

Huntington's New South Wales



**Presymptomatic Testing -
The Consumer Experience**

Presymptomatic Testing – The Consumer Experience

Suzanne, Michele and Bronwyn told their individual stories at the Fourth National Huntington's Disease Conference held in Brisbane in August 1994.

Irina's story was first printed in "Gateway", Vol 5 No 3, the newsletter of the Australian Huntington's Disease Association (NSW) Inc in May/June 2002.

We are most grateful to them all for sharing their thoughts and experiences with us.

A Peek Into The Future???

Suzanne is married with two children and is a teacher, originally from Queensland, now living in NSW She has decided not to have the test this time.

Today I have been asked to share with you my reasons for not finding out the answer to the question "Do I carry the gene for Huntington's Disease?"

For generations my family has gone about the business of living and dying - some with and some without Huntington's Disease. Having Huntington's Disease has not been a marker of how successful or unsuccessful their lives have been. It has shortened the time for which people have had control over their own lives.

Today science has given us a choice. We can see how the cards are stacked. We can peep into a possible future (barring earthquakes, wars, car accidents, cancer, heart attacks) but we cannot change it and this is probably my greatest reason for not having the genetic test. Forty-eight years have already been lived. I have a good education, I have travelled the world, I have a family I love. If I am a candidate for Huntington's Disease then the time is fast approaching. It cannot be stopped.

The second factor influencing my decision is time. My children (aged 9 and 10) were born just before linkage testing became available. Realising that with Huntington's in the family we could not cope with another problem, I subjected myself to other then available genetic tests (such as for Down's Syndrome). Fortunately they were negative. I said at the time that I believed people with Huntington's mostly had a quality of life, and I was not sure that termination in the event of a positive test for Huntington's was the answer. I am still not sure. Every couple must decide for themselves.

Huntington's Disease is a way of life in our family. I answer my children's questions honestly. They know they might get Huntington's. As yet they do not see the full implications. My husband knew about Huntington's when we were married, and had three years exposure to it. Major life decisions are made with Huntington's in mind.

In our immediate family the next crucial time will be when my children are planning their adult lives. Their choice will be different from mine.

There are many choices for having children already, and there will be evidence of how successful these choices have been.

With the gene now under control many new possibilities will have been explored and more questions answered. If the situation arises that I need to have the genetic test done to help my children, I will willingly submit to them.

While the present exciting discoveries have come a little too late to help me personally, far too late to help my mother and other affected members of my family, I have faith that for my own children and the future generations, Huntington's will be a controlled disease.

Deciding to go ahead with Predictive Testing - Irina's Story

Predictive testing is a new term in the medical field of genetic illnesses. Very simply put it means to genetically test a person, *not yet* presenting any symptoms of the genetic disease, like Huntington's Disease. It is a very different consideration from the genetic testing of a person already displaying a number of the physical and/or behavioural symptoms. In this case the person and his or her doctor are looking to confirm the diagnosis of the presentation of the symptoms.

Some of the reasons why a person would consider predictive testing are:

- the person would like to start a family,
- the person would like to take out an insurance policy without incurring expensive loadings,
- the person would like to apply for a job that would entail long-term high level of responsibility,
- the person would like to eliminate the possibility of having the disease in their immediate family.

In my case it was the last reason. On the whole people are optimistic and even though they prepare themselves for a positive result of having the HD defective gene, most of the above reasons are framed with the assumption that a negative result will happen. If I am gene negative I can go ahead and have children naturally, I can take the highly responsible job, etc.

My family migrated from a communist eastern European country to Australia just after WWII. The person who carried the HD gene in our family died during the war and our family lost touch with their extended family. This meant that we were ignorant of the existence of HD in our family. My mother was seventy and in the advanced stage of Huntington's Disease,

when through genetic testing we discovered that she had the defective gene. The physical symptoms, we now recognise as HD related, did not present until she was in her late fifties, although she suffered very badly all her adult life with behavioural problems, delusions, hysteria, aggression and abusive behaviour.

By the time my mother was diagnosed, I already had children. When I was referred to the genetic testing unit in the hospital and considering predictive testing for myself I presented with no symptoms, especially no behavioural signs. My main reason for going ahead with predictive testing was because I was so different from my mother and perhaps that indicated that I did not have the defective gene and I could clear my family nucleus from the threat of HD. The decision was not made lightly and I spent many very emotional days swaying from one extreme position to the other. I felt extremely vulnerable and making a decision as important as this was impossible. At the same time I was caring for my very ill mother and I could see the symptoms of Huntington's every day. Every day I could see the anger and frustration that she was faced with. She was refusing to take medication that would lessen the involuntary movements and as we later discovered, the severity of the delusions and other behavioural problems. This was not a good time for me to be making a decision about predictive testing.

Family members were very supportive and were saying all the 'right things'. My husband said that it was not possible for me to have the defective gene. He stressed that even if I had the defective gene, that I was not the same person as my mother and the disease would not manifest itself in the same way. Of course we were concerned with the physical symptoms, but having watched my mother deteriorate over the years I feared the possibility of displaying the psychological symptoms. My husband was the main person who I looked to for advice and comfort. After all he had a genetic stake in this too - we had a desire to clear three primary school age children and two daughters of childbearing age of HD. It was with a very heavy

heart that I agreed to have the genetic test.

The months that followed while waiting for the result were long and arduous. I continued to oscillate between being sure that I was not gene positive and sure that I was. I was very emotional and could not think of anything else. I thought that I could cope with the result, especially the one that I did not want. I would be able to summon my strength and 'deal' with the news somehow. Nothing could have prepared me for the level of emotional grief that followed the gene positive result. I knew what all the issues were prior to receiving the result, but they were so much more relevant and immediate now. Within a month of receiving my result I booked myself in for psychological counseling sessions and it took three goes before I found the right counselor for me. Over the last two years my counselor's support has been wonderful and it meant that I could discuss with her all the issues that my husband or close friends felt uncomfortable and unable to discuss with me. Even with the help of a counselor I went through a very long period of intense grief that I would compare to the grief that follows the death of one's child.

The issues engulfed my thoughts and would not let go. My thoughts were morbid and I was self-engrossed. What would be the time of onset of the illness and how would my family cope? Would I live too long and experience the frustration, the loss of autonomy, the disfigurement and loss of cognitive function that my mother was now experiencing? Would I become a person that my family would find 'difficult'? Would my children interact with me in a different way? Would they resent me for loading them with all the responsibility of caring for me and for siblings? How would they cope with the fifty-fifty chance of becoming ill? How would they be able to cope with the possibility of not being able to have children naturally and without deep consideration of the possible consequences? There are alternatives available, such as PGD, or the donation of egg or sperm and implantation through the IVF program, or the decision to have children despite the risk of HD. How would I be able to tell them about the genetic illness in our family in a way that they would

be able to understand and yet not affect their love of life and sense of fun? Would they see themselves, as I now saw myself, as damaged goods and not worthy of love?

The positive result not only changed the way I saw myself, but also the way I saw my relationship with my children, my friends and my husband. I was engrossed with grief, but I could not discuss any of my issues with them, because they seemed unable to confront these issues for themselves and they were not ready or needing to do that. I was alone in my despair and I felt isolated by it. I isolated myself to enable me to think and grieve without hearing consolations from others that were not appropriate or trivialised my emotions. I needed to feel that my feelings were validated, and I found that I was the only person who could do that. My temporary isolation was for my protection.

My counselor gave me very good advice: to put aside, visually in a drawer, all the issues that were too painful or too difficult, and that we would bring them out one by one at a later stage. I took her advice, but the grief and looking after my very ill mother became too great a burden for me and I became very ill. Although I was in a lot of pain, my illness became a welcomed physical and emotional release from my constant obligation of caring for my mother. Somehow I managed to get my mother into a nursing home and I was able to address my ill health.

I have come through my grief and I am well again and my mother is well settled in her nursing home. I have learnt a great deal during this time. I have learnt that it is highly likely that most people going through predictive testing will go through similar levels of grief. I think that there is no good time to go through predictive testing. Most people having predictive testing would have someone around them in their family going through the more serious stages of the disease. It is not appropriate for them to try and deal with the emotions of seeing a family member deteriorate and try to deal with the possible result that

they will also become ill. The human psyche is not made to withstand the news that they are ill with a disease that, at the moment, has no cure.

If you think back to what was developmentally the most turbulent time of your life, most people will say that it was their teenage years. This is because the person is developing into an adult and questioning all around them and also developing their own intrinsic personality. This is what also happens to a person who has tested gene positive for HD. It is then that you need to arrive at a new way of seeing yourself. A new persona living with HD. This took a great deal of thinking work on my part and with the help of a counselor (she is an oncology counselor) to arrive at a point in my life where I can feel that I can live with the disease and show my children by example how to overcome each phase and personal dilemma that the disease may present.

Our lives are based on a positive and optimistic basis to our future and our future generations. If we take the basis for optimism out of our lives then we impose a frame of mind on ourselves that is completely against our nature. How are we to cope with the devastating news of being gene positive with a disease that as yet has no cure? We are not made to deal with such devastation. We are at our most vulnerable when deciding whether to have predictive testing and are very likely to ask advice from a good friend, a partner, a genetic counselor, or a doctor. These people have the opportunity to encourage the person to protect their right 'not to know' and should at the very least encourage postponement. We all have a right to live our lives to the full and not know how we will become very ill with a degenerative neurological disease and die. We have an inherent right to be optimistic about ourselves, or in the case of Huntington's family members, at least to retain and preserve a very precious iota of optimism.

A Family Perspective - A Negative Result

Michele is 33 and one of four children. She works as an Information Manager for one of Queensland's largest private companies and is expecting her first child in eight weeks. She has had a negative result.

As someone who has had a negative result from presymptomatic testing for Huntington's, I'm speaking to you with great humility. God's wishes, fate, luck, the predetermination of our lives that chance moment on conception that should mean I do not carry the gene while other members of my family do is a powerful consideration. I have wondered long and hard about the implications of this.

We have known that my mother has had Huntington's for about 15 years. When Mum was first diagnosed (and consequently my grandmother's illness - thought to be Parkinson's - was also diagnosed) a medical practitioner sat in front of me, my sister and two brothers and told us we could never have children - end of discussion! I guess I'm proof today that people can be very wrong!

We were then aged from 16 to 22 years and I must say it was a fairly heartless and quite devastating way to give us the news that we had an inheritable disease.

Mum experienced a dramatic decline about four years ago and could no longer be by herself during the day. My father resigned his position to look after her full time. My mother has a wonderful attitude to life and even though at 59 years of age she appears to be a weak and frail 80 year old, she continues to face each day with humour and grace. It has been my parents' matter-of-fact approach to Huntington's that has meant the disease has not been an overwhelming focus for our lives. It is a part of our lives and accepted and discussed openly.

I describe our family situation so that you realise that we, my

brothers and sister and I, have grown to adulthood in an environment of open discussion, humour and positiveness about Huntington's and the ramifications for all of us and our extended family.

I always felt that I would take the test - if I had the chance to know part of my future I could plan for it. And I always believed that if I could do something to stop the progression of Huntington's that I would do it. The testing procedure is part of that.

My brothers each have a son - they undertook prenatal testing before the presymptomatic testing was available. I would have done the same but circumstances meant that our timing was right when the presymptomatic testing became available (in Queensland) last year .

As we were part of the first group, the testing period took about nine months. I was certain that I wanted to know and initially felt that we could go straight to the result. However, as you know, that is not the way of it, and I must say the counselling period was very valuable. We also appreciated the knowledge and often insight into our characters we gained during this period.

My husband and I believed we had prepared ourselves for a positive result - with our eyes open to the type of feelings we could expect. We had many things to look forward to and could put our knowledge to good use. In particular I knew that I would be no different after receiving a positive result than I was for the preceding 32 years. What would change was my knowledge - a glimpse of the future that few of us gain in our lives.

I believe that the testing procedure is invaluable - it provided a future for our family where there was none.

My husband and I were shocked, thrilled and excited as the wonderful implications of receiving a negative result hit us:

- We could immediately start our family (which we did!) and

have as many children as we choose

- I can work for as long as I wish in the future
- We perhaps don't have to worry so much about future financial implications
- Many aspects of life, like obtaining life insurance, will be easier .

These and more things continued to surface through our initial disbelief.

Our families and friends were very pleased for us also. It seems that while we did not discuss the testing procedure much outside our family, our close friends were very aware of our situation.

Every time I think of our result, my excitement is always balanced, and checked by a very deep concern for my sister and brothers. I felt that very strongly when I told them and my parents of our result. I could not stop thinking about them for the first weeks (and still don't) worrying that they would be positive and desperately hoping that they, too, would be negative.

I know we are all innocent of our respective outcomes but I do feel some guilt. Here I was telling them that I don't have it - and despite the fact that we know it is a 50% chance for each individual - they must have all felt that their own chances of a negative result had diminished.

The relief that I feel to not carry the gene is always tempered with the knowledge that this may not be the case for the rest of my family. We are one unit and what affects them affects me.

However with the knowledge that I am free of Huntington's I have come to realise that I will be in a strong position to help my family in the future should they carry the gene.

I'm sure the impact of receiving a result is different for

everyone. For me the negative result, while wonderful, is also sobering and I often wonder that, if it had been positive, if I would have been as accepting as anticipated.

Taking A Positive Approach

Bronwyn is married with one child and is a teacher from NSW. She has had a positive result.

I am 37 years old and in February 1994 I received a positive HD result.

My initial motivation for having the test came from a desire to have more children, hopefully on our own. Brendon and I have a beautiful little daughter, Sophie, now 2 years old, who was an IVF donor egg baby, so she has no risk of inheriting my HD gene.

The procedure for the HD testing began with a series of interviews with Professor Sillence and our longtime social worker, Fiona Richards. During these interviews they were able to ascertain Brendon's and my knowledge and both our abilities to cope with a positive or negative result. The process, whilst very professional, was caring and supportive. I was presented with some lengthy questionnaires - one rather tricky question with a tick the box selection, posed a problem for me. It was with regard to our marriage and how things were going, and the tick-a-box selection seemed to range from perfect to poor but didn't have one for 'fairly normal with the usual ups and downs', or even 'ask me tomorrow'.

The assessment process included a physical check-up with Dr McCusker who informed me she couldn't see any signs - Fiona told me I was the stillest person in the room. Secondly there was a psychological assessment which I regarded with an attitude that, no matter how I went, I had nothing to lose, as it may serve in future years as a benchmark for comparison. Just as well as I collapsed in giggles when it became a little too challenging.

During the few months before my result, I wrote down a few thoughts to try to clear my anxious mind and give myself some

direction for coping. They naturally fell under positive and negative headings.

If I received a positive result then we would go on, as we had, trusting and relying in the researchers and their dedication to unlocking the mystery of the repetitive pattern in the gene and its effect. We're well informed that they have cloned the gene into mice, and the various groups are researching into different areas and continue to share their knowledge and advances. I would continue to keep up to date through the Association newsletter. And our IVF options are still there.

If it's a negative then what can I do for my brothers. I would feel very heavy if I was the only one to escape HD. We would be able to go ahead with our family plans. Continuing support for the Association.

The following day Bren and I went for the result.

My elder brother was able to join us at the Children's Hospital. Travelling there I realised that my feelings of incredible frustration and helplessness came from the fact that this was one of the few situations in my life where I was totally unable to influence the outcome. No action, or energy would help.

The result was positive.

While taking this on board we asked questions about research, medication and how we could be involved.

We decided to have a coffee and sit in the garden playground at the hospital. This was the greatest leveller of all times - seeing so many poor kids who hadn't even been given a decent start - and I'm 37. This gave us a much wider perspective.

I phoned my other brother and then we three sat and chatted, which really helped to give a little time and space to absorb the implications of the result. Our dad passed away last year so I

felt grateful that he hadn't had to cope with this.

Later that day when we arrived home on the Central Coast I wrote down an expression of hope and trust in the dedication of our researchers and phoned my extended family and friends. I hoped to show them that I was coping, approachable and taking it in my stride and I was buoyed by their concern and loving support. I believe knowledge leads to understanding and hoped that this was laying some seeds for the future, when symptoms appear, as I don't want my family, friends or neighbours shying away from me as I love companionship and conversation.

That night Bren and I talked a lot, and were glad that we had chosen IVF - it was the right choice for us - in having Sophie. My mixed feelings about having more faded with time and we're continuing with the donor egg program through IVF. I hoped that I'd be the only one and that my brothers would be negative.

Bren and I then focussed on how we could help ourselves, at home, in our lives and in reducing any impact on our children and lessen sudden changes. So we began a list for adjustments which we would make gradually before I became affected. We'd buy a pop up caravan for weekends and holidays.

Our List

- Finishing those little nagging things around home
- Non-slip mats
- Level block of land for our home
- Soft furniture
- Lid for the bath
- Kitchen carpet
- Fence the front yard, a safe yard
- No sharp edges
- Elastic, rather than buttons or zips
- Routine for running the house

- Partner learning to cook meals now, that can later be blended: casseroles, bolognaise
- Talking to friends and neighbours so they will understand
- Ironing lady, occasional cleaner (later on)
- Photos up to date
- Take pleasure in the little things
- Clothes that fold or dry on hangers
- Resolve those nagging differences
- Enjoy the romance
- Make things - being busy and satisfying sense of being present and productive.

Within a couple of days Fiona, our social worker, came to visit us at home. I was able to ask more questions about those little things that had occurred to me. Fiona has the greatest knowledge and understanding of what HD is and how it affects families and she was a great support. I felt buoyed by an incredible sense of relief when Fiona reminded me of her follow-up visits, and suddenly I began to have a greater sense of future, and less of loss. We feel very fortunate to have such a devoted professional and caring person, Fiona Richards.

With hindsight Bren and I began to realise that we had come to terms with HD and its implications for us, some years ago. Now we were doing our best to take the negative result on board as positively as we could.

Some three to four months after my result, my hairdresser explained to me that my increased loss of hair was possibly due to the effect of some stressful situation some three to four months prior. She's another who understands my situation.

Straight from the shoulder - the result scares the hell out of me, but I'll do my best to cope.

I draw strength from the spirit my Mum had, even through her frustrations and disability, her quiet strength, dignity and loving trust in our devoted Dad, and our family support.

This is a spirit which I see in other affected HD people, an amazing strength to cope and make the most with their situation.

At the previous conference in 1991, in Sydney, when listening to Dr Susan Folstein, I was overwhelmed by the brilliant knowledge, skill and dedication of the research doctors and teams who have since identified the gene and have now turned their untiring quest toward managing and hopefully containing its effect. I'd like to express our high regard for them and our heartfelt appreciation for all that they do.

Our other tremendous support over the years has come through our HD Association, with its level, unbiased approach and its caring, always helpful presence and devoted family members and friends.

I feel it's appropriate too, to mention the quiet presence of a large body of family members who, at home, eagerly await news of medical breakthroughs, and rely on newsletters for updates in care and advances in research.

Hopefully our generation will be the last to know the real impact of HD. Thank you to all who help our families. It is indeed better to light a candle.

Fact Sheets

- Huntington's Disease in the Family – A Booklet for Young Children
- Huntington's Disease – A Fact Sheet for Teenagers
- Talking to Children about Huntington's Disease
- Presymptomatic Testing – The Consumer Experience
- Understanding Challenging Behaviour in Huntington's Disease
- Living with Someone who has Huntington Disease
- Making a Decision About Residential Care
- Guidelines for Meeting the Nutritional Needs of People with Huntington's Disease
- Eating and Swallowing Difficulties in Huntington Disease
- The Importance of Dental Care in Huntington Disease
- Communication and Huntington Disease

*These Fact Sheets are available from Huntington's NSW
Please see the back page for contact details.*

Helpful Contacts

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For further information on Huntington Disease, a membership form, or details of HD services in NSW, please contact Huntington's NSW