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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Today's presentation

- 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 is it 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



Time matters: A call to prioritize brain health

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





Health promotion recommendations

- 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





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



Direct-to-consumer genetic testing





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

For most health conditions (like diabetes or cancers), each person has some risk of developing the condition during their life. This diagram represents this as a bucket – a person experiences the condition when their bucket gets full up. Most health conditions arise due to a combination of genetic and other risk factors. Genetic factors Other factors Everyone starts life with some genetic risk. factors for the condition. Some people have very few, some people have a lot. You are born with this risk and you can't change it. Direct-to-consumer genetic tests aim to measure this risk (though their measurement is often far from perfect). Over the course of life, people are exposed to other factors that eg smoking increase the chance of them experiencing the condition. If you had more genetic risk factors in your bucket to begin with, it takes fewer other risk factors to fill the bucket and result in the health condition. Some of the other factors, you can't change. like your age. Some factors, you might be able to work on, like smoking, or alcohol intake. Even if you have a 'high genetic risk' of a particular condition, for most health conditions, having a high genetic risk does not mean you'll get it. By working on the factors you can change, you might be able to reduce the chance of developing a health condition, even if you have a high genetic risk.

- 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









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

The General Public's Understanding and Perception of Direct-to-Consumer Genetic Test Results. Leighton et al. Public Health Genomics 2012;15:11–21.

Regulatory and clinical perspectives on DTC genetic tests





What are the current regulations around DTC genetic tests?







PRIVACY BEST PRACTICES FOR CONSUMER GENETIC TESTING SERVICES

💟 @FutureofPrivacy	www.fpf.org	FutureofPrivacy
Annual transparency reports describing law enforcement requests	Request destruction of biological samples	Employ strong data security practices and privacy by design
 Educational resources about the basics, benefits, and risks of genetic testing 	 Access, correct, and delete their Genetic Data 	 Require valid legal process for disclosing Genetic Data to law enforcement
practices and detailed explaination of how Genetic Data is collected, used, and shared	and use of Genetic Data Give informed consent for research	insurance companies, and educational institutions without consent
A high level overview of key privacy	Consumers can: Give express consent for the collection	Companies will: NOT share Genetic Data with employers.
PROMOTES TRANSPARENCY	PROVIDES CHOICES	ENHANCES PROTECTIONS

J Community Genet (2018) 9:117–132 https://doi.org/10.1007/s12687-017-0344-2

ORIGINAL ARTICLE

Legislation of direct-to-consumer genetic testing in Europe: a fragmented regulatory landscape

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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**. Expert Commentary, Oct 31, 2017

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**?

Article/Courses, Mar 26, 2018

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**.

Article/Courses, Nov 19, 2018



23andMe for Healthcare Professionals Advancing patient health. Together.



The DTC Genetics Talk Show Finding the Advantage for Patients



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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Disclosures and acknowledgement

Alastair Noyce has received the following

- Grants
 - Preventive Neurology Unit Barts Charity
 - PREDICT-PD Parkinson's UK
 - East London PD Project Virginia Keiley Benefaction
 - Global Parkinson's Genetics Program (GP2) Aligning Science Across Parkinson's
- Consultancy fees
 - Britannia Pharmaceuticals
 - Global Kinetics Corporation
 - Profile Pharmaceuticals
 - Bial Pharmaceuticals
 - Biogen
 - F. Hoffmann-La Roche

Thank you

