PREVENTIVE NEUROLOGY: How should healthcare professionals and health systems respond to the likely demand for genetic testing to assess disease risk?

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• Health systems are not equipped to manage large numbers of people at potentially high risk of long-term neurodegenerative conditions
• Increasingly, individuals are keen to know their risk of a disease, or have sought out their risk using private ‘screening’ methods, and are seeking answers
• As the demand to assess risk via genetic testing grows, how should healthcare professionals and health systems prepare to help individuals interpret their results?
Overview

- An introduction to preventive neurology and the report *Time matters: a call to prioritize brain health*
- A focus on direct-to-consumer (DTC) genetic testing
  - Background
  - Regulatory and clinical perspectives on DTC tests
  - Future directions
Society needs to understand and talk more about brain health

**Society**
Brain disease is generally regarded as a problem for the elderly, but there is a need to understand that neurodegeneration begins in middle age, even though it is not apparent until later.

**Change**
There is a window of opportunity for change in midlife, during which behaviour changes are beneficial before the onset of symptoms.

**Early intervention**
Policies are needed to improve knowledge about risk reduction strategies and maximize the scope for prevention and early treatment.

**Knowledge mobilization**
We must learn how to share information about brain health in a timely and effective manner.
An expert group developed a **policy report outlining a series of recommendations and a call to action** encouraging positive behaviours and new policies to promote brain health and to aid prevention of neurodegenerative diseases.
The message “what’s good for your heart is generally good for your brain” needs to be widely communicated and understood.

Policymakers and public health bodies should act on these recommendations:

- Protect and provide budget to improve public understanding of how to preserve brain health and promote a positive approach to prevention.
- Encourage behaviours at all ages to improve brain health.
- Provide a supportive environment, including guidance and legislation that empowers people to make important lifestyle changes.
- Prepare for the likely growth in the demand for genetic testing.
- Provide access to available and effective treatments in a timely manner.
Risk factors that affect brain health

Preparation of the *Time matters* report and supporting materials has been funded by grants from Biogen and F. Hoffmann-La Roche, who had no influence on the content.
The ‘at-risk’ phase
What does the general public need to know about risk factors for neurodegenerative diseases?
Increased appetite to find out about individual risk

Which, if any, of the following health conditions do you think it’s possible for people to reduce their risk of developing?

- Diabetes: 81%
- Heart disease: 77%
- Stroke: 60%
- Cancer: 52%
- Dementia: 34%

34% of respondents think it is possible for people to reduce their risk of developing dementia.

Adapted from Alzheimer's Research UK. Dementia Attitudes Monitor 2018
Direct-to-consumer genetic testing

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What do we mean by direct-to-consumer (DTC) genetic testing?

• This is genetic testing available to the public without assistance from a healthcare professional
• The number of companies providing this is growing rapidly, along with the range of health conditions they cover
• Testing kits typically contain instructions and equipment for collecting a saliva sample, which customers post to the DTC company for analysis
• Consumers are notified of their results by post, over the telephone or online
What information do people receive?

• The most popular tests use common genetic variation to make predictions about:
  • physical traits like lactose intolerance and male pattern baldness
  • how a person might respond to specific medications
  • whether there is a higher than average risk of developing certain diseases like cancer or neurodegenerative disease
  • if the person is a carrier of genetic alterations that are associated with a number of rare genetic conditions

• DTC genetic tests are not meant to diagnose, prevent or treat any disease or health condition
Types of results

• Most common results are an estimate of variation in a gene or locus associated with a given disease, based on variation at a single nucleotide polymorphism (SNP), but not from direct sequencing of a mutation
  • Presence of SNP does not mean that you will get the disease
  • Absence of SNP does not mean that you will not get the disease

• Some companies also report polygenic risk scores (PRS)
  • These scores are calculated from a large number of genetic variants scattered throughout the genome
  • There is substantial overlap between PRS in patients and controls, such that these do not amount to good screening tests
Consent and data use

• A person’s genetic data represent personal, private health information

• Where consumers expressly grant permission and provide an informed consent, they can choose to share their genetic data with responsible researchers
  • This is to support important breakthroughs in biomedical research, healthcare and personalized medicine

• Each company has different protocols for safeguarding and usage of data; it remains inconsistently regulated
Misconceptions with results interpretation

- The ability of the general public to understand DTC genetic test results depends on their ability to understand and interpret the complex risk values associated with the results.

- Misunderstanding of the results could have negative consequences, including unnecessary concern, false reassurance or unwarranted changes in behaviour (healthy or unhealthy).

- Confusion about positive results may result in advice being sought from a healthcare professional.

Regulatory and clinical perspectives on DTC genetic tests
What are the current regulations around DTC genetic tests?
Opportunities and challenges for HCPs when discussing genetic test results

**Opportunities of DTC genetic testing**
- Healthcare professionals could use the discussion of results for health promotion
- DTC tests help to promote awareness of diseases that have (in part) a genetic basis

**Challenges of DTC genetic testing**
- Inadequate support or training to enable HCPs to provide effective counselling after risk disclosure
- Lack of oversight and consistent regulation
- Genetics are only part of the story; lifestyle factors must be considered
- Tests are not validated to provide a medical diagnosis
Guidance and training for HCPs

Direct-to-Consumer Genetic Testing: What Clinicians Need to Know
Join Drs McCarthy and Vassy in a discussion of direct-to-consumer genetic testing.

Direct-to-Consumer Genetic Testing: Successfully Navigating Patient Encounters
What do you discuss with your patient when she comes in with results from direct-to-consumer genetic testing?

The DTC Genetics Talk Show: Finding the Advantage for Patients
Join Drs Vega, Palaniappan, and Vassy as they take an interactive, patient-centered approach to new insights in DTC genetic testing.
Future directions

• More **training resources** are needed for clinicians around the world to improve preparedness for increased uptake of DTC genetic tests.

• Healthcare professionals and administrators will continue to play a **key role** in the management of people with, or at risk of, a neurodegenerative disease.

• Further work is needed to **validate diagnostic tools** to identify people at risk and to develop effective treatments.
Feedback

• We would like to hear from you about issues related to the following
  1. Have many patients come to you with DTC genetic test results in your clinic/practice?
  2. What resources would you find helpful to support these patients?
  3. What resources would your patients find helpful regarding DTC genetic tests?

• Give us your feedback and get in touch to find out more!

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