

BCNF Stories



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12 YEAR OLD BRENDON WAS DIAGNOSED WITH NF WHEN 3 MONTHS OLD

By *Brendon Pommier*

My name is Brendon Pommier, I am 12 years old and I have NF. This is my story. I was born in Vancouver, but my family and I moved to Kelowna when I was almost 1 year old. I like living in Kelowna because two of my favourite things to do, skiing and fishing, are near by. I live with my Dad, Ken, and my step-mom, Valerie. I have two brothers, Marlon, who is 20 and Daylon who is 14. I also have a step-sister, Cassy, who is 12. We all get along good. My Mom and Dad got divorced about 6 years ago. That was a really hard time for me, and it still bugs me. My Mom lives in Kelowna and I see her on weekends.

About my NF, my Dad tells me I was diagnosed when I was about 3 months old. I had a plexiform fibroma in my neck area and lots of café-au-lait spots. Not everyone with NF has a plexiform, and it's a drag that I have it in my face region. My grandma suspected what I had because her sister, my Aunt Gina, also has NF. She has one daughter, Karen who also has NF. I like Karen. She is really nice and fun to talk to. Her two little girls also have NF. I am the only one in my immediate family that has NF.

I have had 6 surgeries to deal with. My 7th is scheduled for March 19th in Seattle. My first surgery was when I was 6 months old. When I was 3, the plexiform had worn out parts of my neck. Two Doctors at Children's Hospital in Vancouver, Dr. Treadwell and Dr. Spinbok (sp) operated on me. It lasted about 13 hours, and when it was done I had a fused neck from C2-C5. I lost about 50% of movement in my neck. This had affected me a lot. There are two things I really want to do, play hockey and snowboard & now I can't because my neck is not strong enough. But I play floor hockey



and I ski, so that is ok. I also have a shunt so my brain so it can drain its fluid.

I go and see a Plastic Surgeon in Seattle named Dr. Gruss. He is very good and really nice to me. I go see him because the Plastic Surgeons in Vancouver told my Dad that it was better to wait and let my plexiform grow. My Dad didn't believe them so he researched and found out about Dr. Gruss. He has helped me a lot. Surgeries are a very scary time for me. My Dad and I discuss them a lot. He likes for me to understand everything and make my own decisions.

I think about my NF every day. It is hard to make friends and I wish I didn't have NF. Everyone stares at me and some people make fun of me. I can't play all the sports I want to and I always have to worry about my neck. I worry about girlfriends and the rest of my life. I have trouble in school too. I don't have any bumps on my body and I hope I don't get any.

My Dad and I are started an NF Support Group in Kelowna. I don't know any other kids with NF and I would really like to meet some. I hope I get to go to the NF Camp this summer. That would be great. I don't like having NF but I have a great deal of family and I can walk and talk, so I guess I am lucky.

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IVAN'S STORY

Reprinted from the Fall 1998 issue of iNFo, the BCNF newsletter

Ivan has served on the BCNF Board of Directors, volunteered at many events – and serves up a mean spaghetti dinner! Here is his story:

My name is Ivan Junek and I am affected with an unwanted spontaneous form of NF. My first symptoms of NF appeared when I was ten years old. That time NF was known as Von Recklinghausen's. Kids teased me until my facial fibromas were removed. Then I was free of visible face fibromas for some time.

All my problems started in 1970 when I discovered a small neurofibroma the size of a hazelnut growing in the left underarm lymphatic system. In three weeks this fibroma grew into the size of fist, and started to paralyze my arm. Treatment for this physical condition was surgery. When the doctor discovered that my neurofibroma had become malignant cancer, he recommended the removal of my entire left arm, shoulder and part of the collar bone. At That time, I did not know that if I had not chosen to have my arm removed, my life would probably have ended within three months. The doctor had told this to my sister.

After recuperation from surgery, I went for rehabilitation at the G.F. Strong Centre where it was decided that they would send me to school, Vancouver Community College-Langara, to study in the Food Service Technical Program. Between the first and second year of college, a large neurofibroma started growing in the area of my sternum, and during surgery I had a larger part of my rib removed. Since 1970 to now doctors removed more than two hundred fibromas from my body; what a pleasure to live with NF.

When I graduated from the program my hopes were to start working immediately in some large hospital. But my advanced spread of NF made it impossible. My skin appearance combined with missing left arm, were working against me. I was not able to find work in any large hospital with possibilities of more learning and growth.

During 1990-94, I was involved in the Food Service Supervisors Technician Association in the function as a director on the board. I was an organizer of educational lectures for the Vancouver and Lower Mainland, notifying members about monthly and annual meetings, as well as calling on special speakers. I held this position for four years.

At the present time, my desire is to help the BCNF, to support medical research and to be one of the one of chain links to help find a cure and help to people with NF.

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LIZ, THE MOTHER OF A CHILD WITH NF1, SHARES HER EXPERIENCE

Reprinted from the Summer 2000 issue of iNFo, the BCNF newsletter

Rebekah is our first born daughter. I remember that when I found out I was pregnant, I cried. I was not sure we were ready to be parents. Talking with my husband James about the news, I said, “choosing to have children is choosing to enter an emotional roller coaster, are we ready?” He gently assured me that we would be able to handle whatever happened when we needed to.

When Rebekah was 9 months old, I showed our doctor a brown spot that I hadn’t noticed at birth. Our doctor said it could be the Elephant Man’s disease, but probably not. Our ability to handle the emotional roller coaster was tested. Over the next year, we were referred to a variety of specialists. The paediatrician assured us it was not the Elephant Man’s disease, but that it could be something called Neurofibromatosis. He didn’t feel she had NF but he referred us to an ophthalmologist. He reassured us she didn’t have NF but he referred us to a dermatologist. She felt it wasn’t NF but sent us to a Genetics Specialist. I went to the genetics clinic without James, either because I believed it was just a routine appointment or because I was in denial.

We received the official diagnosis that day: Rebekah had NF1. Panic, grief, devastation were just some of the emotions we felt. Then we quickly moved into research mode and accessed as many articles as we could from the UBC library. As our emotions calmed down and we returned to the paediatrician, the hope we were given was that she “probably has a mild case”. She was followed up routinely but was asymptomatic. At five years old, an MRI was completed as a “baseline”. This

revealed numerous fibromas on her spine, an optic glioma on her optic nerve and scoliosis. Rebekah now wears a body brace 23 hours a day to try and arrest the growth of the curve and is followed up regularly by specialists at Children’s.

When asked how I cope with Rebekah’s disorder, I say it depends on the day. At my most difficult moments I project every possible symptom that I have read about or seen in pictures on to her. I worry about her being accepted by friends, her possible struggle with education, her possible job limitations, and whether she will ever marry or have children.

This is not a good place to be. My husband helps me gain perspective and reminds me that we all will have the strength to handle issues as they come one day at a time. In my saddest moments I grieve. I watch the freedom other children have to somersault and do the monkey bars and I grieve for Rebekah in her brace. I see her struggling with co-ordination and physical stamina and I grieve. I see the value the world places on “the perfect body and on physical beauty” and I grieve. During those times I cry, I talk to my husband or family and I pray. She too will grieve, but I know that hope comes through. At my most frustrating moments, I become an advocate for Rebekah. Unsatisfied with the waiting game, we go back to the Internet and library again looking for more leads to help us do something. We go to naturopaths to go beyond how the medical community

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thinks. We insist on getting the medical attention necessary.

At other moments I move into a healthy state of denial. Life is rich and full and we can go through most days without worry or agony. We are again like any family with three normal children who need us to not put too much focus on the NF diagnosis. Finally at some moments I can look at the gift this has been in our lives. My husband and I have had to struggle with our values and prejudices as to what we see as worthwhile in people, this is changing me. As a counsellor this has given me a new level of empathy for the many difficulties people have to face. It is a gift because it is only NF. My family of origin has grieved the tragic loss of two of my brothers. Rebekah is alive. A friend's child was diagnosed with Muscular Dystrophy and will spend much of her life in a wheelchair. Rebekah runs and walks and dances. It is a gift to us and to her because of the depth of character we pray it will create. We long for our children to become people of compassion and depth but we never want our children to experience the suffering that creates those kinds of people.

When asked how Rebekah handles her NF, I would say better than we do. We used the NF language from a very early age, so it is simply part of her. She has cried, like the day she got her brace and cried for an hour. She has been frustrated and expressed how much she hates her brace at times. She has been excited when she sees Thrifty bags and coin cans that advertise NF. "This is raising money for me!" she tells the store clerks. She is curious about other disabilities people have and is keen to have an NF pen pal. She is growing in compassion and often helps push another student (who is in a wheelchair) around the school. She accepts that she has NF and shows her brace for show and tell. Mostly, however, she loves life and doesn't let NF affect the joy she leaps into activities with. Thus we try to live one day at a time with the unknown of the future but with the known sense that we all can make it.



FIVE YEAR-OLD NOAH WAS DIAGNOSED WITH NF WHEN HE WAS ONLY A FEW MONTHS OLD

Originally Printed in the Globe and Mail, May 27th, 2003 and reproduced with author Robert Mackwood's consent

Very few international donors gave to Canadians—those deemed a perfect match. They are lottery odds and we inexplicably won.

The Call came in mid-afternoon. Perhaps we've all wondered at some point in our lives where we would be when really astonishing news that could change a life forever is first announced. How would we react? What would we say? Maybe the first thing to say is: "Could you repeat that?"

This wonderment became reality for me a few weeks ago. I was truly speechless. The odds against, we were told, were monumental.

The chance of this happening seemed so remote that we had all but given up hope. In fact, I'll confess, I'd forgotten about it.

But on the other end of the phone that day was my wife Diane saying the words that rendered me numb, stunned, and shocked. "Our doctor just called and they found a match for Noah."

Noah is our four-and-a-half-year-old son with a medical history that fills any super-sized three-ring binder. Here's my attempt at the Coles Notes version: Noah was diagnosed with neurofibromatosis when he was only months old and the doctors told us to expect something to happen. Neurofibromatosis (NF for short) is a genetic disorder of the nervous system that causes tumours to form on the nerves anywhere in the body at any time. NF is one of the most common genetic disorders in the United States, (one in every 3,000 to 4,000 births), even though not many people are

familiar with it. This is from the American NF website and I assume the ratios are similar in Canada.

Something happened during a family holiday staying with friends on beautiful Gambier Island in August, 2001. Noah started contracting regular fevers that were more than we could handle and he was admitted to hospital. Watching the tragedy of 9/11 was only a blur for us as Noah puzzled doctors with his condition.

Eventually it was agreed that his spleen was causing this and should be removed. While it helped quell his fevers, it also started rumbling of a different kind and we were told that he was now diagnosed with something called Myelodysplastic Syndrome, which is a precursor to childhood leukemia. Many bouts of chemotherapy over the spring of 2002 led to a period of remission and for the first time in his young life, Noah could swim in a pool-free of drugs and tubes sticking out of his body. Our summer vacation in Osoyoos, B.C., that year is a wonderful memory, but its optimism quickly evaporated. The fevers started to return and the doctors confirmed what we secretly suspected: the leukemia was back. He had been given all the chemo his body could take and there was nothing more they could do without a bone-marrow transplant.

Our catch-22? There was no match. More than one million potential donors had been screened by the

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international registries of unrelated blood donors and so far, nothing matched Noah. Noah's brothers Daniel and Aidan would have been the best candidates as related donors but were not close.

Through the wonderful Make-a-Wish Foundation, we boarded a plane and headed to Disneyworld for a week of almost complete distraction after Noah had been declared terminal.

October turned to November and we even started thinking about Christmas plans. Sure enough Noah made it there-even banged pots and pans about on News Years' Eve. We dared think about his fourth birthday in early February. A wonderful celebration, complete with a purple-iced cake and Star-Wars gifts, helped us all forget-for a bit.

We knew that time wasn't on our side. In March, our amazing doctor Mason Bond at Vancouver Children's Hospital started some alternative drug therapy, although Noah didn't react particularly well. In fact, his cancer-cell count soared and again all were alerted to what could be his final days. Then, just as mysteriously, the counts declined.

The search for a bone-marrow match continued. We've come to understand that the screening for a potential unrelated match is now so intense as to try and eliminate graft-versus-host disease (the body's organ reaction to new bone marrow which can cause devastating results) that it can take a long time. For us, it's been almost two years. We were also told that the registry was looking at the next million possibilities, having eliminated all but a few that were close; ultimately they were not considered candidates.

Then our lottery call happened. I don't know why it happened-why the donor was found now. I cannot comprehend the odds of this happening. Isn't this how Hollywood ends movies? I now tell people-slowly-savouring this mind-boggling news. Those who have followed Noah's progress are so supportive that it feels wonderful to now give something positive back. To properly thank those in the medial community who have helped Noah is impossible.

We've since found out a bit about the identical match although the identity and country of the potential donor is not disclosed to the family (for good reason) for at least two years.

We do know this: somewhere in Europe there is a young woman and I would kiss the ground she walks on forever for what she is doing for Noah. With light saber in hand, Noah is scheduled for the bone-marrow transplant in early June.

We have no illusions about the battle that lies ahead but if he stays reasonably healthy he may get the chance to live.

Final note: I'm writing about this to encourage you-yes, you, reading this newspaper-to become a registered blood donor and join the Unrelated Bone Marrow Donor Registry. A recent newsletter from Canadian Blood Services pointed out that in 2002, there were more than eight million potential registered donors worldwide. Canada has just over 200,000. The really remarkable statistic is this: only 142 international donors donated to Canadian patients-those deemed a perfect match. Those are lottery odds and we inexplicably won.

Become a registered donor. You might help someone in the world get the call of a lifetime.

Robert Mackwood is a literary agent based in Vancouver.

UPDATE: July 11, 2003

Noah had the transplant done on June 4th, 2003 and the doctors and the Mackwood family report everything is going as well as can be expected. We just spoke with his brother, Daniel, and he shared that Noah is coming home today! The BCNF sends our best wishes to Noah and his family.

UPDATE: July 5, 2004

Noah's blood is now Type B, completely female cells, thanks to his generous donor from Europe. (We thought we had to worry about trans-gender things now, but apparently that has nothing to do with it!) The drugs made his appetite insatiable, gave him the Moon Face typical of transplant recipients, and caused body hair growth, his on his head mainly. All of this kept us laughing, with

him, not at him – he was proud to say that he was going to be shaving with Dad, he would warn us to be careful with the “fur” on his back when taking off his shirt, and at his first hair cut he asked if he could please get his forehead trimmed as well. His medications -eleven at first-are down to four as of a couple of weeks ago, and his central line catheter is out for the first time in nearly two years. Every little step is a huge victory for him!

Daily, Noah is behaving more and more like other children of his age-running and playing more than resting on the couch, growing stronger and taller while losing the chubbiness of toddler-hood (and Prednisone!), getting bumps and scrapes and colds just like everyone does. In September he will be in Kindergarten. All the while, Noah has kept his cheerful, grown-up demeanour and made us stronger and better people. Quite a feat for a little five year-old!



A PARENT'S PERSPECTIVE ON LIVING WITH NEUROFIBROMATOSIS

By Susan Wood

Once asked to offer my perspective of life with Neurofibromatosis (NF), I thought “that will be easy. Well, I quickly remembered it is not always easy raising a child with NF. We are a family of five. Mom, Dad, Danny is now seventeen, Chris is fifteen (I can hardly believe that!) and Steven, our youngest, is eleven years old. Life is busy, work, school, sports, friends, hobbies, general family growing pains, life is busy under normal circumstances. Throw in life with NF and it can get pretty complicated. I worked as an LPN before children. My husband is a teacher. After the birth of our first child, I became a stay-at-home Mom, revelling in the dream of care-taking a home and raising a family. Life was good and we eagerly anticipated baby number two. Enter Chris. This time around was far different. He was a very unsettled baby and seemed to be in pain, couldn't latch to breast-feed, vomited and cried and cried. That's when we began our search for answers. At seven months old, Chris was diagnosed with NF. My world seemed to stop. This thing, this disorder I couldn't even pronounce let alone understand, left me desperately afraid, for him and for my family's future.

So began our journey with Neurofibromatosis a genetic disorder causing tumours to grow along the nerve endings in the body and/or on the surface of the body. NF affects over 10,000 Canadians in a myriad of ways from learning disabilities, to blindness, deafness, physical disability, facial disfigurement, and even cancer. So little is known about NF and there is no cure. It would take me far too long to list our personal experiences with this complex disorder, so to make a long and complicated story short, I'll tell you how NF has so far affected Chris. He has many cafe-au-lait spots (coffee coloured

birth marks) and many neurofibromas (benign surface tumours) all over his body. I stopped counting a few years ago and now only search for changes in his two plexiforms (tumour clusters) and possible new ones. Chris also presents with short stature, enlarged orbit, three small brain tumours doctors tell me are of no consequence (they might as well be mountains to me), a few small spinal tumours, again I'm told not much to worry about. He also has learning disabilities that challenge him and us in every aspect of life. Added to his challenges around the age of 11 was a diagnosis of pseudoarthrosis of the right tibia and fibula the fibula already being fractured, and scoliosis. At 13 years, the diagnosis of Tourette's syndrome and ADHD, in addition to NF, was made. With all of this, he is also, and most importantly, a child meeting the challenges of life as every child does. He is an amazing person and it is privilege to parent him, roller coaster and all.

Parenting a child with NF can be hard. It hurts. There is guilt, fear, confusion, frustration, anger, uncertainty and grieving. I have grieved for the healthy baby I thought I had and have grieved with the appearance of every new NF symptom. Just when I think we can be “normal”, at least for awhile, some aspect of it rears up again and the process repeats itself. In an idealistic world, families pull together through the tough times. In a realistic world, raising a child with challenges can pull families apart. I've learned about strengths and limitations of each family member, each friend, and of myself. I've learned to choose

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situations cautiously, always trying to protect, knowing I am limiting the experience of another, too often questioning decisions I'm forced to make affecting the future of my child, and in turn, those who love him. Physical health, social, emotional, mental development all become a focus. What part of his challenges are NF related and which ones are not? At times I have been so focussed on NF and its assault on Chris' body, I fear I forget to see the child. The lines can easily blur. It can be emotional turmoil to love a child so much, as he is, and yet hate the disorder that in reality has shaped who he is and will always be a part of him.

The Medical System presents other challenges to navigate. How many doctors have seen Chris since the beginning. I could figure it out, but overall, too many. How many were actually knowledgeable in treating NF and its complications. Only a few. What one physician has seen as nothing to be concerned about, another has deemed important. Before NF we had a family doctor and a dentist. Now we include the paediatrician, ophthalmologist, orthopaedist, cardiologist, geneticist, psychiatrist, orthotics specialist, development programs, speech therapy, O.Ts, MRIs, X-rays and more, some local, some not, each specialist looking only at one aspect of Chris' body and sometimes I fear, not the whole of my son.

As well, NF has brought many educational challenges. So often, a child's learning disabilities are for the most part invisible to the eye. In hectic classrooms of 25 plus children, it is easy to get lost in the shuffle. Extra help is carefully rationed and not easily attainable. Sometimes a staff member is more concerned with liabilities than building a child's self-esteem and offering flexibility in the classroom can be a tall order in a tight schedule. The school system is certainly a maze and as I discovered, it is necessary to develop the skill of advocating for a child with special needs. For a well rounded school experience, it is important for a child to develop good social skills another challenge for those with learning disabilities. The need to fit in and make friends can be devastating for a child with learning disabilities, physical limitations

and the insecurity of looking different because of a brace and spots. Friends are sometimes in short supply because kids are kids and not always understanding. It is especially heartbreaking to have to explain that team sports are too risky; always searching for alternatives that are not his desire.

For all the negatives life with NF can bring, I would be remiss in not acknowledging the gifts received in the form of experiences and people who would otherwise not have touched our lives. NF gave us the daycare worker who remains a trusted friend. When Chris was two, she began to work to maximize his basic skills and provided a safe and loving environment to begin the merge with other children away from home. The kindergarten teacher who nurtured and guided us through his often chaotic first school year and warned me of the possible school maze pitfalls ahead. With mixed feeling, the grade one teacher whose narrow view forced me to find my voice and learn to advocate for my child. The Independent School, whose philosophy celebrated my child's individuality and helped him to cope in a diverse and fast paced world. The grade six teacher who supported my son's integration back into the public school system. He helped us through the very emotional process of having Chris labelled under a Chronic Health Disability in order to access much needed resources. He helped Chris find the courage to accept his limitations and encouraged him to reach for his full potential and have fun doing it. Each of the challenges we have experienced has some how impacted our ways of being. The upside of living with a chronic condition is that through the necessary soul searching it usually brings, it does provide opportunity for personal growth on many levels and with an open mind, create an appreciation for the small things in life that can benefit all situations.

My first contact with another person with NF was when my son was about 2 years old. I'll never forget the relief and comfort of actually talking to someone with NF. Within a short while an NF Support Group formed on Vancouver Island. Truly now we were not alone. The value of being able

to ask questions, and more importantly, the value of being heard by someone who understands is immeasurable. We have good times and not so good times and the presence of NF in our lives has certainly given it a roller coaster quality. Coping strategies in all areas of daily living have had to be sought and implemented much of the time. The years have passed, fourteen since the initial diagnosis, and still the fear of moving through uncertainty is always present. I can honestly say we now have more good days than bad, but when the dark days arrive and they likely always will, I still have to reach out to someone who understands and who will give me an ear or a little push to keep moving forward.

It was at an NF Symposium that I was able to hear the experts and the experience of so many others about life with NF. For us, that weekend resulted in finding some direction to finally put all the parts of my child into one whole human being, provided information for coordinated care and gave us a solid medical source to call on. What a gift – it is such relief when knowledge and understanding can be obtained.

It is difficult to condense all the information and experiences of my son's lifetime to this point, down to what matters most. For us, life with NF is definitely a journey wrought with physical, emotional, mental and spiritual highs and lows. There is pain and there is joy. My part in this journey has provided me an increased appreciation for another's circumstance and taught me that I need to take care of myself in order to take care of another. I've found it necessary to re-evaluate my personal values on many levels and develop coping strategies that work for me. Each family member reacts and deals differently with the implications of living with a chronic condition. For me, knowledge of possible complications is important – sort of a "forewarned is forearmed" stance in dealing with an uncertain future. For other family members, this information simply creates stress and is not desired.

We each face our fears of an unknown future differently and we each cope with what is, differently. My hope for each of us is to achieve understanding and acceptance of Chris' condition, keep moving forward and maximize quality of life and appreciation of the challenges in life with NF, his and ours.



HOLLY IS AN ADULT WITH NF AND THE MOTHER OF TWO CHILDREN WITH NF...

Reproduced with permission from the Canadian Nurse, Vol. 96, No.6



I have neurofibromatosis (NF), type 1. When I was first diagnosed, in my early twenties, my doctor mentioned the words “elephant man disease,” I know now that this was the wrong diagnosis but, at the time, it was very frightening for me and still is, I was newly married, planning a family, and there was little information available about my disorder. NF and elephant man disease (Proteus Syndrome) are two different disorders. Some 20 years later, I know that NF – also called von Recklinghausen’s disease – is a genetic disorder and that NF type 1 and NF type 2 are two separate disorders that are still not well understood.

NF type 2 or Bilateral Acoustic Neurofibromatosis involves the mutation of a gene on the 22nd chromosome and affects approximately one in 40,000 people. The gene was discovered in 1993. Usually diagnosed in the teen years, or older, the symptoms include internal tumours on both sides of the head - affecting the nerves related to balance and hearing - and other benign tumours in the brain on the spinal cord.

NF type 1, discovered in 1990, is more common than type 2 and involves the mutation of a gene on the 17th chromosome. Affecting approximately one in 3,500 births of both sexes and all races, it can be inherited from either parent or it can be spontaneous occurrence. Fifty per cent of the time it is inherited; the other fifty per cent, it is spontaneous.

A diagnosis of NF type 1 is often made through physical signs and symptoms but genetic testing will provide confirmation. The cosmetic appearance of small tumours, sometimes in the hundreds, on

the surface of the skin can also affect body image and cause emotional trauma.

It is unpredictable how NF type 1 will affect any one individual, but he or she is not likely to exhibit all of the above symptoms. Two-thirds of those affected will be mildly-to-moderately affected, and one-third more severely affected. Tumours may be removed, but not always effectively. Depending on the province, removal of skin surface tumours may or may not be covered by Medicare, as the disorder is equated with warts and moles. Complications can occur at any age and frequently worsen during pregnancy or puberty. More severe complications are seen in younger children. Occasionally, NF type 1 can become cancerous. It is not contagious and there is no cure.

My story

On a scale from one to ten, with one being the least severe case of NF type 1, I am a two. This makes me feel very fortunate. I was told my case was a spontaneous occurrence, but both of my children have inherited NF type 1 from me. They are now 23 and 21 and are mildly affected. But this does not mean that their children will be mildly affected. They may decide not to have children, rather than risk the chance of passing it on. My daughter recently told me she was frightened of what might happen to her in the future, as NF type 1 can get worse with time.

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Now in my forties, I have many cafe-au-lait spots and hundreds of very small tumours on the surface of my skin. I am uncertain and nervous about how many more I will develop. Most of my friends and colleagues do not even notice these, but sometimes I am asked if I have a heat rash. I'm learning to be open and honest with my replies. And I currently belong to a wonderful support group here in Victoria and have many friends with NF. For this sake, as well as my own, I hope a cure is found soon.

Reaching out

Proper diagnosis and counselling by a paediatrician and a specialist in genetic disorders is the first step. Health care professionals need to be aware of the symptoms of NF so that they can provide education and counselling to broaden the patient's understanding - particularly that of a newly diagnosed patient.

A young adult, or newly diagnosed person, may have fears that their chosen career may not be feasible, or that their marriage plans are in jeopardy. They may have low self-esteem and feel that life is not worth living. Feeling of denial, anger, sadness, guilt and anxiety may come up.

Health professionals need to help them through these stages, letting them know that support is available. Self-help groups, web sites, provincial foundations and national organizations are also out there to provide information. Health professionals should also be aware that a diagnosis of NF can affect not only the individual, but their family. Parents can be devastated by the knowledge that their healthy and active baby could develop learning disabilities, bone growth abnormalities and tumours anywhere in their body - as well as complications that can be life threatening. They often ask how to explain NF to their child. They should be counselled to stay positive and optimistic, be aware of the latest research and focus on what is going well in their child's life. There is always hope.

Open communication between individuals and various health care agencies such as neurology,

orthopaedics, ophthalmology, audiology, psychology and social services is also important, so that the patient receives continuity of care.

And lastly, healthcare professionals need to be careful not to double victimize these patients by avoiding or withholding basic human touch. A handshake or a backrub say a lot about acceptance of their condition and of them as a human being.

Holly Meadows is a past board member of the BCNF and a staff nurse, maternity, at the Victoria General Hospital, Victoria.



TO RAISE AWARENESS OF NF, WENDY WRITES ABOUT HER SISTER EVA

By Wendy Vowles

May is Neurofibromatosis (NF) month and to raise awareness of this disease I wanted to write a story about my sister, Eva, who has NF. As I started to write this story, however, it quickly became apparent to me that I'm simply not qualified to write about my sister's experience with NF. All I can really tell you about is my experience having a sister with this disease, and how it has affected her life as well as mine, although I know that I will never fully realize how deeply it has changed us both. I guess, in the end, it's only my story I can tell. I don't recall too much of significance in our childhood aside from wondering why my sister was always falling and unusually accident prone. She was not always as happy as other children either, and looking back now I can identify factors such as the migraine headaches that she suffered since birth which would impact the carefree attitude of a child. It just seemed as though things were always a little harder for my sister but I still wish that I could have held time in motion back then, keep her safe from the disease and extend the time of innocence and freedom from worries that were to come.

I found that society has a bad habit of expecting everyone to be the same and it is often challenging to have different abilities accommodated, especially if you don't come with a label that people understand. As a child, for example, my sister was asked to quit her Brownies group because she couldn't learn to skip - not the flagship gesture of compassion. My own experience with this sort of group was short lived as well because I never forgot the rejection of my sister.

Adolescence of course brought the usual troubles we all face; the challenge of adjusting to the

natural changes that take place in our lives while simultaneously experiencing perhaps our greatest need for social acceptance. For my sister this was an exceptionally distressing time because it was in her teen years that the disease really began to announce its presence. She was just 15 years old when we first noticed the tumors growing in her neck.

A visit to a specialist just before Christmas that year resulted in an initial diagnosis of some sort of cancer, grave looks around the household that holiday season told me that the future did not look bright for our family. A biopsy followed and a very long six months later the waiting finally ended. It wasn't cancer causing the growing tumors - it was instead Neurofibromatosis (NF).

The doctor announced this news to my sister. He then quietly backed up and left the room. And that was it. That was all the information she was given.

It was, we eventually deduced, all the doctor knew about the subject. Diagnose and run was the extent of his abilities and the best of his bedside-manner. I was pleased that no one was talking about cancer anymore, but what did NF mean, and what would happen to my sister now? What would happen to my family?

I was eleven years old then. Now I'm in my thirties and have access to a world of information via the Internet, libraries, etc. I know things about NF now. I know that it is a genetic disorder involving the growth of tumors throughout the nervous system, as well as tissue such as bone and skin, causing

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disfigurement, deafness, blindness, cancer, bone deformation, and mental retardation.

Learning disabilities are commonly seen with NF and patients are prone to cardiovascular problems such as hypertension and congenital heart disease, as well as epilepsy, deformities of the bones and scoliosis. The disease progresses with time and the tumors continue to grow throughout the body causing life threatening or severely disfiguring complications. Emotional trauma and affected body image follow suite. Although some of the tumors can be safely removed surgically, I understand that these procedures are not covered by our medical system unless cancer is diagnosed.

[Editor's note: the removal of benign neurofibromas is covered by MSP if they appear in the head and neck region, but not if they appear on other parts of the body UNLESS they cause significant pathophysiologic dysfunction, and then these are decided on a case-by-case basis.]

NF affects one in 2500 to 4000 people. Half of the occurrences of NF are caused by heredity, half are spontaneous - simply out of the blue with no apparent link to heredity. This disease isn't picky about gender, race or ethnic group. NF is currently unpreventable, incurable and the only goal in treatment of this disease is to try to relieve the symptoms.

Sometimes I miss being eleven years old and not knowing anything about NF. Years ago, however, I realized that while ignorance can seem blissful for a time, it becomes selfish and pointless all too quickly. NF didn't happen just to my sister. NF happened to my family and so I want to be informed. I have found that information can help to give me back a feeling of control, even when the facts are hard to handle. The shock of the possibilities eventually wears off and then I'm left with a less mysterious cloud over the future.

As I've read personal stories of people with NF I've noticed that even young children with the disease tend to be extremely well informed. It is not unusual for NF patients to know more about this disease than some physicians they encounter and

when my sister visits a clinic, emergency room, etc., she must always be prepared to educate. Imagine having to be vigilant, patient, and clearly communicate your medical situation when you are at your weakest, most vulnerable state and most in need of someone to simply care for you. I considered this the last time I was feeling quite ill. I was needy, grumpy, weak, impatient, and felt quite incapable of tolerance towards uninformed medical care. And I only had a cold.

Eva has always tried harder at everything than anyone else I've known. In spite of the physical challenges caused by NF, she has earned two honours degrees from UVIC. She always follows the rules, whatever they are, because she somehow believes in them. Perhaps never breaking the rules should ensure success and acceptance, I'm not sure. Whatever the situation, my sister always seems able to do what's right and what's expected of her. She doesn't get mad often, is more patient than I'll ever be, and is always willing to help anyone who needs it.

It drives me crazy.

I used to hope that she would toughen up and get selfish - give somebody what-for once in a while! If I had my way I would send her out into the world dressed like a hockey goalie, padding from head to foot, so nothing could hurt her. But then, if I had my way NF simply wouldn't exist either. Evidently, you can't always get your wish, so I continue to work on just accepting that she is going to continue to be herself - giving, selfless and often overlooked.

As the years pass, my outrage grows. I seem to feel the anger that my sister doesn't. From my perspective, the medical community has been astoundingly disappointing in meeting my sister's needs. All too often my sister would return from a physiotherapist visit feeling utterly crushed and hurt. Unable to provide any sort of relief to my sister's pain, and likely frustrated with their own failure, medical professionals would conclude that treatment was not successful due to my sister's attitude. Evidently, negative thoughts were

preventing physiotherapy from relieving the pain. Certainly a person suffering from NF, in constant and increasing pain, is going to feel overwhelmed and frustrated at times. Call me idealistic but I feel that vulnerable people should be cared for, rather than victimized by the health care system.

This damaging cycle has stopped for Eva since she no longer subjects herself to the abuse. In 1994, my sister was finally able to have an MRI and the source of her increasing physical pain was clearly identified.

I recall phoning home for the news. At the time I was in the middle of an extended backpacking trip, happily involved in my own world of adventure, travel and irresponsibility. Reality came rushing back with the news – the MRI had revealed two tumors growing around nerve endings in my sister’s body: one in her spine and another in her shoulder.

Surgical removal of these tumors is simply too risky as she could lose the use of her legs or her left arm (she is very left-handed) and they could grow back. I continue to hope that some relief will result from the fast paced medical research into NF.

Identification of the genes causing NF just occurred in the early 1990s and since that time there has been much activity in the research community. The US Department of Defense has been involved in the congressionally-directed Neurofibromatosis Research Program (NFRP) since 1996, appropriating over \$130 million to basic and clinical research. The goal of this research has been to substantially improve our understanding, diagnosis and treatment of NF.

I understand that clinical and translational research for NF is now feasible. This increases the hope for real treatments to emerge from the laboratory environment and be translated to practice in the foreseeable future. My fingers and toes are crossed.

However, ten years after my sister’s MRI, I’m sure the tumors have grown and I want to know more.

NF increases a person’s risk of cancerous tumors by up to 15% and thus reduces life expectancy by an estimated 15 years. A biopsy has never been done on the tumors in my sister’s spine or shoulder.

I am told that NF often causes psychological and social issues for patients and their families. Whether it is psychological, social, animal or mineral, I worry about my sister’s health and her happiness. Unfortunately as time goes by she tends to keep more and more things from us, thinking that she is protecting us. As the disease advances, the burden of protecting us becomes greater for her to bear. Personally, I want to know every change that occurs; I want her to see a doctor about every new development, every new tumor and every new pain she feels.

Understandably she is reluctant to cater to my whims. I try to respect her privacy, but I’ve never been extremely good at leaving her alone. I am, after all, her little sister. I want the best of care for her and I want to see vigilance displayed in monitoring this disease. I don’t want her to fall through the cracks of the medical system or society’s compassion.

While I know that things could be worse for Eva, I will always want her life to be better. I’m hopeful that research will continue to advance our understanding of how NF functions and soon leads to effective symptom relief and ultimately a cure.

In recognition of Neurofibromatosis month I would like to ask medical professionals to learn just a little more about this disease. As NF can affect any part of the body from brain tumors to bone deformities, all areas of medicine are applicable. It would make a huge difference to the quality of life for NF patients and reduce their burden just a little to be able to deal with a community that better understands this disease. I would really appreciate your help and I know that all of the families of people suffering from NF would thank you for your effort also.

TRACY'S STORY

Like many others, neurofibromatosis was not a word my husband or I had ever heard of. However, the first term we needed to learn was pseudoarthrosis.

Our second born son was sent to the Children's Hospital in Winnipeg to see an orthopedic surgeon at three months of age. This was because of an obvious bow in his lower left leg. We were told he would need to be braced to protect and straighten his leg and that if it ever broke, it was unlikely to heal. We were also told that pseudoarthrosis is often linked to neurofibromatosis, but very few details were disclosed at that first appointment. When we arrived home, a quick trip to the computer to do our own research on NF literally scared me to tears. It just didn't seem fair that my sweet little boy would not only need to wear a brace, but possibly face other trials as well.

It seemed that the moment we learned he might have NF, the cafe-au-lait spots appeared. Next, the fibromas started to develop, every few months we'd find another little bump. By six months of age Seth's fibula broke and as we were warned, it has never healed.

By the age of three, Seth had bigger concerns. The bone in his chest was continually becoming more disfigured and was x-rayed. This led us to his first MRI where we learned of a large plexiform tumour in his chest region extending into his neck and brachial plexus. We also learned of two lesions in his brain.

To this day we continue to play the "wait and see" game with MRIs every three months. We are told surgery would be risky and possibly leave some sort of deficit in our son.

This approach left us feeling drained and helpless. As parents we wanted to fix any pain or problem our child endured, only that wasn't possible.

However, we felt the needed to do SOMETHING.

This began the second part of our journey.

I signed up with the NF Marathon Team for the Children's Tumor Foundation (CTF). I had frequently talked with other parents dealing with NF on the CTF bulletin board. I wasn't a runner, but I was willing to try. It wasn't long before we had an article in the paper and a website up and running. This got the ball rolling on spreading awareness and raising funds for NF. We were finally doing something.

In January 2007 I completed running my first half-marathon in Disneyworld with the NF Team. Our small town helped in raising over \$15,000 which the Children's Tumor Foundation allowed to stay in Canada and go to BCNF research. This also inspired us to organize an annual RUN WITH ME FOR NF event in our town. The proceeds raised will go to different local charities that have helped our family. We raised over \$4500 for the Winnipeg Ronald McDonald House. Over the course of a year, 15 articles have been printed and three TV news reports about NF have aired.

The best thing about doing something is that it has introduced us to many amazing friends and other families also living with NF. It is our hope to start a Support Group in Manitoba. I'm also equally excited to be attending my first NF Symposium in BC.

I know God has a purpose for our lives. If it wasn't for Seth's NF we would never have met so many wonderful people. We are blessed Seth functions



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as a normal little boy. He does well in school and music and enjoys playing with friends and family.

I now think of our lives as a marathon. We have to put one foot in front of the other, even when we are completely exhausted; knowing as long as we stay on the road God has led us to, He will guide us through.



TARA'S STORY

*Speech PRESENTED AT THE JEANS FOR GENES DINNER & AUCTION
ON MAY 23, 1998*

Hi, my name is Tara Turley-Dean. I'm eleven years old, and I know a lot of words I shouldn't know. I'm going to tell them to you. Don't worry Mom – not those words!

Neurofibromatosis. That's one word I wish I didn't know. Chromosome 17. Scoliosis. Tumors. Plexiform Fibroma. Learning Disabilities. MRI. CT scan. Don't you think those are words no kid should have to know? Chromosome 17. That's the chromosome that the NF1 gene is located on. Scoliosis. That's what happened to my spine because of NF. NF made my spine grow crooked. I had to have an operation on my back to fuse eight of my vertebrae. I have a scar on my back. I had 23 staples in my back. I have a scar on my hip where Dr. Tredwell at Children's Hospital took some hip bone to help support my spine. I have a rod in my back.

That's all because of NF. Sometimes NF means "No Fun." NF makes tumors grow - that's the "fibroma" part of NF – they grow around your nerves. When they're big, they're called "plexiform". I have a plexiform fibroma on my neck. Since I talked to you at the last Jeans for Genes, this tumour's has grown bigger. I agreed to be the NF poster girl on the Safeway milk cartons so we could find a cure. I started 5 years ago – and we still don't have a cure. I also have learning disabilities because of NF. I've had to have tutoring to help with language and math. I'd like to tell you what a learning disability is. It doesn't mean you're dumb. Actually, I'm quite smart. A learning disability is the difference between your intelligence and your ability to do something. Like, I know the meaning of a lot of words but I have trouble remembering how to spell them sometimes. The good news is that I've overcome a lot of the challenges I was having at

school. I'm getting C's and B's on my report card just like most other kids. That's good news.

When you have NF you get used to going to a lot of doctors and to the hospital for tests. I go see Dr. Tredwell who is my surgeon. I see Dr. Baumgard who knows a lot about NF. My eye doctor is Dr. Cline. My family doctor is Dr. Irwin. And I've seen a lot of specialists at Children's Hospital. I've given so much blood I could start my own blood bank. Some people think that once you have NF, that's it. The truth is, NF changes as you get older. Usually it gets worse.

Since I talked to you last time at Jeans for Genes, I have good news and bad news. The doctors were worried because I was growing slower than a lot of kids in my class. They thought there was something wrong with my growth hormones. Like a lot of things, they don't know the answers for sure, but they think now that my hormones are okay. It looks like I just might not turn out to be too tall. A lot of people with NF are not too tall. I think I'm just right for being Tara. Besides, I'd rather just go watch the Grizzlies than be on the team anyway. Just this week, Dr. Tredwell examined me and said that my bone graft in my spine looks good - and that it's the best he's seen – and he's seen a lot. That's the good news. The bad news is that the plexiform tumor on my neck is bigger. It looks like it may be damaging the bones in my neck. I have to go back for an MRI and a CT scan. Sometimes I have to lie very still in the machine for hours. That's very hard for a kid. X-rays are nothing. I've

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had tons of X-rays. But the MRI and CT Scans show the doctors what's going on in the nerves and soft tissue of my neck. If the tumor is pressing on the nerves in my neck, they may have to operate to take out some of the tumor. That's the bad news.

And that's why all of you are so important. You could be doing lots of other things tonight: but you spent your money to be here and to help support the BCNF. A lot of other people will be thanking you for coming tonight. But I'm a kid, and when I grow up, I want to see a world without NF. So I want to thank each one of you for being here tonight. Your help makes a difference. Thank you.



UPDATE ON TARA

By Tara Turley-Dean, age 18

Wow! I have not written about living with NF in a long time. When they asked me to update my situation and I saw my old photographs from the last time I did this, I realized a lot has happened. I am now 18 and a Fine Arts student at Simon Fraser University in Burnaby. As of this time of writing I am heading off to live in residence and starting my studies. I intend to major in theatre. Acting has been my passion for as long as I can remember. This transition in my life, from high school to university, has been exciting, nerve-racking, stressful and daunting. I am not exactly sure if I am ready for the challenge, but I am doing it anyway.

High school years were not particularly the “best years of my life”, but I managed to escape mostly unscathed. From Grade 8 to Grade 11, I had four surgeries. The first one in Grade 8 was to further correct my scoliosis, with a posterior fusion of my upper thoracic and lower cervical spine. (Note: those of you that remember me will know that I had my first spinal surgery when I was 8 years old). Dr. Tredwell, of Children’s Hospital, removed my old rod and placed a newer one higher up. The post-operative result was a back/neck brace that I had to wear for four months. This being my first year of high school was traumatic enough, but with the brace I made the executive decision to remain at home for the rest of the school year and be home-schooled.

Time for the out-of-left-field bizarre operation, which we were told had nothing to do with NF; I noticed wearing my glasses one day that it was time for a new prescription because I was not seeing so well. It turned out that I had a partially detached retina in my left eye. This is usually caused by trauma to the eye. However, I had never had any kind of trauma save for the time when I

was three years old and I nosedived into the marble coffee table. The operation was a success and the recovery time was minimal.

Thinking I was clear of operations for a few years I went about my business of being a teenager. Barely five months later, which was less than a year after my last spinal surgery, I found out that I was facing even more drastic spinal surgery. In November of grade 10, Dr. Tredwell performed a nine-hour surgery on me, with an anterior incision from neck through sternum to stabilize the front of my spine using mesh cages. My Mom and I joked that “it must be serious”, considering for the first time we got the gold member’s private premier room, right across from the nurses’ station. This surgery required my longest stay in the hospital to date – nine days – including two full days in the ICU. On the last night neither my mom nor I could sleep and my mum ended up packing the car at 4 am - we were that excited to get me home! I also had to wear another back/neck brace for four months. However, I only did home schooling for two months while I was recovering and returned to school wearing the “frick’n” brace. And not enough that I was a freak already, I was not allowed to wear a backpack, so I felt like a flight attendant pulling it on my trolley behind me. “Please fasten your seat belts and put your chair in the up right position.” I think nearly everyone in my school, including myself, tripped over the trolley at least once.

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Last but not least, my vocal cords were damaged from this most recent surgery, so I had another minor operation a year later to restore my voice as much as possible. In some ways this was the toughest thing to recover from; it slowly dawned on me and my family that my voice was going to stay as a hoarse whisper. It was bad enough that I could not be heard at the family dinner table, let alone on a big stage. However, the Vancouver Youth Theatre (VYT), where I performed, was very supportive and let me act even when it was not a guarantee that the surgery would be a success.

Perhaps the best part about having NF has been the NNFF Annual Summer Camp in Utah that I have attended for the past five years. It is kind of hard to explain, but getting to know other people my age that had some idea, if not more of an idea what it is like to be “different” made a world of difference to me. For the first time I actually felt “normal”. I have made many, hopefully, life long friendships. My friends and I make an effort, since we are so spread apart geographically, to keep in touch in any way we can.

Like my mom, former president of BCNF, it has been a number of years since I have been very involved in the NF cause. For a long time I was the BCNF poster child, appearing on Vancouver buses, milk cartons and coin banks and giving speeches at fund raisers. Since I am venturing off to university into something huge and unknown all by myself, this has been a really good time for me to reflect on all I have been through since you last heard from me. Leaving home is a new feeling for me. If I had to have another surgery, sure, I know I would be ticked off, but I have done it so many times I know what it is and that I could probably handle it. University, on the other hand, is something entirely new to me. I do not know what to expect, what to do and if I will be successful in my endeavours. However, if I look at it like I have grown to look at my life – it is just another step – another tier on the cake, then why not? Bring it on.