

Chapter 1

Family experiences: Part I, Diagnosis and early stages

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Chapters 1 and 2 are based on real accounts from families and are extremely valuable for setting the clinical and scientific chapters in context. We chose to publish them with minimal editing. Whilst the perceptions of the writers are real, where others are mentioned it has to be borne in mind that the perceptions of these third parties are not known.

The rightful authors of the first two chapters of this book are the families who have made a huge personal sacrifice to allow us to share, with at times such complete honesty, their challenges. These first two chapters give the reader some insight into the impact that juvenile Huntington's disease (JHD) has on the whole family over a long period of time. Families feel that contributing to this book has given them a great opportunity to open people's eyes, minds, and hearts to try to get across the impact this devastating disease has on them.

Families also speak about those whom they have had contact with throughout their journey with JHD. In many cases families talk about when this contact has been a positive experience for them and how much they have been helped by other people, and in some cases less so. By picking this book up, the reader is already someone who is interested and cares enough to do so. These chapters are important for all readers, to help put later clinical and scientific chapters into context, and we are sure that everyone will gain something by reading them.

We begin this book by hearing from those whose lives have been so irrevocably and relentlessly altered by JHD; the young people with JHD and their families who care for them. It is perhaps a testimony to the challenges young people face living with JHD that their contribution to

this book is given only by one young man. Columb was 26 years old when he died as a consequence of JHD in 2006.

Columb's contribution

My name is Columb. I'm 23 years old.

I can't feed myself anymore because my hands shake and jerk, I can't feed myself and I also can't walk very far anymore.

I used to play rugby; I used to love playing rugby. I got a trial for my country's under 18's but I didn't get in. I used to play prop, I used to love tackling people in rugby.

I can't go out anymore because I don't know anybody of my own age. I go out with my mum and dad. I go to the pub with my Mum and Dad every Friday night and I take Stroma [Columb's dog] with us as well.

I have a scooter that I use and I go down to town. I go into the town and into the cafés and I have a Coke.

On Monday I like going to the walled garden which is in the local hospital. I do reception work there. I work in the shop/café which is in the garden and I also do lunch there as well. Wednesday and Friday I go to another centre. I do art on Wednesday morning and I do music on Monday and Friday and I do art again on Friday.

As I said before I can't walk very far, which makes me very angry; and, as I said before, I can't feed myself, which also makes me feel very angry.

I have lost all my friends, but if they came to see me I would ask them to just give me a chance, like just give me a chance, just to see how I get on with, how I get on with just meeting up with other people.

Columb

The remaining contributions are from the parents and one sibling of children and young people living with JHD. We have chosen to divide these contributions into those that describe the earlier stages, including experiences of diagnosis, and those that describe the later stages. Many families living with JHD describe facing each challenge as it is presented to them, and not looking too far ahead. We hope that by dividing the two chapters as described readers can be better prepared for the areas that they may wish to find out more about. Those contributions that talk about the later stages have been put together in Chapter 2.

Contribution 1

As a mother my worst fear has always been to lose a child. I recall years ago our community was holding a fundraiser to help a young child with a brain tumour; the fear and heartache I felt just imagining how difficult that would be was almost too much. At that time I did not know that young children could have early onset Huntington's disease. It was my misunderstanding that only girls could have this disease and it could not skip a generation.¹ What a devastating mistruth that was indeed!

I find it sad that secrecy about Huntington's disease ever needed to exist. My husband didn't even know that he was at risk until I learned this truth, at a genetics clinic, and shared it with him. I was expecting our second child at this time, my third. I naively told myself that this was why he was having so many changes in personality and behaviours. After all, how could he be OK with this news when he was watching his mother slowly die from this disease?

It was watching and hearing about the abuse to which my mother in law was subjected by people in our community, their lack of understanding and the family's inability to care for her due to a variety of reasons. Not to mention the difficulties her children faced while other kids mocked her for always looking drunk and changing clothes on the street and all her different behaviours from the norm. This is when I decided I would never keep the secrets of Huntington's disease from my own children. I encountered resistance on many occasions by my husband and others around me but I am confident today it was best for my family in the whole picture of our lives that I was inclined to be open about this family disease.

I all too often retreated from the thought and fear that one day this could be my husband's fate that I could lose him at all too early of an age and that my adult children could potentially face this fact as well. I tried to close my eyes tight to the point that when I was expecting our last child I didn't go to the doctor until I was a week shy of 3 months pregnant. This was a serious time of true soul-searching for me. By this time in our lives my husband's behaviors were quite erratic and frightening to me at times. He was becoming a different person and growing sterner with the girls, my eldest in particular. I still held tight to the thought this was due to his own fear of this disease and having another child only enhanced this for him.

That year, while we were expecting Isobel and shortly after her birth, there was a lot of heartache within my husband's family. Chuck's youngest brother was killed in a car accident, his mother passed away with late stage complications from HD, his oldest brother whom we all adored as well committed suicide in the building in our backyard one day just prior to the school bus dropping off our girls; he had recently had his wife leave him and was diagnosed with HD. Isobel was a very welcome love and ray of sunshine in the midst of all the turmoil.

I recall my husband's cousin telling me while I was expecting Isobel that my husband had HD. I was quite angry that she would imply such a harsh statement. I thought he was fine. Well, other than his recent losses at that time and excessive drinking, that is.

It was shortly before Chuck lost his job that he shared with me that he was afraid he was showing symptoms of HD. He shared with me that it was getting far too difficult to do simple nuts and bolts and he was having numerous accidents at work. He even cut off his thumb in the running lawn mower blade. His temper was quite frightening to me at this time as well, I never could do anything right in his eyes and often thought of just leaving him. I was, however, too afraid and lacked confidence, not to mention I took my vows very seriously.

By the time Isobel, our youngest, was getting ready to begin school I was working full time as sole support to my family. Isobel was 4 years old when I began to see something changed in her appearance. She would hold her left side stiff and postured when she concentrated. By the time Isobel turned 5 the school was having issues with her behaviour and noticing what I was seeing as well. This was the time we began to go from doctor to doctor seeking an answer. I heard autism, dystonia, small strokes, Friedreich's ataxia; you name it, they sought it. One day we met a paediatric neurologist and within 15 minutes I was told juvenile Huntington's disease. I had never heard this before. Kids don't get HD, even our family doctor stated this along with several others, so how could that be?

I found myself trying to choose a diagnosis, anything but a terminal one! Despite all my attempts Isobel was still losing her ability to walk, jump, hop, speak; she required constant supervision. The only way to help her was to test her. My husband was not tested at this time; he did not wish to know his fate.

This was the beginning of many times I have been placed between my husband and our children. In the end Isobel was tested. It was 4th February, after her sixth birthday, that I heard the words, 'beyond a doubt, Isobel does have juvenile onset Huntington's disease'. I recall wishing the world would stop right there. I cried, I prayed, I talked to several people, and then I decided to stand back up and help my family, help my child and husband, to redefine itself. This wasn't the end; it was a new beginning for all of us. Broken family ties due to this disease were beginning to be restored. Since this time, family ties that were strong have crumbled.

Our sweet little Isobel, who did everything early, even spoke sentences by her first birthday, knew simple sign language at age four, was learning a second language by age five, was now disappearing one piece at a time. My husband, who was once the world to us all, had become paranoid, angry, unmotivated, and restless. Yet I still could not realize fully what my family was facing. I don't think it's possible to know until you walk those steps.

We have had some incredible special beautiful moments and some terrifying painful times also. I try to hold on to the positive things and not look too far ahead or back so that the fear and pain doesn't consume my family. We have been blessed to meet

many wonderful people who have been able to look beyond the terminal diagnosis of HD and look at how can their lives be best and most comfortable; how can they be their best person in spite of this monstrous disease?

I know now that with juvenile Huntington's disease, just because there is a change it doesn't mean that change is permanent. Our kids can decline and improve skills equally as quickly. Finding the cause of the discomfort and the best way to treat the issue so as to keep them comfortable and offer them the best quality of life is our main focus now. Not focusing on what could be rather what is and what is realistic to achieve for them each individually; having the least regrets.

Katie, our then 10-year-old, reminded me that it's never safe to say never and it cannot get any larger than it is. Our sweet compassionate quiet Katie, who talked to angels, and loved people and the world around her in such an innocent unconditional way, and loved to learn and did it so well, was now beginning to change.

I noticed the summer before her 11th birthday Katie had begun to slow down. She raised her voice for the first time to me. Katie began this odd walk, step, step, shuffle, shuffle. This loveable little girl for no reason one evening took the rubber guard from the vacuum and began to whip me with it as she laughed. My heart sank and I was fearing. I was looking back at what we just went through with Chuck and Isobel. Doctors and family didn't want to see this. I kept saying, 'I'm her mother. I of all people wouldn't wish this on any of my children'.

It was not until Katie was almost 12 that she too was tested. Once again we needed to know in order to help Katie be her best person. It was the day following her 12th birthday, and once again I heard the words, 'Katie is definitely showing early onset of Huntington's disease'. They say God doesn't give you more than you can handle. I don't believe God gave this to them. In the midst of all those who are around us and love us it's still so easy all too often to feel so alone and afraid.

I have heard so many misunderstanding speculations about my family members that I wish could be taken back. However, it takes educating others and an open ear to change this. Not everyone has been able to get close enough. Often they are overwhelmed by our story; possibly facing our own mortality is something we fear?

Thoughts like 'these kids are just spoiled and lazy' can make feeling validated, understood, or accepted difficult for our children. They have a difficult time processing and are very rigid in their thinking. *JHD kids don't adapt to the world around them; at some point in progression we have to adapt the world to meet their needs.* Not all medications are safe or effective, so what's best for them to be comfortable and be their best person as long as possible is still the goal for all of us.

Often others would see my husband as a bad, or mean, selfish person, because when he isn't in a managed care situation he cannot control his anger and responses. His thinking also becomes more narrow-scoped as Huntington's take over and processing becomes more difficult. I'd be lying if I said there was a time I didn't understand this myself. Imagine for a moment being a dad, brother,

son, husband and losing all your independent skills; to still love your family and know this is happening, losing control over your own body and world around you. Huntington's has created a different way for their entire body to function. My husband overproduces adrenalin which creates a physical response of aggression in everyday situations that we wouldn't have ourselves.² Isobel has this issue as well, but it is expressed as fear or anxiety; both irrational to us. However, our brains regulate the body as a whole and their messages are often jumbled up as their brains atrophy and go through chemical changes.

It has been 2 years since Katie was diagnosed and several months ago I needed to place my husband into a nursing home where he could receive that safe, controlled medical care he needs during the end stage of his disease. He still lights up when the girls and I walk in; we can now just love him and enjoy him for the time we have left. Life isn't ideal but we have to do the best with what we are given and I feel successful that I have managed to care for everyone thus far without any harm or injury to them or anyone else. In a world that is created to thinking in a reactive, rehabilitative manner this is not easy.

There have been a lot of moments that Isobel nearly left us this past year. She developed a progressive seizure disorder and recently has had a second feeding tube placed after she became seizure free for the first time since they began over a year ago. JHD seizures are not always typical epileptic seizures in appearance and presentation they have progressively robbed her of so much quality and comfort that not even JHD has imposed on her. I have held her in my arms on more than one occasion when she would convulse severely and her airways would spasm until she quit breathing. She has had multiple feeding issues all creating multiple hospitalizations and she has dropped to as low as 42 pounds in weight.

I have heard several people ask, when will I just let her go? I find that question hard to understand. It's not all about letting go; it's about keeping Isobel comfortable in her own body and helping her to have the best quality of life as long as we are able. She is our little girl, we love her dearly. She still has a world of thoughts to share and she still looks forward to each new day and experience. When she is having such strong chorea that she flops out of a chair or bed and she is beginning to have seizures around the clock, how do we not try to help her? This time, once again, the doctors were successful in restoring Isobel's comfort and quality of life. They not only stopped the seizures, but the chorea is gone and she is being fed and hydrated. For how long we do not know, but we still have today to enjoy one another and that is a gift today. *I appreciate those who are able to say what can we do to help rather than looking at this as sad and hopeless. They are worth more to my family than words could ever express.*

Huntington's disease is cruel but the people who are being ravaged are like every other person; they love, cry, smile, hug, and think like the rest of us. Hope has a new meaning to our family. Without hope there is no life. While many people seek a cure, look for a future, we have hope today that all their suffering will be

minimal and the world that they touch will be a bit better when they leave us so that their struggles aren't in vain; that they will know how much they are loved and not feel abandoned. We all are dying. None of us know what tomorrow holds; when you live with a terminal illness it's just more prevalent in each day.

I believe that being open with the two older girls who are surviving this disease has helped them to be more successful in each choice they make and today they both stay at home with me to take care of our family and home. It always seems much more difficult for those who will not have Huntington's disease; rather, they seem to feel guilt and sadness, sometimes afraid, seeking out why they are spared to go on. Trying to give those girls some feeling of choice and empower them to grow and move on with a somewhat normal life is not always easy. This is in part the most important growing experience that the girls can have, a lesson many people don't have; the opportunity to grow. Each day it's a community effort literally to care for one another.

For those few who believe that our children are conceived by selfish people who do not feel the pain it's a complete mistruth, another which I cannot understand. I have told my girls and husband countless times I'd take their pain, their losses, if I could just see them again to be okay to continue where life began to rob them. Isobel always says 'oh poor mommy, why?' Katie being a teenager just tells me, 'You're weird'. She also says she isn't afraid to die she is 'afraid of the life un-lived'.

We are walking a tightrope between living and dying and they are our teachers! I try to live with as little regret and take each day, sometimes each minute, at a time. *If we look beneath the devastation of Huntington's disease we can see the individuals that they are.* This disease doesn't present itself in one fashion; even within the same family it's as individual as they are. We seldom fit anywhere outside the medical community or Huntington's disease family and some days I find isolation a way of life just for self preservation. I have heard that I am so strong but the truth is it's a strong foundation that holds us up; a foundation of many wonderful compassionate people. And, as the issues grow, days become harder, and decisions are larger, that foundation of community support grows and that is our strength—each other.

Contribution 2

Twelve years ago we had never heard of JHD, even though HD was already a part of our lives at that point. My husband and I had been together since we were teenagers, but we had no idea about HD until we were told about it being in his family while at his grandmother's funeral. We soon figured out that would also be the explanation for all the emotional and physical problems that had been plaguing his mother. Shortly after, I found out I was pregnant with our third child. We had already been blessed with a 3-year-old, a 1-year-old, and his teenage brother (who had moved in and become part of our family after we had to put their mother in a nursing home).

Our story with juvenile Huntington's disease began about 10 years ago. Our oldest child was five and had been having problems with falling. He was a very coordinated child up to that point, yet he was actually injuring himself from the awkward positions he would fall. His doctor referred him to several different paediatric specialists who did all sorts of tests, looking for something orthopaedic wrong. After about a year of tests and still no answers, our doctor sent him to yet another specialist. After examining him and watching him walk, he said this was something neurological and asked if there were any neuro problems in the family. I said 'no, just Huntington's, but that of course is an adult disease'. (Little did I know!) He looked at me and said that I needed to get him to a paediatric neurologist because there is a rare juvenile form of Huntington's. It would be several more stressful years before he was actually diagnosed. We chose not to tell him right away, because the children had been very involved in the care of their grandmother, who had just passed away from HD. Also, my husband did not handle any of this well at all. He is an amazing husband and father, but when it came to this topic, it was off limits. I honestly don't know which was harder to deal with the news of our son having JHD, or the devastated look on my husband's face when I told him. Around the same time, our second child started having problems in school. The teacher was concerned and wanted testing done for learning disorders. It never occurred to me that it might be JHD as well. But, after 2 years of testing and symptoms that started to include motor skills, eye tremors, behaviour, etc., our daughter was diagnosed too.

I have to say that without our Christian faith, I personally would have crawled in a hole long ago! I trust God to take care of things that I can't. I honestly believe things could be much worse and do my best to look at all the wonderful things we are blessed with and how well the children are doing. My hubby has come a long way and now we joke a lot about 'HD things'. I have tried very hard to keep things as 'normal' as possible for the family, while at the same time, acknowledging their struggles.

My experience has been that when you have someone that is supportive (whether it's a doctor, teacher, or even family) that it makes a huge difference in how easy it is to deal with. It seems to me that it is especially hard during the early stages because, while things may seem to be fine to the outside world, you are dealing with so many issues that people who aren't living it just don't understand. Over the years, I have dealt with a few very difficult teachers, doctors, and friends that make it so hard on the kids. Most though, have been so caring and kind that it makes such a difference in the stress level of our family. It has been a process of being comfortable with letting others know about our situation. I really believe that we do have to educate our communities about Huntington's because with education comes understanding, then hopefully support. We stay involved in our local fundraising efforts and it is so fun. It is a balance though because *I don't want HD/JHD to be the definition of our family. We are so much more than that!*

Contribution 3

Like most people I was totally ignorant about Huntington's disease. I learnt about this disease after I got married in the early 1980s, not aware that my husband was carrying the gene and not showing any signs of it at the time. Later on I found out that his maternal side of the family had been affected by Huntington's disease. His mother, grandmother, and three aunts had all died of the disease. In the earlier years of my married life I noticed a change in the behaviour pattern of my husband. The violent and stubbornness side of it could not be explained or understood by close family members including myself. He was finally diagnosed as suffering from the disease. At the same time his younger brother was also diagnosed with the disease.

Having been gifted with two wonderful children, Jane and Peter, life was a struggle coping with the every day needs of my husband. It got to a point where, against all my beliefs and wishes, I had no choice but to separate from my husband. Life was normal, but there was always a worry at the back of my mind about one of my children developing the disease. As years went by, I noticed some signs in Peter at 10 years of age. Trying to convince myself that there was nothing wrong with my beautiful son Peter gave me many a sleepless night. Finally the bombshell dropped when he was diagnosed at the age of 14 years. My life fell apart. At the time I felt everything around me had stopped. I felt angry and emotional. This cannot be happening to me. I cannot describe in words how I felt at that time. The only thought in my mind was how I would cope seeing my son suffering with a disease which has no cure. It took me a great deal of courage and time to come to terms with it. As a parent I felt totally helpless and somewhat guilty to see my son suffering and not leading a normal life like most other teenagers do at his age.

As the disease has progressed I have noticed many changes in Peter's condition. Compared to his father's condition, Peter's is quite the opposite. The stubbornness is common in both but, where as my ex-husband would resort to violence, Peter would go totally quiet when things did not quite work out as they would like to. As expected with this disease, Peter has difficulty balancing whilst walking and the pain factor is being managed by drugs. Swallowing is getting more of a struggle. His speech is barely recognizable. He has accepted the fact that he has got to live with it and get on with life. We as a family persuaded him to go and see his father while he was in the final stages of the disease. Looking at his father's condition Peter made a comment which tore my heart apart: 'Mum, I will be like that one day'. That's what he managed to say, looking at his father in the hospital bed. He even mentioned that he would not like to be tube fed like his father when the time comes. I believe he is not willing to share a lot of his feelings. Sadly their father passed away in early 2005 which did have an affect on both my son and daughter.

Over the past few months I have noticed that Peter's condition has got worse. The disease has affected his mobility and he needs constant support. He is now unable to swallow solid foods. Every thing needs to be liquidized. Even then this is a

struggle for him. He cannot get a good night's rest because of the pain. I would be awake in the middle of the night and encourage him to do some yoga which I have taught him. This gives him some relief.

Despite of all his suffering he has a great willpower and determination to enjoy life as normal as possible achieving all his goals. His achievements at his sixth form college have been highly praised and commended by all those professional and caring people at his college. This came in the form of being presented with an award. In his spare time he is hooked to his electronic games or watching his favourite wrestling programmes on the television when he is not attending other activities such as drama and club outings organized by Social Services. He fulfilled his lifetime ambition to meet his wrestling heroes when he visited Washington, courtesy of a children's wish charity. He has also been granted wishes to visit Disney World, to meet the England football team (just before the World Cup) and to visit the new Arsenal stadium. As a mother, besides caring for Peter, I am constantly engaged in organizing outings and events that keep him happy and give him something to look forward to. He would be counting the days to the event and reminding me of it every day. Although Peter is unable to express his feelings at the best of the times, the occasional smile on his face shows his satisfaction with life. I am hoping and praying that God keeps on giving Peter the will and the strength to carry on living for a long time to come.

The other worry I have is my daughter Jane who I feel is bottling up and less inclined to talk about it openly. I try and give her space and pray that she will be okay. She does worry and care about her brother a lot. My life has been made a lot easier with a lot of help from all the carers and information I receive from the local HD charity. It has given me a lifeline to cope in my difficult times. I feel that I am not alone. It organizes events such as family gatherings and general meetings where I find a lot of helpful information about the disease.

I live in hope and pray that one day a cure is found for this devil known as the 'Huntington's disease' to save a lot of suffering which we see today.

Contribution 4

Our journey into the world of juvenile Huntington's disease (HD) began back in April 1984. It started with a phone call from my doctor. He knew that my husband and I were unable to have biological children. We had adopted a daughter nearly 5½ years prior and wanted to add at least one other child to our family. The reason for the doctor's phone call was to let us know that he had a young woman in his office who was expecting a baby in 6 to 8 weeks. This woman wanted to give her baby up for adoption. We were told that the biological grandfather had HD and that this baby's father was beginning to show symptoms also even though he was in his 20s. At the time, neither my husband nor I had any knowledge of this disease. Yet, we knew we wanted another child and for the past 5 years had

prayed daily for one. So we said 'yes'. Later that day an attorney contacted us and once again emphasized to us that since there was a family history of HD, this child would be "at risk". Still we didn't really know what that meant. Believing that God is in control of our lives and this situation, we pursued the adoption of this baby.

One evening late in May we got the phone call that the baby, a boy, had been born. We were told we could pick him up from the hospital in 2 days. We were ecstatic! Our baby boy was here. No thoughts of HD entered our minds. We were going to get our boy! Our attorney met us at the hospital, led us to the nursery, and the nurse allowed me to dress this precious and beautiful child. Tears of joy and thankfulness welled in my eyes. Immediately after the hospital released the baby to our care, we walked with our attorney to the County Courthouse to initiate the adoption process. We met with a Court Commissioner in her chambers. As she began to review the paperwork regarding the biological family, a look of concern crossed her face. She looked up and said, 'I can't allow you to proceed with this adoption'. She paused and our hearts sank. Why? What could be wrong? She then looked at us and said, 'This child is at risk for Huntington's disease. This is a devastating disease. Do you know anything about it?' We really didn't. We knew just that it was hereditary but nothing more. Our basic knowledge of hereditary disorders was that sometimes they skipped a generation or only affected a certain percentage of individuals. She continued on telling us that due to this child being at risk, we needed to realize all the implications of what we were potentially facing in the future if we chose to follow through with the adoption. My husband and I looked at one another in disbelief. The court commissioner told us, 'You have a choice here. You don't have to adopt this child. You need to become more informed and then decide if you want to follow through with the adoption process. You can take the baby home for now, but you must undergo genetic counselling before you can proceed'. Our hearts broke. I promptly told her that if I had been able to get pregnant and had something happen in my pregnancy, I would keep the child. How was this any different? She looked up at me and said, 'This is an adoption and you have a choice whether to proceed or give this baby back. You need to become educated about what the future may hold. I have had personal experience with this disease and it is devastating. You will need to decide'.

As we left the courthouse, what had started as a joyous day began to have clouds of concern and uncertainty float into our hearts and mind, and yet we were hopeful. We each have a deep faith in God and His plan and control in our lives. He would get us through this. During the next couple of weeks we prayed, cried, sought counsel, and went through the required genetic counselling about the risk factors and what we were facing. We were told that generally HD strikes people during their mid-life, some earlier and some later and rarely as juveniles. At that time, there was no definitive medical test to validate whether a person had the disease or not. All they could tell us was the history of the disease, risk factors, and that they knew it was on the fourth chromosome. The best science could do at that

time was to take blood samples of the biological family members and compare them with those who were demonstrating symptoms and those who weren't to see how they compared. To us, the question about whether to keep our son was obvious. Of course we would. God had answered our prayers and brought this child into our lives, hearts, and home.

Throughout his childhood R.D. struggled in school, was diagnosed with attention deficit disorder, but was a generally a happy-go-lucky kid. As he entered his teen years he struggled more and more in school. One day when I came home from work he told me that he was going to die. I was shocked and asked him what he meant. He said that he had gone through our files and found his adoption paperwork. In reading them he learned he was 'at risk' for HD. From the time he had found those papers and the reference to HD he began to research it on the Internet. What he learned is that people die from this disease eventually. But to a 14-year-old it seemed like a pre-determined life sentence. I carefully explained that we didn't know if he had HD, but that he had a 'chance' that he might have it and nothing was certain. It was no more certain that he was going to get HD than I was going to get in my car and have an accident or end up with cancer. Yet, the fear of the diagnosis was instilled in him no matter was I said.

When he was about 16 or 17 the youth pastor at our church mentioned to my husband that he thought R.D. had some unusual actions and traits and asked if he had Tourette's. In the business of life, we hadn't noticed these movements and the odd things he said were just him. What teen doesn't say some odd things at times? So, life went on. Through the help and guidance of a Special Education Counsellor at the high school R.D. was attending, he was able to graduate from high school. Our minds were never too far from the fear of HD and questions as to whether or not his learning difficulties or erratic body movements were the onset of HD or something else.

A few months later R.D. enrolled at a local community college. Since learning had always been such a struggle we recommended he take just one course. He chose one related to film making since he has such a strong interest in movies. Even though the course was something he was very interested in, he quit going about 4 weeks into the semester. He misunderstood that when a long-term assignment was given in the course that this still meant he needed to attend class. Regardless of our insistence that he still needed to attend the class, he was convinced he didn't need to attend.

So we consulted a learning coach. She didn't have any answers and suggested we take R.D. to a neuropsychologist to see if he could give us further insights into him and why learning and functioning in life was so hard for him. After a thorough battery of tests the neuropsychologist suggested Huntington's disease. We immediately contacted the centre of excellence for Huntington's and arranged an appointment.

Our appointment was with a doctor, a social worker, nurse, medical students, etc. R.D. was not happy to be at this appointment or with the potential that he may

have HD. When the doctor explained the symptoms and likelihood of the disease demonstrating itself and offered to do a physical test of R.D., R.D. was not willing to participate. At 19 years of age, we couldn't force him. The doctor went on to explain to him about the DNA test that could be done to confirm whether or not someone 'at risk' had the HD gene. R.D. was not interested. While he feared the diagnosis, he also didn't want it confirmed. The doctor shared with him that if he didn't have the test, he might wonder each time he stumbled or fell, if that was an HD symptom. At 19 though, he wasn't ready to learn he had Huntington's. Even though the test might turn out negative, he wasn't open to finding out he was positive. So we left.

R.D. did not go back to college and was depressed. We thought he might be showing symptoms of HD; he wasn't receiving any calls back on his employment applications, and couldn't drive. We didn't know what to do. Without a diagnosis, what could or should we do to guide him to do with his future? He wasn't seeing success anywhere he turned.

One day shortly after the visit with the doctor, R.D. met with his psychologist, with whom he had been getting counselling with for depression. In the course of the counselling session, his psychologist asked R.D. if he ever wanted to get married. He said he did. Next, his psychologist told him that he couldn't marry or have any relationship because he had a secret. What secret? The secret that he might have HD. His psychologist shared with R.D. that all relationships are built on trust and without honesty in a relationship he wouldn't have the basis for a strong and long-lasting relationship. This information got to R.D. He wanted to date and someday marry and have a family. He came out of the session and told my husband that he was ready to have the DNA test for Huntington's. He wanted to clear up this secret in his life. The mystery of whether or not he was carrying the HD gene. For better or for worse, he wanted to know. In a sense, we were all relieved. Now we would KNOW. At the same time, what if the news was the confirmation of our worst fears? Also, once we knew, if R.D. didn't have HD, we could pursue other research and testing to find out what was behind the symptoms we had seen in him from the time he was an older teen. The body movements coupled with his learning issues were evidence of something.

So at 19 years of age, in October 2004, R.D. was tested. Next came the weeks of waiting for the results. Weeks we continued to pray that the results would not confirm HD for R.D., but early in November the results did confirm that he definitely had HD. Upon further consultation with the doctor it was verified that his symptoms and positive DNA test validated a juvenile Huntington's diagnosis. WOW! What did that mean to R.D. and to us? Everything we had learned previously had said that juvenile onset was very rare. And it is. Now we were learning that instead of onset happening during the typical mid-life onset with a 20 or more year time frame of how long its course would run, the onset happened in our son's teen years and that might potentially only have 10 years or so of life left to live.³ All the hopes and dreams that we as parents had for our son and that he had for himself had suddenly been altered.

In the intervening years since that news we all have coped in a variety of ways. R.D. deals with his diagnosis by denial. He doesn't like to talk about HD. My husband and I each have grieved in our own ways. We cope by talking with one another about our concerns, read what we can, pray, and work to give him the best life he can have for as long as possible.

The 3-year anniversary of the diagnosis is approaching. Looking back we see the steady progression of the disease in R.D.'s life. He struggles with depression. His handwriting is getting more and more difficult to read. His body movements are increasingly erratic and more pronounced. A small patch on his head is balding from where he repeatedly hits his head with his fist as a result of the chorea. He chokes more when he eats. He stumbles and trips easily. He has few friends. He can't find a job. He has no girlfriend or prospects for a wife or family.

We don't know how long we will have him with us. We wonder how long before he will lose more of his abilities. No one can tell us that. Everything is a guess at this point. So we pray each and every day for God to heal him. God could do that! Will He? We don't know. But, we can ask and ask we do.

Contribution 5

Olivia is now 17 years old, and although biologically Emily's aunt Olivia has grown up as big sister to Emily. Olivia was 8 years old when Emily came to stay at 1 year old.

My little sister Emily has JHD. I am now 17 years old and Emily is 9 years old. I think it's important to understand as you read my story that although I am not at risk of HD my life is completely different because of it. Emily came to live with us when she was 1 year old and I was 8 years old. Emily's dad had HD and my oldest sister was her mum. They both lived really chaotic lives and had made some really bad choices. I was so often really angry and sad with both of them as I grew up because of how they continued to live their lives and cause so much havoc in ours. Their lifestyle made it impossible for them to parent Emily.

When Emily came to live with us she didn't know what a stable loving home was, she didn't know what a normal routine was, she was lost when she came to live with us. So I was an important part of trying to help offer her the stability she needed. But I was just a child. I was the youngest and had until then been the centre of my mum and dad's world. It felt like the life I had just seemed to stop and I had to grow up very quickly. Emily's needs were, and continue to be, paramount. Looking back I remember not liking this new person in my family; Emily was really, really active and needed a lot of attention, but I realize now that I probably was jealous because of my parents necessary change of focus. Emily was funny and adorable and I grew to love her very, very much more than I can put into words. When Emily was 3 years she was really clever, loving, beautiful, and a bit wild.

She melted everyone's heart. I remember about then she started to have problems because my mum and dad started to worry more and more about her. The one thing I do recall was having to pad the doors in case Emily banged into them. I didn't really fully understand then the enormity of what was about to happen.

Our whole family struggled on as Emily was seen by lots of doctors. I know now my dad thought she had HD from the age of about 3½ years. The day before Emily's sixth birthday (I was 10 years old by then) she was diagnosed with JHD. When I first found out I never dreamt it would be anything like that but I had no idea what JHD was and was completely naive to what lay ahead.

The diagnosis made; no one seemed to know what JHD was or what to expect. I just had this sense from the way people acted that this was really serious. The news hit us as a family, powerfully, almost like someone had punched you really hard and you'd got winded by the blow. I didn't understand why we felt this way. My sister and I got onto the Internet (and were never off it) to find out more about JHD. From what I read and maybe because of my age the bits about dying were at the forefront of my mind. I was unable to think of how long her life would last; all I could think of was that Emily, my little sister whom I cared for, protected and loved unconditionally, was going to die.

I stopped going to school. I had this thing in my head, what if I left her and that something might happen when I was gone? I cried lots, and was angry at different people. Our family was introduced to a lady called Jo who knew other families with this condition and she had learned lots from them and in turn was able to help us understand a bit more about the disease and what might happen. I remember one night just before Christmas Jo came to see us and I was really crying, sobbing. She asked me very gently what was wrong. I asked her to please tell me if Emily, even when she got really bad, would always know that I and my family loved her. Yes, from what she understood she would always know. I think we all cried then. It all felt so overwhelming and sad.

Looking back I really had no interest in other things and think I was a bit down. Once I realized that Emily was not going to die immediately my thoughts for her turned to the future. I would think a lot about what Emily might not be able to do, like get married and have children, go to the pub for her first drink (imagine thinking of that when you're 10 years old). My head was full of these things. We had plans as well, my sister, Emily and me. We were all going to go to Spain when we were 16; that was our most exciting plan. I realized that couldn't happen so I began to change my plans and I thought I would need to get married when I was 16 because I wanted Emily to walk me down the aisle. Little did I know that when I got to 16 years old Emily was going to be totally dependent on all care, confined to a wheelchair, fed through a tube and that our life would revolve around hospitals, the hospice and appointments. My oldest sister (Emily's mum) is now dead and Emily's dad is living in supported accommodation.

Contribution 6

My name is Ruth. I am the primary caregiver for my husband and daughter. Huntington's disease was first diagnosed with my husband's mother approximately 18 years ago. My husband was diagnosed in 2001 with a CAG count of 55.⁴ My daughter, Anne, was diagnosed with juvenile Huntington's at the age of 6 in the year 2006 with a CAG count of 100. Our family accounts take place in the USA.

When asked to provide information for the collective experiences of juvenile Huntington's disease (JHD) I did not know where to start. After quite some thought, I was able to decipher what would have been helpful to me as my daughter and I began to travel the road. Although there are many areas that need to be addressed, I have narrowed my input; advocacy and behavioural changes.

As with adults, children experience so many life-altering changes. Adults, usually, can start their own testing process, medication regimen, social and behavioural supports. As parents, it is up to us to advocate what is in our child's best interest. We do not have the time to wait and hope the professionals will take the correct or most effective action. For this is such a rare disease doctors, schools, and other professionals do not know where to start or even know who to contact for information. It is up to us to provide them with the appropriate resources.

I would suggest a starting point in the medical arena, contacting the centre of excellence (COE), and a treating neurologist, paediatrician, and psychiatrist that are willing to admit that they have limited knowledge of the disease and are willing to keep open communication between you and the other medical providers. Once you have your medical support system in place, you can now advocate for your child in all areas of life. The most difficult for us was the school system. However, once I had the medical 'team' in place, I obtained letter after letter from the doctors to back up every request I made. Low and behold all requests have now been met. Third, make yourself and child known. Keep fighting until you feel comfortable that your child's needs are being met. When you feel new systems or devices need to be implemented copy all letters to each member of the hierarchy. It all comes down to a degree of financial control versus liability.

In my experience, young children do not have the ability to recognize symptoms; they just 'go with the flow'. Usually their inability to rationalize and verbalize what their body and mind are going through comes out in behavioural outbursts. Understanding this is the first step to increasing the quality of life for you and your child as well as the entire family. Don't get me wrong, the battle is ever changing, what works today may not work tomorrow, but will work again next week. What I mean by this is, keep a whole bag of tricks. Don't just depend on one or two tactics—have many!

Anne's first symptoms were behavioural. The once seemingly well-behaved, rule and boundary-accepting (age appropriate outbursts, of course), had turned into this aggressive, oppositional child who had the will of 15 men and the

adrenaline of 40. This 34-pound, blonde-haired, blue-eyed, dainty appearing girl could really throw you off at a moment's notice.

The true definition of discipline is teaching a child right from wrong, helping him or her realize socially acceptable behaviours, and understanding boundaries and rules. Punishment, whether through consequences or physical (which I don't believe in), is just one side of the barrel. Reward and praise is the other. Well, with JHD, throw most of what you know about punishment out the window and focus on the reward and praise system. Yes, this sounds a bit one-sided but you will run yourself ragged trying to reason with someone who has lost the ability to reason, or does not have the ability to reason when 'enraged'. For Anne, the pendulum tilts so quickly that she can literally go one second to the next from happy to enraged.

Yes medication is helpful and sometimes necessary, and this will need to be something you explore with the neurologist and psychiatrist, but let's make sure our own expectations are in tact. We cannot expect our children with JHD to act in a 'socially acceptable' way at all times. We cannot expect our children to understand in-depth concepts of cause and effect, so let's focus on what we can. It is necessary to start picking your battles! Before reacting to your child, is his or her behaviour life threatening? Will his or her behaviour cause harm to himself/herself or others? If the answer to this question is yes then immediate action and intervention is necessary. Otherwise, take the time to think, walk away, say the alphabet; I don't care, pre-occupy yourself. None of this is easy. We as parents need to retrain ourselves and our whole life upbringing, but it can be done for the love of our children.

Anne has had poor impulse control, aggressive outbursts (hitting others, throwing everything in sight/reach, etc); and patience, wow, that's almost non-existent. So, what have I done to help minimize this? We did seek help with medication but we also made some changes in the home and environment. We came up with what may seem like bizarre tactics to the 'real world'. But we get through everyday. We enjoy life, and we accept and let go of that which we cannot change.

(1) A reward system: A visual tactile poster hangs in the living room. Each aspect of routine clearly and definitively defined. For example, the morning routine does not say 'get ready for school'. Instead each step is defined, i.e. wake up without arguing, use the restroom, eat breakfast, brush teeth, take medicine, get dressed, etc. We also added some things she truly enjoys to ensure success, such as feeding the fish. Each goal has a space for a star sticker. Make sure, if you utilize this approach, your child has input in creating the chart and comes up with some of the goals. Anne's success is measured by the number of stars. Once she counts 22 stars she can choose something from the surprise box, (items purchased from the dollar store). Once she reaches 44 stars she can pick an activity, i.e. ice cream parlour, movie, game to play, visit someone special. And the top number (which we haven't met yet) of 64 stars is \$5 toward a trip to the toy store. Anne helped come up with the rewards to ensure it was worth working towards! Below are our most useful tactics. Please use what is appropriate for your child and come up with your own.

(2) Sensory: When aggression starts, I have noticed, holding Anne (even if she is trying to wiggle away) and rubbing her back often calms her. Yes it takes up to 10 minutes but you can sense in the first minute or so if it is going to work. The wiggles decrease and the screaming has some breaks of silence.

(3) Change in environmental temperature: I have noticed that removing Anne from a warm or hot temperature to cool air can help relieve irritability and aggression. Again, it's not immediate, but it works.

(4) Shower: We have a hand-held shower massager that often comes in handy. If Anne is aggressive and my touch is only irritating her more, I can often put her in the shower and turn on the shower massager. Obviously, this is not something you want to do when other people are there. It is a given that she will fight me every step of the way, so removing clothing doesn't look so pretty!

(5) Change in caregiver: It is not uncommon for Anne to target the caregiver who has 'made' her angry. I have experienced that if I call my sister for help I can walk out the door, collect my own thoughts and give Anne a break from me. Usually, my sister can use the exact same tactics and they will work.

I have learned so much from Anne, and hope our experiences can help others cope. *Yes, Juvenile Huntington's is a tragic, life-altering, and inevitable end-of-life disease. However, if we all sat and focused on the negative, what kind of life would that be? Anne has and continues to experience extreme life changes but she continues to fight her battle with strength, courage, and dignity. She does not allow this disease to take over her life, why should I? It just reminds us that we need to be flexible and appreciate all that we have and every moment we spend together.*

End Notes

- ¹ Editors' comments: Families frequently have false perceptions, but in fact males and females can both be affected with Huntington's disease (HD). HD does not skip generations. The only way this can appear to happen is if a grandparent is affected, a parent dies very young before the onset of problems, and a child is then affected. There is a further complication in that, for very young onset, the parent who transmits HD is more often the father; even so most fathers with HD are not likely to have a child with JHD.
- ² Editors' comments: This is most likely to be a description of the behavioural and thinking problems which are part of the condition rather than an actual overproduction of adrenalin. See Chapter 5 for a description of the pathology of HD.
- ³ Editors' comments: This is discussed in more detail in Chapter 4.
- ⁴ Editors' comment: See Chapters 6 and 9 for a description of the CAG repeat size.