



Making a Life Following a Positive Gene Test

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Huntington's Disease
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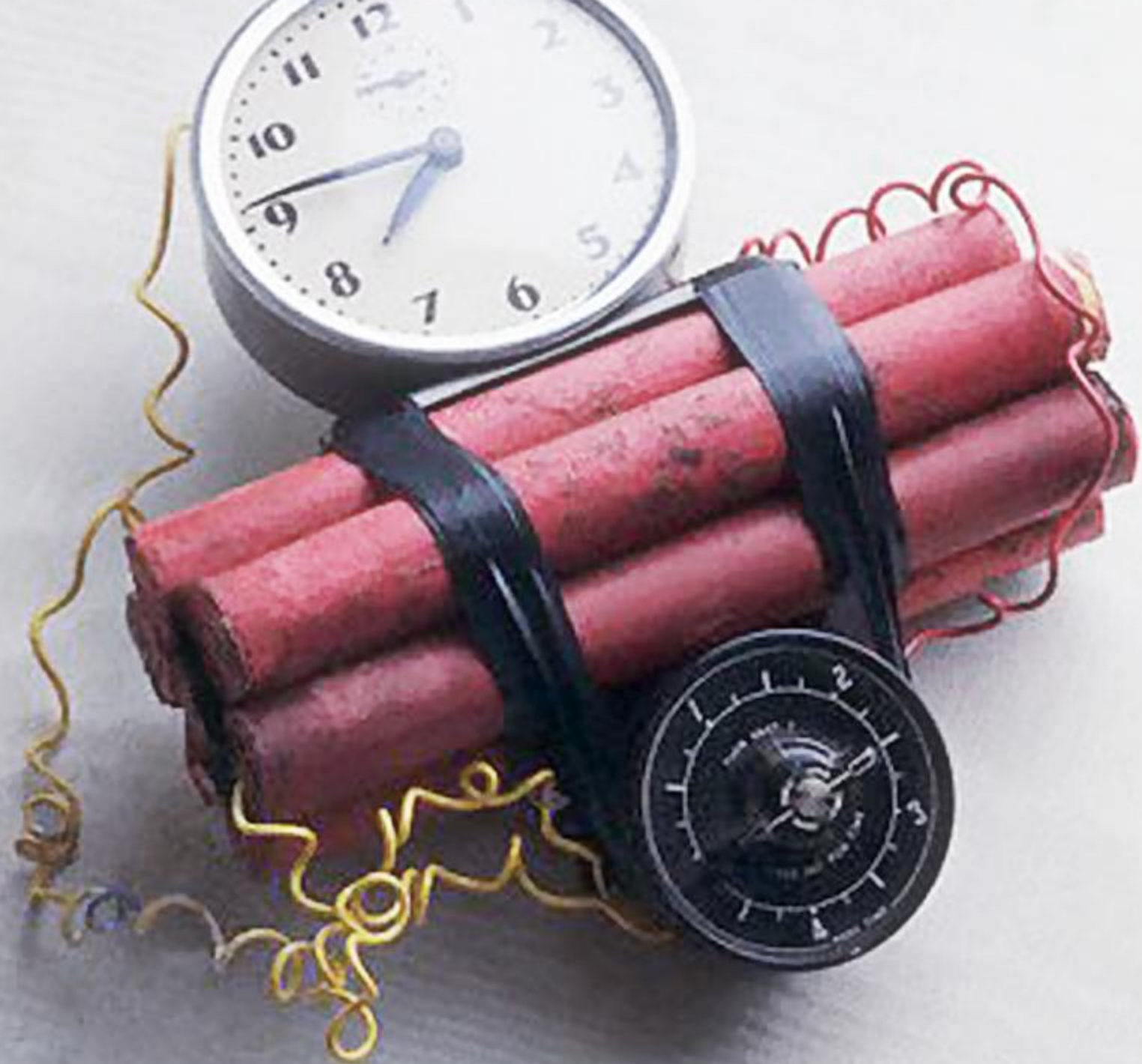
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“Being at risk is dying a little each time you drop a spoon. Being at risk is looking at your brothers and sisters, wondering which one of you will be the first to go. This constant pressure, day in and day out, year after year takes a heavy toll..I feel at times that being at risk has taken away my right to love and be loved.”

Walking the Tightrope
Randi Jones

"All the News
That's Fit to Print"

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Photographs by Suzanne DeChillo/The New York Times

Katharine Moser, below, and above at a nursing home in Manhattan with her cousin James Dowd, who has Huntington's disease.

Confronting Life With a Lethal Gene

A Young Woman's DNA Test Points to an Inevitably Grim Fate

What runs in your own family and would you REALLY want to know?

“Soon everyone is going to have an option like this. You make the decision to test, you have to live with the consequences.”

Nancy Wexler, PhD
President, HDF

Testing of AR Individuals: What are the Implications?

- Departure from family norms
- “Pre-selection” often exists within families
- Adults with normal results generally have prepared for the worst
- Following testing, how to tell and who to tell?
- Generally takes at least six months to two years following testing to redefine life goals and purpose

“Some argue that even a diagnosis of HD is LESS stressful than the uncertainty of 50% risk status.”

Meissen et al, New England Journal of Medicine,
1992: 328

The “Cons” associated with testing

- Employment and or insurability maybe be jeopardized, less likely now due to GINA but life insurance and long term care insurance a worry
- Quality of life during remaining years of normal neurological functioning maybe jeopardized
- If negative result, may question the accuracy of the test, want to be retested. Survivors guilt, no longer “ a member of the club.”
- If gene expanded result, the immediate personal and family burden of **EXPECTING HD** may take its toll on a person’s psychological health and relationships

Common Reasons for seeking genetic testing can be overt and covert

- Life Event Planning
- Personal planning..if it should be positive than avoid being a burden to others
- Long term financial and employment planning
- Finding out for the sake of one's children
- The NOT knowing has outweighed the knowing
- Why wouldn't you want to know?...generational shift

One woman's first response to getting a positive gene test

“ Well, I was just numb. My ears were ringing and everything was kind of far away for a couple of minutes. I was just trying to register the news. It wasn't so much a blow as it was a feeling that something had died...a hope, I guess. Like a death, this awful knowledge cause a period of mourning a emotional volatility...A big chunk of my life had gotten changed and I was ANGRY.”

Three years later this same woman reported:

“The turbulence has lessened in my life. I’m less angry and less depressed. We are planning and making preparations for what lies ahead.”

After the “News”, Now What?

“The world breaks everyone,” wrote Hemmingway. “And afterward, some are strong at the broken places.”

Common Life themes five or more years later

- Changes in practical life
- Emotional responses
- Social relations
- Informing the children
- Thoughts about the inevitable disease

Based on the work of Hagberg et al, “More Appreciation of life or Regretting the test?”, 2011

Changes in practical life

- Often influenced decisions about pursuing a career or may decide different decisions about education, etc.

“I couldn’t get promoted at work. When I got the test results it was like a push to make the best of my life.”

- Those who didn’t have children at the time of testing, often wanted the opportunity to have a child. Those who had a child when tested often had contradictory feelings

“ I got angry at myself for having a child but at the same time I am very, very, very happy that I had a baby.”

EMOTIONAL RESPONSES

- Relief from uncertainty
 - Adapting to test results-often hard to cope for at least the first 2 years but the burden of coping often decreased as the years go on, despite the onset of the disease getting closer
 - GRIEF reactions
 - REGRETTING the test-
Getting the results was much harder than could have been imagined
Now all hope was gone or feels like it has “disappeared”
- ** More appreciation of life-feel an improvement in the quality of life after the adjustment, both at work and during spare time. Life had become more precious!

SOCIAL RELATIONS

- Bringing the family closer together
- No difference in relationships toward the family
- Negative impact-relationship with partners in particular often strained, sometimes linked to anger following testing (by both) and fears of abandonment

INFORMING CHILDREN

- Most folks have informed family, friends and sometimes employers
- For those with children under the age of teenagers, often decide to “hold off”

“It’s hard. It’s so hard. I am thinking I want to tell, I’m thinking I don’t want to tell. How should I say it? What should I say? I know that I was so angry at my mother because she concealed it and it was hushed up.”

- ** Feelings of guilt in passing along the gene. Often a parent will feel that their children’s risk is much harder to cope with, rather than their own gene status.

Thoughts about the inevitable disease

- Knowing gene status can give rise to new uncertainties, and more anxiety about what the future holds.
- Your own experience of affected relatives contributes to anxiety levels and coping strategies
- Use of denial-avoidance behaviors-seeing the “HD doctor” was often a unpleasant reminder about having the gene, also seeing other people with HD was at times quite difficult
- Searching for symptoms “now it’s starting, now it’s coming”
- Planning-many folks decide to test to help with planning, but for a number of folks the planning just doesn’t seem to happen

Some Take Aways- Negative experiences

Regretting the test

Learn ways to shift focus from what has happened to the now

Difficulties in Adapting to a “new status”

Be Kind to yourself; time does seem to help to find “a place for HD” in your life

Seek help for chronic feelings of hopelessness/sadness

Try to have at least one other soul you can talk to about your fears about HD and what may lie ahead

The Impact on the future

It is obvious that knowing that one has the gene for HD can either be a motivator or an obstacle to invest in one’s self.

THAT IS YOUR CHOICE

And some final Take A Ways-Positive Experiences

More appreciation of life-don't lose sight of the present, find the time to enjoy the things that keep life sweet now

In the presence of a major threat (like the HD gene) some people can thrive and adapt-How?

Develop greater compassion and empathy

Find your new strengths(yes, you do have them)

Have some fun (everyday if you can!)

Take care of what you can control (keep pushing your brain and body, get involved in HD research (it just feels good!))

“When you learn to die, you learn to live”

Wayne's story

