Improving Dementia Diagnosis and Brain Health in Down syndrome

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Acknowledgement of Country

Monash Health respectfully acknowledges the Bunurong/Boonwurrung and Wurundjeri Woi-wurrung peoples, the Traditional Custodians and Owners of the lands where our healthcare facilities are located and programs operate.

We pay our respects to their culture and their Elders past, present and future.

I also acknowledge the Bidjigal/Bedegal people, the Traditional Custodians and Owners of the Land on which we meet today

Ngarra-jarra-noun artwork by Dixon Patten

BACKGROUND

95% percent of people with Down syndrome develop Alzheimer’s disease at a young age (around 54) (McCarron 2017)

There are 13,000-15,000 Australians with Down syndrome (Down syndrome Australia 2025) and 6 million people globally.

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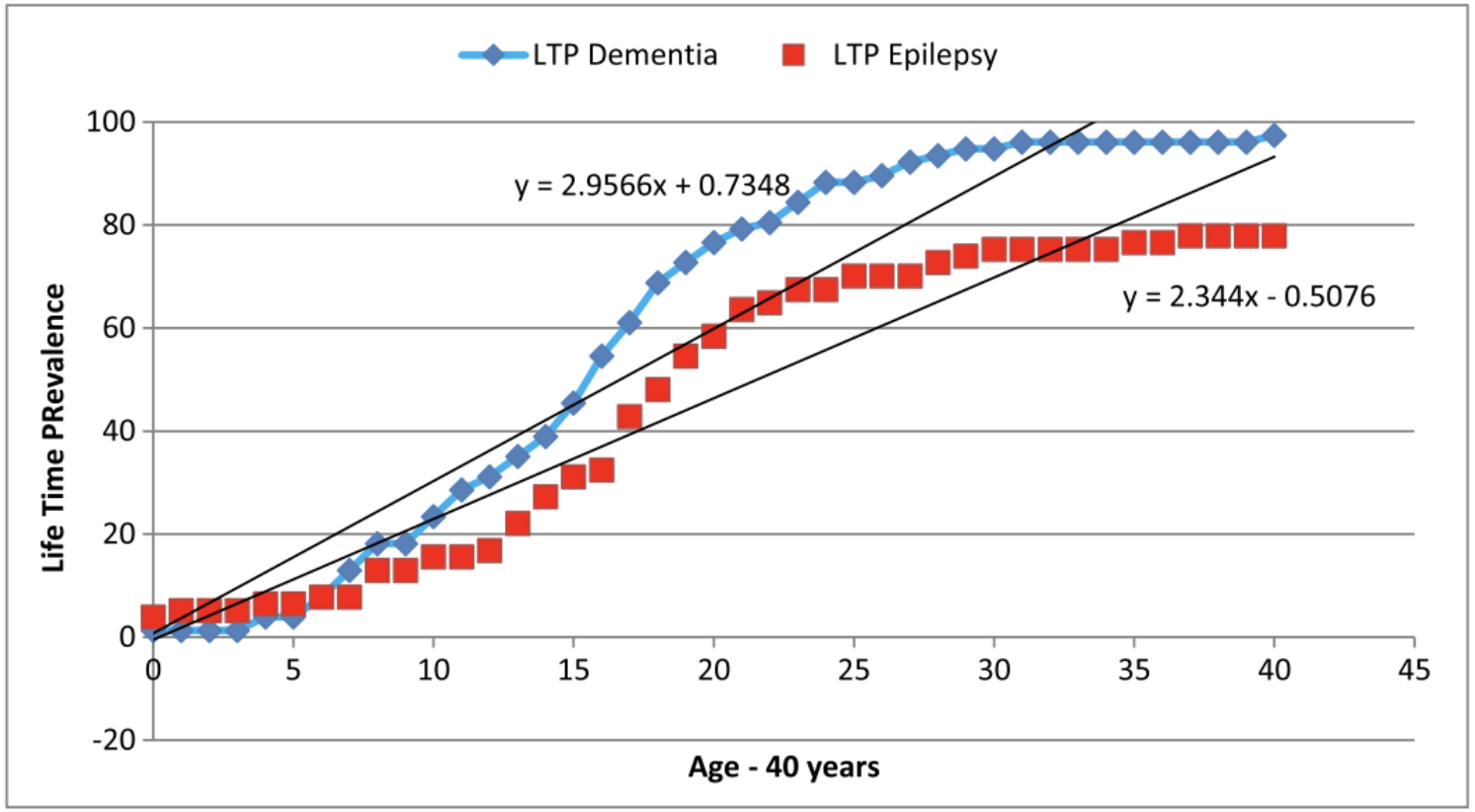


Figure 3 Life time prevalence (n = 77) of dementia (75 of 77) and of epilepsy (60 of 77). [Colour figure can be viewed at wileyonlinelibrary.com]

McCarron et al 2017 Over 20 years of follow up 97.4% (75 of 77) of participants developed dementia. Mean diagnosis was 55 years

Changing landscape of Down Syndrome Alzheimer Disease (DSAD)

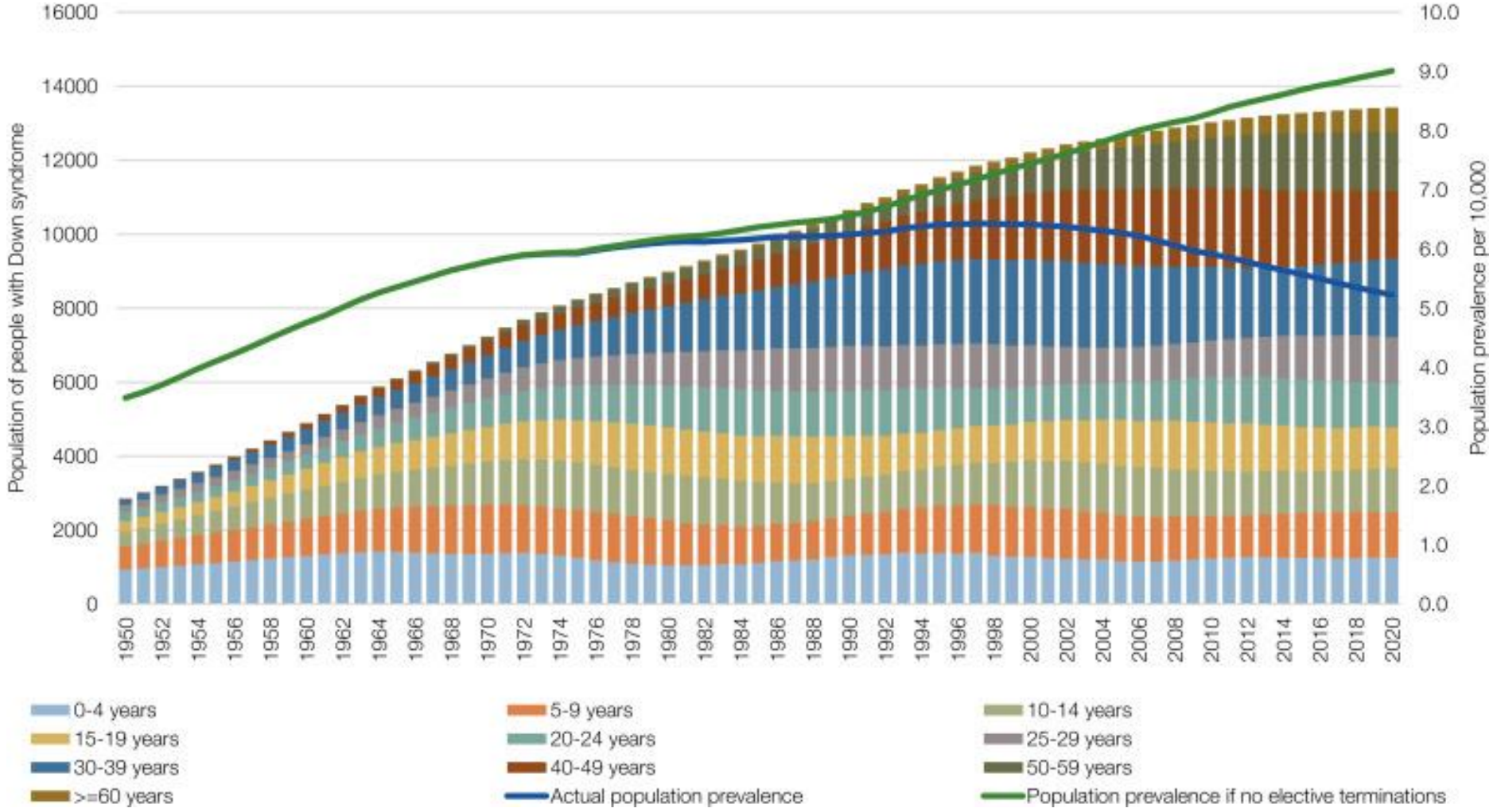
Many countries have established specialized memory services/pathways, established longitudinal studies, research groups.

There are emerging international prevention and treatment trials including disease modifying therapies.

Down syndrome Alzheimer Disease is a fully penetrant genetically determined form of Alzheimer disease. There is a faster rate of progression with mean time from symptom onset to death of 4.6 years

Dramatic improvements in healthcare have improved life expectancy – people with Down syndrome now have a life expectancy limited by Alzheimer Disease

Increasing Ageing Population



De Graff, 2022

People with Down syndrome are ageing

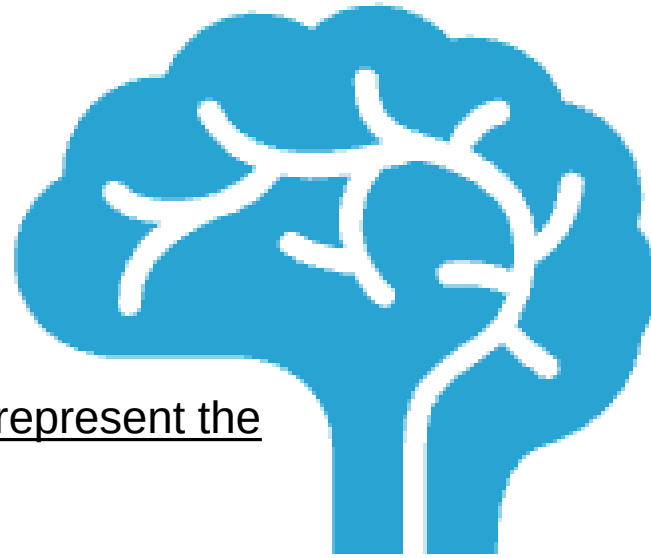
A child born in 1950 was expected to live less than 10 years, with congenital heart disease being leading cause of death (Montoliu-Gaya et al 2021)

Lulita et al 2022 In US death certificate data median age of death has increased “remarkably” from median age of 1 year in 1968 to 57 years in 2019. However, there is a ceiling effect over the last two decades

In 1950, 79% of people with Down syndrome were aged less than 20, and 1% were aged 40 or older. In 2020 data 36% people were aged less than 20, and 30% are over 40.

- This is attributed partly to advances in healthcare and improved equity in access as well as increased quality of life
- Western Australian data indicates that the life expectancy in Australia for someone with Down syndrome is 60 years of age. The same data showed that by 2000, 75% of people in Western Australia had survived to age 50, 50% to 58.6% and 25% to age 62.9.

Closing the Gap



This is a fully penetrant, genetically determined dementia, and people with Down syndrome represent the biggest group of people in the world with genetically determined dementia.

Alzheimer disease is now the leading cause of death for adults with Down Syndrome. There is an ethical imperative to improve diagnosis and management.

Closing the mortality gap with general population will require improved prevention and risk modification, and eventually treatment and cure of Alzheimer's disease

Background work

- Churchill Fellowship in 2024
  - National Intellectual Disability Memory service/TCAID (Dublin, Ireland)
  - Unit Alzheimer Down (Barcelona, Spain)
  - Cambridge Intellectual & Developmental Disabilities Research Group (UK)
  - LonDownS Kings college London (UK)
  - Institute Jerome La Jeune France (virtual)
  - Alzheimer's Therapeutic Research Institute, University of Southern California
  - University of California
  - Queen Square London UCL

Winston Churchill Trust
Learn globally, inspire locally.

MONASH University

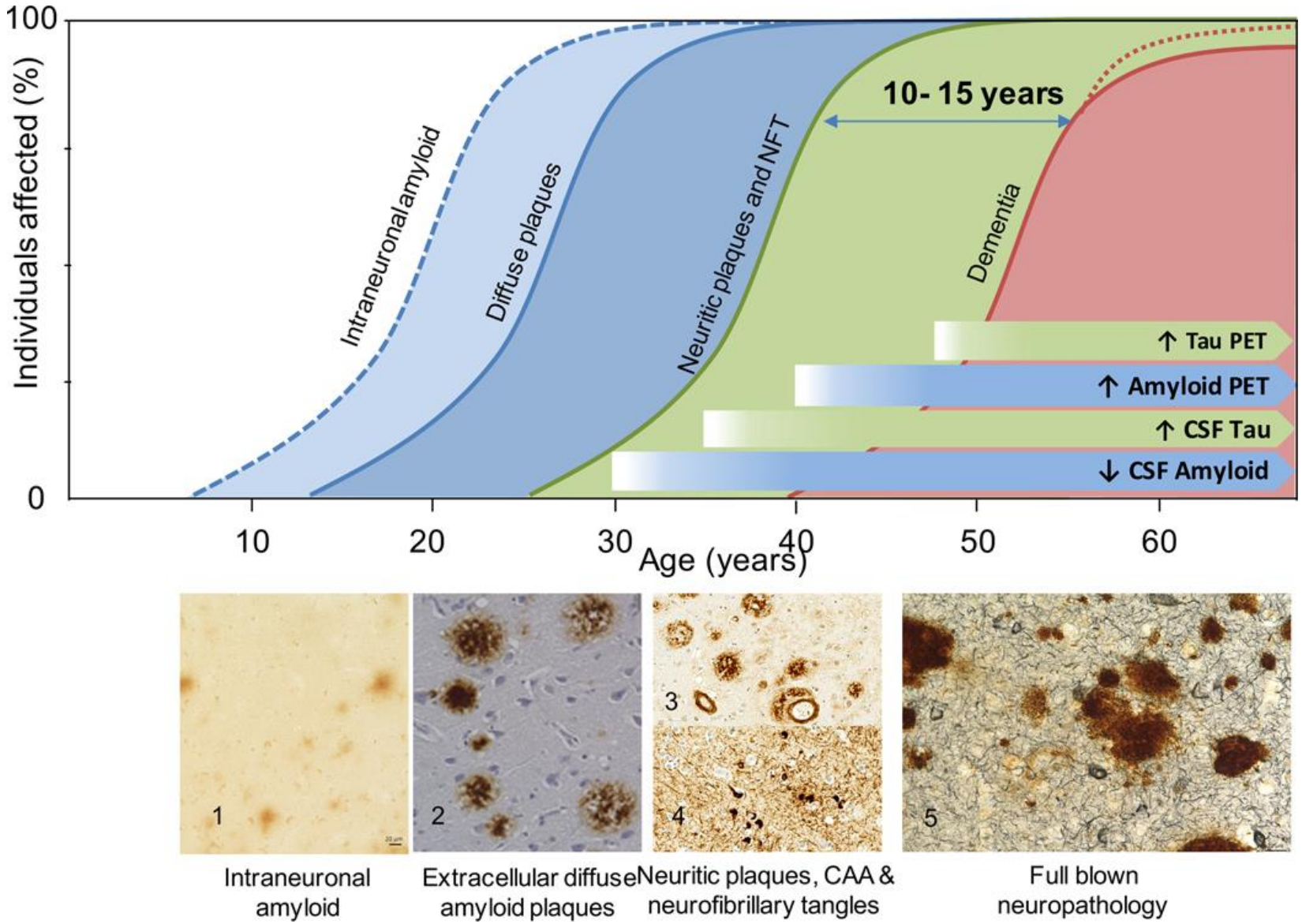
Reasons for increased AD risk

Amyloid precursor protein gene is located on chromosome 21, so people with Down Syndrome generally have the APP gene in triplicate.

There is an overproduction of amyloid from birth onwards.

There are case reports of individuals with partial trisomy who do not go on to develop dementia clinically or post-mortem (Doran 2017), due to lack of triplication of chromosome 21

- Other risk factors: ApoE4 haplotype accelerates amyloid deposition and associated with earlier symptom onset by 2 years, medical comorbidities, poor sleep, depression.
- Pathologically similar to sporadic and autosomal dominant AD
- Also note that cerebral amyloidopathy is also more common



Fortea et al 2022

Benefits of Early Diagnosis

Prompt diagnosis of dementia provides improved access to services and avoids reaching "crisis point"

Promotes brain health and mitigation of risk factors

Prompt diagnosis may enable the person to continue to live in their home with support

Prompt diagnosis empowers support workers, family and friends to understand the person's changing needs

Treatments are coming for all people living with dementia – accurate diagnosis will become critical in weighing up when to treat

Enables participation and access to clinical trials, equal with the general population.

Hearing/vision

Hypothyroidism

Depression/anxiety

Down syndrome regression disorder

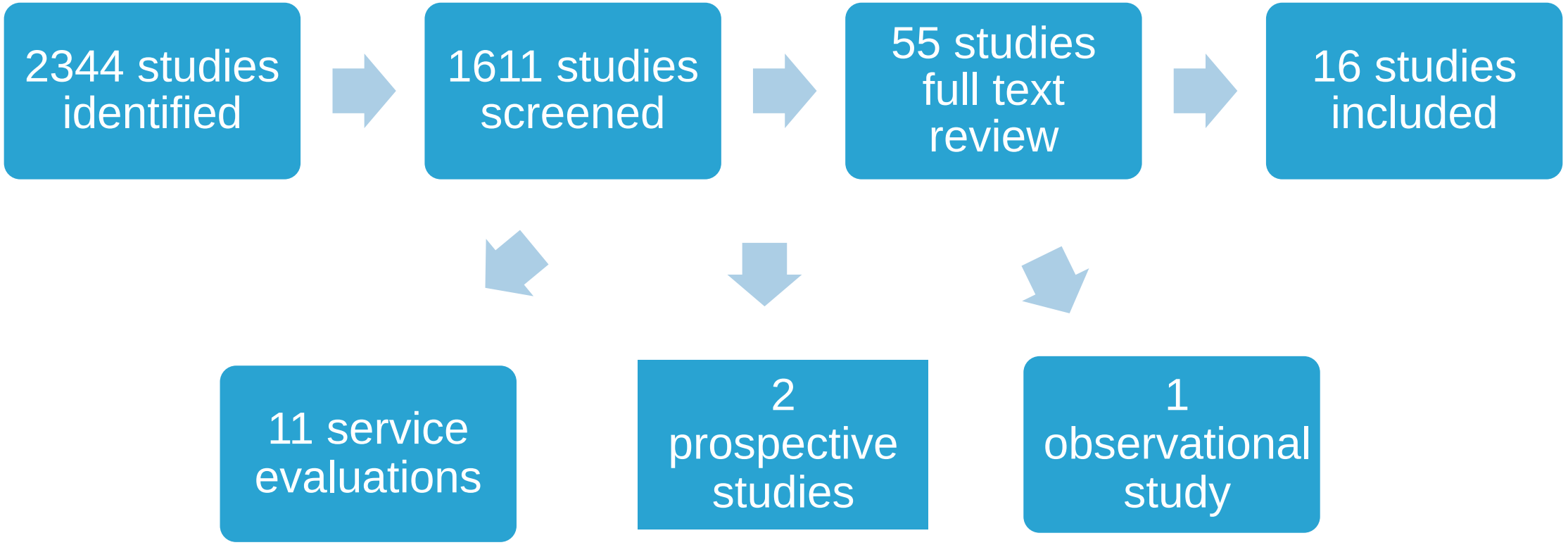
Obstructive sleep apnoea

Scoping Review: “Memory Services for People with Down syndrome: A scoping review exploring models

Aim: To examine existing models of delivery of memory services and dementia diagnostic services for adults with Down syndrome

Methods Scoping review, four (Ovid MEDLINE, Ovid EMBASE + EMBASE Classic, Ovid PsycINFO and EBSCO CINAHL Plus) were searched using the search terms Alzheimer Disease, Down syndrome and Memory Service. All primary study types were included from January 1 2005 to March 2026.

RESULTS


2344 studies identified
1611 studies screened
55 studies full text review
16 studies included
11 service evaluations
2 prospective studies
1 observational study

What is the evidence informing specialist clinic design- literature review

- Clinics need to have a multi-disciplinary team: a social worker, clinic coordinator, psychologist, speech pathology, occupational therapy, nursing, doctors
- People should have a routine baseline assessment 30 and then screening from 40.
- There are a range of tests:
    - Informant: DLD, DSQUID, NTG-EDSD, CAMDEX DS-II
    - Direct assessment: CAMCOG-DS II, mCRT, DS-MSE, TSI

AIMS

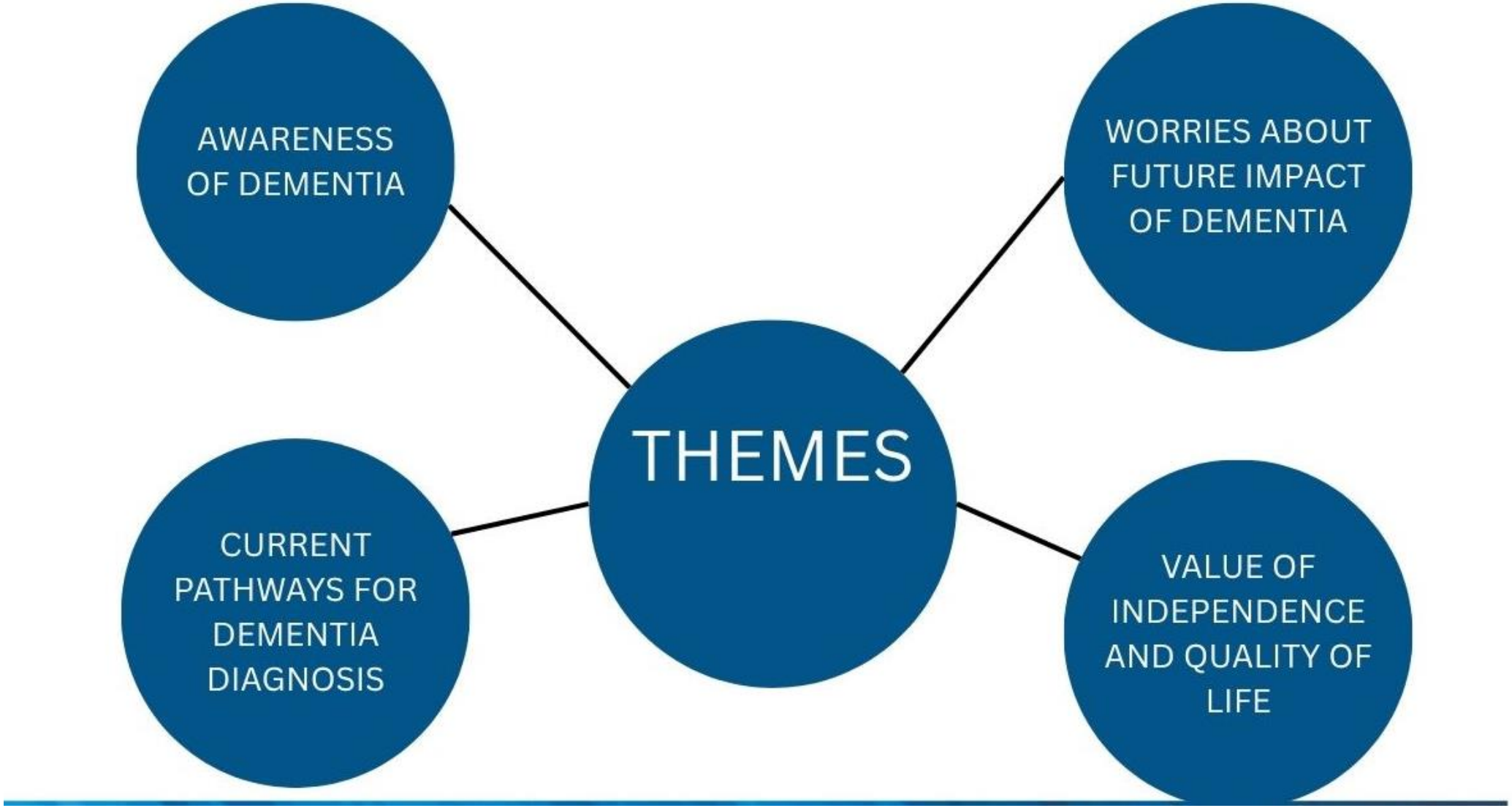
1. To design a memory and brain health service for individuals with Down syndrome, with consumer involvement from people with Down syndrome and their families

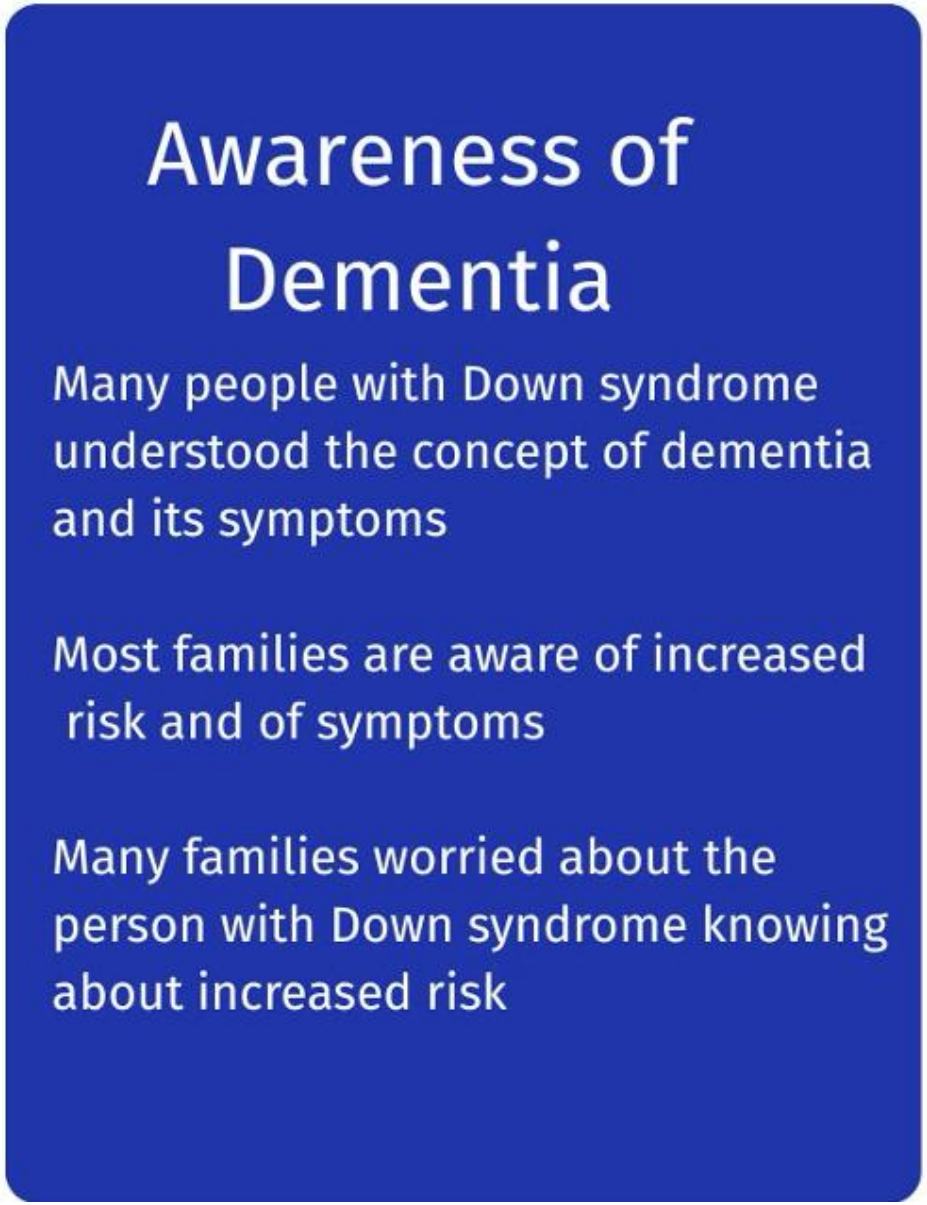
2. To implement this into practice through a 12-month pilot clinic.

3. To evaluate the pilot clinic with consumers.

Methods

- Qualitative descriptive study with semi-structured online interviews
- 25 family members and 8 individuals with Down syndrome
- Applied thematic analysis conducted with inductive and deductive coding


AWARENESS OF DEMENTIA
WORRIES ABOUT FUTURE IMPACT OF DEMENTIA
THEMES
CURRENT PATHWAYS FOR DEMENTIA DIAGNOSIS
VALUE OF INDEPENDENCE AND QUALITY OF LIFE



People with Down syndrome understood the concept of dementia, often knowing someone living with dementia, especially older relatives. This health awareness highlights an opportunity for education around brain health.

“My auntie..she lost her memory”

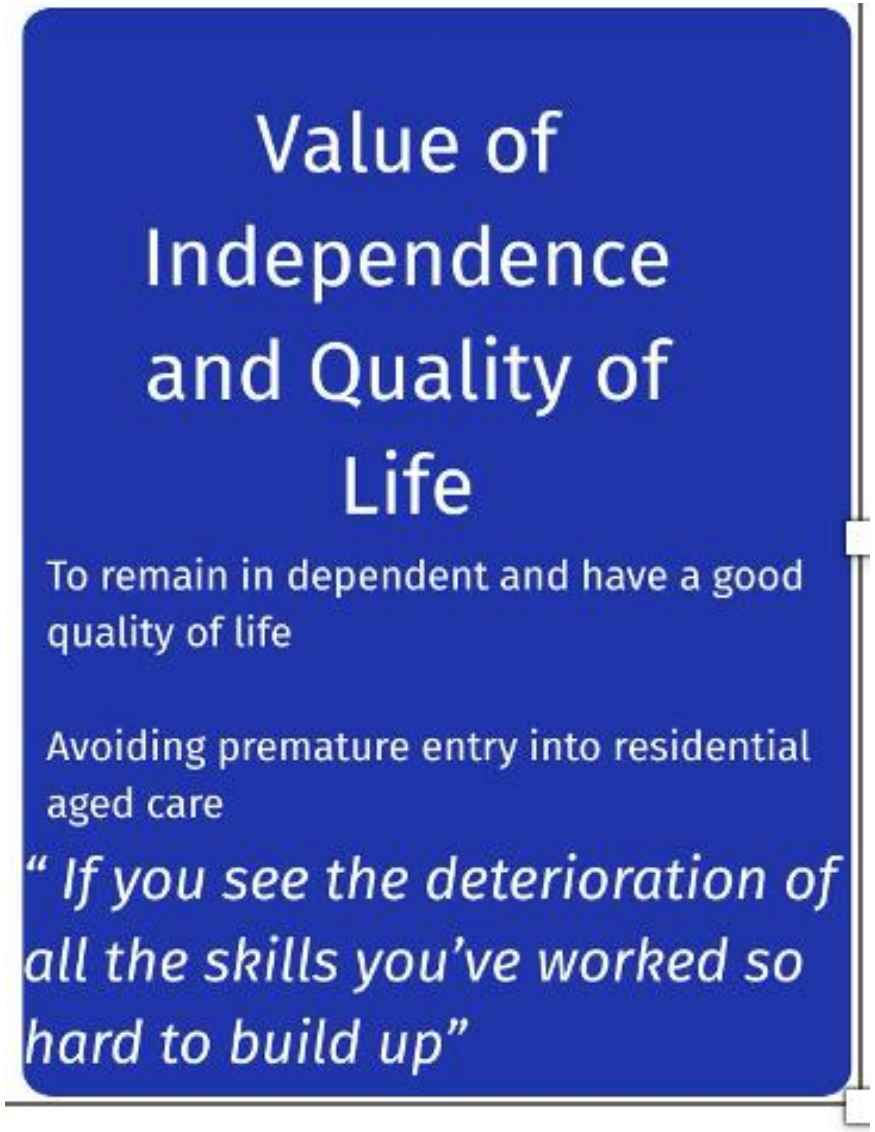
All families were aware of the risk of AD, and many volunteered knowledge of the age of onset.

“Just I think that most parents with adults with Down syndrome realize that dementia is coming for them, really, and it’s inevitable. You know, like there doesn't seem to be anybody really who doesn't get dementia”

Families worried about the person with Down syndrome learning of the increased risk and had mixed feelings about a future clinic giving a dementia diagnosis directly to the person. This is well documented in existing research.

Families worried about future care needs and not being there to provide the support. Six family members expressed their worry that they would not be well enough, or alive, to support their loved one through dementia: *"My worry is what happens after we go"*

6 siblings had become the main support person after the death of parents: *"We are an invested family…my brother and I have not missed on appointment"*



Families placed a high value on independence and quality of life:

Maintenance of independence was a common motivation for seeking early diagnosis to access improved services.

Two families expressed concern that a dementia diagnosis would lead to loss of NDIS funding.

"IT's the loss of quality of life that would happen if he developed Alzheimer's, I think it would be a nursing home situation for him"

"You don't want to put plans in place in a crisis. You never want to do that. So you therefore want to have time,"

“In our Lived experience, there were no pathways”

Specialist pathways are unclear

Public Hospitals are difficult to Navigate

Good news - General Practice is the central point of care

“Almost everyone we have dealt with has been interested but not necessarily skilled or feeling skilled enough, so where do you get that kind of expertise?” Family member

Is a specialized Intellectual Disability Memory Service Needed?

Participants generally felt that a specialized memory service was worthwhile, although some felt that the responsibility sat with general practice or general clinics and they needed to upskill.

“Dealing with people with intellectual disability, and Down syndrome in particular is a specialist area, I can’t see every GP to know everything about every condition. I want them to know enough to know when to refer me when we need something far more specific. I mean I wouldn’t expect my GP to do my heart surgery, so you know I’m more than happy to be referred to a cardiologist for that” (Family member 11)

Designing a clinic - considerations through qualitative research

CLINIC DESIGN

environment should reduce stress

Accessible information
before appointment
and tests

Telehealth is acceptable and
feasible
when necessary

COMMUNICATION

Talk to me, not my family

choose simple language and check for understanding

easy read or video information

FEEDBACK OF DIAGNOSIS

Honest and Comprehensive Information

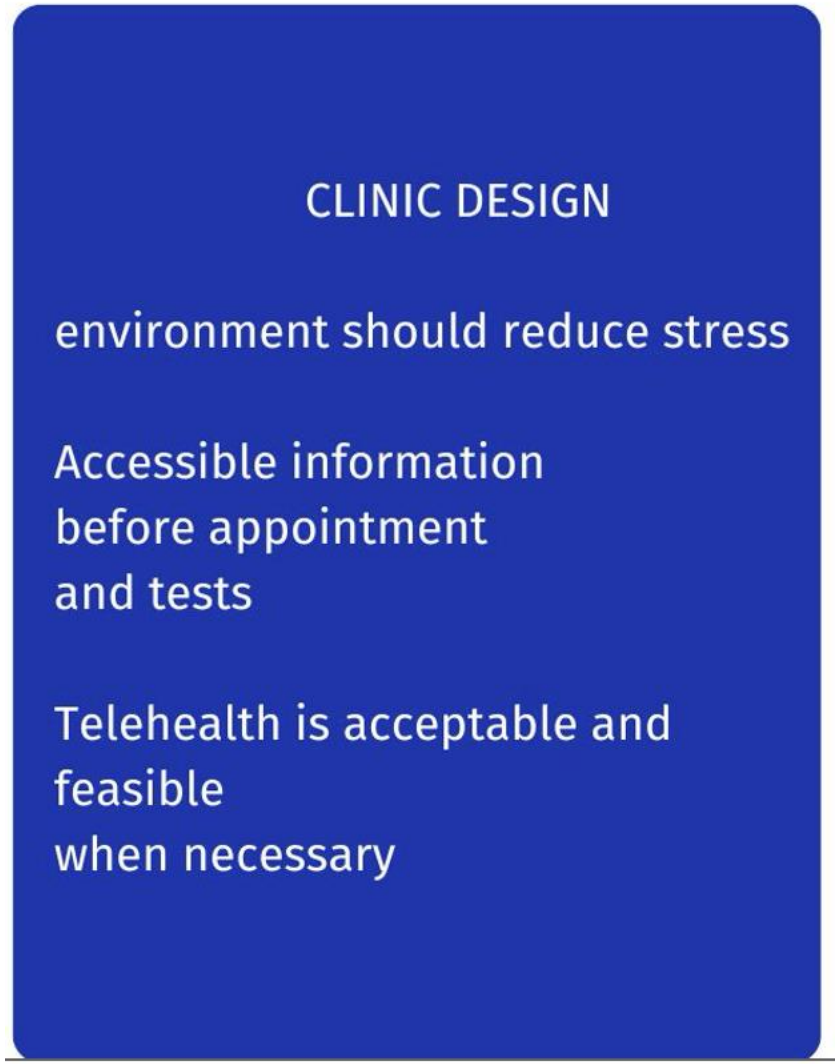
People with Down syndrome and their families want to know about treatment

Early diagnosis Is essential for planning

- *"I've lost count of the amount of times I've taken someone to a GP and the GP is talking to me, not even acknowledging the person I'm with and referring to the person in conversation. That happens all the time. Yeah it still happens now" (family member)*

- *"miscommunications and not understanding what's been said and not keeping up with the conversation. It can be hard to balance the pacing of the information that's provided" (participant)*

- *"Having a doctor who doesn't ask my any questions but asks mum and dad" (participant)*

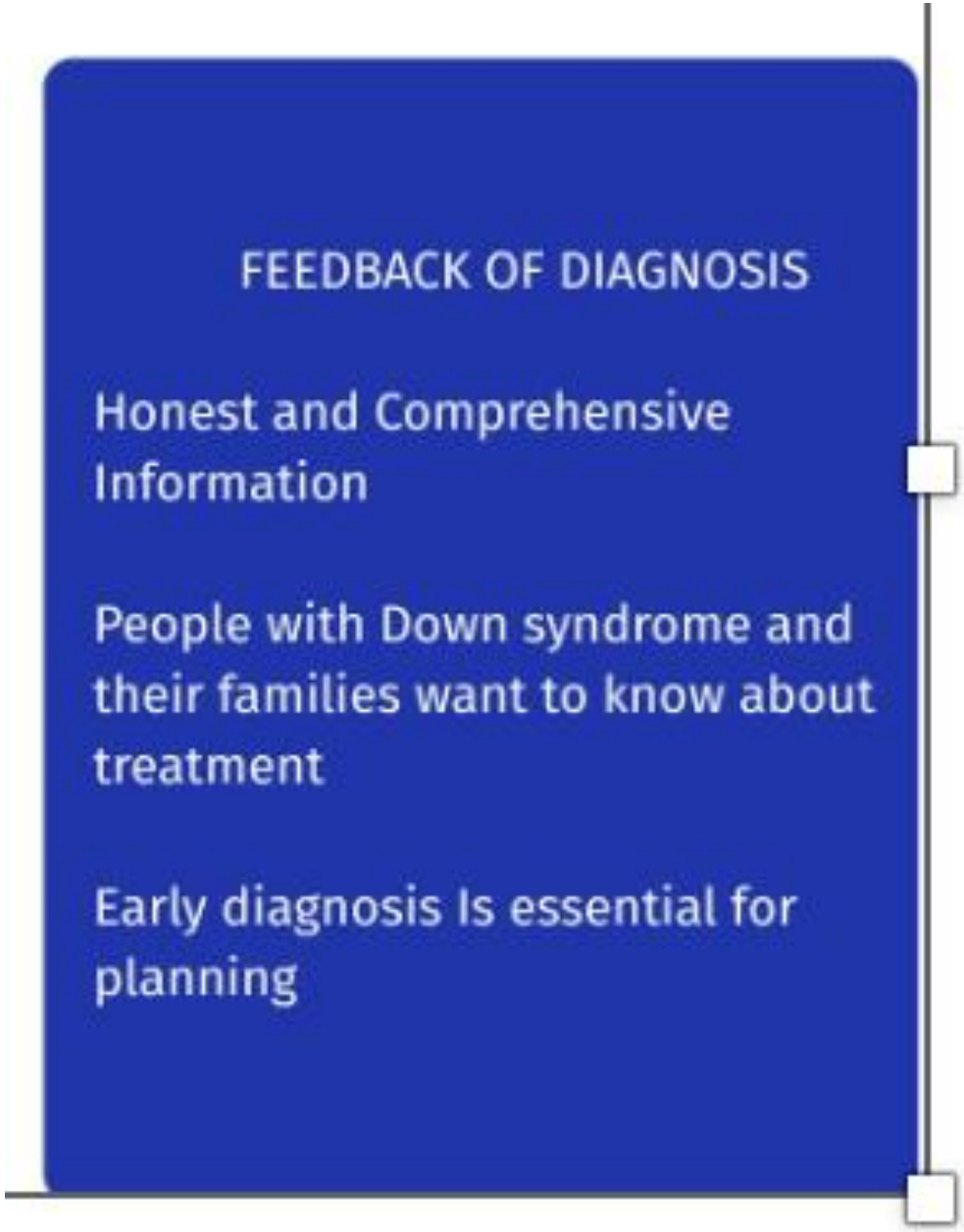


For that, this will say. If you have a wheelchair, be hard" (Participant)

So I personally think something a little more comfortable and less clinical would be better for people..There's a lot of other people like that who would be anxious and worried about it" (Family member)

"The waiting room is nice and quiet" (participant)

"I would like to know what is going to happen" (Participant)
"A handout so I can understand it written down" (participant)



Receiving a diagnosis was perceived as confronting but important, and families requested honestly and comprehensive information

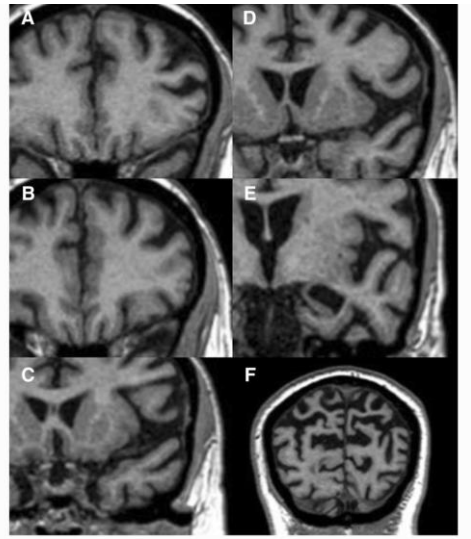
"You want to know the truth. You want to know the hard truth" (family member)

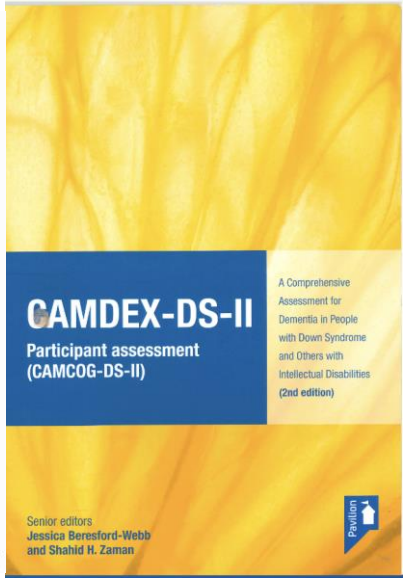
I think it's important, we spoke to participant 10 about her Alzheimer's…you've got a disease called Alzheimer's and that's in your brain, and sometimes you forget how to spell your name, or you can't remember things..that's just your brain….definitely didn't tell her the end result because that wouldn't be it….They have a right to know" (family member)

"My intense interest is in cures..a cure for Alzheimer's because I mean participant 11 is a prime candidate" (family member)

"The feeling about participant 15 having dementia wouldn't be that we've got to do everything we can to stop him having dementia. It would be more well, he's got dementia and this is part of him now. " (family member)

Implementation





Location: Monash Health, Melbourne

Eligibility: People with Down syndrome aged 18 and over

Assessment Tools:

Informant history: “CAMDEX-DS II informant tool, medical history, NPI”

Direct assessment using “CAMCOG-DS II, mCRT +/- TSI, MRI brain

Implementation (cont.)

- Discussion with multidisciplinary team
- Personalized brain health plan
- Feedback of diagnosis
- Post diagnostic referrals

Stage 3

- **Evaluation:** qualitative interviews with participants and their families about experience of clinic and post diagnostic impact
- **Collaboration with consumer groups** Down Syndrome Medical Interest Group Australia, Down Syndrome Australia and Down Syndrome Victoria
- **Funding applications**

THANKYOU

- My patients and families!
- Churchill Trust
- Host teams in UK, US, Europe
- Down Syndrome Victoria/Australia
- Monash Health/NHRMC