

Personal Perspectives on Genetic Testing for Huntington Disease

A collection of stories
compiled and edited by
Susan M. Cox, Ph.D.

Reproduction of any or all sections of this booklet is strictly prohibited without the express permission of the Huntington Society of Canada.

Part II of this booklet is based on material from Dr. Cox's doctoral dissertation, "*It's Not a Secret But ...*" *Predictive Testing and Patterns of Communication about Genetic Information in Families at Risk for Huntington Disease* (The University of British Columbia, 1999)
copyright © Susan M. Cox.

Design and Layout: Real World Graphic Design

Table of Contents

Foreword

Introduction

Part I: Do I want to know?

Overview

No need to know

Who I am at this time

I'd rather wait

Being a coward has nothing to do with this

Now I find myself wanting to know

Part II: The process of
predictive testing: before,
during and after

Overview

When your load is lightened

Why walk on egg shells?

Allow your family to help

A task to deal with

A new direction

Part III: Now that I know...

Overview

I won't be ambushed

Uncertainty

Alone

Sometimes I feel cheated

Never give up hoping

Reflections

Appendix 1: Genetic Testing

Resources



Personal Perspectives on Genetic Testing for Huntington Disease

Foreword

This is an important collection of personal accounts of predictive testing for Huntington disease. It is not always easy reading, because it is frank and realistic in its depiction of the challenging questions and difficult feelings associated with predictive testing. Do I want to know my future? How could I accept knowing that I will end up just like my father? Will my husband leave me? What about my children? Am I able to live with more fear and loss?

At the same time, it leaves an indelible sense of the courage that characterizes individuals and families affected by Huntington disease, whether or not they elect to pursue testing – the determination to be in control of their own lives, the role of family and faith, and the process of coming to peace with the results, positive or negative.

We are very grateful to Dr. Susan Cox of the University of British Columbia, who has assembled and edited this publication with such skill and insight (and in so doing has provided an outstanding companion piece to another Huntington Society of Canada booklet, *Genetic Testing for Huntington Disease*). Many of the accounts in this collection emerged from research conducted by Dr. Cox, in collaboration with Dr. William McKellin. Thanks are due as well to Jessica Easton, whose thoughtful reflections serve as the conclusion to this booklet.

Above all, we are indebted to all those members of the Huntington's community who have so generously elected to share their thoughts and feelings. By doing so, they have extended a supportive hand to all those who have an interest in the human implications of new genetic technologies.

Huntington Society of Canada
2002

Introduction

The decision about whether or not to undergo predictive testing is not easy. For some, the test provides desired information about the future. For others, such information seems unnecessary or undesirable. It is, in either case, a very personal decision.

It was for many years impossible for persons with a family history of HD to know in advance if they would develop the disease. With the 1983 discovery of markers linked to the gene causing HD, a presymptomatic test was developed. In 1986, centres in Canada, the U.S. and U.K. began to offer this test. With the 1993 discovery of the genetic mutation causing HD, a “direct” test was developed. This test permits persons at risk for HD to learn, with near 100% certainty, whether or not they will develop the disease later in life. The same test may also be used to confirm a diagnosis of HD in persons who are already showing symptoms. Thus far, the majority of persons at risk for HD, have decided not to proceed with the test. This decision is an equally valid, thoughtful and courageous decision.

The personal accounts in this booklet offer a range of perspectives on the experience of predictive testing. Some are from persons who are *considering* whether or not they wish to have the genetic test. Other stories are from persons who are in the process of *undergoing* testing or *reflecting* upon what the experience was like for them. Although identifying details have been removed, each person is the author of their own story in that they are telling, in their own words and from their own point of view, what the experience was like for them. Family members may have different perspectives on the experience and, though their stories do not appear here, it is important to acknowledge that the process of predictive testing has many significant implications for the spouse/partners, children, siblings, parents and other family of the person being tested.

The word “stories” seems a modest way of describing the personal accounts that appear in this collection. Stories are, however, a vital part of life. They allow us to convey our personal experiences to others and, at the same time, they help us to make sense of our experiences. Stories also preserve family history, foster new awareness and help to build community.

This collection of stories about the experience of predictive testing for Huntington disease has grown out of the Huntington’s community. Many people contributed by sharing their experiences in either written or oral form. Others, such as Ralph Walker, Rod Morrison and Shawn Mitchell played an important role in recognizing the need for such a collection and in nurturing it through production.

Stories are a vital part of life. They allow us to convey our personal experiences to others and, at the same time, they help us to make sense of our experiences. Stories also preserve family history, foster new awareness and help to build community.



The stories appearing in this collection were selected from several different sources. Those that appear in **Part I** were, with one exception, submitted in response to a request for personal perspectives on genetic testing that appeared in the Fall 1999 issue of *Horizon*, the national newsletter of the Huntington Society of Canada. The stories in **Part II** derive from a series of in-depth interviews that I conducted with persons who were going through the process of predictive testing in British Columbia in the mid 1990's. The stories in **Part III** were adopted from a collection titled "Personal Experiences with Predictive Testing in Huntington's Disease" compiled by the Southern Alberta Chapter of the Huntington Society in 1996. There were more submissions than could possibly be accommodated in this collection and it was a difficult task indeed to winnow them down to a cross-section of 15 stories that would speak to many, if not all, of the most important implications of predictive testing.

Some of the stories that follow are not easy to read. They confront tough issues head on. Others present a point of view that may be difficult to understand or relate to. Yet others may call up familiar feelings or painful memories. Some offer guidance and advice, others pose unanswerable questions and a few incorporate humour as an antidote to what might otherwise be overwhelming. All, however, contribute with great insight and clarity to our collective understanding of the experience of predictive testing.

I would like to thank Jessica Easton, a psychotherapist who is conducting a qualitative study of adolescent's experiences of HD, for her thoughtful reflections on the collection and Sandra Funk, Sandra Russell and Susan Tolley (Huntington Society of Canada HD Resource Centre Directors), for their careful reading of the manuscript and practical suggestions.

In closing, I would be very pleased to hear from readers near and far about their impressions of this booklet. I am honoured to have been invited to compile and edit the collection and grateful for the opportunity to continue my involvement with the Huntington's community.

Susan M. Cox, Research Associate
Centre for Applied Ethics
The University of British Columbia
suecox@interchange.ubc.ca

Some of the stories that follow are not easy to read. They confront tough issues head on.

Part I

Do I Want To Know?

The five stories that appear in **Part I** are written from the perspective of persons who are considering having predictive testing. Four were submitted in response to a request for personal perspectives on genetic testing that appeared in the Fall 1999 issue of *Horizon*, the national newsletter of the Huntington Society of Canada. A fifth story was selected from a collection compiled by the Southern Alberta Chapter of the Huntington Society of Canada.

As the first and fourth stories suggest, predictive testing is not for everyone. Although it is sometimes assumed that the majority of persons at risk for HD have had predictive testing, it is only about 10% of persons at risk who have actually decided to have the test. It is also helpful to recognize, as the second and fifth stories do, that there is no time limit on the decision about whether or not to have predictive testing. The experience of being at risk for HD may change throughout someone's lifetime. Consequently, it sometimes happens that persons who do not initially want to have predictive testing later change their minds. The openness of the decision is also reflected in how predictive testing is offered. Canadian genetic testing centres provide pre-test counselling and support to ensure that the decision is well-informed and appropriate for the person considering having the test. Further, as the third story in this collection illustrates, it is always possible to stop the process and not obtain the test results if the person decides that they would rather not know.

Predictive testing is not for everyone. Although it is sometimes assumed that the majority of persons at risk for HD have had predictive testing, it is only about 10% of persons at risk who have actually decided to have the test.

“We felt and still feel no need to know what the future may hold.”

No need to know

I never considered predictive testing was for myself or my family. Yes, Huntington's is alive and well in my family history — that is, my grandfather, two great-aunts, one great-uncle, my father and my aunt — but fortunately the debilitating effects of the disease do not appear until approximately age 55 or later. At the initial availability of predictive testing, I was already married and had two small children. We felt and still feel no need to know what the future may hold. I cannot imagine trading a life with kids for one without.

I only pray that before my father dies, before myself or my brother, or before my children or my niece and nephew are diagnosed, that there will be a control or cure for Huntington's. I cannot think that either knowing I have or have not inherited the gene for Huntington's would change the way we live and are raising our children. My father and mother do not have the quality of life one would like to think a retired couple in their late 60's deserve, however, they still enjoy following their grandchildren's activities, church fellowship, curling games and the like. Huntington's is not the only debilitating condition one can suffer with. Perhaps, as my children become adults, they may feel the need for predictive testing and more definite information. Perhaps, a cure is on the horizon.

Who I am at this time

My maternal grandmother, at approximately age 65, started having a limited amount of chorea and my grandfather spoke of her personality shifting. I was an adolescent at the time and she was just old and Grandma.

At age 18, shortly after giving a male child up for adoption, (I felt strongly a child needed two parents) my mother and siblings were asked to meet with a geneticist. It was impossible to confirm (at that time HD could only be confirmed at death) but Grandma was suspected of having Huntington's. How did we feel about it? My response — well she raised her family, had a good life, we don't even know if it is HD, much less if Mom has the gene, I'm not going to let the possibility of HD rob me of my life.

I married at 20. Our first child, a boy, was born three years later. Grandma died and, yes, she did have HD. Feelings — she raised her family, had a good life, we don't know if Mom has the gene. *Avoidance.* Another child, another male arrived, life went on. Mother could no longer sit still — well she raised her family, she's into her 60's. (When you are not yet 30, 60 plus seems old.) There is no reason to panic here we don't know Mom has the gene. *Hiding my head in the sand.*

Time passes, Mother moves a lot more, another consultation with the geneticist, 'Do you think your mother has HD?' He basically confirms all our fears about Mother having HD but doesn't tell her. My feelings — well I've already had my children, I can't send them back, I'll just hope I don't have the gene. *Anger, resentment, unfairness of life.*

Blessings of blessings, a child, a daughter is born. She was not asked or longed for (at least not consciously) but she completes our family. A marker gene is discovered — they can tell you if you have the gene or not — 90% accurate. Do you want to take the test? My response — what would it prove? Am I going to live my life differently if I know? I don't want to know — it is too frightening? *Fear of the unknown.*

New and improved predictive testing that is 99.99% accurate is available. Do I want to be tested? Why should I? The HD strain in our family is later onset, a person has the opportunity to raise a family, live a life, do things, see things. Look at Mom she's still going strong, HD hasn't slowed her down much. Do we have the right to look into the crystal ball of our future?

The Huntington Society asks me to be a delegate at the National Conference in Vancouver in 1994. While there I learn that the HD gene behaves differently in males than it does in females. A male carrier will have the same age onset as his HD mother BUT his children may be all over the board. HORROR — I DIDN'T KNOW THAT. IF I HAVE THE GENE, WHAT HAVE I DONE TO MY SONS? GOD HELP ME. WHAT DO I DO NOW?

My father is diagnosed with terminal cancer, life gets chaotic, my mother's stress accelerates her HD. Too much to think about, too much to do. A year goes by — Father dies, Mother stops living. Another year and Mother, still in her own home with live-in care, is trying to learn to live with an ever decreasing amount of control over her own body. It hurts. It breaks my heart. How can life be so unfair? I can no longer deal with it all. I stay away. I can no longer bear to see my mother. Is this how I am going to end up? Who will be there for me? Will there be anyone? Will I develop HD and slowly lose control over my body and/or mind? I want to retreat. No, I don't want to talk about Huntington's, I want it to go away. What might I have done to my children? *Guilt, fear of the unknown, anger, resentment.*

Tired, stressed out, the negatives weigh heavy. Stress feeds on stress and one sinks. Another day, the sun shines and life isn't so bleak. I attend a Huntington Society meeting and receive encouragement in that others are present. (I am not alone.) My doctor hears from me after three years and gives a word of encouragement — if a child has the gene, a cure is so near now that their chance of a full,

“If I have the gene, what have I done to my sons? God help me. What do I do now?”

**‘I am the
Lord, your
God, who
takes hold
of your right
hand and
says to you,
Do not fear;
I will help
you.’**

Isaiah 41:13

healthy life with children is good. (There is hope.) I do visit Mother and a sister tells me how much Mother enjoyed our visit. (That cheers me.) I pray. I attend prayer times. My Heavenly Father, my Saviour, the Lord Jesus Christ, my companion, the Holy Spirit, friends and fellow Christians, they all minister to me.

The Word, a double edged sword slices through the darkness to reveal the light, the truth:

‘Perfect love drives out fear’ 1 John 4:18.

‘I am the Lord, your God, who takes hold of your right hand and says to you, Do not fear; I will help you.’ Isaiah 41:13

‘He who dwells in the shelter of the Most High will rest in the shadow of the Almighty.’ Psalm 91:1

‘I am God’s child for I am born again of the incorruptible seed of the Word of God which lives and abides forever.’ 1 Peter 1:23

‘I am raised up with Christ and seated in heavenly places.’ Col 2:12

‘I am complete in Christ.’ Col 2:10

When I keep my eyes on Jesus, my attention on others, my thoughts above all else fades and ‘I have the peace of God which passes understanding’ Phil 4:7. Because I know that nothing, not illness, not loss of control over my body or my mind, not Huntington’s Disease, not even death can separate me from the love of God that is in Christ Jesus my Lord. Romans 8:39 (my paraphrase).

I am at a crossroads. Knowing what I know about how the gene behaves in the male sperm and considering I have three sons, do I go through predictive testing to spare them the fear, the dread, the unknown? Knowing what I know about my life with God, I don’t need to know for myself. As fortune telling is prohibited in the scriptures I am repulsed by the thought. And yet? Do I owe it to my children to spare them or is it just my guilt being displayed? I believe it is my duty as a mother to do what I can for my children. I have a 50/50 chance of having a gene that, if passed to any/all my children, will affect their lives, some more immediately than others as they may be considering parenthood now or in the near future. Alternately, if I do nothing and a child steps forward for testing and receives a positive test — will I be in a position to support that child when his revelation will diagnose myself? I think not.

Living at risk of developing an incurable disease. That is who I am at this time. Am I willing to risk predictive testing that I may possibly spare those I love the most from this one horror of the unknown? Yes. Will it make a difference? It will to me.

I'd rather wait

Mom had HD. She passed away in 1985, at age 69. I knew something was wrong with her in her 40's. Her mother passed away earlier from HD. I didn't become aware it could be passed on to me until the late 1960's. My wife and I decided to have children in spite of being at risk myself. I reasoned that my mother had a reasonably good life until her late 40's, so I should have similar such chances, as well as my children. Also, research may have found a 'cure' before my children became affected.

My parents had five children. The oldest was diagnosed in the early 1990's with HD. The youngest was diagnosed in the late 1980's.

I have taken all of the predictive testing, except I have not asked for the results. The reason is because my wife does not want to know the results as she feels it would adversely affect our future. I would like to find out the results prior to my children having children, for their benefit as well as my own piece of mind.

The medical staff advised the testing could be terminated at my/our request at any time. I'm more at ease not knowing the results for myself as each year passes by, but I'd still like to know before I pass on, for my children's sake. The medical staff were very considerate of my feelings.

I can't see any good in telling my employer that I'm at risk at this time. If I knew that I was not at risk, I would be more open at work about Huntington's, but for now I keep a low key approach at the office. (This hurts Amaryliss sales.)

My wife and I had a will made up 21 years ago before our children were born. We are making changes to the will to reflect the fact that our children are now young adults and it requires some alterations. Our financial planner has agreed to act as executor in the event that both my wife and I suddenly perish together. At this time we both have signed our driver's license to donate useful organs upon our death. We will ask a lawyer about power of attorney requirements.

We are considering the purchase of a new house in the next couple of years, a bungalow with a walkout basement. I have thought about wheelchair access somewhat but have not seriously discussed it with my wife at this time.

As I'm three years away from being eligible for early retirement, my main goal or objective is to keep working for my employer until that time — to protect my wife and family for our later years. Work beyond age 55 depends, obviously, on my health (which is pretty good right now), my enjoyment of working in this pressure packed work environment, and our financial needs for the future (assuming the company still wants/requires my services).

“If I knew that I was not at risk, I would be more open at work about Huntington's, but for now I keep a low key approach at the office.”

Being a coward has nothing to do with this

This is about my experiences living with Huntington's disease and why I do not want predictive testing.

My father had Huntington's disease. I figure he started showing signs when I was 11 years old. He was diagnosed 10 years ago and has been dead for four years.

HD to me is cruel and unsympathetic to both the victim and the family, and therefore is very much a family disease in every sense of the word. It is a very difficult thing to stand by and watch a strong, independent, proud man lose everything. He lost his ability to drive, work, handle his money. He did not care how he looked, he aged rapidly, had trouble swallowing and speaking to a point, and lost his balance easily. This wrenching disease is like watching a full grown adult revert to childhood.

Before my father was diagnosed, I just thought he was a hateful, abusive man and I hated him. I often wished he was dead. I used to get so frustrated with my father because nothing I did was right or good enough in his eyes. I got to the point where I didn't even care. You get totally lost in HD. The disease takes a front seat to everything else. HD does not get worked into your life, your life gets worked around it!

I hated school but it was a place to go where he wasn't. My friends never knew I had a father and I never made any effort to let them know differently. I remember in Grade 9, my first year of high school, going to gym class and having bruises that I never explained — but I could see the other kids staring — I always tried to ignore it. The physical and mental abuse of the disease is absolutely unbelievable. Looking back now, if today's laws had been in effect then, he would have been in jail.

There is one incident that happened in my early teens that I'll never forget. My father had no concept of time, if it wasn't for himself, and he always kept us waiting. If anyone told him to hurry up we were going to be late, he got angry and started a fight and this took even more time. One of these times, we were going to my grandparents and we got there two hours later than we could have. My grandfather asked me, "How come you people are always so late going anywhere?" I was 13 years old. How do I explain it, especially when you are told, "What happens in this house stays in this house."

People would never have believed us if we told them about my father and his ways. He was Mr. Nice Guy in front of people and a paranoid abusive person in the confines of his own home.

After my father died, everyone outside the household would say what a good man he was and what a good father. Bull! How do you tell

people this quiet, gentle, kind man they knew was not the same violent, abusive man you lived with.

The outside world had such a different picture of my dad, and that was great! We've probably made this image possible by not telling about the monster that lived in our house. We never painted the gloom and doom that lived within, on the outside walls. But let me tell you, if the inside walls could talk... People would stand back shocked to hear the tales of 10-15 years of living hell.

The first 10 years of my life I scarcely remember, but the next 10 years I remember vividly being yelled at, kicked at, blamed, suspected and not trusted. Finally, a diagnosis of HD was given and I realized that all of this happened because of an illness. Now all of a sudden magically, we must forgive and forget. In the last 10 years some types of medications and help were available and this HD victim that we lived with would not accept them because he thought someone was out to get him.

“On March 22, 1993, it was great news to hear they found the gene that causes HD. The implications of this are great, but for me predictive testing is something I really don’t want. I really don’t worry about getting HD.”

While living with HD my time was not my own. When not working, the whole day was spent running with and for Dad. As his short term memory failed, much time was spent constantly repeating and explaining simple things over and over. As a result of this, I can be quite selfish about my time, and am very impatient when doing things. I get extremely frustrated when people interfere with what I am doing, especially on the job. While I was helping to look after my father, HD had control over the events and happenings in my life, and there was absolutely nothing I could do about this. But now I'll be damned if I'll allow HD to control and dictate my life today.

On March 22, 1993, it was great news to hear they found the gene that causes HD. The implications of this are great, but for me predictive testing is something I really don't want. I really don't worry about getting HD. I also don't want to know if or when I could develop this disease. There's no cure for HD so why sit and wait for my hands to shake?

Because HD has no effective treatment or cure, this is one reason I do not want children. Due to this decision, I have run into two separate occasions where two different professionals made predictive testing an issue. They thought testing should be done before they would agree to my decision. Because I did not agree with this, one comment was, "Why not, are you a coward?" I was so angry I walked out of this specialist's office. Being a coward has nothing to do with this. I'm now in control of my life, and I will decide if I want to know or not know. Maybe someday this will change, but for now I'm the one in charge of making my own decisions.

My greatest fear is not HD itself, but the fact that my doctor will not know anymore about HD if I should get it than he did when my father had it.

I recently heard a most appropriate comment that sums up predictive testing in my mind, "If genetic testing becomes the basis for daily decisions, it will become a new form of prejudice." I personally feel we cannot allow this to happen.

Years ago, I remember being so angry at my mom for going to all the places my dad frequented and informing these places that he had HD. But it really did help because in the end it made these people more understanding and patient towards him, taking some of the burden off of us and allowing him some independence.

In closing, the one thing my life's experiences have taught me is not to keep this disease quiet, because when people become more knowledgeable about illnesses, they become more sensitive.

Now I find myself wanting to know

I got married fairly young, around 20 years. At that time no testing was available. My husband-to-be was completely informed on Huntington's disease, all that we knew at that time. There were no secrets. He could see all the full effects in my mother. At that time my mother was quite advanced and exhibited all the symptoms of HD.

My husband, the special man that he is, has great insight and even with the twisting movements, and ever changing moods and behavior, he could see a flicker of my mother's beautiful character. She was a special lady and my husband had, and still holds, high regards toward her. She passed away in 1987. We still remember her.

This same man told my parents, as we sat together discussing wedding plans and HD, if we have 20 good years, it will be worth marrying your daughter. If we have 10 good years, it will be worth it. Boy! How proud I was of him to be so courageous in the face of such a horrible disease. This gave me courage and strength to be able to live our life together, no matter what.

At that time, in my mind, HD was far away. It seemed it would take an eternity to ever show up. I felt that I could have my family and raise my children without ever having any signs of HD.

Nevertheless, we talked about having children, because we felt it was an important part of our life together. We tried to plan them early in our marriage. This was to enable myself to raise my children without too much threat from HD. One of the reasons I chose to get married and have children is because I believe in hope and quality of life. For me a life without children was not complete. I had hopes for a better life for myself and for my children.

Besides, there were no guarantees that I was absolutely going to have HD or that my children would inherit the gene. I had a 50% chance. I took a chance for the better. If the worse came, I still prepared myself in the event HD would show up.

Soon after the birth of my second daughter, I gave my life to Jesus. I was born again, and this made a big difference in my life. Jesus gave me a new hope and a new strength in my life. This changed my outlook on my life and HD. I see things from a new perspective.

Families with HD hide behind a lot of shame and fear. This shame and fear seems to take over our life, till there 'ain't no more' life to look forward to. People with HD in the family are not 'nothing people.' We can accomplish many things. We are people who can recognize people's hurt and sympathize with them. I've never seen a group of people so dedicated, warm-hearted and willing to help those in need.

Isn't that what living is all about? Caring and helping one another in our time of need? Who can say for sure that they will *never* need anyone's care or help? There will always be a time when you will appreciate a kind heart and warm smile. People who face the risk of having HD every day are such people of concern.

You will never know if you get into an accident and get paralyzed or handicapped, and there are other diseases like heart disease, stroke, Alzheimer's, multiple sclerosis. We can all be victims of such horrible circumstances. People at risk are real people, people with feelings, needs and something to give.

There are people who will criticize a couple at risk for having children. Why? Our society would like to have the perfect society. In some societies, if their first born is a female, they will get an abortion until they are pregnant for a boy. Hitler tried to make a perfect society by eliminating all those people who had any defects. Is society any better today? Our society will not be better by murdering babies before they are born. I don't believe in abortion. I feel it's wrong and God calls it sin. I ask you who has the right to rob another person's life because it is less than perfect?

“People with HD can accomplish many things. We are people who can recognize people’s hurt and sympathize with them. I’ve never seen a group of people so dedicated, warm-hearted and willing to help those in need.”

Now at 36 years old, and celebrating our 17th anniversary, I count myself lucky. My beautiful daughters are 12 and 13 and a half. I love my husband, I love my daughters and I love my life with them. I feel good about having my family, whether I have HD or not.

I don't know if at 20 years I would have been tested, but now I find myself wanting to know the truth. I feel positive about getting tested; it will give me a sense of control over my life. It will help me to make some important decisions about my future and my family's future. This will help me prepare mentally and physically for the challenge that lies ahead. To anyone who is at risk of HD, I would like to say that a different perspective is needed, one that will accommodate a challenge to change, and a challenge to meet the needs of a different stage in your life and to do the best you can with what you have.

Whatever happens, if I test positive or negative, I don't regret my decisions, and my trust and hope is in God Almighty. I know that God doesn't throw away people because they're less than perfect. He loves them greatly.

Who can stand and say you have no right to fulfill your life? Who has the right to take away your hopes and your dreams for a better life? It's not a matter of eliminating those who are less than perfect. It's a matter of hope, caring, sharing and love.

Part II

The Process of Predictive Testing: Before, During and After

The five stories in **Part II** are from persons going through the process of predictive testing. They derive from a series of in-depth research interviews with persons who proceeded with predictive testing in British Columbia in the mid 1990's. These interviews were held in people's homes several weeks before, as well as several months after, the test results. With the permission of research participants, the interviews were tape-recorded and transcribed.

The transcripts were edited by the researcher (Dr. Susan M. Cox) in order to produce a highly condensed version of each person's story. The researcher then provided a draft copy of the story to each participant in order to ensure that it was factually correct and a fair representation of their experience. Though the passage of time sometimes meant that participants wanted to retell their stories in order to reflect their more recent understandings, all agreed that it was important that readers of this booklet have the opportunity to learn what the experience was like for them at the time they were going through it.

Pseudonyms have been used where necessary to maintain the anonymity of participants and identifying details have been removed. Longer versions of each of these stories appeared in Susan Cox's doctoral dissertation, "*It's Not a Secret But...*" *Predictive Testing and Patterns of Communication about Genetic Information in Families at Risk for Huntington Disease* (The University of British Columbia, 1999).

The research project "Individual Experiences and Social Meanings of Predictive Testing for Huntington Disease" (Dr. William McKellin, Principal Investigator) was funded by the British Columbia Medical Services Foundation, the University of British Columbia Hampton Fund and the Huntington Society of Canada. The project received ethical approval from the University of British Columbia Behavioural Sciences Screening Committee and was carried out with the collaboration of Dr. Michael Hayden and the Predictive Testing Research Group.

When your load is lightened

One week before his test results, Colin described how he first learned about Huntington Disease and eventually arrived at the decision to proceed with testing...

I grew up in Ontario. I have one brother, five years older than me. We had a normal family life until my Dad took sick when I was 13. That was in 1966 and for a few years we thought his illness was something to do with a serious car accident he was involved in. In hindsight we can see that it was the onset of Huntington's. It wasn't until 1970 that we actually found out it was Huntington's. My Dad would have been in his mid-50's.

The year that my Dad was diagnosed was about the same time as the Huntington Society was being formed. I was there at the initial meeting in Toronto. Marjorie Guthrie was there too. In those days there were quite a few myths about Huntington's, but one thing I always knew was that there was a 50:50 chance of inheriting the disease.

“There were fewer options when my Dad was sick. No one understood. There was no Huntington Society going out to the care homes and giving them specific training on how to understand and deal with someone with the disease. There was none of that.”

I moved out to BC in 1972, and met my wife Emily in 1974. I told her that I was at risk for Huntington's before we were engaged. And before we got married we had genetic counselling so that she could get a professional point of view. I don't think there's anything that the doctor could have said that would have scared her off. We talked about it. I tried to get her eyes as wide open as possible, but when you're in love it doesn't matter. It is not until later in life that you find out what your love is made of.

Seventeen years whizzed by for us. We have three children aged 13, 11 and 8. We're pretty busy, but I've always tried to find a balance. Huntington's aside, you never know how many years you have together. Our philosophy has always been that we try to have fun along the way because you just never know.

My brother Brad was diagnosed with Huntington's about 10 years ago, and he's not doing very well. It's time for him to go into a group home. That's a hard thing to do. There were fewer options when my Dad was sick. No one understood. There was no Huntington Society going out to the care homes and giving them specific training on how to understand and deal with someone with the disease. There was none of that.

I first heard about the predictive test through the newsletters of the Huntington Society and I thought it was stupid. Why in the world would you want to know? There was just no way that I would entertain something like that. I figured that you're not supposed to know what's going on in the future. You live by faith, so why dwell on it? But I was in my early 30's then so I had a different perspective on life. You get to 40 and you start to look at life differently, whether you have Huntington's hanging over your head or not. You start to look at where you've been and where you want to go.

All through this last five years I thought that I should at least, for my children's sake, get my blood banked in case I get bumped off by a bus or something. That way, if they ever wanted to know whether they're going to get it or not, there's some blood on hand and they can get the predictive test done. That started me thinking. At the same time, the Huntington's cloud was gathering on the horizon and getting closer and closer. Then it started hovering over me. I didn't think about it heavily every day but there were more and more days where I was thinking about my risk situation. Having your own business, you have to plan ahead. And then there are the children. I was thinking about all this and my thinking went from not wanting the test at all, to at least getting my blood banked, to thinking that maybe I would get the test so that I could use it as an instrument for planning my future.

At the same time, it looked a lot better to go back to Ontario, start fresh and try to enhance my brother's and my mother's lives. But I thought I can't do that right now. I need to know whether I'm going to get the disease because I wouldn't want to take my family and my wife away from family and friends especially. She would need her support system if I should I get the disease. That's what brought me around to wanting to know for sure. Over two years I did a complete 180 degree turn in my thinking.

My decision to have the test was something I came to on my own. I didn't talk to Emily about it. I haven't discussed my feelings because I don't want her to be concerned. It's my way of protecting her. I mean if I really shared my inner-most thoughts over the last few years, I would tell her that, at times, I'm scared. And I am scared when I trip. Sometimes it does concern me, but I'd never say that to her or to anybody else because then they'd just start looking for it.

“My decision to have the test was something I came to on my own.”

When I told Emily about deciding to have the test she was glad because she had thought about it too. And the friends that she's told are quite happy that I'm doing it for our children's sake. I could live without the knowledge but you get to the point where you feel you owe it to your kids. I don't think my kids know that they are at risk. I don't think they're old enough. They know Granddad had it and that their uncle has it. They know I can get it. Depending on the results of the tests, we'll handle it with them as they get older. I remember it was pretty hard when I found out about my dad. You are confronted with mortality in a new and different way. I remember going through that process myself.

We will make whatever lifestyle adjustments we need to make sure we have a good quality of life and enjoy each other. So in that way, this test is a real opportunity. We view it as an opportunity even if the results aren't the way we want them. We know we have a time frame. It's not like somebody with colon cancer, that's got two months to live and has to go into chemotherapy the day after they are diagnosed. But I only look at it that way because I've gone through the process of thinking about what the test really means to me.

“Testing sounds like a good idea but you've got to think it through.”

When you are young you have your health and everything looks great. But you really have to come to terms with mortality. When you have the possibility of something like this you need to look into it and know as much as you can about the disease. You have to come to terms with the reality in your life. All of a sudden things that you've been believing all your life go out the window. It is quite amazing. The technology is coming faster than the psychological understanding. Testing sounds like a good idea but you've got to think it through.

I think the only thing that I would hope for other people is that they're able to go through the process with somebody else, and if they have access to it, the Huntington Society has an at risk support group. It's really good to go there. You can share your feelings. People can relate to it, they can empathize with you because they've had the same feelings. There's a real camaraderie in talking to people in the same situation.

I had an idea that the pre-counselling would have more to it than it does. I don't feel like I've garnered anything from it other than I know exactly what to expect with the process of testing. It hasn't helped me in my thinking, but from what they tell me, Emily and I are more prepared for this than almost anybody else they've seen.

I'm ready now but on results' day it might be a whole different story. I haven't really thought of what it will be like. Every time I think about it, I don't want to think about it. On a good day I figure knowing for sure is better than not knowing. But that's easy to say

when you don't know for sure. I certainly could adjust real easy if I don't have it. I can adjust to that news so I want to make sure that I can adjust for the other reality, if that's what it's going to be.

Colin's predictive test results indicated that he did not inherit the gene that causes HD. About six months after receiving this news, Colin explained that he and his wife Emily felt relieved to know that their children were not at risk for HD. Colin said it did, however, take a long time for the meaning of his test results to really sink in.

The last few days before we went to hear the news was a busy time. It wasn't a good time to be going through predictive testing, but when is a good time? I wanted to go through the process and I wanted to know as quickly as I could.

I was getting pretty testy before results' day. It was really hard for Emily because we both came to the conclusion during the last few days that I had the gene. We could deal with it for us; it was the children. We were told before we got married that we shouldn't have kids, but you hope that, first of all you're not going to get it; and secondly, if you do get it, there will be treatment, and by the time you have kids there will be a cure. And on top of that, you still have a life until it happens, even with kids. But when you're confronted with it and you realize that you've done something that you could have prevented, you've had three children and you could have prevented passing it on. It wasn't a nice feeling for us as parents.

We didn't tell the children what we were doing, but the little darlings figured it out. I wasn't there, but on the morning that Emily and I were going in for the results, they were at breakfast and the little one was saying grace and asking God "please don't let Dad have Huntington's disease." So how they figured this out I don't know; we had tried to be so careful.

It was a long drive in to the clinic, literally and figuratively. We still had a certain peace because we had prepared ourselves for the worst and honestly felt that we were ready for it. And we had looked at the positive side of the worst and felt that we could deal with it. So when I got the results I was not ready, it just went right over my head. The doctor started off by coming into the room and saying that he had some news that he thought I would be pleased with. I didn't say it out loud, but my mind said, "then he'll tell me the bad news." So he told me that I did not have the gene and it didn't fizzle on me at all. I looked at him and the genetic counsellor and wondered what they were thinking. Should I be going up and down the hall doing cartwheels and shrieking and hollering? I had no reaction, it was nothing.

For people that know me, my reaction was probably predictable. I just sat there and listened. I'm not an excitable type of person as a

“We still had a certain peace because we had prepared ourselves for the worst and honestly felt that we were ready for it.”

rule. I was relieved, but we honestly weren't expecting that result. I've been carrying this monkey on my back for about 24 years and it has been such a big part of me that I can't just throw it off my shoulder, walk out and do a few cartwheels.

The results certainly didn't sink in that night. It sunk in a little bit each time we told somebody. We walked out of there kind of numb, not knowing really how we should feel. But we went out and had a nice dinner. I don't even know what we talked about, but it was just nice to go and enjoy ourselves and unwind. Then we came home and stopped at a neighbour's place to tell them because they were standing by waiting for the results too. They were probably wondering why I wasn't giddy and ecstatic. It would be easy to say I should be the life of the party, but it didn't go that way for me. But it was nice to have shared it with them. Then we went home and I don't know if we opened a bottle of wine or not, we were pretty tired and emotionally exhausted.

“I’ve been carrying this monkey on my back for about 24 years and it has been such a big part of me that I can’t just throw it off my shoulder, walk out and do a few cartwheels.”

After that, it was business as usual. I got up the next morning and went to work. I don't think I was any cheerier than usual. Part of it is that I've just been too busy to really think about it. The other reason is that we have very few people we can share the news with. Because of the nature of this disease you don't tell just anybody that it is in your family. If they understand that it's hereditary they put two and two together, and you're really jeopardizing your position in work or insurance matters.

I figured the less people know, the better. But when I got the results, the doctor asked, “well, who are you going to tell?” I said, “I'm not telling my brother or my mother.” He asked why and I said, “Because I feel guilty, especially in my brother's case, I feel guilty.” The only reason for not telling my mom was that I was afraid that she'd unintentionally tell my brother. But the doctor said to me, “What you received today is a gift. You have the gift of knowledge, you know.” We were looking at it that way but the doctor said “Would you not want to give that same gift to your brother and mother?”

My mother passed away quite suddenly a few months later, so we all went back to Ontario to deal with that. My brother was there as well. He's still independent but probably not for more than another year. I found that with the disease not being a threat to me now, it's easier for me to deal with it with him. Before, when we'd spend time together, he used to tell me what he was experiencing at each stage. I could only take a certain amount of that because I was still at risk myself. I didn't want any other reminders of what my future could be. But going back this last time it was a lot easier to deal with. I think it will help me to be more understanding because I don't have that emotion or threat. The only thing I have to deal with is survivor's guilt, and I have a great deal of that.

I did tell my mom and I'm glad. I phoned her within a week of my results and she was thrilled. I had planned to tell her the next time I saw her in person, but it would have been too late. So I was able to pass the gift along and it was probably good for her because she had one less kid and three grandchildren not to worry about. In that way it was a very real gift to her.

There are certain people you can't tell afterwards even though you have good results. They will feel bad that you never thought enough of them to share it with them before. You always have to be thinking who you can tell and who you can't, even afterwards.

The other thing that I'm living with right now is that I haven't grieved my mother's death. I had to go back East to do the funeral because my brother wasn't capable. I wanted to take an hour to go down to the cemetery and just sit there, say goodbye in my own way, all by myself. It's sort of just below the surface, but you are so busy that you keep pushing it back, pushing it back and then one day it won't be pushed back anymore, it'll come. So I guess that's the way it'll happen. And probably the results of the test will happen the same way. One day I will turn a cartwheel.

I would like to move back to Ontario and really try to give my brother a better life in his remaining years, let our family be his family. Emily is quite prepared to do that. Since I learned my results it would be easy for me to turn my back and say, "I'm out of here. Twenty-four years I've lived with this, it's history. I don't have to pay my dues to the Huntington Society anymore. I don't have to go to the meetings anymore." But I don't feel that way at all. I feel a sense of duty. I don't have a lot of time, but if I can be available for somebody that wants to have a chin-wag, especially at risk people, I think the story that I have to tell could really help a lot. I'm not trying to brag, but I've never been so aware of how a person can go 180 degrees in their thinking. I've never done that on any other issue in my life, and I think it is important to understand how each person and family affected gets through these things.

I wish I could have taken two or three days after the results to think about what happened. I would have liked to have had some quiet time to process everything and put it to rest. I also wish I could talk about it to more people. But again, I think most of my pleasure from the whole thing is for my children. It's nice that they can look at their uncle and realize that they don't feel threatened just as I don't feel threatened. I can look at him or I can go to the Huntington's convention and see people and realize that that's not me in five years or two years. I don't have that fear anymore. That's when you feel like your load's lightened.

Postscript: Colin and his family moved back to Ontario to assist with caring for Colin's brother.

Why walk on egg shells?

Two weeks before her predictive test results, Regina described how she first learned about Huntington disease and eventually arrived at the decision to proceed with testing...

I was born in Ontario. I have one brother and one sister, both older than me. My grandfather died of Huntington's the day after Mom and Dad got married.

I knew about the family history of Huntington's as soon as I was old enough to understand, because my grandma was the type of person that ate, slept and breathed HD. Even when we were very young, HD was always in the forefront when Grandma was around our house.

My dad was in the military so we moved around a lot. He started showing signs of HD about 13 years ago. At first it was nothing really major. You start seeing little signs—the handwriting goes, you lack coordination in some things. Mom was the first one to notice it but there wasn't a big "to do" made about it. We had a totally different attitude from what I was raised with around my grandma. Things were always done as much as possible in the way Dad wanted. Nobody had the attitude "No Dad, you can't do that 'cause you got Huntington's." It was a matter of finding a way to do it.

I was very close to my dad; I was probably Daddy's little girl. We were it, him and me. So losing Dad hit me very hard. Dad was a very special man. I never heard anyone say a negative thing about him. He would do anything for anybody. He would give the shirt off his back.

I love my mother because she's my mother, but I don't have a close relationship with her. All of us kids moved out of home very early. We were 16, 17, 18, in there. Since then each of us has, at one point or another, been on the outs with my mother. As far back as I can remember, the only time we've all been speaking was just before Dad died, during that process.

“We decided that we were willing to take the risk with one child, and we felt that risk was okay, but we weren't willing to take it with more than one.”

My husband Mark and I probably would've had two children if it wasn't for the Huntington's. My ultimate was always to have two children. As it was, I had my tubes tied when Geoffrey was six months old. We decided that we were willing to take the risk with one child, and we felt that risk was okay, but we weren't willing to take it with more than one.

Huntington's is not something we talk about a lot. It's there but it's not there. When Dad was quite ill, I did however do one thing that was probably very strange. I told Mark that, if he had any second thoughts, the opportunity for him to get out of his relationship with me was now. Once I got sick it would be too late. He said he was in for the duration. People would say to me, "You're so mean for putting him in that position," and I said I have to know in my heart that if I get sick he's there because he wants to be, not because he feels obligated or trapped. That means a lot to me. The last thing I want from anybody is pity.

I found out that there was a predictive test a couple of years ago. At first I really didn't care. I wasn't willing to make that decision in my life. I don't know why it's changed, maybe it's because my life was so busy for such a long time that I never had time to think about it. I know my mom was upset when my brother had his test done. I told her he deserved a lot of credit because he had made a very hard decision, whereas I was riding the fence. I never decided whether I wanted it done or didn't. Then all of a sudden it was bugging me all the time and I wasn't sleeping. Not being a very religious person, I thought somebody somewhere is telling me that it's time I made the decision in my life. Instead of being in my subconscious it was kind of consciously there all the time. I thought there's got to be a reason for it.

I'm a very spontaneous person, so as soon as I decided, I called. Then I was on a waiting list for about six months. That seemed pretty long because I'm very impatient. If I go shopping and I want to buy something, and they don't have it, God help you 'cause I'll go somewhere else and buy it. I'm very much impulse on some things. I want it now; don't wait.

My husband was totally against me having the test and it finally came down to "well, it's my body, my life." It's not that I didn't listen to what he had to say. I did. I weighed the pros and cons. He said he was afraid. You see he was a very big part of my dad's life. He was very close to my dad and he had a very hard time when Dad passed away. He said he didn't want to know. I told him, "Fine, I won't tell you. If and when you're ready to know you can ask me and I'll tell you." He's not going to be here when I get my results since I didn't get my appointment until after his work commitments were set. He did go with me to my counselling appointment, and he did voice his

concerns. That's fine. I listened to them, but I've made my decision and that's where it stands. Nothing anybody says or does is going to make me change my mind.

There wasn't a lot that people at the clinic told me that I didn't know already. I knew about all the research that had developed because they found the gene. That was about a month before Dad died. When Dad passed away, the Huntington's research received whatever tissues they wanted to use for their studies. That seems to help me a lot because I know that Dad was the type of person who always wanted to help if he could. Knowing that he would feel that he went through this for a reason has helped me get through a lot. It's like when I go to the clinic and they ask if a student in genetic counselling can sit in on my appointment. I don't mind because it just brings more people in that know about it. How else are they going to learn?

Geoffrey has grown up knowing about HD. We've always been firm believers that anytime he asks a question it's answered honestly. If he doesn't ask, we don't volunteer the information because we figure when he gets older he can ask then. Geoffrey doesn't know anything about my test. What am I going to tell him to make him understand? Right now it hasn't come up, so there's no sense bringing it up. He's only eight years old.

“Everybody’s got their own circumstances, their own reasons as to why they want to know what they want to know. I think it would make a very big difference if somebody was making this decision and they didn’t have some kind of support around them. Then I don’t think it would be a smart move.”

If my test comes back negative we don't ever have to worry. The only thing we have to tell him is it's done with and you don't have to worry about it. If it comes back positive, then he is going to have questions and they'll be answered at the time he asks. We've never lied to him about anything, but we wait and let him do the initiating.

Either way the test is going to answer a lot of questions about Geoffrey and for Geoffrey, but also there's little things that Mark and I always decided. One of the things I always wanted to do is go on a

cruise. I'd like to go when I'm in my 30's rather than when I'm 50 because I plan on having a hell of a good time. Also, we have always agreed on the fact we can comfortably afford to help Geoffrey through university without going into a great deal of debt because we set our goals for paying our house off.

Everybody's got their own circumstances, their own reasons as to why they want to know what they want to know. I think it would make a very big difference if somebody was making this decision and they didn't have some kind of support around them. Then I don't think it would be a smart move.

There are other people that know I'm having the test. It's not like I've ever tried to hide it, but I also don't volunteer the information because I don't think it's something people want to hear about. I told my friend Denise when I first called to get the test done. I don't think it ever came up after that, but she knew roughly when my appointments were.

Results' day is no big thing. I have to work till I go for my appointment, then I'll drive into the clinic and if somebody wants to come with me that's fine. It's kind of hard to get somebody to go with me though, as most people work Monday to Friday, and it's a 2 hour drive. I'm fine with it, though. I had originally intended on doing it on my own anyway so nothing has changed. It's not like anything's going to change.

Two weeks later, Regina went to the clinic and received her predictive test results. Her friend Denise went with her. When Regina learned that she had inherited the gene that causes HD, she did not have a strong emotional reaction. About a year later, Regina explained why this was the case.

Just before going to the clinic, I felt good about my test results. I'm a fairly positive person, but if there's something negative to consider, I'll usually find it. Or at least, I used to be like that. So it was really strange that with something like this there were no negative thoughts. I was convinced that maybe it would be good news.

Once they told me my test results, I just said "okay." My friend Denise and I started joking back and forth and talking about booking our cruise and that was basically it. They kept asking questions and drawing the session out. When we left, Denise and I almost said at the same time that it felt like there was something that was supposed to happen that didn't happen. I didn't have an emotional reaction, I didn't break down. And I still haven't. I thought about that and I thought why? Usually when you have an emotional reaction there's something that's caused the emotion. Like when you stub your toe, you cry because it hurts. Or somebody says something to you that hurts your feelings. It upsets you, but it's usually relevant at the time. Well, the news wasn't great, but I'm

“It felt like there was something that was supposed to happen that didn’t happen. I didn’t have an emotional reaction, I didn’t break down. And I still haven’t.”

wondering if maybe the reason I didn’t have this big emotional reaction I was supposed to have was because it’s really something that’s not relevant right now. Maybe the time will come when I’ll start showing signs and symptoms and that’s when I’ll feel that it’s an appropriate time to have a reaction. Right now I guess I could break down and cry, but what am I going to be crying for? Nothing’s really happened to make me react that way. I control my life, not HD.

They were telling me the future, that’s all they were doing. It’s not now. Giving birth to Geoffrey, that was emotional! But there was a reason for the emotions. With this, there hasn’t been a reason for the emotions yet. Maybe when there is that’s when it’ll hit me. Crying over something that hasn’t happened yet is kind of like putting the cart before the horse.

A couple of weeks after I got my test results I got a phone call from the genetic counsellor. They wanted to know how I was doing and if I was okay. And I’m thinking, “What the hell’s going here?” Is there something wrong with me? Am I going to be in the shopping mall and all of a sudden be an emotional basket case? What are they looking for?

Are they really looking realistically at what people are doing and dealing with and how it’s affecting their lives? Before your test results you have to fill out a package of questionnaires. I might understand the relevance of the questionnaires if they were personalized but I don’t like being forced to choose one or the other answer if it doesn’t relate to me. I guess I deal with things very differently than most people. You just play the cards that you’re dealt and go on. I’m not one to talk about the problems that are going on in my life. There’s always somebody else out there that’s having worse ones. I guess maybe I have a very callous attitude. I don’t know what it’s called. I guess I was never going to be the textbook patient.

It’s not denial. It’s called going on with your life and dealing with reality and not allowing Huntington’s to run your life. I have my life to think about, and I have Geoffrey’s life to think about. Nobody says what I feel is correct. Some people agree with me, some people don’t.

Mark did come to me and say that he was ready to know my test results. When he asked me, I said, “What do you think my results were?” He said, “Well, I just thought it was good news. You’re fine and nothing’s been bothering you.” And I said, “Well, actually you’re wrong.” He didn’t quite know what to say, and basically from that point on he just totally withdrew. He was a totally different person than what I expected. He did everything with Dad. And he was right there by his side no matter what he needed, any time of

day; moral support, emotional support, everything. You couldn't have asked for a better person. And he just said that he went through it once and he didn't think he could go through it again.

I'm pretty happy with my life right now. I'm involved with somebody new named David, and I have Geoffrey. It doesn't look too shabby. My relationship with Mark, my ex-husband, has its ups and downs. I've made a lot of sacrifices for Geoffrey and put up with a lot for him, but I figure I have broader shoulders than he does and I'll bounce back. Geoffrey is coping pretty well, and no matter how Mark and I feel about each other, Geoffrey is still number one. He spends a lot of time with his Dad and that will only change if his Dad wants to change it.

I have a new job working in reception for a law firm. It's okay but I will never be a lifelong receptionist. To change careers you have to start somewhere. So it's definitely just a starting point. I also just finished six months of school. I dedicated probably 99% of my time to it and I came out with a 95.8% overall average. I took computer courses and accounting courses. You have to branch off and start somewhere.

So bad has turned into good. I got laid off and I got separated, but because I got laid off and I got separated I knew had to go back to school. If those things had not happened I wouldn't have been in a situation where I could do it. So I'm starting to believe more and more that everything happens for a reason. You have to be optimistic somewhere in there.

The biggest hurdle I have had to cope with was telling David about HD and my test results. I guess it was a big hurdle because of the reaction I got from Mark when I told him. David had been really sick, and I knew that I was going to have to tell him sooner or later. He was almost better and we were sitting at the kitchen table at his place and we were talking. We hadn't been dating very long, maybe two or three months. I said, "Well now that I've nursed you back to health, are there any other medical conditions that I should know about you?" He has a degenerative tissue disease and he's got rheumatoid arthritis. Maybe that's why he can relate so well. So when I told him about HD he just said "Okay." And I said, "Well do you know what it is?" And he said, "Yes, but you can get killed walking out in the street tomorrow by a bus. Cancer can hit you, Alzheimer's can hit you, a gazillion other things can hit you." He said, "This just tells us that if nothing else does, this will." So that's kind of his attitude. Very pragmatic. That's David.

I don't really remember telling anyone else about my test results. The friends that knew I was having predictive testing listened to see if I had anything to say, and if I didn't, they wouldn't pump me for more information. So it's on a "how I feel comfortable" basis. That's

“It’s not denial. It’s called going on with your life and dealing with reality and not allowing Huntington’s to run your life.”

“I’m still the same person, I just have this bit of information that other people don’t have. It doesn’t make me any different. That’s what I’ve told myself.”

how it works and that’s fine with me. People that have to know, know. I don’t go around advertising it to the world. Some people knew about Dad and they know the disease. They’ll ask about my risk and I have no problem telling. I’m still the same person, I just have this bit of information that other people don’t have. It doesn’t make me any different. That’s what I’ve told myself.

I wouldn’t tell my mother about my test results until I was ready. She didn’t know until at least a couple of months after. I think I was mad at her when I told her. She hasn’t been there for me. I’ve been through everything on my own with my friends. I am lucky in that I have some really good friends. I also know who I am more than I ever have before. I’m quite happy with my life and the way it’s going. I’m not out there for everybody’s approval. What I’m doing makes me happy and it makes Geoffrey happy, so that’s what counts.

Maybe that’s why my attitude is different when it comes to HD, too. Going to the clinic causes a lot of uneasiness for me. It brings so much back again that I’ve already dealt with and put behind me, stuff that I don’t think needs to be brought up again right now. For some reason, I seem to be the abnormal one and I don’t quite understand why. I go in to the clinic and they want to know what’s happened in my life in the last year and a half. I say, “Oh, not much.” “Why do you say it’s not much? You got laid off, and you got separated, and you have a new relationship.” Yeah, I do, but it’s my life; it’s no big thing. Do they want me to say all of this is caused by HD? It’s not. It could have happened if there wasn’t HD in the family. I’m just too damn stubborn to allow HD to alter my life and change who I am. Maybe that will be to my disadvantage down the road, but a round peg just doesn’t fit into a square hole.

I still don’t really know why I wanted the test. I haven’t altered the things that I do in my life. The only thing I might change is I don’t worry about putting money into an RRSP anymore. I’m optimistic, though, that they are going to find a treatment. And then the information from my test might come in handy. They might find a drug they think can prevent onset of HD. If they didn’t know who was going to get it, they wouldn’t know who to give it to. So there’s an advantage. If that test wasn’t there, they would only have a 50:50 chance of finding people that fit into the right categories.

Knowing what I know, I just sit back and think about it realistically. Is it altering my life now? I say no. Is there anything I can't do? No. Well then why walk on egg shells? Why not live your life the same way you were living it before? Nothing's changed. Nothing physically has changed at all. You have one little bit of information that not everybody has the opportunity to get, and that's it. That's the only thing you've got that's different. So why should it change your life? I don't think it should. I don't know if that's right, but it makes sense to me.

Let your family help

Two days before her results day, Helen described how she first learned about Huntington disease and arrived at the decision to proceed with testing...

I grew up on the coast, went through school, married, had kids and divorced. I've been living common-law with Duane for about 12 years. I have three sons, all in their twenties.

I have one sister, Norma, who is 18 months older than me. We were always close. When she and her husband split up, she and her son moved in with us for a while.

I have worked as a bookkeeper for all of my adult life, but recently I went back to school. I've been training to do troubleshooting in computers. I love it, but for this last while predictive testing has been an excuse not to go back to work.

I didn't know there was Huntington's in my family until about seven months ago. I got a letter from my cousin in England telling me that his sister had been diagnosed with Huntington's. That was the first either my sister or I ever heard about it. I don't think my cousin felt that my sister and I could be at risk, because as far as we knew my mother had never had any symptoms and she was 67 when she died from cancer.

In the letter, my cousin talked about his memories of his mother and her illness. We never knew anything about his mother's illness. It was very much a secret that my aunt had even been ill. My mother made such an effort to find out, that I think she must have suspected a lot more than she told us. It's strange to us, but I don't think the doctors in England tell people as much as our doctors do. My cousins were shocked when they found out how much I knew about the cancer my mother had.

When I got my cousin's letter, I didn't know anything about Huntington's. I naturally assumed that the doctor's office would be able to tell me something, but they couldn't. Nothing. I went to the library and looked at the medical books and it was not pleasant.

“I was absolutely up and down. ‘I want to know,’ and, ‘I don’t want to know,’ and, ‘It’s happening and it’s not happening.’ Very little was getting in.”

When I came out of there, the one thing that stuck in my head was the fact that they really didn’t know what the normal cause of death was because most people with HD committed suicide. That may or may not be true, but that was the one thing I learned from my little foray into the library. I kind of bounced off the wall for a few days.

I talked to the receptionist at the doctor’s office. I said, “How do I get tested? Can I get tested?” And she said, “We don’t know.” The next day she told me that they knew that there was research being done in Vancouver at The University of British Columbia. They called, and I called, and finally I got through to one of the genetic counsellors. She made an appointment for me right away and that was probably the first sanity I had. I was going pretty crazy by this time. I remember bouncing off the walls. I was absolutely up and down. “I want to know,” and, “I don’t want to know,” and, “It’s happening and it’s not happening.” Very little was getting in.

When I got my cousin’s letter it was January. My sister Norma was to be married at the end of the month, and I didn’t know what to do. I’ve always been considered the strong one in the family. I’ve always felt that it’s up to me to look after everybody and make sure that they’re fine. Knowing that Norma has been through a major depression, I thought she would call off her wedding if she knew and I didn’t see what good that would do for anyone. I had very little information, and I couldn’t see any sense in telling her, but I felt very strongly that her fiancé Ken had to know. He said that it didn’t make any difference what was in the family, he wanted to go through with the wedding regardless. I don’t know what I would have done if he’d backed out.

About two days after the wedding, the genetics counsellor called me back with an appointment date. Then I had to tell Norma. I thought she should know. She was not particularly impressed, but she went to Vancouver with me. It really wasn’t until I went to Vancouver that I really knew anything. A little bit of knowledge made things easier to deal with.

I would have liked to talk to Duane when I first got the letter, but I felt that he wasn’t as supportive as he should have been. But then we talked about it and he said that he knew my Mum and that she didn’t have any symptoms, so there was nothing to worry about. He is absolutely 100% certain that things will be fine and there’s not much sense discussing it, which is the way he is with all things; it’s not just this.

The idea that I could have predictive testing was probably my salvation. That was the big thing for me, just to find out. I wanted to take the test long before I even knew there was one. That was my first response, I have to know. I wanted to know for the sake of my sons.

I've had two visits to the clinic in Vancouver. Norma went with me to the first session. It was the first information we had. The genetics counsellor also sent information to my doctor so he'd have an idea what was going on. She said, "Think about if you want the testing done." I said I would have the testing done immediately, if I could. The biggest shock to me was the fact that everybody didn't automatically get tested, that people could live without knowing.

I don't usually share what I'm thinking with anybody. I'm very closed. Even five years ago, just before my Mum died, I never really shared with her how I felt. It's just something that I've never been able to do. I'm practicing it a little bit more now. Being alone fighting something is really tough. It is important to have somebody you can talk to. My immediate reaction was I don't want anybody to know, but as soon as I came to terms with the fact that it's no different than any other disease, I felt much better about it.

Mentally, I'm very strong. Stronger than most of my friends. What I need is the tools to work with, and the information. Without the information I can't do anything. I can handle almost anything as long as I know what I'm dealing with.

Most of my friends know that I'm being tested, and they all say, "There's absolutely no way it could be positive, we know that you're okay." This is nice, but what else could they say? Very few people know anything about HD. I get questions on what it is, and why I'm at risk, but nothing very detailed. I have two girlfriends who I have really discussed it with. One of them had breast cancer a few years ago, and I talk to her when I get the "what-if's." I know that she gets the "what-if's," too. I'm very lucky in having a sounding board there when I get these feelings. I call her and she says, "I feel that way, too."

I am really pleased with my family doctor. I've been with him for fifteen years or more, and I have great confidence in him. I found myself feeling very sorry for him with my coming appointment to get my test results. He said, "You know, this is the first time I've ever had to do anything like this. I don't know how it's going to be. You're going to walk in and take one look at me, and if I'm grinning from ear-to-ear, you'll know it's good. If I won't look at you, you'll know it's bad. You won't have to wait, you'll know." I actually found myself feeling very badly, thinking, "Why did I do this to him? This is stupid." I care that I'm giving him a dirty job. I know if I tell him he's a quack and he can't read the results, he will know that it's just nerves. I feel very comfortable with him. For me that is very important.

My results are two days away. I'm not sleeping as well as I would like to. I worry about this testing on and off. I'd like to just say that it doesn't bother me, but it does. The day before my last appointment

“The biggest shock to me was the fact that everybody didn't automatically get tested, that people could live without knowing.”

with my doctor, I cried all morning because I was scared. I have these ups and downs but at the moment I'm feeling really good. I told myself from the start that crying didn't do any good, and that I would not allow myself to be overcome by something that was only a maybe. I wouldn't allow myself to cry until the day before my test. And the day before my test I could sit down and have a damn good cry, and that way it would be over and done with, no matter what happened. Whether or not I actually will, I don't know. At this point I feel that it's too late. My target date for breaking down is tomorrow, but because I've allowed myself to do it, I probably won't.

I don't want anybody with me when I get my results. Before I start my appointment, I will have my doctor's receptionist call a cab so that it will be there for me when I come out. I want to get home right away. The house will probably be empty, which is what I'd prefer. I will deal with it myself before I talk to anyone. After that I have no idea. I'm not going to go out on a shopping spree to celebrate or anything strange like that. I'll tell Duane. He and my few closest friends will know first.

If the results are positive, I will probably look for work that is similar to what I've always done, and I know that I'm comfortable with. If the results are positive, I will definitely want to know more. I would expect my doctor to get more information because at this point I know more than he does. I know there is a Huntington's book for doctors; I've seen it advertised. There are a lot of things I want to do so I probably would make other changes in my life too. Nothing radical. I have twice before dealt with the fact that I could die. I had cervical cancer at age 30, and I was in a quite severe car accident. I have dealt with the idea that life doesn't go on forever, so this doesn't really make a huge psychological difference. It's renewing things that I already know.

Under the rural protocol, Helen was able to receive her predictive test results from her family doctor. About six months after Helen learned that she had not inherited the gene that causes HD, she explained that it had been very important to her to receive her test results in familiar surroundings. As her story demonstrates, even "good news" can unleash a flood of unexpected emotions.

Before I got the test results, I was quite numb. I was scared but I was in a lot better shape than I thought I would be. Duane wanted to come with me, and I told him I was planning to go by myself. I didn't realize at the time that he wanted to be there with me. I felt that I was strong enough to handle it by myself and that he should be working. I think I really hurt him by that, by not bringing him into it. But I went in and got the results by myself.

My doctor was a little nervous about giving me my test results. His face was a blank. Other than the initial, "Sit down," he told me

right off the top, “I got your results. They’re good. You’re fine.” There was no beating around the bush and I’m glad. Then after he told me, he went over the fax that he received from the clinic in Vancouver, and asked me how I felt. There was a short piece of paper with about four lines on it. I wasn’t even curious enough to have a look. My doctor wanted to know if Duane was waiting for me, and I said I sent Duane off to work and that I was okay by myself. I really didn’t have any questions. I had nothing to say. I felt I should be ecstatic but I didn’t feel anything. I was no different than I had been walking in.

I took a taxi home, and I lay outside in the hammock for about two hours. Then Duane came home. He thought I wasn’t home, so he went out again. I was furious with him, just furious. I went into the house and doubled up and cried. Everything hit me at that time. I had told myself that I wasn’t going to get upset, but I guess I don’t have as much control over my mind as I thought. When Duane came home I was probably the meanest I’ve ever been in my entire life. I was really nasty. I accused him of not caring enough to be there to hear the answers. He asked what happened and I said, “If you don’t care enough to be here then you don’t deserve to know.” I wouldn’t tell him. I went in the bedroom and I cried, and I asked him to pack his bags and leave. I can’t believe I actually did that. I was absolutely vile. He said that he was going to stay for 24 hours and if still I wanted him to leave he would go. It was probably an hour or so later when I finally got my head together enough to come out and tell him the results. Still not feeling guilty, still thinking he’s deserving everything I’m doing to him. I can’t explain it. I just completely snapped and I went through the rest of that day crying, and telling him how horrible he was, and that I didn’t want him around.

By the next day I was beginning to see the light a little bit and realize what I had done. He was very calm through the whole thing, and he’s never said anything other than that he understands I was under a lot of pressure. I really hurt him, but he knew that I wasn’t in my right mind. I told my doctor this, and he told me that I had to talk it out with Duane. I told Duane I was sorry, and that I was very lucky to have him here. He said that was fine. Other than that we’ve never talked about it. I don’t think that is particularly good. I’ve got this terribly guilty feeling, but I guess I deserve that.

I think my doctor realized I wasn’t as together as I thought I was, because he called me that night and the next day. He stayed very close, making sure that I was going to be all right. I think he expected my reaction more than I did. I was astounded by it. I think he handled it very well. It mustn’t have been a pleasant experience for him, but then doctors have lots of unpleasant experiences they have to deal with.

“I think my doctor realized I wasn’t as together as I thought I was, because he called me that night and the next day. He stayed very close, making sure that I was going to be all right.”

Things started to shift after about the third day, but it was another two weeks before I was on an even keel. I told each of my sons separately. I waited till they dropped by the house so I could talk to them. They both said thank you for not telling us until you had the results. They took it a lot better than I thought they would. They don't plan to have kids anyway, but they were both adamant that unless there was some cure they wouldn't have had kids if it had turned out that I had the gene.

I told my sister Norma my results within a week. She had decided not to have the test, but I told her that she had to let her son know right away. I have a pamphlet that describes Huntington's, and has the address and phone number on the back. I told her that I wanted to give it to Larry. I feel that he, like my kids, had the right to know where to go for information. Family is not the best place to go. Norma told me that I had no business talking to him, and we haven't spoken properly since. It has caused a real rift. Norma and I have both told each other how we feel and why we feel that way. And other than that there's not much that can be said.

“My advice is never to be over confident that you're handling it well. You really don't know.”

As soon as I was over the initial week or two of shock, it's almost like I dreamed it; like it didn't happen; it's in the past. It feels more like a bad dream now, and I have to remind myself it actually did happen. It's not really something I think about too much anymore. I've got too many other things to worry about.

I approve of the fact that I had predictive testing. That's a strange way of putting it, but I feel that it has given me a bit of confidence that I can face up to things even though they're not pleasant. I can do what I know is right. I think I've always faced up to what had to be done, but this one was particularly hard. Even though I knew I had to do it, it was hard for me to deal with. I realize that now.

I wish I could have had my results faster. It was almost nine months and that was a long time to wait. The initial lack of information was the main frustration, though. I also think I would have been in serious trouble if I hadn't been able to get my results in my home town. I was feeling so confident that I would have gone to Vancouver by myself. And I would have left the clinic saying, “I'm just fine. I don't feel anything.” And then when I got somewhere where there was no help, nobody to stand by me, I think I would have been in serious trouble. I mean if I had been really vile to a stranger they couldn't have put up with me.

My advice is never to be over confident that you're handling it well. You really don't know. I was sure that I was fine, and it was a shock to me to find out that I wasn't. There's really no advice I could give to anybody other than to allow your family to help you. I felt that I could handle it, and I didn't want to burden Duane. But looking back on it, I was shutting him out. If I had it to do over again, I would let him in on how I was feeling. But it's too late now; I had tunnel vision. I was looking at how it was going to affect me, and not considering that it was affecting everybody around me as well.

A task to deal with

Three weeks before test results, Gabriella described how she first learned about Huntington disease and eventually arrived at the decision to proceed with testing...

I grew up in Alberta in the 1940's and 50's. I have two older sisters, Karen and Annette. When I was young, we always went to church. Now I go to church with my sister Karen when I visit as she really is a strong, religious person. I'm not an atheist. I do have some kind of spiritual belief, but I'm not attached to any particular church.

My grandfather probably had Huntington's, but he was never diagnosed because he died of cancer. After he died, my mother went on an amazing search for her roots. My grandfather would never tell my mother anything, so she went back to England to search for information about her father's and her mother's families. People said her father had a sister that was crazy, and that she would dance through the fields. We figured that she must have had Huntington's too.

My mother would have been about 40 years old when her symptoms appeared, so I was seven. When I was a teenager, we were told she had schizophrenia. When I was 20, she was admitted to a mental hospital, and she was never released on a permanent basis after that. I feel very sorry because she didn't have a clue what was wrong with her.

After my daughter Suzanne was born, I wrote a letter to the psychiatrist at the mental hospital to ask if they knew what my mother had. I told the psychiatrist that I thought my aunt in Ontario had an illness which was quite similar to my mother's. The psychiatrist did not write back to me, so that got me nowhere. Then my mother took a terrible downturn. I was living in Ontario at the time and was pregnant with my son Jason. That's when the doctor told us that they thought my mother had an inherited disease, but they wouldn't know for sure until they did an autopsy.

My mother died two weeks after my son was born. I can remember walking the streets thinking, "What am I doing?" having had this

“I’m a bit worried that I’m not understanding myself well enough, that I might react to the negative news more strongly than I’m anticipating.”

baby. At that time we were told that Huntington’s was inherited through females only. My doctor in Ontario actually got a letter from someone at the Alberta mental hospital saying that it was inherited through the female and that it skipped generations. I knew that this wasn’t true, and so did my doctor, so he wrote back and said, “Aren’t you incorrect about this?”

When the autopsy results confirmed that my mother had had Huntington’s, I asked my uncle if my Auntie Sylvia had been diagnosed with Huntington’s, because I was still hoping that my aunt and mother had different diseases. My uncle said he didn’t know. Then, several years later when my Auntie Sylvia died of cancer, my uncle wrote me a letter saying, “I don’t know why I didn’t tell you before, but your aunt was actually diagnosed with Huntington’s in 1962.” In his letter he said, “I don’t understand my reasons now, but I’m writing to tell you that I’m really sorry.” I could never figure it out. Unfortunately he died about three or four months later, so I never had the chance to talk to him. I still have his letter, but I don’t understand his reasoning. People are funny about this kind of thing.

Huntington’s was one of the reasons why I went to teach school for two years in West Africa. I said to myself, “You better get on and do something with your life.” In 1970 I had my tubes tied, and I really regret it now. I had gone to see a geneticist because I wanted some counselling about being at risk for Huntington’s. All I wanted to do was get some advice, but when I went into her office and said I wanted to talk about the idea of having my tubes tied, she just said, “There’s absolutely no question about it, of course you should get your tubes tied.”

About a year later, my husband and I separated. I met my current husband, Brian, and we started living together in 1973. After we got married, I tried to get my tubes untied. I had gone through all of the preliminaries and it was all set in my mind. But when I walked into the doctor’s office he said, “I can’t do it. You’re 36 years old. I just came back from a conference in New York where I said the cut off date for this kind of thing was 35.” I was crestfallen. That was in 1976, and given that a lot of women are now having their first child at age 40, 36 seems like an unreasonable cut off date. So times change.

I remember exactly when I told Suzanne and Jason about Huntington’s. They were 10 and 12, old enough to have some comprehension. We were driving along and I remember saying to them, “You know the blood test that we just had? It was part of a research study on a disease that grandma died of, and it’s a disease that’s inherited.” I just explained it. I don’t remember a horrendous discussion, or crying or lying awake worrying. Of course, they had never lived

with anybody with Huntington's, so I don't think it was real to them until they got older and realized what it was.

I'm not sure when I started going to the Huntington Society meetings. I was very involved in the 1970's. I also went to the medical library to read about the latest research. My life was dominated by Huntington's for a long time, but the most amazing relief for me was when I had a PET scan. This was when PET scans first came out as a diagnostic tool. I can remember exactly what I felt like just before I got the result, and how exhilarated I was afterwards. It was my first 24 hour period of freedom from Huntington's since I'd found out that my mother died of it. Before I had the PET scans, I could lose my temper three times in a row and people would say, "Oh, oh, she's probably got Huntington's." Even normal behaviour didn't get treated as normal, and that retarded some of my own personal growth.

When researchers first started offering the predictive testing (using linked markers), our family had trouble obtaining enough blood samples. Also, I didn't really want to go ahead with it then because of the uncertainty. Once they discovered the gene and the 100% results were available, the road seemed clear. I talked about having the test with Brian, Suzanne and Jason. I needed their okay. I don't know what I would have done if Suzanne said, "Don't do it because I can't handle it." I might not have gone ahead. But they said go ahead. Jason doesn't seem to make a major thing out of it, and Suzanne said "Mother, I think it's really important because if you've got the gene we need to plan our lives with the possibility of helping you." That's a pretty warm thing to say, but that was her attitude.

I think the results are going to be okay. But how do I know? I've been hanging around the house for two days working and doing some of the things I like, but I've got myself in this huge state. I'm busy making jam. I need to clean out the basement. I've also been thinking about taking an extra course, but what I really need to do is unwind. It seems to be hitting me in a heavier way than I had anticipated, and I'm a bit worried that I'm not understanding myself well enough, that I might react to the negative news more strongly than I'm anticipating.

If I don't have the gene, I'm going to wish I was in Alberta to give my sister Karen a hug. I feel to some extent that I'm cheating her if I don't have it, because then she's the only one. On the other hand, if I do have the gene, that's going to be hard for my Dad. It'll make him, as well as a lot of other people, sad.

What we'll do with the news if it's not good, I'm not sure. I honestly cannot predict how I'm going to react. If the news is bad, it isn't different than it was yesterday because I've always had it. I'll do my best to forget it until I have to not forget it, but I certainly won't like

“I just wanted to get it over with.”

it for my kids. I know that. I might have to go away by myself for a couple of days. I don't know. I'm aware that I've missed out on a lot of support because I have this independence thing. I don't seek help unless I'm desperate. I don't find it easy to ask, “Gee, what have other people done in this situation?” But I am learning that maybe that's a jolly good idea. In this certain circumstance, I might really benefit from doing that.

Gabriella's predictive test results indicated that she had inherited an intermediate form of the mutation associated with HD. As such, it was unclear whether she would eventually experience onset of HD. As she explained several months after learning this news, it was a good thing that her husband, daughter and son went with her to the clinic. Everyone had a lot of questions.

During the week before my results, we went to an island where friends of ours own property. It's very barren, but it's an absolutely beautiful place. Our friends were using dynamite to clear the area where they're going to build their house, and so I ended up moving a lot of rocks. I worked for the whole week. It was so therapeutic.

We came home on Sunday night and I got the results on Tuesday. I was very anxious on the morning of my results. I just wanted to get it over with. But I always do all the things that are going to make me feel good, like wash my hair and wear something I like. I always do that for these things. Suzanne and Jason and Brian went with me. I can see everybody and where they were sitting, but I'm quite blank about the visit. Getting the news was a shock, I don't know how else to describe it. I expected to hear that I didn't have the gene.

The geneticist told me how many repeats I had. Then he said, “We don't usually tell people how many repeats they have, but we're telling you that you have 36 repeats because you're in this odd category of people.” He said it was complicated. The clinical team didn't think it was going to be like this. He said, “We've looked at all of the data and there are only about 15 people who have between 36 and 38 repeats. Some of them don't have Huntington's and they're in their 80's, there are some who are in their 70's, some in their 60's and some in their 50's.” We had a long discussion about this. We also discussed the inheritance pattern as it differs between males and females, what that meant for Jason and Suzanne, and how odd it is that my sister and I have quite a difference between our number of repeats. It seemed that we had a bit of an odd pattern in my family. It was interesting, too, in that if I'd had the predictive testing several years ago (with the linkage test), I probably would have been told that I had very high expectations of having Huntington's. The geneticist said he felt like he was giving a university lecture; we had so many technical questions. He gave us a few articles to read because we like knowing what there is to know.

“After I got the test result, it was almost like a high. It was so odd. I guess it was just the fact that the anxiety was over.”

Jason was really upset. I was amazed and worried at how upset he was. He started crying not very long after I got my test results. Suzanne was fine. In fact, she was stronger than I thought she would be.

After I got the test result, it was almost like a high. It was so odd. I guess it was just the fact that the anxiety was over. It was also peculiar because I had these odd results. It's like you have it, but you don't have it. So I had to focus on the positive, the fact that it was kind of bad news with a good flag. I phoned all of our kids that night. I spent half that night on the phone, I remember that.

What I remember about the week after is that you sure know that you've got some good friends. One friend gave me a gift certificate for a manicure, and some other friends came over with a big bouquet of flowers. And on the same day that I got the results, I went to the drugstore and bumped into my allergist. He just happened to be there, and I saw him and told him. He's a very comforting person. I know a lot of really genuinely caring people, and it wasn't hard to tell them.

After I got my results, Suzanne, Jason and I went to Alberta for three days. I told my sister Karen my results before I went to Alberta, so everybody there knew. One night while we were visiting, our whole family went out for dinner. My niece was great. She got up and said, "Well, there's been a lot of tension in this family, there's been a lot of phone calls back and forth." And she said, "When I get tense, I shop." She bought a goofy present for everybody there. It was such a great way to make everything lighter. I really appreciated it because I thought, "It's not me, it's our whole family."

I had many good conversations with my middle sister Annette, more open conversations about Huntington's than I've ever had with her. I never knew that she worried about it because she never wanted to talk about it. My Dad hasn't dealt with it. He doesn't understand it, and I don't think he wants to. That's the odd thing. He told my aunt in England that the news was good, and so I had to write and tell her that it wasn't quite that way.

The second time that the lab ran my test, the result was the same. Maybe I had a little hope that it would be different, but I was really expecting it to be the same. You don't have any control over what

“One of the good things about Huntington’s is that it encourages you not to waste a day. Your life has to be meaningful.”

the results are, obviously, so you’ve got to take what you get. The other thing is that everything is not known. I know that it’s 36 repeats, and so I know there isn’t some sort of fine line between 36 and a half and 35 and a half, that it is actually 36 repeats. But I also know that I don’t believe every bit of research that I hear. So I just say to myself, “I’m not even ready to believe that it’s 36; it could be 35; it could be 37.” I’ll just think in the most positive way I can.

I made up my mind that I didn’t want to have anything to do with Huntington’s for a year, so I said to Suzanne and Jason, “Let’s not talk about it every time we see each other. Every three months we’ll just say how are you doing, anything new?” In a year from now, I may go out to the medical library to read the latest research, but not right now. I want to have fun. I also need a break from HD. My Dad and his wife are not well. And the time will come when Karen will need a lot more support from me than she does right now, and I’ll be willing to do that. I’ve also made up my mind that I might have to work longer because I don’t want to give up the chance for disability insurance. Brian has to have a hip replacement operation, and if I get Huntington’s we will need care. It’s a natural process that you might have to put some effort into caring for your parents, but I don’t want our kids to have more than their share of the load.

My gut reaction to the idea of Jason or Suzanne having the predictive test is, “Don’t bother, it’s too yucky...you’re too young.” Obviously they’re going to do what they want to do, and maybe it would be good for them to know one way or the other. But I don’t hear them talking or worrying about it. If the thing’s nagging at you, that’s different. Then you’ve got to do something about it. But if it’s not nagging at you, why bother?

One of the good things about Huntington’s is that it encourages you not to waste a day. Your life has to be meaningful. At school, I think it’s a benefit to the kids I work with because I have an intense need to have an honest relationship with them. I have an intense need to have positive energy in my classroom, and I really encourage them. This need to have what I call “meaningful” rather than “superficial” relationships with my students is good, and I would say that part of it is due to my confrontation with Huntington’s.

I think Brian’s like me. He just made up his mind to look at the positive aspects of it. I need to ask him, though, if it is something that he’s worrying about. To some extent I’m too scared to. The PET scans were all good news, and now I’ve got to deal with something closer to the real thing. The PET results were great. The genetic result was not so great. It is a kind of task to deal with. It is work, this one.

A new direction

Several weeks before her test results, Carla talked about how she learned that Huntington disease was in the family, and how she arrived at the decision to request predictive testing.

I was born in 1952. At the age of six months, I went to live with my aunt and uncle because my mother was not capable of looking after all of us. They raised me, and when I turned 12, I went to live with my father, my sisters and brother.

When I was 15, my mother passed away. My father hadn't explained to us that she had Huntington's. I don't think he really knew she did at that time. She was in a mental hospital for about 12 years before she passed away.

When my Mum married my Dad, some of my aunts and uncles tried to tell him that maybe it wasn't a good marriage, that maybe my Mum wasn't a good candidate for marriage because she just didn't seem to be all there. But my Dad didn't really listen to them. I don't know whether or not he had any inclination as to what was happening, because he'd tend to overlook things and pretend that things were okay.

I didn't find out about Huntington's until I was 30 years old. My aunts and uncles had assumed that we knew that Huntington's was in the family because none of us married, or had children. They figured that this was a choice we had made and so nobody said anything to us. I couldn't ever quite figure out why things were happening the way they were because it just didn't make sense.

When I found out about Huntington's being in the family, I was visiting my mother's sister. We were sitting around the table, and she was telling us about how her father had passed away. He was found hanging in the barn at his farm, and my mother was the one that found him. They don't know whether or not he had Huntington's, but he probably did. When my aunt told us about the family history, I felt kind of relieved because we could carry on and have some answers. I could make some choices about my own life from that point on.

I thought it over for a few years and decided to have a tubal ligation. I didn't want to see children suffering anymore than they had to. It helped to talk to my friend Eva about it. I've always had women friends that I'm really close to. I know I need that. I was always uncomfortable with talking to men about those sort of things. I think it comes from having to kind of deal with my Dad. If I could deal with that, I think my life would probably be a lot easier. But it's really hard for me. He doesn't really change.

“When I found out that you could have a definitive answer, I knew that was the choice I wanted to make.”

One of my sisters has Huntington's. She was diagnosed 10 years ago. It's really interesting, because if you sit my Dad down, and you look him straight in the eye and say, "Nicki has Huntington's." He won't look at you. It's like, "No, she's got something else." It's that kind of denial. So I never even bother to bring it up with him. I talk to him about things that he enjoys like his gardening. I'm sure he's done as best he can for us.

I found out that there was a test three years ago. I came home with some pamphlets and my friend Eva had an article that she found. When I found out that you could have a definitive answer, I knew that was the choice I wanted to make. My family physician recommended me to the clinic, so she knows I'm having it done.

The results will definitely affect my future. I feel they're going to affect my life in terms of my plans for what I'll be doing in the next five years, in terms of my career, in terms of my relationship with my boyfriend. If I do have Huntington's, my lifestyle will definitely change.

I don't find myself thinking about having the disease because I really don't feel I have it, but I do think about how I'm dealing with it. I feel like I'm a little bit unclear at times, and I haven't really talked a lot about it... I feel like those things, those thoughts will just be verified, and cleared up, and I can get on with my life. I feel good about how I'm dealing with it. Very good. Getting outdoors helps a lot. Exercising. Being with my boyfriend, Tom. I've done a lot of great things in my life so it's like now I really want to just stop and smell the roses.

Several weeks later, Carla went to the clinic and received her predictive test results. Her partner Tom went with her. Carla was devastated to learn that she had the gene that causes HD. Several months later she described the emotional impact of this news and her growing concern that she might already be showing signs and symptoms of the disease.

I was devastated. Hearing my test results was like hitting rock bottom. I don't know. I guess it didn't surprise me in one respect. I did feel like there were certain things happening in my life that hadn't ever happened before, yet I wasn't quite sure where they were coming from. I was pretty upset emotionally, and I do have days like that now. Whenever I can't cope with something or I have to tell people and be really clear about something or make a decision, I just kind of fall apart. I get worried because it must be happening.

Tom went to the clinic with me. He drove, and we stopped at the liquor store on the way home. I think it was Valentine's day, or the day after, so we bought a big box of chocolates, and came home and ate it, and drank a bottle of brandy. I was crying. I was pretty upset

for a couple of days. I was supposed to phone people after I received my results and I didn't. I just found I could only phone about one person a day after that, and talk to them, and I would get pretty upset. But then I got stronger. As the healing process took place, I got stronger.

The first person that I called was Eva. She is kind of like my long lost sister that I never really had. She and I . . . we talked a lot about our upbringing, and shared a lot of good times together. She's a very "up" person, very positive. I've had a lot of laughs with her. I think I probably had the most fun in my whole life with Eva. She's having a hard time dealing with this, with me, because she notices some differences. She was pretty upset too. But she was really positive, and she said, "Well don't worry about it. Be optimistic and a lot of things can change. There's a lot of new changes and new developments, so you never know." And she's been really supportive of me, and booked me in to see a Tibetan healer.

The next person I phoned was my girlfriend Pat. She came over that day and we went for a walk. Then I phoned my brother and I spoke with his wife. She was very upset. Very, very upset. She's having a real problem living with Huntington's in the family. It's just been really bleak for her. She made a lot of choices in her life because of this "monster" as she calls it. Then I spoke with my brother. My brother is wearing a lot of hats. Plus, he's living with two kids and a wife who are all very worried about this whole monster of Huntington's being in the family. So he's trying, and he wants to have the test done, but there's no way that he would because he just feels it would change his life too much. He's seeing all of us go through changes.

I spoke with my step—Mum too, and she said, "Ah, hell, it's not any big deal. I mean, we all have problems. We all have weird genes." And I wrote my Dad this long letter and I didn't hear from him for about six months. I ended up phoning him and talking to him myself. He never did really address it. That was before I went down there to visit. It's typical. It's nothing unusual. So I wasn't really overly surprised, but I was hurt. I mean . . . at a time when you need to have support from somebody. Just for somebody to say they care. Just, "How are you doing? Sorry to hear this or that with the results."

Tom couldn't believe that I was phoning all these people and he said, "You shouldn't be doing that," because I was all upset and crying. And I said, "Oh, it's okay. I feel better. At least I've talked to somebody." It felt good. It felt better. I felt like I was getting stronger after each one, because for me it's really important to talk to people. It was therapeutic for me and I needed that. It's good for me to be having that kind of contact with people. That's what my needs are at this time. A lot of friends and family.

“I could only phone about one person a day after that, and talk to them, and I would get pretty upset. But then I got stronger. As the healing process took place, I got stronger.”

I'm definitely feeling some changes. I always used to just get on with life, and have a good time, and put a smile on my face. That's always been my direction in life, and my family sees that's changed. But the physical part of it they don't really see very much, whereas I'm feeling some changes happening in my body, myself. I find my balance isn't as good as it used to be and I have this really kind of twitchy feeling inside, like there's something in me that just wants to keep moving around. It's a really bizarre feeling inside.

It could be that I'm not as strong as I used to be, but I've been walking an hour to three hours a day, sometimes. So I don't know whether I'm going through changes that way, or whether it's because this gene is showing, or the Huntington's is showing its effect on me. I've also noticed a lot of changes in my own management level. I find I'm having problems with doing things like paperwork or balancing my checkbook, and I get really freaked out. I'm quite emotional about it because it's not ever been something I've enjoyed doing, but it never used to bother me. I used to just do it and get on with something else but now it bothers me.

“I'm happy that I've had the test done. I don't have any regrets. It's been painful, but it gives reason to what's been occurring in my life over the last few years.”

Those are things my family noticed. And that I would get upset by things. I notice a big change in my memory because I used to be able to remember people's names, or dog's names, or horse's names, and now it's like I'm stuck. It's like I can't come up with it, and then I get frustrated because I know that I used to know that person's name or that person's kids. It's very frustrating. That part is something I noticed in a big way.

I think it took a lot of courage for me to go ahead and get the test results. It was devastating, but at the same time it wasn't. I definitely think more now about how I am coping on an everyday level. I am more dependent on other people to make choices for me, and I want somebody to just take control. That's nothing unusual though. I've always been that way, but it's definitely very comfortable letting somebody else run my own ship. That's no change for me.

It's been quite painful at times. The truth has been painful for me. And it's had a lot of disadvantages for a lot of my friends. I think that's what kind of hurts me the worst—having to tell my friends

that I have it. It's the pain it causes them. But I'm happy that I've had the test done. I don't have any regrets. It's been painful, but it gives reason to what's been occurring in my life over the last few years. I've had a lot of problems keeping jobs, and when I phoned up some of my ex-employers they said, "That doesn't surprise me. It seems to me you were having some problems when you were here and we were worried about your health." It was feedback that I'd never sort of had before.

People don't usually get to know what's going on inside the heads of people that have it. The doctors are very clinical and they're into that kind of clinical arena. They never really touch the emotional impact or what's happening inside a person's head or heart. I feel like there's a very clinical way that it was treated in terms of receiving the results. I just felt that I was in this very clinical environment, and I just wanted to go and run into the skirts of the nearest woman and cry my eyes out or something. I felt like it was everywhere I looked. It was those white walls. But I think the doctor has done a great job. He's got a support staff that's really very empathetic.

I was in dentistry for 25 years, and in one of the offices my job involved being the support person for all the staff and patients, so a lot of people used to phone me up and talk. Whenever there was some concern or somebody had not had a good experience, I'd always phone them up just to check and make sure that they were okay, just so that they knew that there was someone there that they could talk to. A lot of people change when they are in the home environment. They end up telling you things that you might never have known before. So I thought it was great when the genetics counsellor phoned me back two days after my results session, but I could have used that everyday. "How are you doing?" "It's okay, we're thinking about you."

Right now I'm not working at all, so I'm thinking about getting income assistance or something like that. And that's hard for me. I've always worked. It's very humiliating for me. I need something to do so I've been thinking about going to the Huntington Society and doing some volunteer work. I have also made an appointment to go back out to the clinic and have the neurological tests done. I've seen changes, and that's why I decided to have the genetic test done originally, and I want to have the follow-up. I just want to get on with the next part of it. So I'm ready to have the next set of results done.

Nearly two years later, Carla had found a new sense of direction in her life.

For me the whole experience has been kind of like a gift. I've taken everyday and made that day special for myself.

“For me the whole experience has been kind of like a gift. I’ve taken everyday and made that day special for myself.”

One of the things that I did was I left the man I was with living with. I wasn't really getting a lot of support from that relationship. I just felt like I was needing to be in an environment where I'm supported, and healthy, and happy. I was getting quite pulled down. He was very negative and very upsetting to me, telling me things like, "You deserve to have Huntington's," and, "You deserve to be in the position that you're in because you're not really doing anything to help yourself." So that's when I started to go and have some counselling. The counsellor kept saying, "Carla, you don't have a problem. It's not your problem. You don't have to keep all the fish that are swimming in the sea, you know, you can throw the odd one back." So I said, "Okay fine."

I also saw a Tibetan healer, and he said, "Well, you know, it's one thing to have whatever it is you have. That's okay but you shouldn't be afraid. You should be accepting it. You should be enjoying your life, not running away from people. If you're not getting the support, then change it."

So I gave up the man and I bought a dog. She and I walk almost everyday. We'll walk eight or nine miles a day sometimes. We go hiking in the woods or around the sea wall a few times. I talk to her and tell her everything that's going on, and she's pretty perceptive about what's going on in my mind. So I've moved on and moved through that relationship, and I feel a lot better for it. I have enjoyed the changes that I've arranged for myself. I just feel like I've done a wonderful job of making changes that are good for me. I have placed myself at the top of the ladder.

I had always wanted to go to Africa, so I ended up buying a ticket as a treat for myself. Many people in my support group were very supportive of me and said, "Yeah, you can do it, you'll have so much fun." So I ended going on a five month overland trip that went from Cairo to Capetown in Africa. And that was fun. I really enjoyed it. There were 20 or 30 people on the trip with me, and that was nice because there were always people to go out with. I think that's what I find the hardest with the Huntington's: it's not having a network of people and not having people to work with. When you're working you have that whole circle of people. I've been in touch with a lot of my friends, but everyone is so busy with their lives.

But I'm getting better and healing on another level. When I returned from Africa, I met a new friend at one of the support groups. She was diagnosed with HD last summer. Both of us have been sharing a lot of concerns that we have in dealing with Huntington's and the world. A lot of times there's a lot of shock out there. I find that once people find out that you have some sort of medical concern, they treat you totally differently. They back off. They really do.

People just need to be treated with some normal respect. I remember going a couple of times with my sister when she went to see her neurologist. And he was very cold. He made her feel awful, so edgy. The next time they wanted her to go in to see him, she wouldn't go. I don't blame her. When I went with her to see the neurologist he kept looking at me and said, "Oh, so you've been diagnosed. Oh, well I don't see any symptoms there." And then he said, "I guess you'd better go home and get ready for the almighty hell to hit." I just looked at him and said, "No, I'm going to get ready for my life. I'm going to start living."

When I had the test done, I knew I'd be taking a chance, but I really didn't feel like I had the symptoms yet and I still feel the same. But now I'm not working and then I was, so that's been the big impact for me really, the lack of work. But I've found other ways to fulfill my life, and I have a new direction. You've got to do what you do in your life if you're going to do it. There's no point in doing it half-heartedly. You make the best out of what you've got. That's what the experience has been for me.

Part III

Now that I know...

The stories in **Part III** offer personal reflections on predictive testing from the perspectives of five persons who learned their test results some time within the last decade. Four stories were adopted from a collection titled “*Personal Experiences with Predictive Testing in Huntington’s Disease*” compiled by the Southern Alberta Chapter of the Huntington Society in 1996 while a fifth was selected from the submissions to *Horizon*.

All five stories offer retrospective accounts of the experience of predictive testing. This retrospective point of view allows the reader to understand how people looking back on the experience of predictive testing continue to assess the meaning and significance of their test results.

“I will not have any sort of “assassin” sneaking up on me, if I can help it.”

I won't be ambushed

I had the test in 1994, after my elderly mother was diagnosed with HD.

I wanted to have genetic testing and learn the results, mainly because of a general curiosity and a desire to learn whatever I can about myself and my life. I fear ignorance, not knowledge.

I wanted to be able to inform my children whether they might also be at risk, so that they could decide whether or not to have their own tests, or plan families.

I wanted to be able to plan as far ahead as possible in order to be able to do what I could to provide for my wife, and to make appropriate financial plans and make certain decisions about existing life insurance.

I will not have any sort of “assassin” sneaking up on me, if I can help it. In the end, I may lose the battle, but at least I won't be ambushed by my own ignorance.

When my test came back positive, I had the benefit of excellent counselling, and the battery of neurological tests done at that time have provided a benchmark that will help me and my doctors to monitor the progress of my condition. What I have learned has generated a curiosity about the disease, and a corresponding interest in the remarkable efforts being made by HD researchers. Encouraging developments I would probably have been unaware of if I had been unwilling to take genetic testing and learn the results.

I have found all of this very reassuring. I would urge all those who are vacillating to have the test. Even if the results are not favourable, I believe you can recover your equilibrium and make some positive use of the information about yourself to aid in your own future plans or treatment, or to plan for loved ones. The courage to face the truth, whatever it is, will give you strength to deal with the rest of your life more fully and effectively.

Uncertainty . . .

As a little boy, I remember my grandmother only as a figure confined to her bed, then a nursing home. She was sick, but specifics were never discussed. Four years after I married, on a family visit in 1979, I was told about the “family secret,” Huntington disease. No one must know — people will be fired from jobs; the family name will become synonymous with “crazy;” future prospects for the children will be non-existent. I remember my mother telling me that she did not have it. But I somehow instinctively knew she did. And as I watched her for the next number

of days, I could see mannerisms and movements that were strange. Her sister, who we visited, had carbon-copy mannerisms as well.

My wife and I had a good cry that night. I look back now and wonder why — was it because of what was going to happen to me and us, or was it because of the uncertainty? There was no way to be tested back then.

When I returned to work after vacation, I immediately discussed the condition with my boss. I would not live in the “dark ages attitude” that my relatives did. I decided that I needed to know if my possible future health risk would actually risk my current job. Since they said absolutely not, that was one worry out of the way, but looking back I suppose this risk-taking indicated that I wanted to know what all the facts were, good or bad, to better my life and to be in control. I didn’t like surprises of that nature.

As my wife and I reflect back to those years before testing, we realize that a wall built up between us, a fear of getting too close. Though our relationship was fine most of the time, the uncertainty every once in a while would beat down the mutual growth that two people require. This was a subtle thing, and not truly apparent at the time. We were also looking at our finances differently. Rather than put money into our house, we took trips. We wanted to have our “retirement” now, in case. “In case.” Two words, often unspoken, that cover us like a family heirloom.

**“‘In case.’ Two words, often unspoken,
that cover us like a family heirloom.”**

When predictive testing became available, I immediately wanted to know. But because many of my relatives were gone, however, I had to wait for the genetic testing in 1993. I found that preliminary meetings with the medical team were essential for the support and information that they gave me. I felt prepared on both a technical and spiritual level for the answer that we had been waiting for after all those years.

My results came back negative. I did not carry the HD gene. Another good cry followed. I remember standing there with my wife, hugging each other, saying, “I’m free, I’m free,” over and over. I don’t think we realized how much of a relief it would be. I don’t know how we would have reacted if bad news had been in the cards, but somehow I suspect we were always thinking HD was there. We assumed the worst without ever saying so.

I think we telephoned and wrote everyone in the world who knew us, and it surprised me how much our concern had been on the minds of so many others. Though I never hid it from my relatives

and friends, I didn't realize how much the uncertainty had affected them as well.

So it is now two years after the test. My wife and I have grown closer, and though we kept children out of the equation (in case), we talk more about our lives together, and especially about future plans, with far more conviction than before. Our house, which I often saw as a place in space, became a home to me, our garden more interesting, and yard work less painful. Though we had decided long ago to pay off our house and other debts before I'd be incapable of working (in case), we now are doing it with a relaxed attitude, taking life in and thinking the sooner the better, not the sooner we're safer. I guess we feel in control of our fate now. No one can be certain what the future holds, but now we know what it doesn't hold, and we can both deal with that.

Alone...

Learning of an Inherited Disease. It has been almost a year since I learned that I'd inherited the abnormal gene that causes Huntington disease. In hindsight, I can say that if I'd known a year ago the way I was going to feel today, I probably wouldn't have chosen to learn the results of the test. At the same time, I should also say that a year ago I would not have accepted an explanation of the risks-of-knowing.

Depression. I always thought that knowing whether or not I had inherited the abnormal gene would only have positives associated with it. On the one hand, if I was certain that I didn't have the gene then I could go about my life normally. I had forgone nothing (except perhaps having a family). On the other hand, I thought that knowing I had the trait would allow me to better plan what would likely be a shortened life. All of this put the most positive spin possible on learning the results of the test — either a small cloud would go away forever, or the small cloud would be there tomorrow. For me, the reality has been that the small cloud has become much larger and much darker, and has cast a shadow over everything.

I have not wanted to tell anyone. I don't want to burden family and friends with this. In addition, the fewer people who know, the less chance of someone else knowing. If I had a spouse, obviously that's one person I would have told.

In fact, I did tell one person. About the time I learned, I was going through a difficult period with a significant other of several years. At this time we weren't seeing one another, but we weren't seeing anyone else, either. Reconciliation was still an option, but only if we both wanted it. Both of our lives were on hold. It seemed to me that disclosure was the right thing to do.

Loss of Self-esteem. This has manifested itself in the way I interact with individuals of the opposite sex to whom I feel attracted. At some point during dating, certainly by the time the relationship became “serious,” I would have to inform the other person of the diagnosis. Although this hasn’t happened yet, I do feel the other person would not want to be involved with someone with this diagnosis. This has resulted in a decreased sense of self-worth. Also, a concern about being completely alone at some point in my life.

Feeling of “What’s the point?” There have been numerous times in the last year that I’ve found myself thinking, “Well, what’s the point of ...” For example, I have been a 90% vegetarian, and I eat low-fat food. I have found myself eating much more junk-food, meat, etc. Predictably my weight has gone up. There have been times I’ve asked myself, “Is there any point in doing something about it?” This isn’t a behaviour I’ve exhibited before.

Changed Priorities. The most significant change I have made in my life is changing the priorities of the things I intended to do in the 10 years either side of “normal” retirement. I find myself placing the highest priority on the activities that have the most personal satisfaction. I had planned to do humanitarian work for the first few years of retirement. I have decided this is something I want to do now. People have tended to view this change with incredulity; obviously, because they don’t know the reason why I’m doing it.

Living With the Symptoms. I have no memory of seeing anyone with the symptoms, so I don’t really know what the symptoms look like. However, I do know that unlike many motor-diseases (e.g. ALS), where thinking seems to be unaffected (exemplified by Stephen Hawking), this is said not to be the case with HD. I find myself wondering whether I would have the desire to live with the symptoms.

Also, I’ve gone from viewing myself as primarily “well” to “not well.” I wonder, and I have concern, that others would view me the same way if I told them.

“I’ve gone from viewing myself as primarily ‘well’ to ‘not well.’ I wonder, and I have concern, that others would view me the same way if I told them.”

Sometimes I feel cheated

There are seven children in my family, and I am the youngest. When I was younger and living at home, I think that me and my next oldest brother experienced the worst of my father’s behaviour. Having to deal with the mood swings is a very difficult thing to do, as well as watching somebody slowly rot away to nothing. I never had the same experiences as my older siblings. They remember a father who took them camping, who took them for family holidays, and all the normal father stuff. I was taking care of my father who was sick and could not do all the fatherly things one would expect of

“I wanted to make sure that if I tested positive for the disease that I would change my life goals accordingly, and try to live the best I can.”

him. In fact, most of the time, I had to watch over him to make sure that he was safe: safe from choking on his own food; or safe from falling off his chair; or safe walking down stairs. I had to cook for him and help feed him. It is not easy work, and I realized that I would never want to go through the same experiences in my life. And yet that is what faces me now. I too must feel the pain and the suffering that my father did.

My father was about 40 when the doctors found out what was wrong with him. He had already had seven kids and on his way to having ten grandchildren. He was completely unaware of what was happening to him. All of a sudden his life changed and there was nothing he could do about it. It was too late to do all the things that he might have wanted to do. That's when I decided that I wanted to know what was going to happen to me ahead of time. I wanted to make sure that if I tested positive for the disease that I would change my life goals accordingly, and try to live the best I can.

I waited until I was 25 and then I got myself tested for Huntington's. Since I left home when I was 18, I worked really hard at putting what could be ahead for me way back in my mind. I went to university and college and graduated. But as I got older, I began to think more about longer term goals, and of course you would have to think about Huntington's too. When I began the testing program I was prepared for being tested positive for the gene. It is difficult to go through your regular workday as if it's just another day. But, it is far from another day. I think that having a handful of friends and family that know you are going to be tested can be helpful when you need to talk about how you are feeling. Going through the sessions with the counsellors and the psychiatrist really helps you vocalize your feelings for the test. This also can strengthen or weaken your ambition to continue with the test. Waiting is one of my least favourite things to do. From the last part of the test, when you say that you are going to get the blood work done, until the results day is a very long wait. It can preoccupy your thoughts a lot. But the day of the results is a very emotional day. When I heard my results were positive I never cried but I was completely numb. Some how I waited all these years for this answer and now it was true.

As time goes by the reality begins to sink in as to what this means for my life. Now there is not a day that goes by that I don't think of how I feel. Sometimes I feel cheated out of a fuller, richer life. It hurts when you hear your friends talk about what we would be like when we retire and you know that it won't be happening for you. I also feel sad when I think about getting a mortgage for a house and realize that I won't be healthy or living at home at the end of the 25 year mortgage. Some days I can get depressed quite easily, and it can stay for a few days each time. Certainly all the things like your job, financial problems (especially planning for the future), family

and all the other stuff sure can affect the way that you feel. I try as best as I can to bring myself out of it, but it is not always easy. Certainly having people to talk to who understand your feelings is the best thing to have.

I have gone on more holidays and made my goal to enjoy these days that I have now. I am not sure how long I have before I get sick. That scares me a lot. Am I going to have ten years, five years or less? It's hard to say. The other hard thing is that I just started a new job, so I worry about the right time to tell them. Do I wait until it is very noticeable, or do I tell them ahead of time and risk my job? For now, I think that I will just leave it for a later time.

Having someone special in your life can really help when you go through the testing. It helps to talk to them first to understand why you want the test to be done and how it will affect your relationship. This can also help them understand what the future may hold for the both of you. You can support one another when things start to get difficult, and that makes a big difference.

Never give up hoping . . .

When my second child was born, my body jerked and twisted for three days. I felt a sense of dread overcome the joy of our healthy newborn. The movement lessened to twitches and grimaces, which I felt I was successfully masking. I watched in certainty as my father developed Huntington disease. I prayed for, "Just five more good years, please Lord," and made sure we would have no more children. My handwriting deteriorated, I refused to eat certain foods in public for fear of disgracing myself. I was so afraid of losing control, I would never let myself relax. When I would make an error at work, I wondered, "How long will I be able to continue?" I sewed Halloween costumes, read stories out loud, and baked cookies because I wanted my children to remember their mother for something. My second child was now twelve years old.

In autumn of 1993, my youngest sister was sure she was developing symptoms and inquired about predictive testing. She could go no further until we had an immediate member of our family confirmed. She was willing to try matrimony again, but would not if she had HD. How to have Dad confirmed? His own doctor was no help. His assessment of a little Parkinson's, or perhaps a bit of forgetfulness was more than exasperating. I engineered a referral to a Movement Disorder Clinic, only to discover that the specialist was proficient in Alzheimer's Disease! We tried again. I had told my father that I wanted to have predictive testing and would he submit to an examination and blood test? He asked why the doctor wouldn't believe him? Anyway, his blood had been stored in Vancouver for fifteen years! My sister tested negative and we rejoiced at her wedding in the summer of 1994.

I admired her resolve and decided to go ahead with predictive testing for myself. In the meantime, my father was visiting the Huntington's clinic monthly and was testing a trial drug. He had another minor car accident, and was warned by the police, but continued to drive. The doctor at the clinic gave him the ultimatum to stop driving before "someone" rather than "something" was his victim. He had been driving for 55 years, but the doctor's reply, "Yes, but you haven't had Huntington's for that long," silenced him. I felt there were some days when I should not be driving and worried that I would be at fault in an accident.

Suddenly, after 30 years of watching Huntington's affect our family, I HAD to know. My children had watched their grandfather and would remember the staggering, the difficulty at mealtime, the difficult speech. They looked at me, and I wondered at what stage was I, and how long would it be?

“Only my husband and two sisters knew I was undergoing testing. I felt my having HD was going to be harder for the people around me; I would be coping well in my own world, thank you.”

My husband and I discussed all recent developments. . . the hope of treatment and cure. My children would be able to choose for the next generation, and Huntington's could be obsolete in their generation. I would support their decision. I felt if I had to be alive on life support systems, and there was no quality of life, I would not want to continue. Even if there was a cure or new treatment the next day, I would have made a decision based on all the information available at the time. We had been through this process twelve years ago before I decided not to risk any more children. I was in good health, working part time, volunteering in my church and community, and felt that I had indeed been given, "Five more good years, thank you, Lord." I wanted my husband to divorce me when I was in institutional care so that he could carry on with life, but he refused to discuss that. Only my husband and two sisters knew I was undergoing testing. I felt my having HD was going to be harder for the people around me; I would be coping well in my own world, thank you.

We were unsure of our finances but we had survived business catastrophes, job loss, career changes and moving halfway across Canada twice. We would cope as we had before. My husband had life and disability insurance, the mortgage on the house would be paid

in a few years. The children were older than I was when my mother died, and I remembered that throughout my whole life, God has surrounded me with wonderful Christians that enrich my life. There wasn't anything else we couldn't handle together with faith. I was ready to have the test results.

I wasn't ready for my father to take his own life. I never believed he could consider death as an option. For him there wouldn't be any tomorrows to go fishing or visit his family or enjoy apple cobbler. He did not want to be dependent. Many years ago he told me that he would never leave or sell his house, "They will have to carry me out." Those words took on new significance. During my counselling for predictive testing I realized that my father had reached a point of no return. He was never going to get any better, and he made his decision. Deep inside of me, I felt responsible for his choice. I had been selfish wanting to know about HD for myself and inconsiderate of my father, who knew and was still trying to live a normal life. In everything he was optimistic, but I could not see past the disease. He knew what was ahead for him, and for us, and made his decision. I knew it could never be my decision. My 12 year old child was worried that I would do the same.

A second sister was now undergoing predictive testing some distance from me. She was having panic attacks in the shower and hyperventilating. I approached my results appointment with finality — now I would know my stage of Huntington's. If I had incurable cancer I would want to know. My cup had always been half empty, now I became aware that it was approaching overflowing!

During the writing of this letter I have gone through the whole process again and I still wouldn't change my decision. Counselling was the most important part of this process. Visiting two hospitals for my appointments and seeing everyone else's problems reminded me of all my blessings. "Life is uncertain, eat dessert first!" seems a flippant remark, but I ALWAYS check the dessert menu before ordering! My right knee always tells me what the weatherman didn't predict, and my children don't hesitate to tell me that they REALLY did learn everything in kindergarten. Life is uncertain, but I will live it one day at a time.

One Christmas after my mother died, there was a very large box beside the tree with my name on it. On Christmas Day I opened it and discovered a nightgown inside. My face must have registered disappointment because the watch I coveted was not inside. I moved to open another gift. Daddy was astounded that I did not empty the box! So I dug a little deeper and found a small package. It was a velvet watch box! Shrieks of joy turned silent when I found it contained a small card that said, "Better luck next time!" Another disappointment! "Don't give up!" my father urged me. Another package was a dressing gown to match the nightgown. I felt that

was a mean trick for Christmas, especially when a small bottle filled with tacks was labelled “Official Noisemaker.” I really had emptied the box. “Never give up hoping!” Daddy was emphatic. Among the crumpled newspapers used was a crumpled ball of holiday wrap. I smoothed it out and there was my watch! In those 10 minutes I had hit all the highs and lows of my 15 years. Daddy kept urging me on, “Never give up hoping!”

Children don’t always remember what their parents tell them. I tried to forget some of the things my father said as his HD progressed. “Never give up hope!” came flashing back to me as I was waiting for my results of predictive testing. For me there was no “downside.” I was already convinced that I had passed the HD gene on to my children. They would be the ones to know there was treatment and hopefully a cure. Watching them and wondering ... if? More often, the question was “When?”

I do not have Huntington disease. Six weeks later my sister phoned to tell me joyously we would grow old together. She also tested negative; we three sisters had all tested negative! Our father gave us all his love. I wish he could have known he gave us life.

Reflections on Reading the Stories

Predictive testing creates a choice for individuals at risk for HD to learn information about their future. Although the stories included in this booklet are wrapped in the experience of predictive testing, what seems to bind them lies beyond this process. These stories reflect the attempts of thoughtful people making meaning of their world and making decisions that carve the pathways through their lives.

Whether one decided to test or decided against testing, the stories spoke of people being engaged in a *self-reflective process*. Part of this process resulted in an awareness of self that lay within the context of family and family history, “My maternal grandmother, at approximately age 65, started having a limited amount of chorea and my grandfather spoke of her personality shifting. I was an adolescent at the time and she was just old and Grandma.”

There were stories that revealed *courage*. “When you have the possibility of something like this you need to look into it and know as much as you can about the disease. You have to come to terms with the reality in your life. All of a sudden things that you’ve been believing all your life go out the window.”

There were stories that revealed a *questioning of the significance of predictive testing*. “I cannot think that either knowing I have or have not inherited the gene for Huntington’s would change the way we live and are raising our children.”

There were also stories that spoke of the strain caused by knowledge of the possibility of having the gene for HD. “As my wife and I reflect back to those years before testing, we realize that a wall built up between us, a fear of getting too close. Though our relationship was fine most of the time, the uncertainty every once in awhile would beat down the mutual growth that two people require.”

Additionally, there were stories that reflected experiences of *enhancement of life*. “One of the good things about Huntington’s is that it encourages you not to waste a day. Your life has to be meaningful . . . This need to have what I call meaningful rather than superficial relationships with my students is good, and I would say that part of it is due to my confrontation with Huntington’s.”

A benefit of these varied and personal reflections is that they may assist people at risk for HD in making sense of their own world. These stories demonstrate that the experience of predictive testing is a personal and individual one. Yet, each story provides potential strategies for understanding and making meaning of the possible impact of predictive testing.

Each story provides potential strategies for understanding the possible impact of predictive testing.

The diversity of insights in this booklet helps to address the concern for different perspectives. “To anyone who is at risk of HD, I would like to say that a different perspective is needed, one that will accommodate a challenge to change and a challenge to meet the needs of a different stage in your life, and to do the best you can with what you have.” These personal stories, coupled with people’s ability to reveal themselves to the reader, provide an opportunity for us all to begin to understand and develop a new awareness of the possible impact of genetic testing in the lives of people at-risk for HD and their families.

Jessica Easton
Centre for Applied Ethics
The University of British Columbia
jessica@easton-snelgrove.com

Appendix 1: Genetic Testing

Genetic Testing (before the 1993 discovery of the gene that causes HD)

In 1983, genetic markers closely linked to the Huntington disease (HD) gene were identified. This discovery, together with the identification of additional genetic markers, led to the development of predictive testing programs for HD.

Blood was taken from the person requesting the test, as well as from several family members – those known to be affected by the disease, and others unaffected by HD. Geneticists then “read” the DNA patterns for the affected and unaffected members of the family to try and establish a pattern unique to the affected members. The results of these comparisons were put through a computer program to calculate the probability that the person requesting the testing was carrying the gene (e.g. “You have an 80% chance of developing Huntington disease”).

Genetic Testing (after the 1993 discovery of the gene that causes HD)

In March, 1993, the gene causing Huntington disease was identified. This means that individuals at risk for HD can now be directly tested to “predict” who will develop this disease. The same test may be used to confirm the presence of the gene that causes HD in individuals already exhibiting clinical symptoms of the disease, especially when the family history is sketchy or unknown.

Because it is now known what gene causes HD, this genetic test is definitive. If you have the gene that causes HD, and if you live long enough, you will develop symptoms. If you don't have the gene that causes HD, you won't develop symptoms.

Additional Notes

Predictive testing refers to the use of an HD gene test in a person who has no symptoms but wants to know whether or not he/she carries the gene that causes HD.

A genetic test can also be used following a diagnosis for HD by a neurologist, as a way of confirming the original diagnosis.

A PET (positron emission tomography) scan uses special radioisotopes to obtain images of the brain's inner structures and information about their function. A substance injected into the bloodstream works its way into the brain structures to measure activity in the brain. In Huntington's, PET scans help researchers identify specific areas of the brain where little or no activity is occurring (thus indicating areas that have been affected by the disease). Typically, PET scans are used for research only, most recently as part of the PREDICT-HD study being run by the Huntington Study Group.

For more information about genetic testing, including specific information about the gene that causes HD, the science relating to how the test is done, as well as the testing process, please refer to the Huntington Society of Canada's *Genetic Testing for Huntington Disease* booklet.

Resources

For additional copies of this or other booklets or information, please contact us (see information on back cover).

The Society has a network of Resource Centres, staffed by trained, professional social workers in locations across the country. The Society's national office will be happy to provide local names and telephone numbers if you are having difficulty locating them.

Contact Us

Huntington Society of Canada

151 Frederick St., Suite 400

Kitchener, ON

N2H2M2

phone: 1-800-998-7398

phone: (519) 749-7063

fax: (519) 749-8965

email: info@huntingtonsociety.ca

website: www.huntingtonsociety.ca

