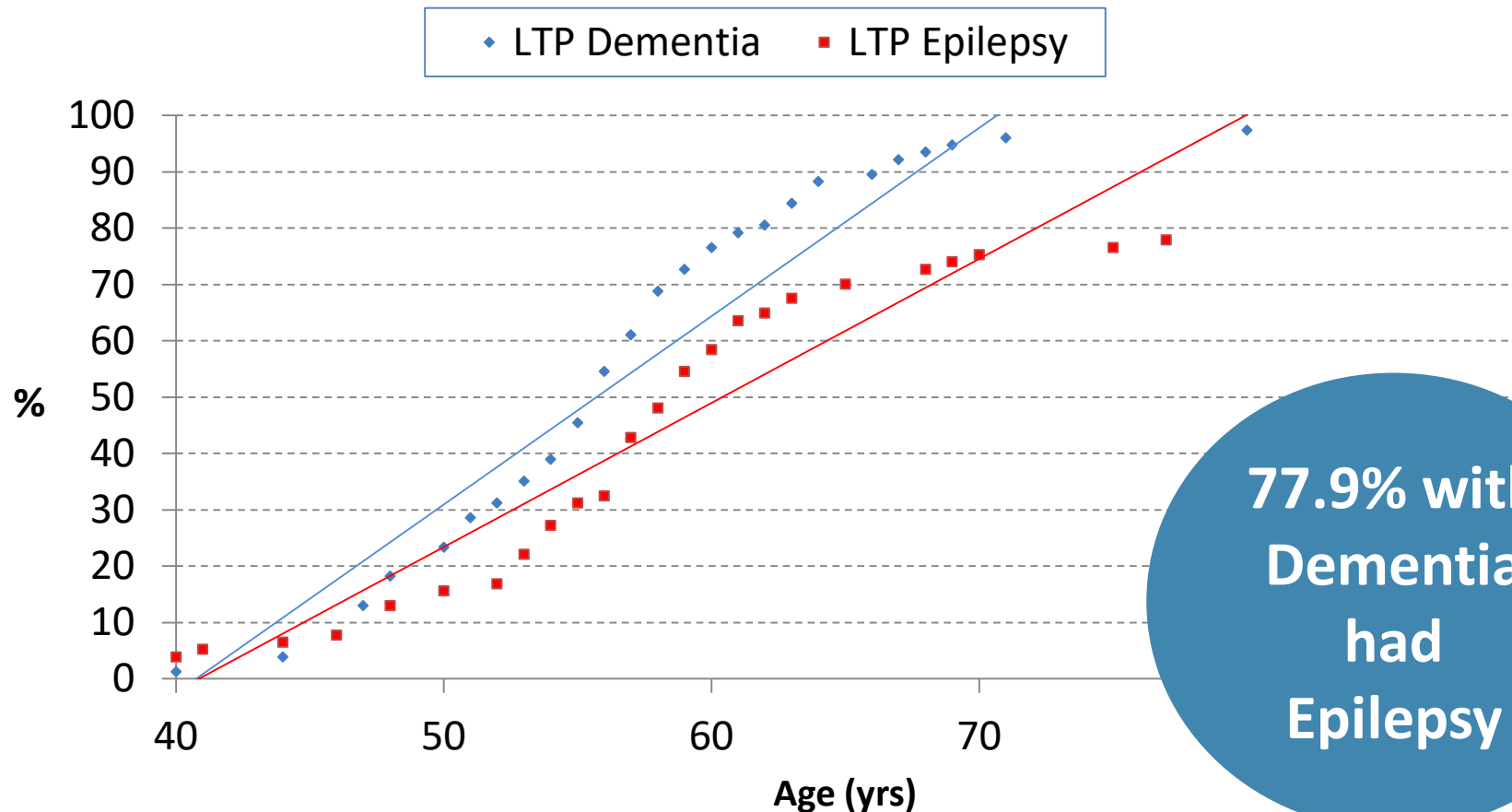
The Intellectual Disability Supplement to
The Irish Longitudinal Study on Ageing
(IDS-TILDA)



Dementia and Epilepsy In Persons with Down syndrome

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Life Time Prevalence

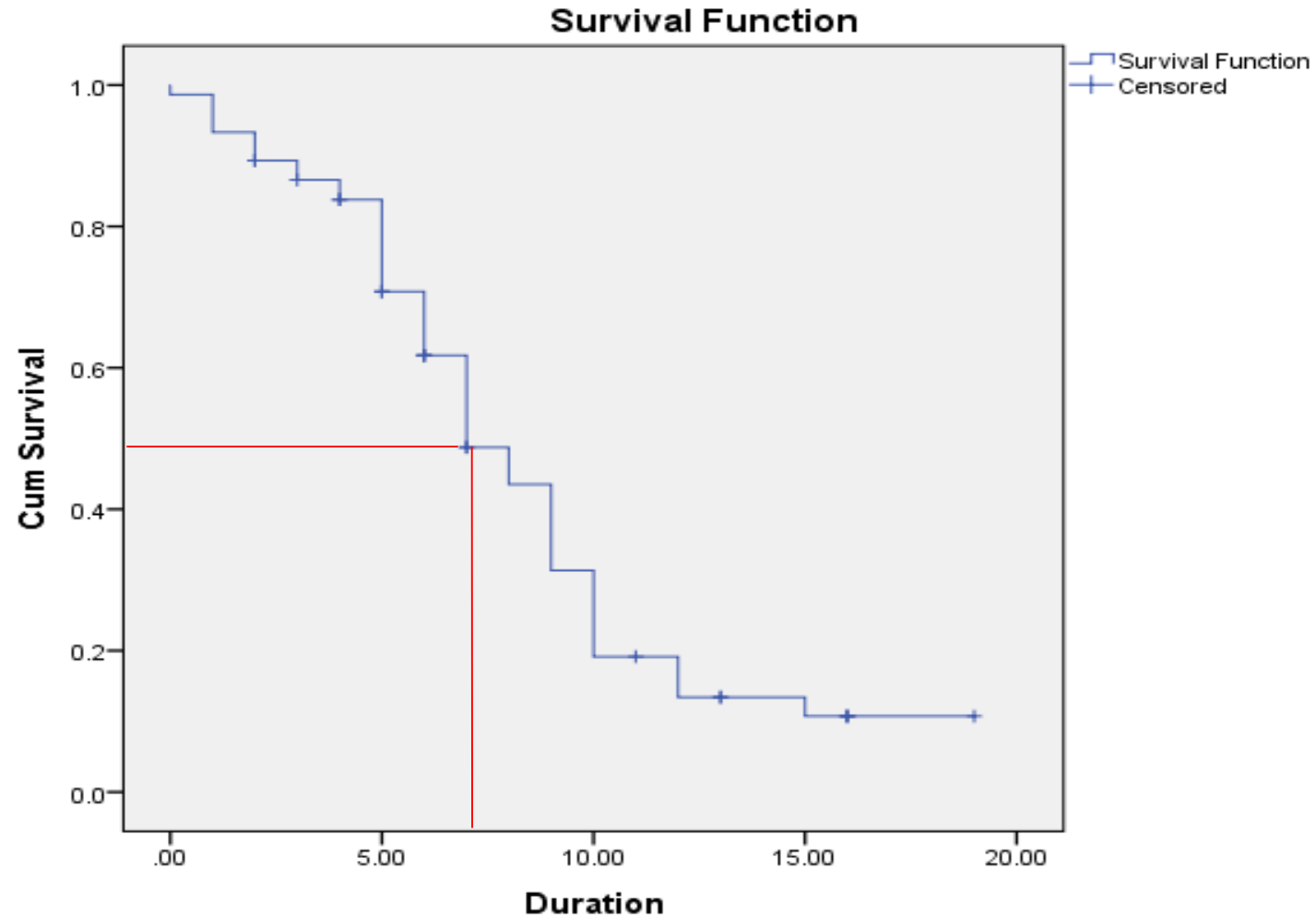


**77.9% with
Dementia
had
Epilepsy**



Dementia Mortality In Persons with Down syndrome

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In Summary

There is a substantial increased risk of dementia >50 years but

1

- Survival **less precipitous** than previously reported

2

- The **rate of progression varies** among individuals

3

- Previous concerns of adults with Down syndrome **'falling off a cliff' are uncommon**

4

- There is a high risk of **new onset epilepsy**

6

- There is **increased survival** at advanced dementia



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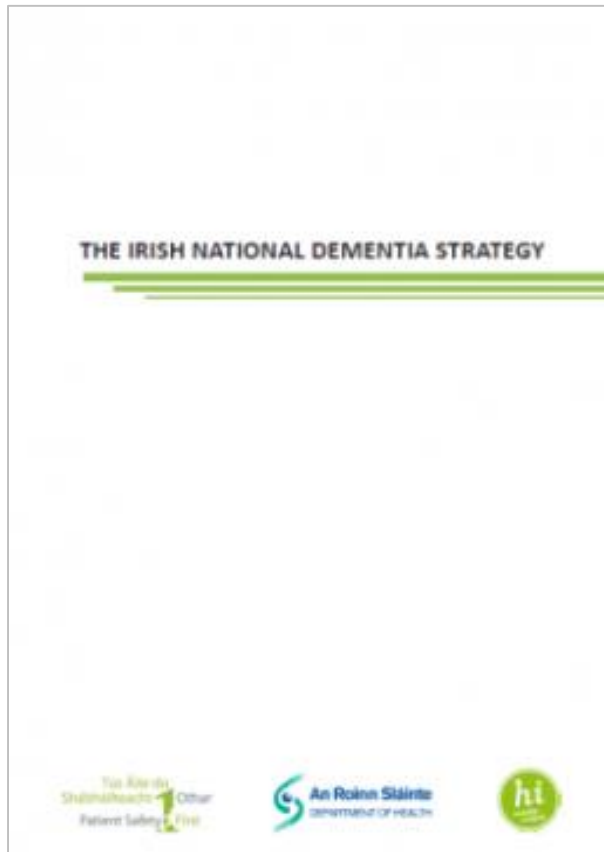
Address inequity in service provision for people with ID with memory concerns

A National Intellectual Disability Memory Service (NIDMS)



Background to the National ID Memory Service

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‘stressed the need for systems, structures and age appropriate services specifically **to promote timely diagnosis of people experiencing early onset dementia, including people with Down Syndrome**’



Translating Findings

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Working with the
Federation of
Voluntary Bodies' 60
member services,
initially exploring
service responses to
dementia



2020 Highlight

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Memorandum of Understanding

TCAID was delighted to sign a Memorandum of Understanding (MoU) between Trinity College Dublin, Tallaght University Hospital and the Daughters of Charity Disability Support Services in September.

The MoU has enabled the development of a new memory service in Ireland specifically for people with an intellectual disability.





The Team and Partnership

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For Ageing and
Intellectual Disability



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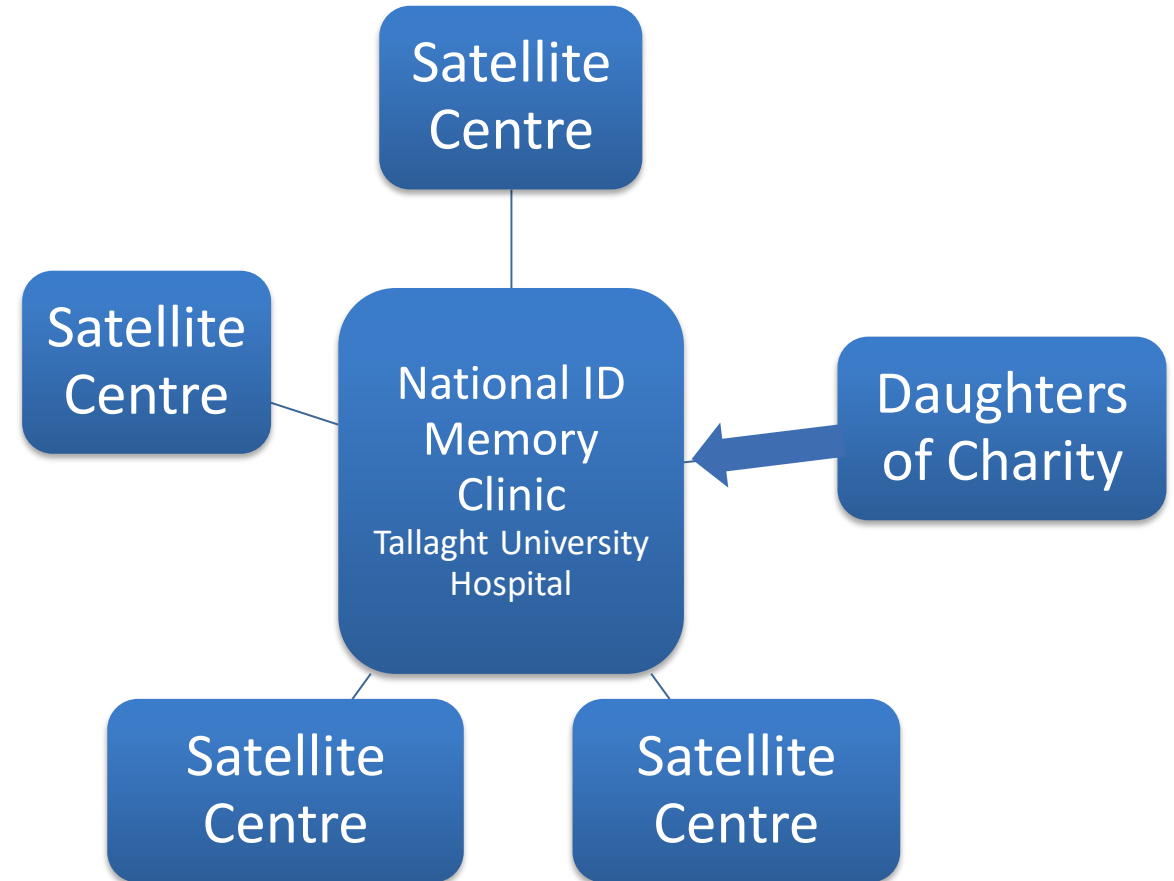


A Hub-and-Spoke Model for the Memory Service

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The diagnosis of Alzheimer's disease in persons with ID requires appropriate evaluation tools and a specialized team

- **Harmonizing diagnosis procedures**(diagnostic work up including clinical investigations, neuropsychology, biochemistry, blood, imaging etc)
- Defining **common criteria for diagnosis of Alzheimer's disease**





Confirming Dementia: Consensus Meetings

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Overall classification established based upon a comprehensive evaluation

No Dementia

- Stable or age-related changes

Questionable with Complication

- Some declines or concerns; may be due to some other underlying aetiology (e.g. mental or physical health condition, environmental, major life event)

*Query Dementia/Red Flag

- Substantial declines of late onset, query MCI requires further follow up

Definite Dementia

- Substantial declines of across several key areas-day to day living, memory, cognition etc > than 6 months




Easy-Read Information for the Memory Service

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What is dementia?

	<p>Dementia is a disease that affects some people as they get older</p> <p>Dementia causes some changes in the brain</p>
	<p>You might have trouble remembering things.</p>
	<p>You might forget people's names.</p> <p>You might forget where you put things.</p>
	<p>Sometimes you might be confused or sad.</p> <p>You family or support staff can help you with these feelings</p>

	<p>You might need someone to help you with things like:</p> <ul style="list-style-type: none">• getting dressed• doing shopping• doing work around the house
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Living Well with Dementia

	<p>You can keep doing all of the things you enjoy!</p> <p>Your family and staff will help you.</p>
	<p>You need to keep your brain and body healthy.</p>
	<p>Eat healthy food</p>



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Future Directions

Addressing questions that remain unanswered



There are HUGE Gaps in Knowledge

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- All adults with DS by age 40 have key neuropathological hallmarks of AD, and yet many will not present with the clinical features of dementia until they are much older-Why ?
- We need to urgently develop skills in dementia recognition and assessment for people with DS, as well as valid diagnostic standards
- Significant opportunities for boosting cognitive reserve education, exercise, cognitive stimulation and social engagement.
- *Most critically, we need to develop responsive and humane services for the changing needs of this increasingly at risk population with dementia.*
- *Masterclasses series will address some of these issues*



Into the Future

Questions that Remain Unanswered

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- Is it excess β amyloid production 'driving' the risk of dementia in people with Down syndrome or are there other factors?
- What role can neuroimaging, amyloid PET scanning and biomarkers play?
- Can a simple blood test help determine the risk for developing dementia in people with Down syndrome or help with clinical diagnosis ?
- What further markers could signal progression and decline in this group?





Include the voices of people with Down syndrome in the conversation on dementia

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“Like everybody here, I have dreams for the future. There are things that I want to do and I know dementia might play a part of my life too.

So, when you start talking about dementia, I want to be included in that conversation. I want to be included in the conversation about my future.

I want to be empowered so that I can choose how to reduce the risk of dementia”

- Mei Lin Yap





We need a **Paradigm Shift** in health care





Future Directions

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Department of Neurology,
University Hospital,
LMU Munich





Future Directions



The Intellectual Disability Supplement to
The Irish Longitudinal Study on Ageing
(IDS-TILDA)

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We want to ensure that people with Down syndrome have the opportunity **to be involved in dementia research.**



- Research in dementia needs to focus on **dementia prevention.**
- We need to **focus research at a much younger age.**



Prevention in the General Population

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- The PREVENT Dementia study aims to identify the earliest signs of dementia
- Scientists believe these may occur in the brain decades before symptoms appear

Prevent Dementia is a UK and Ireland wide study for people aged 40 – 59

Includes thinking and memory assessments, clinical examinations, MRI brain imaging and genetics to track how the brain changes throughout middle age

We hope to find ways to predict who is at greatest risk of dementia, so that we can intervene and prevent the disease taking hold



PREVENT Dementia – DS

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- Aims to complete the same protocol as in the general population
- This will include people with Down syndrome between the ages of 25-40

- Blood pressure readings, an Electrocardiogram (ECG), height and weight

Physical health checks



- Blood, urine, saliva

Biological sample collection



- Memory and thinking tasks are completed which aim to identify subtle changes in cognition.

Memory assessments



- Participants will undergo an MRI scan

Brain Scanning



- Diet, education, personality, history of brain injury, sleep quality, life events.

Lifestyle questionnaires



Dr Eimear McGlinchey



Remember we are all human

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Those with dementia are still people and they still have their stories and they still have their character, and they are all individuals, and they are all unique. And they just need to be interacted with on a human level.

Carey Mulligan





Trinity Centre for Ageing and Intellectual Disability

Stay Connected

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The screenshot shows the website for the Trinity Centre for Ageing and Intellectual Disability. At the top, there is a header with the Trinity College Dublin logo and name, and navigation links for Faculties and Schools, Courses, Research, and A-Z. A search bar is also present. Below the header, a blue navigation bar contains links for Menu, Home, About, People, Research, NIDMS, and Contact, along with a 'Gaeilge' option. The main content area features a section titled 'The National Intellectual Disability Memory Service' with a photograph of a man working on a laptop. To the right of the photo is a text block describing the service as a centre of excellence in proactive dementia assessment and diagnosis for people with an intellectual disability (ID), particularly supporting those with Down syndrome. A 'MORE' button is located below the text. At the bottom, there is a 'News and Events' section with a link to register for a webinar, and a 'Twitter' section showing tweets from @NIDMSIrl.

www.tcd.ie/tcaid



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Thank You

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Website: www.tcd.ie/tcaid/NIDMS

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