Service improvement project: increasing awareness and opportunities for genetic testing in young onset dementia

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INTRODUCTION

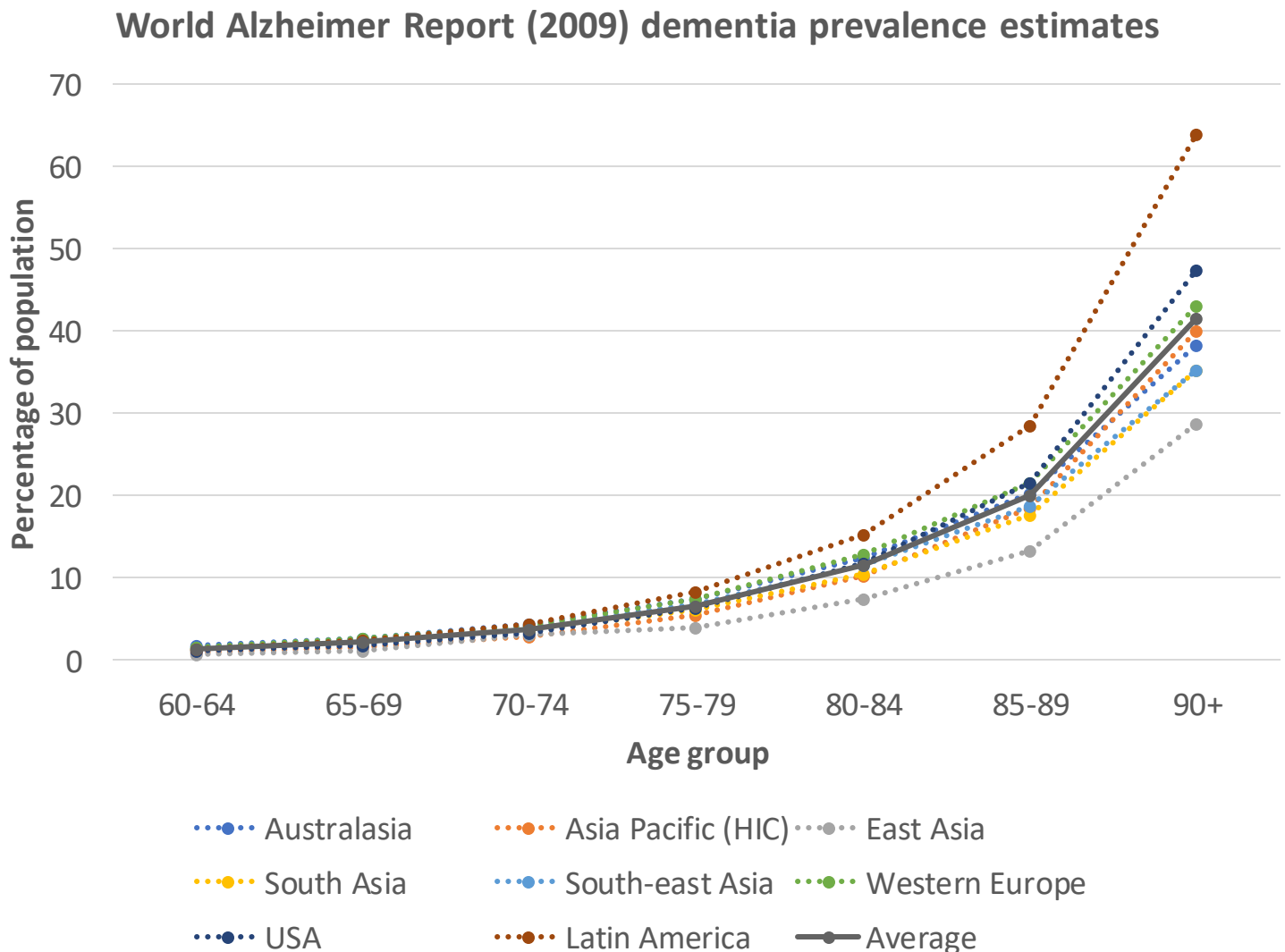
- There is a higher likelihood for people with YOD to have a potential genetic component to their dementia.
- We identified as a service that healthcare professionals were not confident in having discussions around potential genetic risk or testing with people with YOD.
- Admiral Nurses are well situated to have initial and ongoing conversations about genetic factors and the implications of onward referral for genetic testing, especially as many services discharge people at 3 months.

Genetic testing in young onset dementia

- YOD = 7.5% of all dementia
- YOD = >1 in 1000 people aged 30-64

- 75% have no affected first-degree relatives
- Type of dementia influences familial risk

- 15% appears familial
  - 12% YOAD have a genetic cause = 1.6% total
  - 45% YOFTD have a genetic cause = 7.3% total

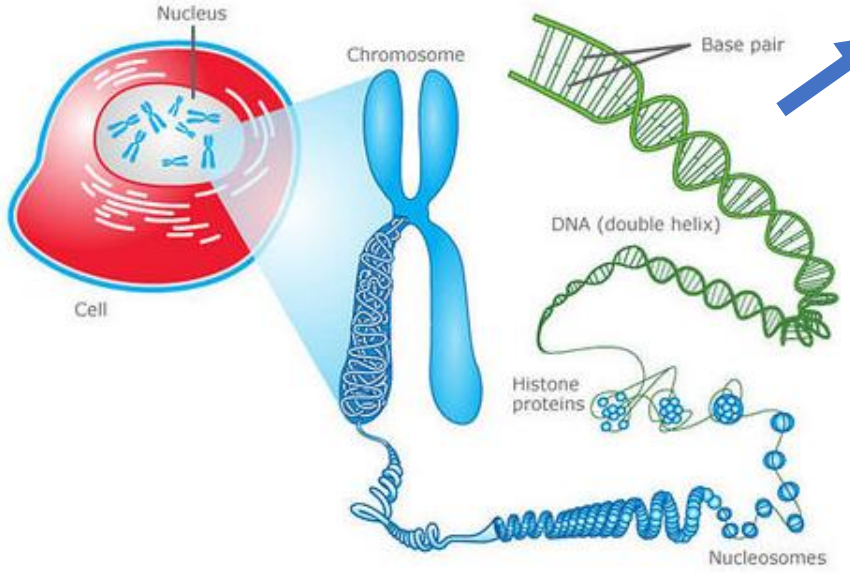


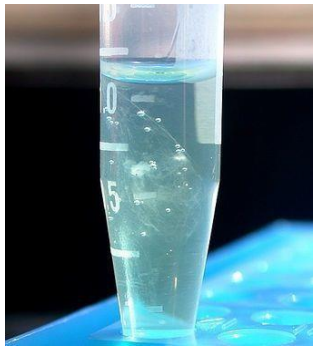
Genetic tests in dementia

- Age of onset <55 years with no acquired cause OR
- Family history of dementia of the same type or of motor neuron disease in first or second degree relative

- Whole-genome sequencing is used:

Blood sample





DNA

Sequencer

Genome sequence "jigsaw"

Neurodegenerative disorder genes (110 genes)

Pros and cons of testing

Pros

- May find a clear genetic cause
- Risks to relatives may be clarified
- Relatives may seek predictive testing
- Gene-specific treatments may become available in future

Cons

- May cause anxiety for relatives
- Can generate feelings of guilt
- Burden of knowledge
- Unable to “un-know” a result
- May get an unclear result
- Risks of dementia often not 100%

- **NB** – DNA storage option

At the Genetics Clinic

- Assess family history information
- Gather and review relevant information (scan results, clinical reports, letters, etc)
- Neurological examination where needed

- Diagnostic test pathway
  - Genetic counselling
  - Genetic test if appropriate

- Predictive test pathway/Unaffected relatives
  - Genetic counselling
  - More than one pre-test appointment needed

Wokingham Memory Clinic

1of 6 Memory Clinics in Berkshire

Embedded in Community Older Adults Mental Health Service

Covers population of 30,000+ >65 year olds

See all age memory referrals – approx. 550/year

Two consultants have clinical interest in YOD

Links with YPWD charity

Admiral Nurses x2 & Dementia Care Advisors for YOD

40 aged <65 years, 95 aged < 75 years

RCPsych College Report Young Onset Dementia

“To optimise diagnostic accuracy, the specialist team should be able to access genetic testing”

Low rates of discussion about genetic causes and no referrals to the regional Genetics Centre.

Approached genetic service

Bimonthly MDT

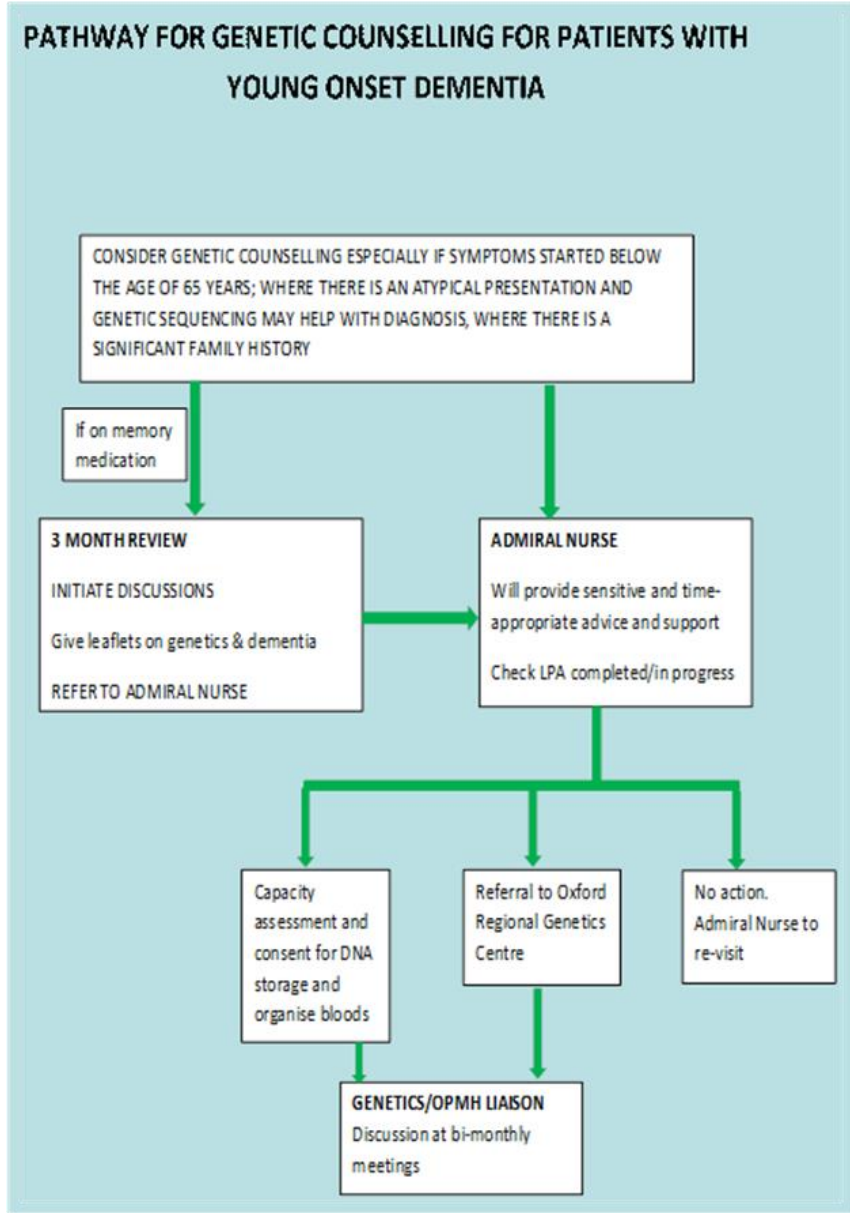
DNA Storage

Referral to Genetics Centre if appropriate

Admiral Nurses

- Discussion about potential genetic factors is not always appropriate at initial assessment or the diagnosis appointment.
- Discharge from Memory Clinic when stable
- Admiral Nurses have unique relationships with people with dementia and their families
- Well placed and highly skilled for conversations about genetic factors, facilitation of DNA storage and the implications of onward referral for genetic testing
- Funding was secured for 2 years of an Admiral Nurse one day per week to increase awareness of genetic factors and access to counselling for people with young onset dementia (YOD) and their families.

PATHWAY FOR GENETIC COUNSELLING FOR PATIENTS WITH YOUNG ONSET DEMENTIA



Outcome 1

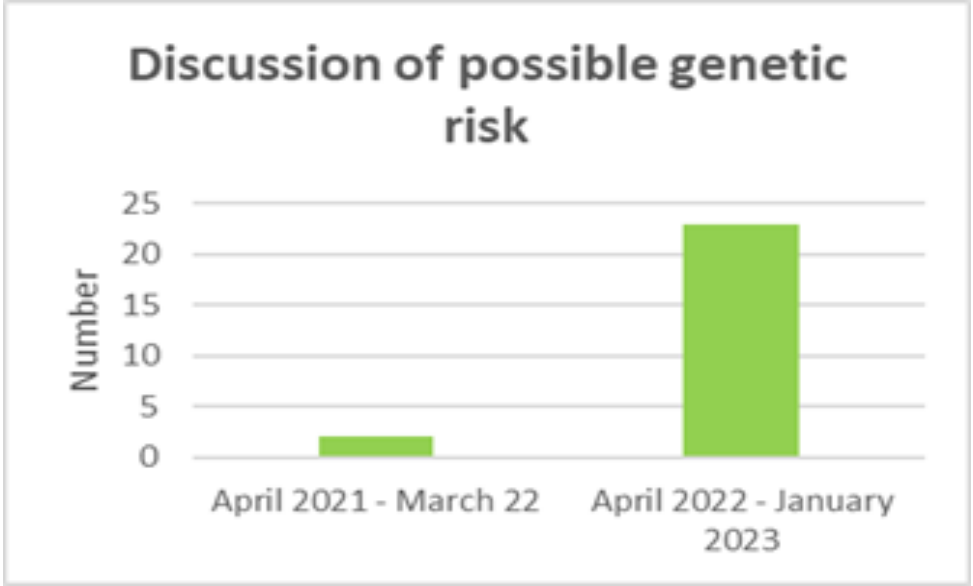
Objective: People are enabled to take an <u>informed and supported decision</u> when considering the implications of genetic testing.

What did we do?:

Produced an information leaflet;

Compared how many conversations about genetic risk before and after (case notes)

Outcome



24 diagnosed per year

Outcome 2

Objective: Professionals have an improved understanding and knowledge around genetic testing for families of people with YOD

Evaluation:

Teaching – before and after knowledge questionnaire.

Audit of recording of genetic history in case notes

Outcome

- 3 training sessions have been delivered. 70% of clinicians reported that their knowledge of genetic testing had improved or greatly improved
- 100% of patient records had family history of 3 generations recorded

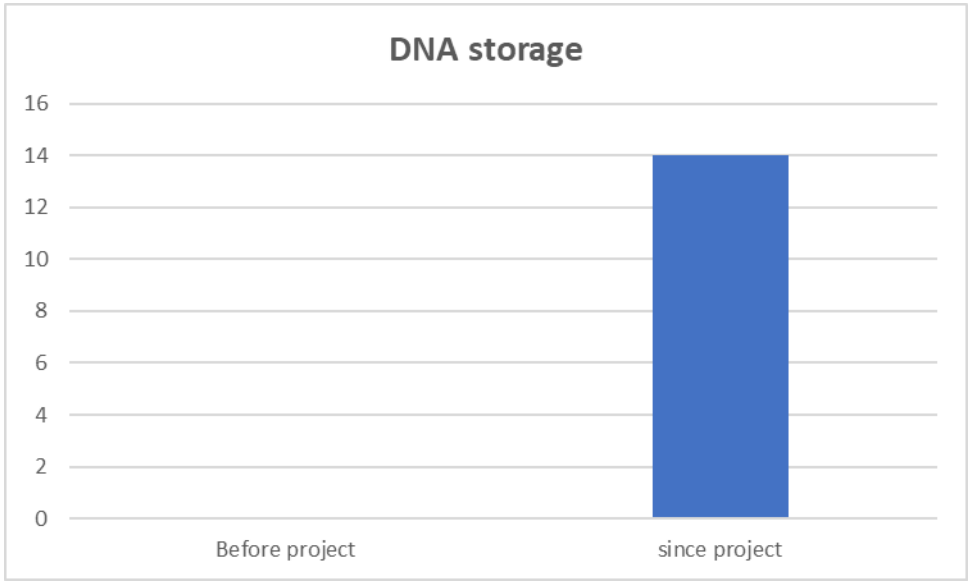
Outcome 3

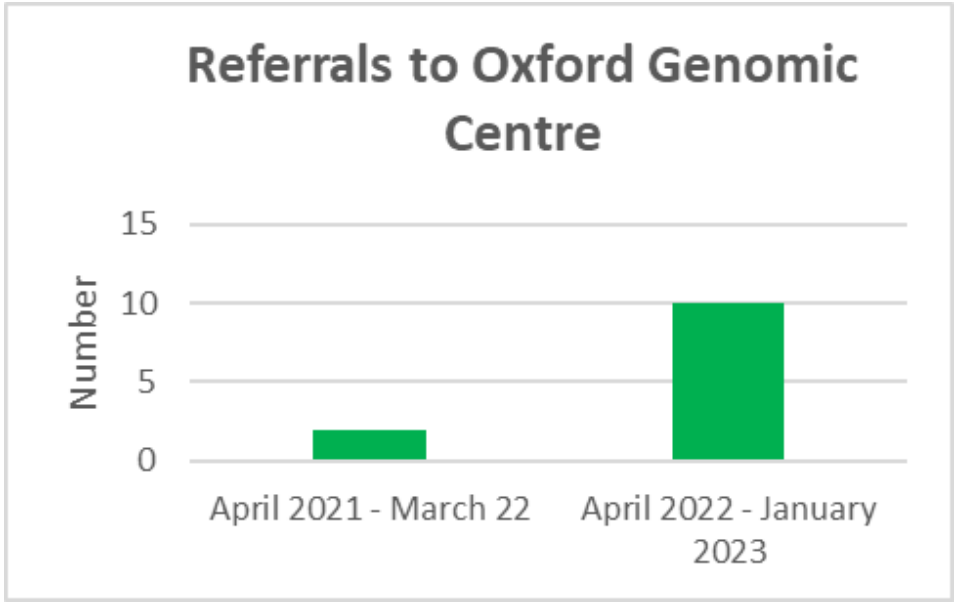
Professionals are more confident in making appropriate referrals for families who may benefit from genetic testing.

Measures:

DNA storage

Referrals to Oxford (including MDT discussion of appropriateness)





Our insights

- Biggest driver has been the link with Genetics Centre – the Admiral Nurse becoming part of the MDT
- Increased knowledge and confidence has come through shadowing & skill sharing
- DNA storage, has been made more accessible for individuals with good capacity assessments.
- Referrals to the Genetics Centre are appropriate and considered

- Genetics consideration should be part of everyday practice for YOD

Time for Questions