Catherine's Story

1. Family History

My family is afflicted with Creutzfeldt-Jakob disease (CJD). Some 20 years ago an uncle died at age 51 from what was assumed to be sporadic CJD. His death, however, was followed years later by the death of another uncle and then, a year later, by the death of an aunt. They were both aged 67. A DNA sample from my aunt was subsequently sent to America, where a specific gene mutation for CJD was identified.

At this time my father, having been in general practice for 39 years, was toying with the notion of retirement. He was extremely concerned about the risk of inheritance for his children and grandchildren, and consequently underwent genetic testing in October 1996. We were shocked to learn that he possessed the mutation, and were devastated when, two months later, he manifested the initial physical symptoms of the disease. His deterioration was swift. In February 1997 he could no longer work, and, after a truly agonizing ordeal, he died in May 1997 shortly after his 67th birthday. Thus, of my father and his seven siblings, half have succumbed to CJD.

Since my paternal grandparents both lived to the age of 93 with no hint of the disease, our genetic mutation is said to have incomplete penetrance.

2. Genetic Testing

2.1 Why test?

The majority of people offered testing for fatal, adult-onset diseases choose not to be tested. I chose to know rather than spend a lifetime wondering anyway. After all, if I hadn't inherited the mutation, there would be no cause for concern. If, on the other hand, I *had* inherited the mutation, some contingency planning would be desirable.

Firstly, retirement age is a major consideration in such planning. If there is the possibility of an early death, I wish to organize my finances accordingly, and retire sooner rather than later.

Secondly, I wish to construct a living will. My father knew death was imminent, but we did not discuss it. It was simply too painful. Consequently, there will always be an element of uncertainty, even guilt, about the decisions we made for him in his final weeks. A living will would spare my family and friends such discomfort.

Finally, I wish to choose a hospice where staff are familiar with the disease and would hopefully be sympathetic to my needs.

Reproductive choices have no place in these plans, since I'm an independent 'career woman' having neither spouse nor children and no plans to change the status quo.

2.2 Testing Procedure

I was not apprehensive about genetic testing, as I had accompanied my parents to Dad's initial consultation and was familiar with the procedure. I had helped Dad research the disease, and knew that my risk of inheriting the mutation was 50/50. I also knew if I did have the mutation, there would be a substantial probability - but not absolute - that I would later develop CJD.

Having read about the significance of dreams and believing in the 'wisdom of the body', I decided to question my body and dream the test result. Although a little far-fetched, I thought it was worth a try, as I had a 50% chance of getting it right! I subsequently had a dream which led me to believe that I did possess the mutation.

Thus, I presented for testing, which included extensive discussion of my probable reactions to either a positive or negative result. This seemed superfluous as my aim was simply to get the result. I didn't see the need for further counselling, and said as much. My opinion was respected, and a blood sample was subsequently taken that day.

2.3 On Getting the Result

I was a little anxious about getting the test result, but eager to know whether my dream was correct. The counsellor confirmed that I had, indeed, inherited the mutation. She also said clients often intuit the result.

I experienced a wonderful sense of euphoria. It seemed as though my life suddenly clicked into place, and all was as it should be. In retrospect, this was perhaps an over-reaction to days of persistent low-level anxiety. However, I was thrilled that my dream was realised, and felt I could trust my body, whatever happened. My inheritance forged a strong bond between Dad and me that, like love, transcends even death. The textbooks say that being at risk can make you feel 'special'. I've always known I'm special, but this confirms just *how* special I am - possibly one in 5-10 million!

2.4 Effects on Family

Of my 4 brothers, one has chosen not to be tested. Of the 3 who have undertaken testing, one brother has tested positive and one negative. The other, concerned about privacy and the stigma of disease, has declined the test result. He has subsequently become a committed vegetarian, and is convinced a healthy diet will reduce the risk of CJD.

Although I think he's living in a fool's paradise, I can appreciate his need to maintain hope and some semblance of control in the face of uncertainty.

3. Follow-Up

It has now been two years since I was tested, and, at age 40, little has changed. My retirement fund is non-existent, but I am more discriminating in my spending. I have discussed my final wishes with family and friends, but there is nothing in writing. I don't obssess about CJD, but am possibly more attuned to my physical and mental condition. If, for example, I experience episodes of unexplained dizziness or increased fortgetfulness, I fleetingly wonder whether this is the beginning of CJD. Common sense, however quickly prevails.

4. Final Words

In **Cannibals, Cows & the CJD Catastrophe** Cooke asserts

"*.....An individual stands more chance of winning lotto than dying of familial CJD".*
Perhaps she's right. For years now I've bought Tattslotto tickets. Although I sometimes fantasize about how I'll spend my winnings, I'm not in the habit of waiting with bated breath for my numbers to come up. I've got better things to do

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