

Chapter 15

Disclosure of results

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Key points

- A timely diagnosis of dementia has many benefits such as post diagnosis support and planning for the future.
- Disclosure of results is the moment most feared by people seeking a diagnosis as well as their family members or friends.
- Although most clinicians are at ease with disclosing a dementia diagnosis, they need to be aware that a risk of catastrophic reaction may exist.
- Clinicians should promote informed decision-making, employ proven health communication techniques and provide guidance on appropriate next steps.
- The COVID-19 pandemic has increased the need for remote clinical assessment and disclosure of the diagnosis of dementia.



General background

No doubt, the moment most feared by people seeking a diagnosis, as well as their friends or family members who accompanied them through the diagnostic journey, is the disclosure of the results. Based on a human rights-based approach, the person with dementia should be informed of their diagnosis. However, many people with dementia due to Alzheimer's disease, have a lack of awareness regarding their cognitive and functional decline (this phenomenon is called 'anosognosia') that makes them uninterested in the diagnosis and its likely causes. On the other end of the spectrum are those people who are so anxious about their diagnosis that a catastrophic reaction such as severe depression, and even suicidal thoughts are possible. At this point, the clinician is usually aware enough about the person's state of mind to use a

stepwise disclosure approach; they may say, for example, 'You do have a memory problem and I am glad that you came to see me, let's check your test results and see how I can help you.' Most clinicians will answer a direct question truthfully when there is a low risk of a catastrophic reaction. This outlook is reflected in the survey results. All clinicians will inform the designated legal representative to initiate post diagnosis management (refer to Chapter 16), but often the person accompanying them is a first-degree relative who also has a vested interest in the genetic risk for themselves. This is addressed at length in the upcoming expert essay. The disclosure is usually conducted in person with the clinician; however, COVID-19 pandemic restrictions has increased the need to disclose a dementia diagnosis remotely.

Survey results

The majority of the 1,111 multidisciplinary clinicians who replied to the survey stated that they were comfortable disclosing a dementia diagnosis in their practice.

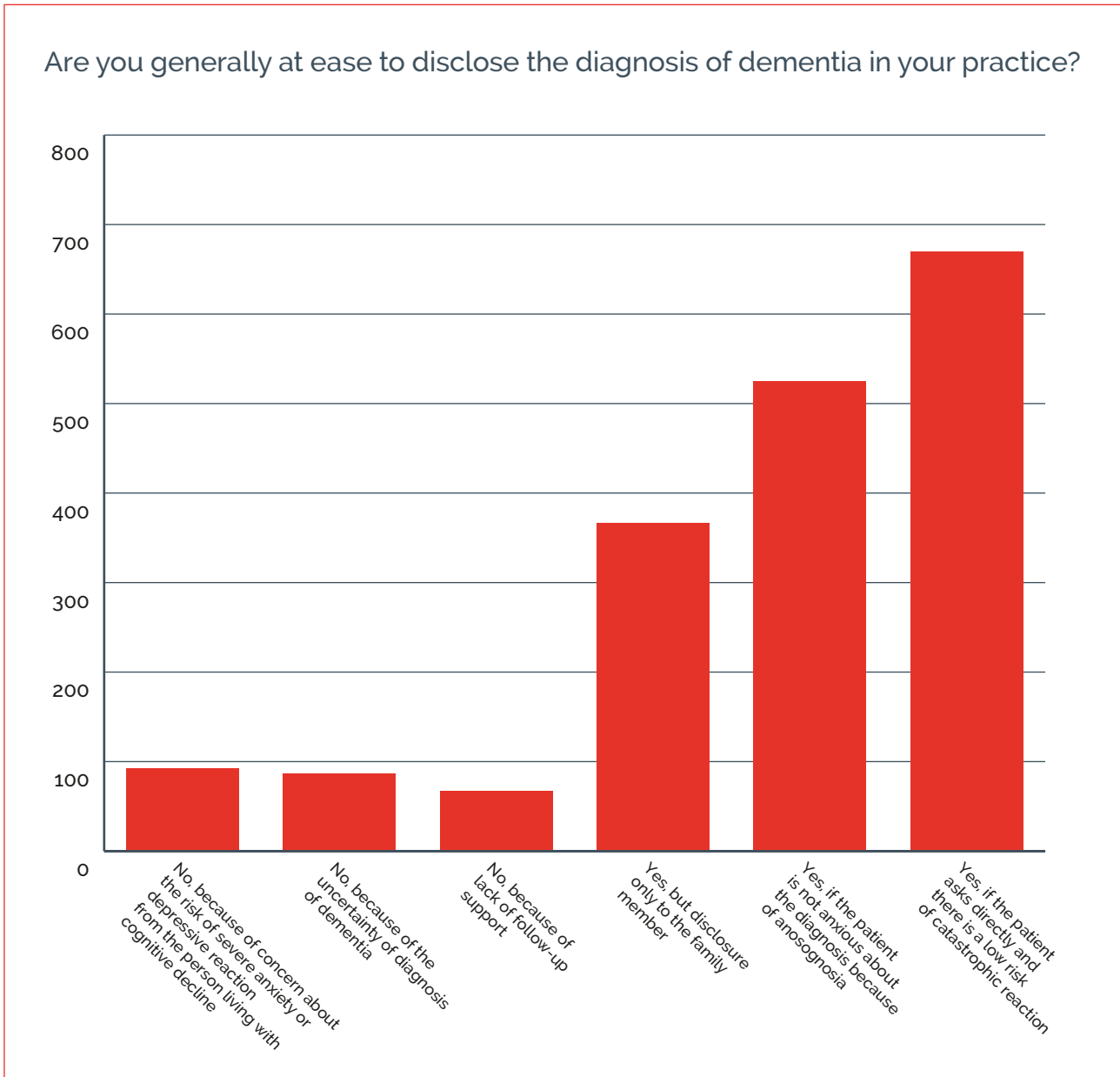


Chart 1. Clinician responses.

The 2,327 people with cognitive complaints dementia and their carers who participated in the survey, indicated that they saw various clinicians during their diagnostic workup. The majority were given the diagnosis by a neurologist.

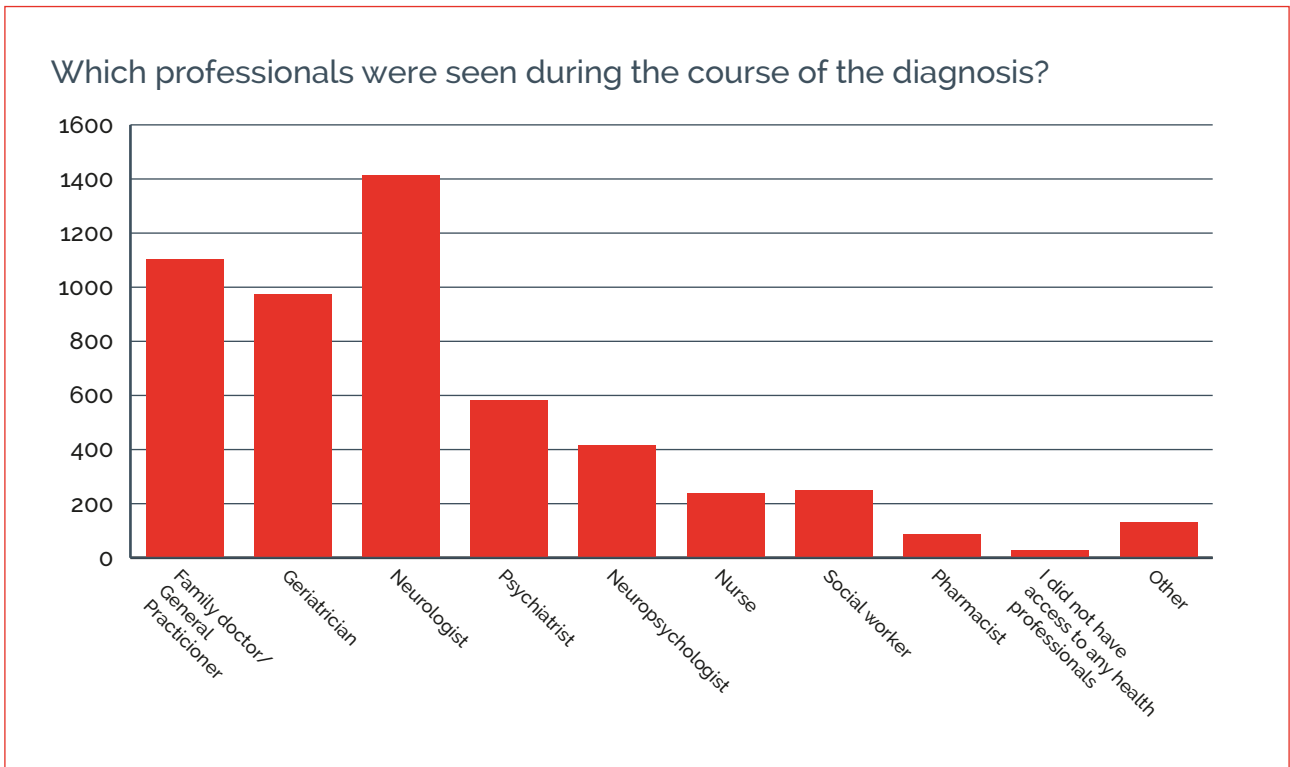


Chart 2. People with dementia and carer responses (multiple answers selected).

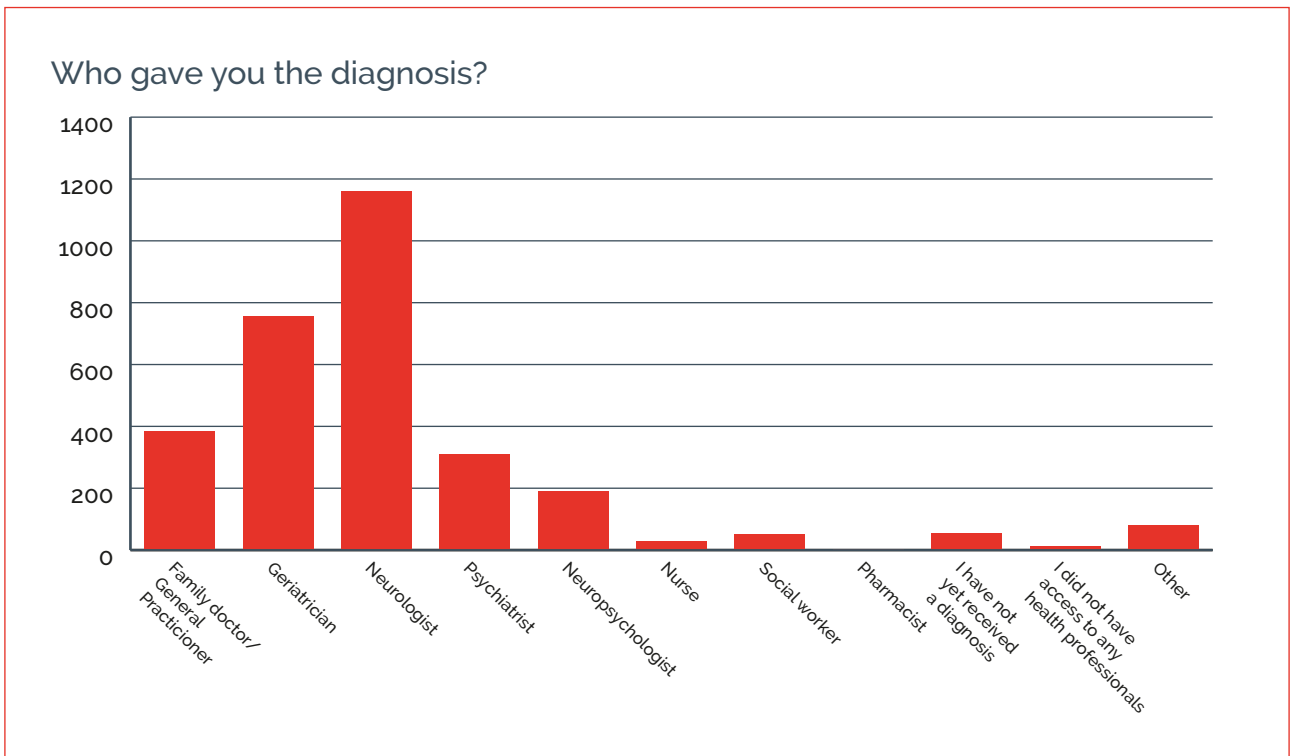


Chart 3. People with dementia and carer responses.

Expert essay

Disclosing APOE genotype to individuals at risk for Alzheimer's disease

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Background

The link between the apolipoprotein E (APOE) gene on chromosome 19 and the risk of Alzheimer's disease dementia has been well-established for decades. Carriers of the e4 allele, who represent approximately a quarter of the general population, are at increased disease risk compared to the general population (where lifetime risk is approximately 10–15%), with e4 homozygotes presenting a particularly high risk (1). However, the e4 allele is neither necessary nor sufficient to cause Alzheimer's disease, and a recent pooled analysis of four large population-based cohort studies of older adults found that lifetime Alzheimer's disease risk in e4 homozygotes is less than 50%, a lower estimate than previous research had suggested (2).

Given its limitations in the predictive value and lack of proven Alzheimer's disease prevention options, APOE genotyping for susceptibility testing in asymptomatic individuals has generally been discouraged by medical experts. For example, a 2011 consensus statement from the American College of Medical Genetics and the National Society of Genetic Counselors recommended against APOE testing for predictive purposes in both clinical and direct-to-consumer (DTC) genetic testing contexts (3). Nevertheless, there is significant public interest in genetic susceptibility testing for Alzheimer's disease, particularly among those with a family history of the disease. Such individuals perceive numerous potential benefits from testing, including learning results that can inform advance planning (for example, purchasing insurance), decisions regarding medical care and clinical research, and engagement in health behaviours to reduce disease risk (4). In 2017, the DTC genetic testing company 23andMe obtained approval from the US Food & Drug Administration (FDA) to offer APOE testing for Alzheimer's disease risk assessment, which has provided millions of its customers the opportunity to learn their genotype.

APOE disclosure has also taken place as part of research studies of the psychological and behavioural impact of genetic susceptibility testing for Alzheimer's disease. Our REVEAL Study, a series of randomised trials examining

APOE disclosure in populations at risk for Alzheimer's disease (for example, first-degree relatives), has demonstrated methods for successfully communicating genetic risk for Alzheimer's disease using processes that a) minimise risks such as a misunderstanding of results and clinically significant distress reactions, and b) require less time and human resources than traditional predictive genetic testing and counselling protocols for neurodegenerative diseases (such as Huntington's disease) (5).



Prior to undergoing APOE genotyping, individuals should be afforded the opportunity to learn about its potential benefits, risks, and limitations. They should know that testing will not provide them with a simple 'yes/no' answer about whether they will ultimately develop Alzheimer's disease dementia, and they should be mindful that results may have implications for other family members.

Best practices in APOE disclosure

Our experience in disclosing APOE genotype status to over 1,000 individuals has yielded some key recommendations for healthcare professionals considering this practice.

1) Promote informed decision-making

Prior to undergoing APOE genotyping, individuals should be afforded the opportunity to learn about its potential benefits, risks, and limitations. They should know that testing will not provide them with a simple 'yes/no' answer about

whether they will ultimately develop Alzheimer's disease dementia, and they should be mindful that results may have implications for other family members; for example, all children of e4-homozygotes would necessarily be e4 carriers themselves. Concerns about genetic discrimination may be pertinent for some, with legal protections such as the US Genetic Information Non-discrimination Act (which covers health insurers and employers but not life, disability, or long-term care insurers) worthy of consideration. Such issues can be addressed in a variety of formats, including online decision aids (for example, www.genetestornot.org) that do not require involvement of genetic specialists (6).

2) Employ proven health communication techniques in disclosure

Ideally, knowledgeable healthcare professionals experienced in communicating sensitive health risk information should divulge results, with telephone and videoconferencing as acceptable alternatives to in-person disclosure. Given widely varying levels of health literacy and numeracy among laypersons, communication may need to be tailored to individuals receiving risk information (under the auspice that sometimes 'less is more'). Visual aids can enhance understanding of quantitative risk information, especially when

comparing risk across different groups. In the REVEAL Study, we have used pictographs (Figure 1) to simultaneously demonstrate both absolute and relative risk associated with being an APOE4 carrier (7). Limitations of risk estimates should be conveyed. Individuals may possess risk or protective factors for Alzheimer's disease not accounted for in models generating risk estimates. In addition, the studies on which risk estimates are based often lack notable diversity in terms of race/ethnicity.

3) Provide guidance on appropriate next steps

APOE disclosure should be accompanied by recommendations for reducing disease risk. Although there are no proven means of preventing Alzheimer's disease, several health behaviours and interventions show promise in lowering the risk of Alzheimer's disease and related dementias, including regular physical activity and management of hypertension. The World Health Organization (WHO) summarised such approaches in its recently issued guidelines for risk reduction of cognitive decline and dementia (8). Individuals should also be made aware of substantive dementia education resources such as the Alzheimer's Association and the US National Institute on Aging. In addition, encouragement to participate in clinical Alzheimer's disease research may be appropriate

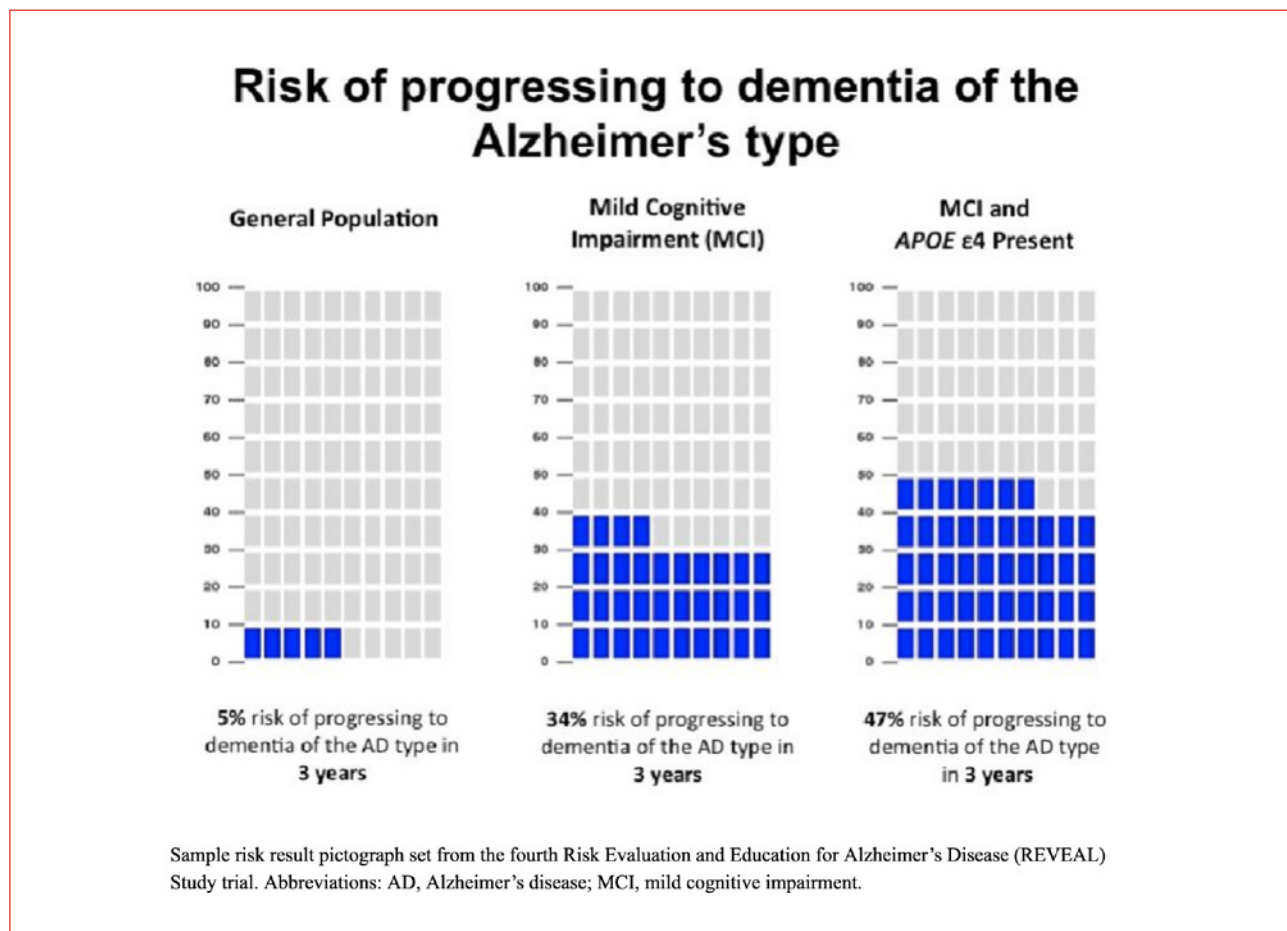


Figure 1. Reprinted from Lautenbach et al, 2013.

in some cases. All key information disclosed to individuals should be concisely summarised in a take-home document for future reference.

Emerging trends

APOE disclosure is increasingly being used or considered for purposes beyond merely informing interested individuals about their chances of developing Alzheimer's disease dementia. For example, APOE genotyping has been employed to help identify asymptomatic, elevated risk participants for Alzheimer's disease prevention drug trials (9). As noted elsewhere in this report (Chapter 24), APOE testing could assist in reducing costs of the Alzheimer's disease

diagnostic process by helping determine which cognitively impaired individuals need (or don't need) expensive follow-up biomarker testing such as amyloid neuroimaging. The recent US FDA approval of aducanumab to treat Alzheimer's disease suggests a potential adjunctive role for APOE testing in informing medical decision-making, given that $\epsilon 4$ carriers are at significantly elevated risk for the side effect of amyloid imaging related abnormalities (ARIA); APOE genotyping has already been used to inform clinical management of a $\epsilon 4$ -homozygote patient experiencing vasogenic oedema (ARIA-E) and intracerebral haemorrhage (ARIA-H) side effects from aducanumab use (10). These developments demonstrate the rapidly evolving uses and implications of APOE testing even three decades after its introduction.

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Expert essay

Sharing the diagnosis of dementia in the post-COVID-19 clinic: patient and practitioner perspectives: dementia assessment and diagnosis during lockdown

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The timely diagnosis of dementia has many benefits for an individual such as accessing medication and post diagnosis support, and planning for the future. Most people with suspected dementia are seen at a memory clinic, but the COVID-19 pandemic resulted in many memory clinics moving to remote consultations using telephone and video-calling.

Receiving a diagnosis of dementia is often a negative experience for the person and their close family (1). There is also a recognition that making a diagnosis is 'nuanced and

challenging' (2) for the clinician; the shift to remote diagnosis has made this even more complex. It has also raised concerns about how remote diagnosis is being experienced by the person with dementia and if it is possible for it to be delivered sensitively with appropriate support.

There are arguably disadvantages to a remote diagnosis including difficulty picking up on the person's non-verbal cues and distress, technical issues such as time lags with connection, and having an uncontrolled environment for the assessment and diagnosis of dementia. Indeed, anecdotal

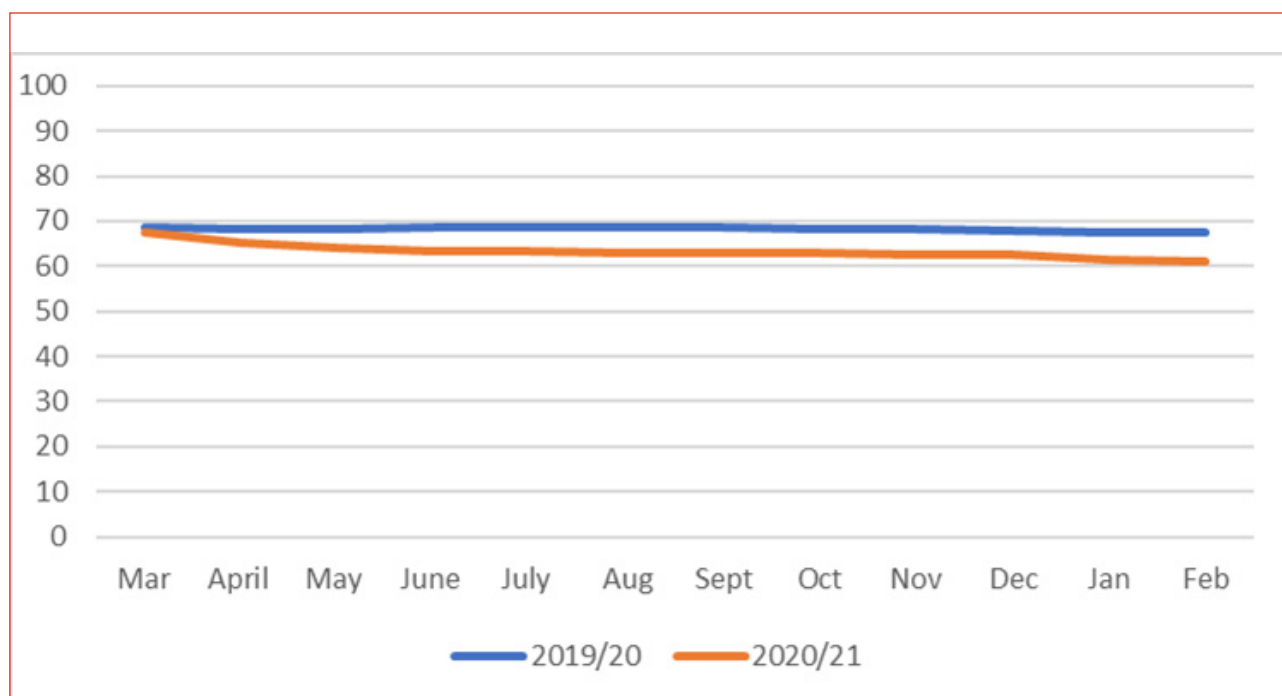


Figure 1. Proportion of the total estimated number of people with dementia who have been diagnosed. Percentage of the total estimated number of people with dementia who received a diagnosis pre-lockdown (2019/20) and during lockdown (2020/21). Source: NHS Digital (4).

experience highlights that not being able to see how someone responds during this discussion makes the process much more challenging for the clinician.

The number of people being diagnosed with dementia has decreased since the beginning of the first UK lockdown in March 2020 compared to the previous year, and this has resulted in a 7.6% drop in the number of people with a diagnosis of dementia for the period (Figure 1). This substantial reduction has been attributed to a range of factors including clinical guidance on reducing the priority of non-urgent primary care, people being fearful of contracting COVID-19 and also not wishing to burden health services during a pandemic (3).

Purpose of study

While standardised assessment tools have been suggested as reliable for the diagnosis of dementia via video-calling (5), the impact this has on the individual remains a key consideration. Being informed that you have dementia is a significant event in a person's life, and how the assessment and diagnosis is experienced will remain with that person for a long time. The rationale for this research is to impact positively on the practice of remote diagnosis and crucially, the experience of the person with dementia and their close family members.

The study arose from discussions with two key groups: 1) debates within clinical services around what is considered ethical and best practice and 2) consultation with the Patient Public Interest Group of the NHS Scotland Neurodegenerative and Dementia Clinical Research Network (6). Delivering a remote diagnosis may become the new normal post-pandemic, but there is a lack of understanding on how this is experienced by the person with dementia (7). It is also disputed whether the practice of remote diagnosis should continue at all.

We want to explore the experience of people given a diagnosis over the phone or video-calling as well as the staff working in memory clinics. This research project will consider the emotional impact, practical implications and ethical considerations of delivering and receiving a remote diagnosis of dementia. The focus will be on the impact on the individual, drawing conclusions from the findings of the interviews and the consensus reached through an Online National Forum to make a recommendation on whether remote diagnosis should continue, and if so, how it should be conducted.

Study approach

A Research Advisory Group has been established comprising people with personal experience of dementia. This Group will inform and advise the research team throughout the research process, meeting regularly to work collaboratively on planning, analysis and reporting.

The research team will interview approximately thirty people who received a remote diagnosis of dementia from the beginning of the first UK lockdown in March 2020. Staff from memory clinics and equivalent services who have been carrying out remote dementia assessment and diagnosis will also be interviewed. Recruitment to the study will be UK-wide, making the findings applicable to all four nations of England, Scotland, Wales and Northern Ireland, and arguably further afield. The findings from these interviews will be analysed in collaboration with the Research Advisory Group, drawing on methods used by research team members in previous co-produced research projects with people living with dementia (8).

Led by the values of co-production, our approach brings together people with personal experience and researchers to work in partnership. To include these perspectives, we are ensuring involvement is accessible and equally valuing the knowledge of everyone involved.

The second phase of the project will bring together a wider range of stakeholders including people with personal experience of dementia, professionals and people working in dementia fields in an online consultation. The findings from the interviews will be presented at this event and discussions held to allow the participants to contribute to the outputs of the research project.

Outputs from the study

Learning from people who have received a diagnosis during a global pandemic will allow us to enhance practice for the future, including a more nuanced understanding of the ethical implications. It is also important that we learn how practitioners have adapted their approaches to meet the challenges of working remotely with their patients.

There will be several outputs from this project to share the learning as extensively as possible. These will include clinical guidelines for practitioners, a briefing paper for policymakers, academic papers to develop the evidence base and a short, animated film and podcasts targeted at a wider audience. These outputs will also be shared with people who have contributed to the research as members of the Research Advisory Group, taking part in the online consultation or being interviewed for the study, as well as being distributed more widely.

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Conclusions

A visit to a healthcare professional to receive diagnostic results can be a nerve-wracking experience. It can elicit fear – fear of the unknown and perhaps also that suspicions may be confirmed. Some people with anosognosia, a lack of awareness about their condition, may appear indifferent or unconcerned while others may feel high levels of anxiety and may have depression or suicidal thoughts. A skilled clinician, while remaining truthful, should be able to discern which way an individual is leaning in their reaction and adapt their responses accordingly during the disclosure process.

When it comes to taking matters into your own hands, the proliferation of genotyping kits has given people the opportunity to explore their probability of developing dementia. Some individuals prefer to know their risk level so they can be prepared and plan for the future. There are, however, predictive limitations to these types of available kits, and most medical professionals discourage their use for this purpose.

The COVID-19 pandemic, and its restrictions, led to changes in the diagnostic process, and how disclosure is conducted. Telephones, and now video-calling, has made remote disclosure a reality. However, constraints are evident, especially as the uncontrolled environment may inhibit the ability of the clinician to pick up on an individual's non-verbal cues, not to mention any technical issues that may interfere. Learning from both the clinicians' experience and people who have received a remote diagnosis should provide direction for an effective reciprocal exchange and development of best practice.