

Risk disclosure and its consequences

Perspectives of asymptomatic research participants in the UK and Spain

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Medicine

Alzheimer's breakthrough seen

Simple eye exam may lead to diagnosis

BOSTON (AP) — The discovery of an apparently unique form of optic nerve damage in victims of Alzheimer's disease may make it possible someday to diagnose the illness with an eye exam, researchers say.

Even though Alzheimer's disease is a major cause of senility in the elderly, with an estimated 2.5 million victims in this country, it is not the only cause. Its symptoms are confusing, and doctors often cannot be entirely sure they have diagnosed it correctly until the victim dies and an autopsy is performed.

In a new study, researchers have found degeneration of the optic nerves in people with Alzheimer's disease. The next step will be to see if this causes specific, unique vision abnormalities that can be measured.

"It would be helpful perhaps in making the diagnosis, if we come up with a battery of tests that sort this out in life, rather than at death,"

said Dr. Carol A. Miller, chief of neuropathology at the University of Southern California.

The study was directed by Dr. David R. Hinton, a researcher in Miller's lab, and was published in Thursday's New England Journal of Medicine.

Alzheimer's disease attacks nerve cells in the brain. Symptoms include memory loss, disorientation and changes in personality.

In the study, doctors examined the optic nerves, which carry visual messages from the eyes deep into the brain.

The actual nerve damage can be seen only during an autopsy. The researchers examined the optic nerves of 10 Alzheimer's victims and found the degeneration in eight of them. However, they found it in none of 10 normal people of the same age.

However, they did not check people with other forms of brain disease. Dr. Peter Davies, an Alzheimer's expert at Albert Einstein College of Medicine in New York, said the significance of the work won't be clear until this is done.

"It is really going to be specific for Alzheimer's disease and not occur in other neurologic diseases?" he said. "That's the real issue here."

It's not distinguishing Alzheimer's from normals. It's picking out Alzheimer's from any one of 40 other diseases."

Miller said another co-author of the research, Dr. Alfredo A. Sadun of the Estelle Doheny Eye Foundation in Los Angeles, is testing Alzheimer's victims to see if the nerve degeneration causes measurable abnormalities in vision. Finding such disturbances will be necessary if the condition is to serve as a signpost of the disease.

The optic nerve functions something like a telephone cable. But instead of being packed with individual wires, it is a bundle of nerve structures called axons. The researchers found a two- or three-fold depletion of these axons in the Alzheimer's victims.

No treatment exists for Alzheimer's disease. A better diagnostic test could help doctors distinguish it from similar-looking diseases that do have a treatment. However, accurate diagnosis may offer little for those who are in early stages of the disease.

"I don't know how much it will help the patient, because who wants to know they will become demented?" said Miller.

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On Not Jumping the Gun: Ethical Aspects of APOE Gene Testing for Alzheimer's Disease^a

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Sunday Review

What if You Knew Alzheimer's Was Coming for You?

Simple blood tests may soon be able to deliver alarming news about your cognitive health.

By PAGAN KENNEDY NOV. 17, 2017

"I don't know how much it will help the patient, because who wants to know they will become demented?" said Miller.

"when it comes down to what we want to use data and AI for, it is early detection and diagnosis. That's, that's one of the big ones for us. And [that raises] questions around, why would you want to know early?"
(digital health tech researcher, interview with RM, SPACE study 2020)

Consequences of risk communication to asymptomatic individuals



- ApoE result communication does not cause distress among the majority of people who do not have major anxiety or depressive disorders, and who display an interest in genetic testing
- REVEAL (11/14 studies)
 - Selected/non-representative populations
 - US based
- Some evidence of change in LTC uptake, behaviour change
- A small % of people did experience psychological effects
- Possible 'nocebo' effect
- Increased fears of employment discrimination
- Focus on short-term psychological effects (anxiety, depression)
- Over time, seen as valuable – but with lasting effects for some (Zallen 2018)

Approaches to the Communication of Alzheimer's dementia Risk (ACAR)

1. What do older adults involved in dementia research expect would be the implications of learning Alzheimer's dementia risk?
2. What expectations would they have of any disclosure process?

Approaches to the Communication of Alzheimer's dementia Risk (ACAR)

Focus group based research in the UK (Milne) and Spain (Diaz) and with EWGPWD

N=61 (32 (20F/12M) in four groups in Spain, 19 (10F/9M) in four groups in the UK), 1 group of EWGPWD.

Information provided on changing approach to Alzheimer's research

Thematic analysis around shared structured protocol



estudi alfa



Background

Discussion influenced by family experiences

Some already felt that they were 'at risk' because of family history

Most people expressed interest in learning their risk status – but didn't feel their desire to know was typical



Risk and time

“you’re not going to have an attack of Alzheimer’s, it’s more of a, it’s a ‘living with’ situation, which sometimes heart disease can be, obviously, but sometimes you don’t get the option of living with it!” (UK2)

At, with and beyond risk: expectations of living with the possibility of future dementia

**Richard Milne¹, Ana Diaz², Shirlene Badger¹,
Eline Bunnik³, Karine Fauria⁴ and Katie Wells⁵**

The risk experience



At risk

**With
risk**

**Beyond
risk**

Living at risk

Reducing risk

“I think it's important to know to act. Although there is no medication, if factors such as diet, environment influence it, you can act somehow” (SP3)

Reducing risk

“What are we going to change? Do we eat better? But we already do that, or we should. Do we exercise more? We should do it. What are we going to change? . . . You will not change anything; I think, honestly.”

(SP4)

Pre-emptive suicide

“I want to know immediately, that is, even a risk of 80%, not 99%, to make my own decisions ... Bump me off before I get to be like my mother.” (SP2)

Living with risk

Informal vigilance

“It would have an impact on how I then lived my life, and how I conducted my relationships with others, and my work. I would be looking for the signs of it . . . I’m bad enough as it is.” (UK1)

Formal monitoring

“For me what is important about knowing the risk is early detection, a serious advantage is that you can monitor to diagnosis.” (SP2)

Living beyond risk

Planning for self and others

I would do many things if I knew that this was going to happen to me. I would spend more time with my family, I have a grandson of a year and a half and one of eight months. I would try to be with them as much as possible and all those things.'

(SP1)

Summary

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Similar conversations in UK and Spain

Learning one's Alzheimer's risk may have medium and long-term effects

These are neither clearly benefits nor harms, but disruptions that need understanding and supporting

Future relationships with families and healthcare need to be accounted for in preventative programmes

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