

GENETIC ALLIANCE MONOGRAPH SERIES NUMBER 2


# GENETIC TESTING STORIES

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PUBLISHED BY GENETIC ALLIANCE



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# Preface

Here is a collection of incredible stories: the stories of men, women, and children who have suffered from uncertainty, misdiagnosis, a lack of options and, in some cases, tragic loss. This is also a collection of stories of hope and the search for answers, truth, understanding, and options. Overall, they reveal the raw interface between knowing and not knowing, and our search as humans to understand and strive for the best.

We give thanks first to the brave individuals who were willing to share their stories. They offer us a window into the world of diagnostic odyssey: a sometimes quick and effective label and other times a very long and frustrating journey. These are the pioneers in a new land of promise. But they are the ultimate pragmatists as well—they need safe and accurate tests, to be used as tools in the healthcare continuum, without undertaking an advocacy role for themselves.

We offer this collection as a panoramic, qualitative view of a small subset of the experiences of real individuals engaged in real testing.

# Carrier Testing

## 22q11.2 Deletion Syndrome

*Adult/Newborn*

In July 2001, my son was tested for the genetic mutation that leads to 22q11.2 deletion syndrome. The doctors hoped that the testing would confirm our son's diagnosis, which had been based on his Tetralogy of Fallot and immune system defects, while we were trying to understand all of the variables and specialists that were reviewing our son's progress. My husband and I were tested later that year to determine whether our child's condition was familial or spontaneous.

The tests required our informed consent. We also spent a great deal of time pleading with our insurance company, requesting that they cover the cost of the tests, which involved fluorescence in situ hybridization to detect the mutation. Our son's results took just a few days, while the results for my husband and me, took about a week. Our son tested positive for the 22q11.2 deletion, while my spouse and I tested negative. We discussed the results with a geneticist, who then offered us further information in the form of research study materials. We were not offered any support group resources at that time.

While we were devastated by the results from our son's test, the compassion we received from medical professionals provided us with the strength to persevere. Our son had open-heart surgery on the day that my spouse and I were tested. Needless to say, we were feeling extremely upset at the time. The quiet competence displayed by the health professionals, along with their willingness to explain the nuances of our son's condition, helped ease our pain.

*Editor's Note: Fluorescence in situ hybridization is a technique that is used to study a particular region of a person's chromosomes, the structures in a cell that carry genes.*

## Adrenoleukodystrophy

*Adult/Child*

In 2003, my son was diagnosed with adrenoleukodystrophy. We had a genetic test performed in order to discover the specific mutation he has. At the same time, my daughter, nephew, and I were all screened for the condition. The following year, my mother and brother were also screened. During my son's initial hospital stay when he received his diagnosis, we first met with a genetic counselor and then ordered a blood plasma test kit (VLCFA). It took about 10-14 days for us to receive the results in the mail. My daughter, mother and I were all identified as carriers, while my nephew and brother are unaffected.

After the testing, the only information we received was a website print out. We did not receive any guidance from the doctors, and I had to educate myself. As a result, I started my own foundation in an effort to educate both medical professionals and the community at large about this condition. While the testing was simple, dealing with the results has been traumatic.

## Albinism

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*Adult/Child*

My husband, son, and I were tested for albinism through a blood draw in 1996. We initiated the testing ourselves as we were seeking more information, particularly for family planning purposes. We spoke with a field expert at a conference and then had our blood drawn by our own doctor.

Because we used the grant program offered by the expert, we had to wait over two years to receive the results by mail. The results were presented in an easy to understand format and indicated that my son had albinism. While neither support nor materials were offered, we did not ask for them or feel they were necessary.

Since we had initiated the testing ourselves, we did not think the experience was a big ordeal. After the testing was completed, we had five more children, two of which have been diagnosed with albinism as well.

## Cleft Palate, Chromosomal Abnormality

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*Newborn/Adult*

My son was born in 2005 with a cleft palate and we had no family history of it. After his birth, my parents, my husband, and I were also tested in an effort to see who carried the cleft and how it was passed down through the family line. A fluorescence in situ hybridization test was used to study our chromosomes. I was unconscious when my son was delivered and did not come to until the next day. Therefore, I did not sign a consent form for the original test, and in fact I am not sure if my mom signed a consent form or not. All of the testing was done in the NICU, but it was not until he was out of the NICU that we found out more of the test results and where to go from there.

Within a few weeks, the results were given back to the genetics team and then we had a meeting afterwards to discuss the results and what we needed to do from there. The fluorescence in situ hybridization test showed that my husband and I have a balanced translocation. (We have an extra piece of chromosome 21 on chromosome 10.) It turns out that I might have had a cleft but via the muscle. The results mainly sent a mixed message because the results were so rare. It was determined that more studies needed to be done and we were given the opportunity to participate in a research study. Fortunately, the genetics team was there to answer my questions every step of the way. I found the experience to be rough and confusing. It has been helpful to know that I have a balanced translocation, but the unknowns that are out there are still scary.

*Editor's note: A balanced translocation refers to the rearrangement of genetic material between at least two of the 23 pairs of chromosomes. In this case, it sounds as if a piece of one chromosome 21 was inserted into one copy of chromosome 10, but because this rearrangement appears even and did not result in any obvious extra or missing genetic material, it is referred to as balanced, rather than unbalanced. Individuals that carry a balanced chromosome translocation usually do not have any associated health complications; however, they are at risk to have children with an unbalanced chromosome translocation and associated birth defects.*



## Cleft Lip

*Adult*

In 2005, my husband and I underwent genetic testing. The tests were performed to determine whether either of us had a mutation that would explain our son's midline cleft lip. We were thinking of having more children and wanted to know whether we were carriers of anything.

The testing was a joke. We were never required to provide informed consent and nothing was explained to us. When our blood was drawn, the hospital staff had to call for confirmation as they didn't even know what tube to put our samples in.

We waited several months for the results, which were eventually delivered over the phone by a doctor. According to the tests, we were fine, but I still feel like something was missed. We didn't receive any support either, even though my son has a rare craniofacial condition. It was as though no one in the medical community really knew what was going on. I had to do all of my own research.

Looking back, the testing was not very helpful. We are still looking for answers. The fact that our insurance company gave us such a hard time didn't make things any easier. We had to pay for most of the testing expenses ourselves. My son also went back to a genetics doctor in August of last year. It was November before the doctor speculated that he had Aarskog Scott syndrome. I was told that it wouldn't be a 'true' diagnosis until he undergoes a genetic test that costs \$3,000. I didn't know when that would happen.

In September of 2006, I switched my son's pediatrician. I felt that this doctor was the person holding us back. He was the one who ordered the genetic testing of my husband and me, but he refused to order the genetic testing for my son. I requested the insurance company cover the test. They denied the request. I appealed and they accepted. We went to a new local doctor who agreed that something was going on. We were referred to a pediatric endocrinologist because of Robert's small stature.

Upon meeting the endocrinologist, we were relieved. We finally had a doctor who would listen to us. Testing came a week later and it was confirmed that my son had diabetes insipidus and growth hormone deficiency. Since there were two areas of his brain affected, an MRI was ordered. They found a deadly secret in my son's head that he had been carrying his entire life: a very large sphenoidal encephalocele. This means that there is a bone missing in his skull that keeps his brain contained in the skull. His brain is filling his sinus cavity and sitting on top of his palate.

We are currently deciding the best course of action for my son. Surgery is a high probability as it is highly dangerous to have your brain in your sinus cavity. Free communication between your brain and your snout is not ideal for anyone.

Robert has since received a high-resolution chromosome test—we are still awaiting the results. My husband and I are trying to have another child. My son has three of the most rare diseases or birth defects currently documented. Despite all of that he is a blessing and a joy to be around. We will only be as lucky if we are blessed with a child JUST like him—issues and all.

## Duchenne Muscular Dystrophy

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### *Adult/Child*

My son was tested for Duchenne Muscular Dystrophy (DMD) when he was three years old (he's 10 now). Only when his diagnosis was confirmed (three tests and four years later), could I be tested to determine whether I was a carrier. The test was to determine a mutation responsible for DMD. It was administered because his CK enzymes from a prior blood test were sky high. Although this is a good indicator of DMD, the genetic testing would offer more information. It was particularly important for us because we had never heard of Duchenne and had no family history anywhere in our lineage. We did a mutation detection test; more specifically, I believe it was a western blot. Both times the test came back negative. My son did not have a "typical" deletion and I was told that this test only picks up about two-thirds of DMD deletions. He was three when we started this process between 1999 and 2000.

After my son was diagnosed at age three, we were sent to a genetic counselor at a local hospital where the tests had been ordered and sent out to another lab. It was still important for us to discover my son's cause of DMD for two reasons. First, we needed to know his deletion before I could be tested to determine if I was a carrier. Although I had a second son without DMD, we were still interested in information to assist us in future family planning. Second of all, there was a lot of research being published that was specific to certain types of mutations.

In 2001, I went to a conference put on by a muscular dystrophy advocacy organization, and I met a researcher from a university. The doctor and his team had developed a testing methodology to sequence the Duchenne gene. I was told the sensitivity of the test would pick up close to 99% of mutations. We agreed to participate as part of the research, and in April 2003, we found out my son had a premature stop codon at exon 26. It provided us with more information as well, including the specific nucleotide, the nucleotide change, the codon, the amino acid and the amino acid change. The same lab tested my blood, and then I was provided with the opportunity to discuss the results at length by phone with the genetic counselor involved in this study. It was determined that I was not a carrier. She further explained that it was still possible to have a germ line in my eggs, so not being a carrier did not necessarily guarantee I wouldn't have more children with Duchenne.

The test, as part of research protocol, did include informed consent. I spoke by phone with both the researcher and the genetic counselor. When the results were first available, the doctor called me to go over the results and explain what it meant for my son. We talked for a good half-hour. It did take several months to get the results from the time we sent the blood in and much longer to receive the results of my carrier status. The latter, I believe, was because they were inundated with samples from around the country at this time. Although the doctor called with the results, I was also sent a report by mail.

My impression of the first two testing experiences and the support I received was not very positive, but of course, I didn't get results. The research study was much more positive and hopeful, although it was time consuming. I felt the information provided was excellent, with knowledge of the mutation being the most beneficial. We parents are hungry for knowledge. We want to do whatever we can to support our kids. The doctors and the genetic counselor were respectful and

professional during and after the study. I am so glad we discovered this methodology was even available. And, of course, as research, there was no cost to our family. Prior to finding this researcher, we were told that we could have his gene sequenced at the only facility doing it, which doesn't accept insurance for that test. The cost would have been upwards of \$1,200 out of our pocket, which we couldn't afford. Information gives us hope and something to work with. Based on the genetic results we received, my son is now participating in a clinical trial with a drug they believe can read through premature stop codons. The hope is that it will turn the stop codon back on.

*Editor's note: A germline mutation refers to a genetic mutation that is only present in the germ cells (egg or sperm) of an individual. It is not possible to obtain and test all of the germ cells a person has to determine which cells and how many cells, if any, carry a mutation. Therefore, the risk of that person passing on a germline mutation to a child cannot be predicted.*

*Usually very large deletions in genes are tested for using a Southern blot, while a Western blot tests for the protein that a gene produces. A premature stop codon is the result of a mutation in a gene that completely interrupts the gene and prevents it from making the normal protein that it should. The hope is probably that this drug will reverse the effect of the stop codon and lead to the production of normal protein again.*

### Fragile X

*Adult/Child*

My family decided to get tested because our children presented some symptoms like dyslexia and developmental delays. My nephew and my son were both tested for fragile X syndrome using a chromosomal test but these tests both resulted in false negatives. Then, in 1997, when the DNA test became available for us, all of my family got tested for fragile X using a DNA-based mutation detection test: my parents, my nephew and niece, my sister (their mother), my other sister and her three children, my two sons and myself, my two aunts, one uncle, and their offspring.

When we first heard about testing for fragile X (from my nephew's neurologist), we asked him a few questions and went on the Internet. Then we had a discussion with the neurologist and the biologists who were in charge of testing us. They were very supportive! About two months after the testing, we learned our results in a session with the biologist who performed the test. We learned that 12 of us were carrying the pre-mutation and that four of the children had the full mutation. The results were clear but as a result our future became unclear, so we asked questions again and again trying to understand what it all means for our children and for other family members. The neurologist was, and is, always available to answer all of our medical questions. The biologists answered our questions, provided us with genetic counseling, and encouraged us to found a fragile X parent support group. (The biologists even invited experts and joined us at a fragile X meeting to launch our Association.) They are always there for us. They also provided us with references and articles about the syndrome.

Learning that we have transmitted a genetic mutation was stressful, but it was a relief to learn the cause for our children's symptoms and to be able to give the disease a name. It gave us the chance to learn about the syndrome, connect with people like us (it helps decrease the sense of aloneness), and gain an increased ability to deal with the day-to-day issues. The problem is that, at present, there is

no cure for fragile X, so we have to face the problem without having a chance to solve it completely. The genetic condition has limited our options, but having knowledge about the condition has expanded our horizons.

## **Hemophilia A**

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*Adolescent/Adult/Child*

In 1986, my son was tested for hemophilia A. My daughter, two of my younger sisters, and I were also tested for carrier status at that time. Very little information was given to us about the test. It was mainly the doctor asking if we wanted to find out if the other females in the family were carriers and we then scheduled a date to have our blood drawn.

After about one month, the results from the test were delivered by mail. Our son's diagnosis was confirmed. I already knew that I was a carrier, my daughter tested positive as a carrier, and one of my younger sisters did as well. My sister's letter mentioned someone else's name though, so I called the hospital and spoke with someone in the testing department. They just brushed it off and said that it was correct, even though the name was mixed up with someone else's. We didn't really receive any support after the results were delivered. My sister was 15 at the time and my daughter was four. There should have been some more information given to my teenage sister because she had questions when she got married and was trying to decide if she should have children or not. I had to be the one giving her advice, suggesting that she talk to a genetic counselor. While it has been beneficial just knowing our statuses, it was also very frustrating to have been given that type of information without much follow-up.

## **Hirschsprung's Disease**

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*Adult*

In 1999, I was pregnant with my second child and we wanted more information about the possibility of a repeat of the unusually severe case of Hirschsprung's Disease in my daughter. I'm not sure what kind of genetic test was performed, but the doctor took a blood sample and determined that there was probably not a genetic connection for my first daughter's case.

The testing process consisted of an informed consent and two meetings, an interview, and a discussion of the results. The results discussion occurred a few weeks after the test. We sat with the doctor for about 45 minutes to an hour, and the doctor was matter of fact but kind and reassuring. Because we knew we were having a girl, the doctor could say that there was a 7-15% chance that our second daughter could have Hirschsprung's—but the chances were even slimmer that if she had the disease it would be as severe. We understood the doctor's discussion of probability, but we already had read this information in the literature. It was good to talk to a professional about our worries, but we were hoping to find out more information than we already knew, and we did not.

## Oto-Palatal-Digital Syndrome, Type II

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*Child/Adolescent/Adult*

Within a few days of my son's birth in 1989, we learned he had oto-palatal-digital syndrome, type II, an X-linked chromosome disorder. Following his diagnosis, we received genetic counseling (from a geneticist who provided full and open information) and did extensive research on the disorder, its problems, the likelihood of carrier status—the full range of issues. The genetic marker was not known at the time of his diagnosis, however, the geneticist pointed out several physical characteristics, in myself and my mother that showed minor manifestations of the disorder. When the actual marker was identified, we were frustrated at the amount of effort required to get the testing and the refusal to test all three of our daughters for their carrier status. My husband and I are well-educated with non-medical Master's degrees, and we feel strongly that we should tell our children their status at what we believe is the appropriate time, rather than having them learn this information from a man or woman in a white coat who spends a few minutes with them in a conference. We have prepared them all of their lives with an understanding of both the risks and benefits of the disorder and have instilled in them the attitude that knowledge is power—knowledge of your body and health, in everything you do. We never stop learning and believe that knowledge of your own body is essential.

When enough information was available for mutation detection and screening for carrier status, my son and I were tested in 2003, and I was not surprised to find out a month later, after more “counseling,” that I was a carrier. After more discussion and counseling and signing of the informed consent forms, my 17-year-old daughter was tested in 2005. After about a month, we were told in person that although I am a carrier for the syndrome, my daughter was not. Due to our family history of the disease and our knowledge base, we found the results to be very clear and were provided with no additional materials other than those we had already seen. However, we were offered future counseling.

Although we found the experience to be incredibly difficult, it was very beneficial for my daughter and me to receive a definitive answer regarding our carrier status. The many counseling sessions were meant to be helpful, but it really seems as though the sessions ended up being a waste of our time and effort in many cases, and we actually felt insulted by the condescending attitudes we sometimes encountered. Parents of a special needs child have to learn much more about medicine than they ever wanted to in an effort to ensure the health and safety of that child. Genetic testing was just one more piece of that maze that we have to navigate when raising a special child.

# Diagnostic Testing

## 18p and 22q Deletions

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### *Prenatal*

Due to my advanced maternal age and concerns about 18p and 22q deletions in my unborn child, I underwent chorionic villi sampling (CVS) in January 2002. I did not have a good impression of the center where the testing took place from the start, as the only preparation I had was the explanation my obstetrician gave to me about what was actually going to happen during the procedure. I did have to fill out some paperwork and answer some general questions.

After waiting one very long week, we received the great news by phone that everything came back normal. I did not find the results to be very clear however, because they did not explain why the females born on my dad's side have clefts. I did not have a chance to ask anyone about this either as no support was given. The testing itself was very scary because of the possibility that my child had an extremely high chance of being physically and mentally challenged due to the fact that I exhibit some traits of the syndrome and have a lot of bad stuff in my family history. It was very comforting to know that my child was going to be okay and that she was a girl. However, I wish the staff at the center had not been so cold and uncaring.

## 47 XXY

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### *Prenatal/Child*

In 1996, my son was diagnosed with 47 XXY via amniocentesis. The amniocentesis was performed as a routine procedure, since I was 35 years old during pregnancy. The results from the amniocentesis were then confirmed with another test when he was six months old. I don't know the exact nature of the tests performed, but they helped to identify whether our son's condition was a mosaic or "full." Both sets of tests included informed consent.

The results from the amniocentesis were received approximately one week after testing, while the results from the second test took about 10 days. Both were delivered by mail. Regarding the results from the amniocentesis, the prompt genetic counseling we received from genetic experts at the hospital within 24 hours was the most beneficial. While we discussed the results from the amniocentesis at length, we already knew the outcome of the second test before the results were delivered. We were encouraged to see our doctor and to join support groups. My impression of the genetic testing via amniocentesis was extremely significant. Pregnant women say, "I won't have amniocentesis because I wouldn't terminate anyway." The purpose of prenatal testing is for identification; it has nothing to do with one's pre-intended responses. Being informed allowed my son to be eligible for early intervention. I encourage all women who are eligible to have prenatal genetic testing.

## Adrenoleukodystrophy

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### *Child*

In 2004 my son was tested for adrenoleukodystrophy (ALD) because my father had a positive diagnosis. I'm not sure exactly what type of test was done but it was done using blood. The preparations for the test included providing informed consent, having a discussion, and receiving printed materials. After about two weeks, we received the results by mail and e-mail. The test was positive for x-linked ALD.

The results were clear and very informative. We were glad to get the results quickly and receive support and information. Unfortunately, the test was expensive. However, we chose to get our second son tested as well. Thankfully, he was not found to have the condition.

## Albinism

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### *Child*

In 1995, my son was tested to determine his particular type of albinism. I really have no idea what kind of test was done but perhaps it was a blood test. To prepare for the test, we gave our consent and met with a geneticist. I do not specifically remember how long we had to wait for the results, but they were delivered during an appointment.

From the test results, it was clear that my son had OCA1 albinism. I do not really think it was necessary for us to find out the specific type of albinism. We were provided with information afterwards, but we did not receive any support, which I did not feel was necessary anyway. When we switched healthcare insurance due to my son having a confirmed diagnosis, we discovered all services related to albinism were not covered.

*Editor's note: OCA1 albinism refers to oculocutaneous albinism, type 1.*

## Albinism

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### *Child*

We adopted our daughter from China in 2003. She was in special need of adoption because of her albinism, for which we had her tested in 2004. She was two years old at the time. Doctors had been unable to make a definite diagnosis because my daughter's eyes were not characteristic of albinism. The genetic test was performed to make a diagnosis and determine whether her condition was caused by a mutation.

We were able to discuss the test ahead of time and provide our informed consent. We waited two months for the results, which were delivered by phone and mail. Unfortunately, the results were inconclusive. She has a gene that is "flipped," but the doctors do not know whether the gene is important or how it might affect her future. They offered to do another test for albinism, but so far we have declined. We would retest to double check the results, but our insurance will not pay for additional testing. We scraped her cheeks for a study at a university, but we have not gotten any results back yet. We were not offered any support after the tests and now feel that our daughter went through it all for nothing. We did not learn anything.

## **Albinism**

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### *Child*

When my son was four months old in 2005, his pediatric ophthalmologist suggested we have him tested to determine his specific type of albinism, as pediatric ophthalmologists only focus on the eyes. In addition to determining the type of albinism, we wanted to use it to rule out any of the possible accompanying conditions. Originally, we thought it was just ocular albinism, since he had so much pigment in his hair (brown eyelashes and eyebrows) and skin.

Before the test, various doctors examined him and looked for other related genetic problems. Then they took blood from his arm. About four weeks later, we received a phone call stating that the results were ready. We returned to the hospital where we met with a geneticist to go over the results. We learned that my son was heterozygous for oculocutaneous albinism (OCA) type 1A/1B and is temperature sensitive. The results were not clear at the time, and I still do not understand all of the different types a year and a half later. The support that I received was all related to the genetics of the condition, such as how it can affect any of my future offspring. The only printed material I received was the report from the genetics lab that tested his blood.

While I wanted to know all of the information that I could during the testing, I no longer think that knowing the type makes any difference because every individual is different. It was helpful to know that he has OCA instead of ocular albinism, since he was born with pigment. It was recommended that we return in a year for follow up screening, but since I do not see the need, we have not and will not do so. A hearing test was also recommended for my son when he turned one, as hearing problems can often accompany his condition.

## **Andersen Syndrome**

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### *Adult/Child*

Three of my children and I were tested for Andersen syndrome. I was tested first because my mother had a diagnosis of Jerville-Lange-Neilson and hypokalemic periodic paralysis (HPP). I found out that I had HPP since birth, but it went unchecked for 30 years until a prenatal scan showed my first child had a pericardial effusion and questions began to be asked. We wanted to have my kids tested for Andersens because the disease carries a risk of fatal and preventable arrhythmias. However, it took a lot of persuading to convince some of the doctors that there was reason enough to test them before they were able to give consent. (They were two, three, and four years old at the time of the test.) The purpose of my test for Andersens in 2000 was to look for common mutations, which allowed them to look for the same mutations in my children in 2005. Because my testing was done as part of a research project, I provided my informed consent and blood sample at the same time. In fact, little counseling was given at that time. I felt the research fellow was excited at the prospect of confirming another diagnosis. However, the results took so long to come back that I had been discharged from the clinic without a diagnosis and had gone on to have another child by the time the news came. It was very hard to organize the testing for my children, as it involved lots of discussions and appointments. I did not receive any written materials in either instance.



While I had to wait over two years for my own test results, the results of my children's tests were delivered in approximately four months. My test results were given in the form of a letter just six weeks after my second child was born. I found this appalling, as they had gone to great pains to say that I did not have this condition clinically and had even discharged me from the cardiac clinic. When I was discharged from the clinic, it made me feel almost relieved, but I do not think I really believed I did not have it. It also felt like a bit of a fraud. It was made worse by this letter coming out of the blue. I did not think it was right to be given this kind of news in a letter. After all of the results were in we learned that I had tested positive along with one of my children. As you can see it has all been a bit of a nightmare, but we now feel we can move forward, and certainly as far as the kids are concerned, it has clarified their management. I accessed counseling through my general practitioner when we were having a hard time getting doctors to test the children and while I was coming to terms with my own diagnosis. No other specific support or literature was offered. Instead, I had to find it myself.

Although we have benefited from having a confirmed diagnosis for myself and my children, including knowing that two of them don't have it, it is a shame that no one knows much about how best to improve the symptoms, though maybe that is for the next generation. After all, my mother—who certainly also had this, but died in 1998—never had the right diagnosis. I hope that my son will grow up in a world that learns how to help him with his symptoms. There are also numerous measures that can be taken to improve this process for those involved, as it has been hard for me to relive my experience to write my story. I was made to feel defrauded by the doctors. Additionally, I did not feel that being informed about the results by letter was appropriate. I also had to do a tremendous amount of fighting to get the tests for the children and to get the doctors to actually listen.

### **Andersen-Tawil Syndrome/Long QT Syndrome**

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#### *Adolescent/Adult*

More than 20 of my family members are affected with a “channelopathy” that causes heart arrhythmia, migraines, and periodic paralysis. In 2002, my family participated in a research study conducted by a cardiologist. His lab tested my husband, my four children, my brothers and sister, my two aunts, my 20 cousins, and me for what I believe is the Kir mutation. The testing was done to determine if my family's symptoms were a result of Andersen-Tawil syndrome or Long QT. I do not know what kind of test was performed but the testing did include electrocardiograms (EKGs). Before the tests, we filled out informed consents, had multiple meetings, and received printed materials about the testing. Within six months to a year, we received our results through a doctor. We were found negative for the known mutations but I don't know what specific mutations were tested. Since this was done for research, there was no support provided. However, the person who conducted the test for the cardiologist was a gem!

Since then, two more doctors have tested us, but no answer or follow-up has happened so far. We are currently waiting on test results for my 14-year-old son. A geneticist is retesting for the most common mutations of periodic paralysis. The last hope is a doctor in Germany, but the cost and preparation of shipping is an obstacle. Our mutation is still unknown and no one seems to be researching it yet. Research leaves too many holes and questions and gives no benefit to the family.

## Aneuploidy

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### *Prenatal*

As a prenatal genetic counselor, I have had numerous experiences with genetic testing, as I am often the one delivering the results. However, in 2004 I underwent genetic testing myself, as my maternal serum screen and ultrasound indicated that I was at an increased risk for aneuploidy. In order to assess this risk more formally, an amniocentesis cytogenetic test was conducted.

To prepare for the test, I had a discussion during which I signed the required informed consent. Within two days, I received the outcome from the FISH test and the finalized results shortly thereafter, which were delivered by my obstetrician. It turns out that the results, which were presented in an appropriate manner, came back normal. Due to the nature of my profession, I received a lot of support throughout the process from my colleagues. I was not supplied with any additional materials, but I did not feel that any were necessary.

Reflecting on the experience, my encounter with genetic testing was a great one for having to be in such a situation. As a genetic counselor myself, I was very fortunate in that I had access to my colleagues for support. Additionally, I leaned on my obstetrician for support more than I would have thought. While I did not feel any additional information was necessary, my husband felt that he did not receive as much information as he would have liked.

*Editor's note: Cytogenetics refers to the study of chromosomes and a cytogenetic test is a test that examines the chromosomes of a person or fetus.*

## Cardio-Facio-Cutaneous Syndrome

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### *Child*

In 2006, my son was tested for cardio-facio-cutaneous (CFC) syndrome. He was 11 years old at the time. Though he displayed characteristics of CFC syndrome early in life, my husband and I had decided to hold off on having him tested. We were waiting until our son needed some sort of medical treatment, at which time we would go ahead with the testing. One day, he fell and broke his two front teeth. Surgery was required to repair the damage. I then contacted his pediatrician, who made all of the plans to have blood drawn and sent for testing. The hospital's anesthesiologist was very helpful in moving the process forward. If it had been left up to the hospital staff to have the blood drawn, it would not have gotten done. The blood was drawn while my son was in surgery and then given to us. It was all very simple, since my son did not have to feel the blood being drawn. We kept the blood refrigerated at home and sent it to be tested the next business day.

Our son's pediatrician received a letter from the lab approximately two weeks later. I then went in to pick up the letter with the results. I don't know what type of test was performed, but the results showed that my son had tested positive for the BRAF gene mutation, confirming his CFC syndrome diagnosis. It has been very beneficial to know the results of his genetic test, though our son had to break his front teeth before we decided to have the test done. I wish that it hadn't come to that.

## **Cardio-Facio-Cutaneous Syndrome/Noonan Syndrome/ Costello Syndrome**

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### *Child*

Earlier this year, my child was tested for cardio-facio-cutaneous syndrome (CFC) as well as Noonan syndrome and Costello syndrome. The goal was to confirm that she had CFC syndrome, while simultaneously ruling out the other two. Preparation for the testing was very easy for me as our geneticist set up all of the necessary procedures. Additionally, the blood draw was performed during another surgery, in order to minimize the impact of the procedure for my daughter.

Three to four weeks after the tests were performed, we received all of the results. I learned the negative result of the CFC test through a letter, whereas I received both a letter and a telephone call to inform me of the negative results for Noonan syndrome and Costello syndrome. Because I received a phone call, the results of the Noonan syndrome and Costello syndrome tests were very clear to me. Conversely, I was left to try and understand the CFC paperwork without any additional explanation and found myself very confused because everyone thought my daughter had CFC, yet the results came back negative.

While I was left with neither an overall positive nor negative impression of the experience, I was not offered any support or materials to help me understand. It was helpful to know that she does not have Noonan syndrome or Costello syndrome, but I found it hard to understand how she could still be considered to have CFC without any proof to support that diagnosis.

## **Cardio-Facio-Cutaneous Syndrome/Breast Cancer**

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### *Adolescent/Adult*

In 2006, my 13-year-old son was tested for cardio-facio-cutaneous syndrome (CFC). He was part of a research study and was tested to confirm that he had the mutation. In preparation for the test, I did my own informed consent and filed it in the files for our organization. About two to three weeks after the test, I received an e-mail from my very own genetic counselor and friend. She told me the results had been faxed. I went to her office the next day to pick them up and she reviewed them with me. No surprises there! My son was positive for the MEK2 mutation. My genetic counselor printed off the list of amino acids and highlighted the two that are missing. The science teachers that work at the high school with me have offered to give me a review lesson on proteins and how amino acids bond, but I am too busy right now.

Knowing that my son is now officially diagnosed with CFC gives me peace of mind. However, dealing with the insurance company has been a very stressful situation. A hospital refused to draw my son's blood even though I had doctor's orders, as well as all of my information from the lab doing the testing, because there was no agreement with my insurance company. My insurance company told me to go and get the testing done with no prior approval! I'm not sure if my insurance company will pay for my son's test. I am finding many parents in the US are paying out of pocket for the CFC testing. This is no longer a problem with only our foreign families! I am going to collect data on our group so we know exactly how many were covered by insurance for testing and how many were not.

With my son's testing completed, I can now start being genetically tested for the variant mutation that my first cousin has in BRCA 1 or BRCA 2. My mom and two of her sisters have all died from breast cancer. The situation with insurance problems continues with this new testing. My insurance has refused to test me for the full gene screening. They will only test me for my familial variant. This is in light of four family members who have had breast cancer and my positive testing for malignant melanoma. Yet despite these statistics, I have been told that I do not meet the medical criteria for the genetic test. My fight for genetic testing has not ended with my son's CFC syndrome. I am now prepared to find out about my own genetic health condition. My son will need assistance with all aspects of life as he ages. I am prepared to be here as long as I can.

## **Cleft Lip and Palate**

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### *Child*

Because of my son's physical characteristics, he underwent testing in 2003 to determine if he had a genetic disorder that was responsible for his cleft lip and palate. Prior to the test, we had a discussion with the doctor and a physical exam was completed. Within just a couple of weeks, I received a letter in the mail explaining that at that point in time there was no particular explanation for why he had a cleft lip and palate. The test also indicated that he was at high risk of having a child with a cleft, and I was also at high risk for having a second child with a cleft due to the severity of his defect. The results did not tell us anything new, although we were offered support after the test.

## **Costello Syndrome**

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### *Child/Postmortem*

Starting in September 1995, my son had multiple genetic tests for multiple conditions. He had evident dysmorphic features and a host of medical issues, but we did not know the cause. Because we had so many tests, we had multiple discussions and informed consents, but there were no printed materials about rare syndromes at that time. In fact, if there were any materials offered about the syndrome my son was eventually diagnosed with, they would be prepared by my organization that I have since created.

The amount of time needed to get the test results varied. For one test, I called the lab back after six weeks—only to discover that we had to provide another cytogenetic sample. We would receive each of the results at a doctor's office visit; every test came back "within normal limits." I appreciated the honesty of the doctors when they shared that they were really stumped. After my son died, and with the recent findings of a mutation associated with the syndrome that my son was clinically diagnosed with, researchers who already had samples of my son's DNA notified me that they had performed a test for the HRAS mutation associated with Costello syndrome posthumously, without my knowledge. This test came back positive. With the notification, I was invited to ask the researchers if I had any questions or concerns.

I would have liked to know up front the step-by-step process involved with each test, how long each test would take, where the samples went to be tested, and who to contact if I had any questions or concerns about the process. I had to find the contact information for the labs on my own. Once, although I hand-delivered the sample to the doctor, it was lost between that doctor and the lab.

## Cyclic Vomiting

### *Child*

My 7-year-old son had cyclical vomiting of an unknown cause accompanied by hypoglycemic episodes. His symptoms appeared between four and seven days of age. When the episodes would occur, his white blood count would be at least 30,000, yet there was absolutely no indication of why he was having prolonged episodes of projectile vomiting. In fact, the abnormal glucose level wasn't discovered until I took him to the emergency room one evening and the nurse grabbed him out of my arms and ran him back to a room right away. I thought he was just sleeping, but it turned out that he was unconscious due to a blood glucose level of just 39. After that day, I took it upon myself to make sure that he always ate a decent dinner and had an extra peanut butter sandwich before bed on his really active days. This seemed to curb the episodes and they would only occur if he had not finished the sandwich, unbeknownst to me, or had been extremely active the day prior. By this time, I had given up trying to find out the cause. I had been dealing with it for so long that I just tried to control the episodes themselves.

Finally, when Joseph entered kindergarten, and missed the first day due to an episode, I told our pediatrician that something had to be done. Although it was a nuisance prior to this, it was now going to affect his education, which I couldn't allow to occur. Starting in the fall of 2005 and continuing into the spring of 2006, my son underwent starvation testing followed by Growth Hormone Stimulation Testing (GHST). The starvation test was used to cause an episode and the GHST was used to verify the blood results from the starvation testing. Before the testing, there was just a discussion between the physician and myself. I got the results within two weeks of each test when the doctors phoned me. The results from the GHST showed IGF-1 levels at three times the normal level, so an abdominal ultrasound and an abdominal CT scan with contrast were performed. So much testing was done without providing an absolute answer that we decided to take two months off and then repeat the lab work.

I was very impressed with the two doctors who did the tests. One of the doctors immediately scheduled my son for starvation testing, which allowed him (and his medical students) to visibly see what physically occurred during an episode. When the doctor was stumped by the test results, he immediately contacted a colleague for a consultation. This was the only time that things got mixed up with my son's care. It was approximately six months before the two doctors were able to meet up and discuss the case, which was still a little unclear to the second doctor when we went to our first appointment. He thought he was just doing the Growth Hormone Stimulation testing, and my husband and I were under the impression that it was a full-consultation with the stimulation testing. However, this didn't prove to be a big problem for him. He saw my son for the consult and then immediately started the testing right afterward.

Recently, my son returned to see the second doctor for a follow-up and a recheck of the labs. This time, nothing was shown to be abnormal. Therefore, the doctor said that he would like to see my son once a year, but if problems occurred during that year, I should just call back and make an appointment for him to be seen sooner.

The results are confusing to all involved, including the geneticist and endocrinologist. Normally, the results from each test would lead to a diagnosis. However, each test led us onto something that was not expected. The doctors had no explanation for an IGF-1 of three times the normal amount without any sign of a pancreatic tumor. The one question that I am hoping to get answered is “Why is his lab-work all over the place during the episodes, yet seem just fine after the glucose has returned to normal?” Of course, when I spoke to the nurse about the results, I couldn’t think of anything to ask. I still need to call and ask the question. Is it possibly a glycogen storage disease that lies dormant until it rears its ugly head? Of course, I do not want to try to force another episode upon my son, but I also fear the effect this could have on him later on in his life.

Since there has been no diagnosis at this point, we were not offered any materials. For now, the plan is to avoid the episodes themselves by avoiding any fasting. We are treating the hypoglycemia itself. However, he cannot use any glycogen (or similar medications) because testing has proven that his body is unable to break it down and use it anyway. Right now he doesn’t play any sports, and I still have to watch and make sure he eats enough. What happens when he starts sports? The teachers at school already state that after he has gym class he is just completely wiped out and has a hard time “coming back” afterward. I really do fear allowing him to do any sports for this reason. Yet I cannot hold him “hostage” from physical extracurricular activities without a good reason. He already thinks that I am just a nagging mom when I make him slow down physically.

If you cannot find anything in the blood work, you just have to hope for the best and stress to your child the importance of eating properly and taking rest periods when needed. However, when they become teens and adults, we cannot hold their hands. Instead, we just have to pray that our children take their own health as seriously as we do when it was totally our responsibility.

As parents, we are lost in a world where we try to control the episodes and keep our children healthy. But we still live with a fear of “the one time” that the normal treatment doesn’t work, and doctors at the local emergency room won’t/don’t listen and our child ends up in a full-blown crisis. Without an absolute diagnosis, my son’s health issues are sometimes ignored. In fact, in January 2006 my son was scheduled to have tubes put into his ears due to chronic ear infections. Our primary doctor made it clear that Joseph could not fast for surgery and needed to have a glucose drip before, during, and after the procedure until it was clear that he was stable, with both his eating and drinking. He said that it was extremely important that the ear, nose, and throat doctor knew this—not only knew about what needed to be done, but why it needed to be done. If the surgeon couldn’t understand the reason behind these conditions, then he may not take Joseph’s health history seriously, therefore putting him at high risk for an episode while under anesthesia. He also told me to have the doctor phone him if he needed clarification or had a problem with the non-fasting and/or use of an I.V. drip at all times during the procedure.

When we arrived the day of surgery, I could absolutely tell that neither the ear, nose, and throat doctor nor the anesthesiologist understood the health issues at hand. They kept trying to point out that the surgery wouldn’t take but five minutes (which our doctor was aware of). They said that people under anesthesia had an increase, and not a decrease of glucose levels. I tried to explain that Joseph wasn’t a “normal person” but you could just tell that they did not understand the health ramifications if my son went into an episode once put under; namely vomiting and inhaling vomit

into his lungs. In the end, they complied because I wasn't willing to just brush it off, and because they had spoken to Joseph's doctor directly. They would be responsible if anything should happen, because they ignored his instructions.

This is the type of situation that, as a parent, scares me about not having a firm diagnosis. If there weren't a geneticist willing to stand up and demand that the doctors listen, would there have been a different outcome? Would I have lost my baby instead of watching him grow into an adult?

In the end, the doctors who decide what testing should be done, and how to go about getting a diagnosis, cannot always give us the answers that we desire. Therefore, frustration and fear sets in. You just hope that you have a pediatrician and/or specialist willing to stand up for you and say, although we can't prove exactly what it is, there is something "not right" with this child and his/her care needs to be taken seriously. It is better to err on the side of caution, rather than risk the child's health, and future. Testing doesn't always make us feel better. Sometimes it leaves us feeling even more frustrated.

### **Down Syndrome**

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#### *Prenatal*

As a result of my unborn child screening positive for Down syndrome through the regular maternal blood test, I underwent genetic screening in the form of amniocentesis for both of my unborn children in 2002 and again in 2005. In both instances, I supplied them with informed consent and had a one-on-one meeting with a genetic counselor. I was supplied with printed materials and had another brief meeting at the test.

Within 14 and 16 days respectively, I received the comforting news over the telephone that no major genetic conditions had been identified, and that the previous result had been a false positive. The results were very clear and were followed up by an appropriate amount of support. It was very helpful to hear that I no longer needed to worry about my children having any major genetic conditions, but it would have made my life easier if I received the results in a more timely manner. It is tough to hide a pregnancy until 20 weeks.

### **Down Syndrome**

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#### *Newborn*

In 2001, my son was tested for Down syndrome because he was born with features indicating the genetic condition. Before the test, we had a discussion and it was assumed that the test needed to be done to confirm the suspicions. At his two-week checkup, we got the results that he did have trisomy 21.

The pediatrician was very cold and unconcerned, and the only information we received was a binder of reference material from a Down syndrome organization. This wasn't very helpful since some of the material seemed dated, and I didn't really want the big picture at that time. We later found a support group on our own, and it was great to find other families going through the same emotions.

## **Duchenne Muscular Dystrophy**

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### *Adolescent*

In 2003, my sons Michael, who was 14 years old, and Sam, who was 12 years old, were tested for Duchenne Muscular Dystrophy (DMD) using a deletion detection method. An MDA clinic neurologist had previously diagnosed both Michael and Sam. However, since they were diagnosed almost two years apart, it had not been determined if they had the same deletion. The youngest did not show the outward characteristics typical for a child affected by DMD when the eldest was diagnosed. If I had read my older son's DNA report more carefully, I may have decided to have the younger tested at that time, as the document recommended. Even so, the doctor told me it was unnecessary to have an analysis completed for my younger child, as he would not be treated any differently by the clinic. Due to the significant variation in the progression of muscle weakness between the boys, I personally sought to know if they shared the same deletion. While this is a single gene disorder, knowing the mutation is important as current research is based on specific mutations. Many families do not know to request this information, yet as researchers identify therapies based on specific mutations, physicians, patient advocacy organizations, and the media should make this known to the affected populations. Michael and Sam were tested under a research study, but as we lived too far from the study site we did not participate in all phases of the study.

In preparation for the testing we received printed materials about the study and testing procedures. I was provided with an informed consent form and spoke with a member of the lab by telephone about the procedure and study. Instructions were provided for having blood samples drawn, both for us, as the people seeking the test, and the lab collecting the sample to be sent to the study lab. After the lab received the blood samples, it took just over three months to obtain the results by mailed letter. I clearly understood the results that both of my boys had the same mutation, the deletion of exons 48-50. The letter explained that the testing had been performed in a research lab and suggested the results should be verified by a commercial laboratory. Michael's DNA had been sequenced at diagnosis, and since the results were identical, I chose not to seek additional testing. I found the experience appropriate for my needs, as I only sought confirmation that my sons had the same deletion. The most beneficial aspect of testing was the peace of mind I got from learning they had the same deletion and that I was dealing with one disease and one mutation that caused the disease. The least beneficial aspect was being unable to participate more fully in the study, yet I do not find it to be a problem.

## **Ectrodactyly Ectodermal Dysplasia-Clefting (EEC) Syndrome**

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### *Child /Newborn*

Our son was tested just after birth in 2002 and then again at three and a half years of age for ectrodactyly ectodermal dysplasia-clefting (EEC) syndrome. The testing involved mutation detection and a chromosome screening. The first test was suggested to confirm our child's diagnosis, and the second to see if we could determine whether there was something within our family's genetics or if our son's condition had developed spontaneously.



We were required to provide informed consent before the tests. We felt comfortable doing so, since we had received tremendous amounts of information and support from pediatricians and geneticists. It took four to six weeks for the tests to be processed. The results were delivered by e-mail and standard mail. The results from the first test came back normal, with no variations or mutations of any kind. Based on the results alone, our son would have been considered physically “normal.” The second time the testing was done in 2005, there still wasn’t a confirmed diagnosis. A few things popped up in the results, which came back stating that our son had some kind of polymorphic variation.

Even though neither test confirmed our son’s diagnosis, his physical features and symptoms continue to point towards EEC syndrome. We’re pretty sure that the diagnosis is correct. We’ve been told that around 10% of all genetic testing on this syndrome doesn’t show up and that our son most likely falls within this category.

After the tests, we received a lot of support from friends, family, doctors, and geneticists. My husband and I also looked towards one another for support. Although I was apprehensive about the second round of genetic testing, my husband felt that it would be beneficial to know whether our son’s condition was inherited. Since our son’s syndrome appears to be the result of a spontaneous mutation, rather than family genetics, our daughter will be able to have children without fearing for their health. We had a great experience with the company that performed the tests. Overall, our testing experience was very positive and informative. However, I do recommend talking directly to a geneticist not just a pediatric developmental doctor. I feel that geneticists know more about what you’re going through, since it’s their area of expertise.

### **Familial Mediterranean Fever**

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#### *Child*

My daughter experienced periodic pains and other disturbing symptoms such as vomiting and a high temperature. She was hospitalized several times in a single month with the same pains. There were different suspects but all were rejected. We finally discussed the necessity of genetic testing to understand the cause of my daughter’s symptoms with a doctor. In 2005, she was tested for Familial Mediterranean Fever (FMF). About 15 days after the test, we received the results in paper form. She was positive for FMF. With the positive diagnosis, we were given regular control over testing every three months as well as free delivery of colchicines.

I didn’t know about the existence of FMF and that it is inherited by genes, since we never had it in our family, even in a distant relative. My daughter inherited her FMF from my husband who I had divorced when my child was two years old. Since then I never see my ex-husband because he is so careless about us, never tries to see or talk to us. I didn’t know what kind of illness it was and I was in a panic as I had heard different descriptions, until I started to read about FMF on the Internet. My worries multiplied after I learned what consequences FMF can have and how dangerous it may be if my 5-year-old daughter does not keep to her diet and follow the instructions. I am depressed and worried. It was good to find out that she had FMF so that we can take the required measures to avoid the bad consequences.

Unfortunately, even after receiving colchicines regularly, my daughter experiences the same periodic spasmodic pains twice a month. Now the doctors can't figure out the cause of these frequent attacks. We live in Armenia and some people say that the local climate might be one of the influencing factors to provoke the pains more frequently. They say that if we leave Armenia for a milder climate, then it may have a positive influence. I am apt to believe it because we were living in Europe for several years after the birth of my child until she was three years old, and she never had similar complaints there.

My daughter is very sensitive and became more nervous when she understood that she was sick. For her, her condition means that she is weaker than others her age. She is worried because she thinks she may not be able to have the same life as normal children. She always asks me why and I really have trouble giving an answer to her question.

I wish to believe that the treatment for this terrible fever will be found soon. I call for all the scientists to invest all their brainpower and intelligence into finding a solution. I wish to learn good news before my daughter's next attack happens.

## **Goldenhar Syndrome**

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### *Child*

My daughter was tested for chromosomal abnormalities in 2005. She was diagnosed with Goldenhar syndrome but the genetics doctor wanted to rule out chromosome issues. To prepare for the test, we met with the geneticist after giving the counselor a ton of family history. After the doctor did a physical exam of my daughter, she told us she believed it to be Goldenhar, but she wanted to send my daughter's blood to be tested for chromosomal abnormalities, specifically 22q11. She also set up appointments for my daughter to have a spinal X-ray, kidney ultrasound, and to see an ophthalmologist. My daughter was also sent for an Auditory Brainstem Response and tested normal.

We received the results of the chromosome analysis four and a half months later by regular mail. The results were clear: the test did not indicate any abnormalities of the chromosomes and the most likely diagnosis was Goldenhar syndrome. When we had the appointment with the doctor, she gave us some suggestions for support. We were also encouraged to call our genetic counselor or the doctor at any time if we had questions. We have, and the response has been excellent.

Overall, the experience was fine. The doctor had a great bedside manner. Perhaps some reading material at the time of initial diagnosis would have been helpful, but I'm really not sure how available it is, considering that the condition is rare. The most beneficial aspect of the experience was the quick response from the genetics clinic to get the ball rolling on a diagnosis. The least beneficial aspect was looking on the Internet myself before and after the diagnosis. This was very scary, and of course, I had no idea what was true and what was exaggerated or completely false.

## Hereditary Hemochromatosis

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*Adult*

I wanted to find out if I had a mutation for hereditary hemochromatosis (HH), so in 2004 I got tested. A saliva test was done through the mail with informed consent and printed materials provided. There were no meetings. Less than a month later, I got the results in the mail. The results clearly showed that I was negative for HH in regards to the three genes tested.

Overall, my experience with genetic testing was easy. It was nice to know that I was negative for HH. I do wish that I could have been tested for other known genes for HH.

## Hereditary Hemorrhagic Telangiectasia

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*Adult/Child*

Between 2003 and 2005, my husband, my son, my two daughters, my nephew, my three cousins, and I all underwent testing for hereditary hemorrhagic telangiectasia (HHT). A mutation detection was done to determine if the children inherited the mutation. Prior to the testing we engaged in a discussion with a genetic counselor and gave informed consent.

After the test was administered it took three weeks to get the results by phone and letter. The results were negative. However, the mutation tested for is a missense mutation so there are currently further studies. A family concordance study is being conducted to determine if the mutation found is the disease causing mutation because there are clinical signs of the disorder. The results were not clear and no materials were offered to help us better understand the results. However, I did receive support from a friend who is a genetic counselor. The most beneficial part of my genetic testing experience was being able to identify the affected chromosome and gene matching to my extended cousins. I was disappointed by the lack of standard techniques in the laboratories and having to pay for the expensive tests out of pocket because of insurance discrimination.

*Editor's note: It is difficult to determine if a missense change in a gene actually causes the disorder because such changes are very mild and do not change the DNA sequence of the gene significantly. Often additional testing of other family members is performed to see if anyone that is unaffected has the mutation(s). If so, this would suggest that the missense change is not associated with the condition, but is just a variant of normal in the family.*

## Hereditary Spastic Paraplegia/Amyotrophic Lateral Sclerosis/ Adrenoleukodystrophy

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*Adult*

Starting in about 2003, I was tested for hereditary spastic paraplegia (HSP). Since then I have been tested at least five times for various forms of HSP as well as amyotrophic lateral sclerosis and adrenoleukodystrophy. The tests have been to determine the mutation that is causing my symptoms. For my first test, a blood test was taken. Then my physician dropped the ball in letting me know the results; I had to call and ask to get my results. I learned that the test was negative, but I did not receive any additional information.

I have since learned that the test for SPG4/Spastin mutations does not detect all possible mutations. New research completed with a different testing technique detects significantly more mutations than the technique used by the lab that did my test. Needless to say, I'm not happy knowing that even though I had a test that showed no mutations, the results might not be accurate. If the new testing technique is ultimately used commercially, how do I convince an insurance company that I need to test for SPG4/Spastin mutations again?

## **Hermansky-Pudlak Syndrome/Chromosome Abnormality**

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*Adult*

My brother and I have a very rare disorder that doesn't currently have a name, and we have been looking to get a diagnosis of this condition. I have albinism, Crohn's disease, and a few blood abnormalities in addition to (or as part of) this unknown condition. I've had many blood samples taken in the past for detection. Doctors suspected that the unknown disorder was Hermansky-Pudlak syndrome (HPS), so I was tested for HPS in the fall of 2005 in Canada. Before the test, I met with my genetics specialist, and we discussed the HPS test as well as the chromosome testing that I was about to undergo.

I received the results from my genetic counselor by telephone about a month later, which was fine because I was working at the time. Both the HPS test and the chromosome test came back negative or normal. The results were very clear, and while there wasn't much in the way of materials, but that was okay because I am very familiar with my condition; I'm just trying to get a diagnosis for it. If I can get a diagnosis, then it will be easier for me to stay on top of my health.

My genetic counselor is very good and easy to talk to. I can call her to ask questions, and she's very interested in helping me learn more about my condition. Throughout the testing process, I was kept informed every step of the way. Right now, I just want to stay on top of the test results so I can find out more about my condition.

## **Hypokalemic Periodic Paralysis**

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*Adult*

My mother and I experience episodic paralysis all day long most days. We also have several other symptoms similar to hypokalemic periodic paralysis (HPP). I first had a muscle biopsy in 2002 after having a discussion about the test and receiving materials on how to take care of the location biopsied and providing informed consent. After about five months, I received incomplete results in the mail. They then did a second analysis, and I received a more in-depth description in the mail two months later. About two years after my initial biopsy, I received an e-mail with a complete description after a third analysis was done. The results showed that there was severe inflammation damage to the muscle tissue perhaps caused by Myasenia Gravis. This has now been ruled out.

Starting in 2004, my whole family underwent genetic testing. This included my mother, father, sister, and me. We read materials about testing from the Internet. Then our blood was drawn to determine if we had a mutation for HPP and sent to Germany for testing. It took a year to get the results back by e-mail. We found out that we had none of the known mutations for HPP.

Overall, my testing experience was hellish. I had to find out myself about periodic paralysis because no doctor told me about it. I had to expend a lot of personal energy proving I had periodic paralysis to the doctors. It took me years of hard work. I was told I was crazy or that the doctors couldn't help me. I could prove scientifically that these attacks had nothing to do with my mind, but doctors would insist I was making it up. You CAN'T fake paralysis. A normal person has to blink sometime. When I am paralyzed, my eyelids can be paralyzed open for several minutes straight. Each time one of the 60 doctors I saw in a six-year period told me that they had given up and couldn't help me, I became more and more scared about how bad this was going to get. I was also worried that I would never be able to do simple things like stand in one place for more than five seconds ever again. After finally proving periodic paralysis to my neurologist through carbohydrate triggered attacks generating low potassium readings, I finally received medication for the condition. My health improved drastically at that point after several years of decline.

General practitioners need to be supportive of their patients with strange and rare conditions. Even if they don't understand the condition, they should be willing to read any documentation the patient finds on their condition. Neurologists should stop trying to pretend they are psychologists. It can be traumatic for someone to be told they are crazy, when they are trying to figure out strange symptoms that make no sense.

### **Hypospadias/Chromosomal Defects**

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#### *Child*

In 2004, my son underwent genetic testing to determine the source of his hypospadias. He was also born with albinism, but that was diagnosed on appearance and did not require testing. The test was performed to identify any chromosomal defects.

We met with a doctor prior to testing, at which time we discussed the possibility of a chromosomal defect. We then went ahead with the test. The results were delivered over the phone about three weeks later. Our son had tested negative for the defects. Since the results were negative, we didn't receive any information or support.

Our genetic doctor was very kind, patient, supportive, and explained things clearly. Our insurance company covered the first test, but it was not clear to me if insurance would have paid for a genetic test to confirm his albinism diagnosis. Dealing with the insurance company was very confusing. We would be interested in testing for albinism to see if we could discover what type of albinism he has, but dealing with the insurance company to determine what is covered is more work than it is worth to us at this time.

### **Klinefelter Syndrome**

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#### *Adult*

I was diagnosed with primary hypogonadism in 1981 at the age of 29. At age 38, in 1990, a genetic test was performed using a blood sample to understand the cause of my hypogonadism. I was supposed to receive counseling before the test but that never happened. About 30 days after the test, I had a doctor's appointment to learn my results. I found out that I have 46 XY, 47 XXY and 48

XXXY chromosomes, which meant I was diagnosed with Klinefelter's.

I received very little support, and the only printed materials I was offered were reprints from outdated medical textbooks. These materials discussed studies that said Klinefelter men are criminals, mentally insane, etc. It was good to know what condition I had because doctors from the 1950s, 60s and early 70s when I was growing up had no clue. It was hard growing up with Klinefelter's.

## Medium Chain Acyl CoA Dehydrogenase Deficiency

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*Adult/Child/Postmortem*

When we were expecting our second child, we had a false positive maternal serum screening test for Down syndrome. We were put on an emotional roller coaster ride—we had genetic counseling, offered amniocentesis, etc. It was stressful. However, when the baby was born and it was confirmed that he didn't have Down syndrome, we were happy to know that our newborn was healthy. It did take a bit of time for the stress to leave us entirely, but as we saw our son grow and develop normally it did go away.

When our third child, who was deemed "healthy" at birth, died suddenly at nine months, it was initially thought that this was caused by Reye syndrome. As no aspirin products had been administered, this didn't make sense to the pathologist, so gene analysis was done which yielded the final diagnosis of death as being caused by an inborn error of metabolism, Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD). A coroner's warrant was issued due to the sudden death, so consent was not obtained. The pathologist is permitted to do the testing when children die suddenly, in efforts to obtain the correct diagnosis of death.

The coroner mailed us some outdated information about the disorder, which he obtained from a medical encyclopedia. He had never heard of the disorder before. We were fortunate to have access to the Internet. We sought out a related support group. They have been an invaluable source of support and information regarding this disorder. Through them we learned about the use of comprehensive newborn screening programs for the early detection of inborn errors of metabolism like MCADD.

My deceased daughter's screening blood spot card was obtained from storage at the public health lab and analyzed for the presence of metabolites related to MCADD. Written consent was requested by the geneticist to obtain the newborn blood spot test card created shortly after birth in February 2002. The analysis of the newborn screening blood spot card revealed that my daughter's levels were approximately 30 times the cutoff for C8 carnitine and the C8 to C10 ratio was 12. The cut off rate for that ratio is 2.0. These results strongly supported the diagnosis of MCADD.

We were initially notified by telephone about four weeks after the test. We were advised that an appointment had been made at our local children's hospital for us to meet with a geneticist regarding the results. The geneticist was thorough in his explanation of the disorder. We requested testing for our other children. We also requested that my husband and I be tested for carrier status of MCADD. We requested this for added reassurance that what we were being told was correct; that we were carriers of the same mutation that our daughter had.

We have peace of mind knowing and understanding exactly what caused our seemingly healthy

nine-month old daughter to die so suddenly. We feel reassured that our other children are not affected by this disorder. We are now aware of our carrier status for this disorder. This is of benefit for the purposes of family planning.

Some provinces in Canada were screening newborns for MCADD at the time that our daughter was born. Unfortunately, the province where we lived only screened newborns for phenylketonuria (PKU) and congenital hypothyroidism (CH) at the time of her birth. These findings suggest that if comprehensive newborn screening had been in place, we would most likely have known about the disorder shortly after our daughter's birth and she may still be alive. Needless to say, the stress from this experience hasn't gone away.

We believe that public education is the key to ensuring that children who are affected by rare inheritable disorders receive equitable access to comprehensive newborn screening services which can assist in early diagnosis of their disorders.

At our request, our local Member of Provincial Parliament (MPP) introduced a bill (101) to legislate the expansion of Ontario's newborn screening program for rare inheritable disorders. The bill passed two readings. It didn't make it to third reading because the MPP who introduced the bill resigned from the provincial legislature to run in the federal election.

On November 2, 2005 the Ontario Ministry of Health and Long-term care pledged \$18 million dollars to expand the province's newborn screening program from two disorders to 27 disorders. The program officially began in April 2006. MCADD was the first disorder added to the program. The other disorders on the expansion list are to be gradually added to the program. The target date for completion of the newborn screening program expansion is December 2006. We have been advised that so far three cases of MCADD have been detected. The benefit of this is that lives will be saved!

It seems very illogical that our child received screening for this disorder upon her sudden death and yet not when she was first born. It is upsetting to know after the fact that we could have acquired supplemental screening services for our child for less than one hundred dollars. Our child has paid a very high price for the lack of knowledge about these disorders and the issues of comprehensive newborn screening!

I understand and can definitely relate to the stress that parents may experience from having a false positive newborn screening test or any other false positive medical test for their child. I can also understand the concerns of some healthcare professionals that perhaps false positive results may lead to "vulnerable" child syndrome. Many "tools" in medicine (MRI, X-Ray, ultrasound etc.) can yield potentially stressful information for patients and their families but this doesn't mean that we don't use them. My husband and I would gladly have traded places with parents whom have experienced a false positive result—and in the end have a healthy child.

## Mental Retardation

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*Adult/Child*

When we were first married fifteen years ago, my husband and I visited a genetic specialist in Boston to see if we might have “special children,” since my husband has two older brothers with mental retardation. My mother-in-law had six pregnancies that resulted in three miscarriages and the birth of my husband and his two brothers. My husband and one of his brothers had their blood drawn but nothing of concern was found at that time. When I was seven months pregnant, the ultrasound showed the baby to have enlarged head ventricles. When she was born, she had good Apgar scores, but we had Early Intervention from our state come to our house once a week for occupational and physical therapy. Now, she gets special services at her public school.

In 2003, we began to notice our then 3-year-old daughter was exhibiting some developmental delays. Because of her father’s family history of two older brothers with mental retardation, both of our daughters, my husband, and myself were tested at that time through a blood screening to look for chromosomal abnormalities. As we were preparing for the test, I think we had one meeting and maybe received a one-page document about chromosomes and possibly gave our informed consent.

Approximately two weeks later, we met with the genetic specialist face-to-face to receive our results. While my older daughter was shown to be unaffected, my younger daughter expressed a genetic chromosome abnormality in that she has extra material on chromosome one and that she is missing material on her sixth chromosome. It turns out that my daughter received this abnormality from her father (who has a balanced translocation), and he is very sad that he passed this along to her. The results were much clearer to my husband than to me, as I am very confused by chromosomes. I would have found it more helpful if more than a single page about chromosomes was offered to me because I ended up seeking out further information on my own.

Having the genetic testing completed allowed us to find the reason for my daughter’s delays and understand why she was experiencing them. Additionally, it was selfishly reassuring to know that I did not do anything during my pregnancy to cause her current delays as my amniocentesis had come back fine. While such information is helpful to explain her current state, it does not have the ability to predict her unknown future and the progress we can expect, which is not very helpful. The results benefited my other daughter as well, since we learned that she is neither affected nor a carrier.

*Editor’s note: While amniocentesis does examine the baby’s chromosomes, it cannot identify all chromosome abnormalities, especially one that is too small to see, even with the current technology. To identify such changes would require additional testing; however, additional testing is not typically done if everything else looks normal.*



## Mental Retardation

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*Adolescent*

In 2003, my son started getting tested to discover the reason for his mental retardation. My son and I were both curious to know the cause. I'm not really sure what kinds of tests were performed but they have been going on for several years and have included several different tests. We have had multiple discussions and signed multiple informed consents over the years.

We got the results of the first test back within a couple of weeks, but he was still undiagnosed. We had found out nothing, so no information was provided. We appreciated the thorough exam that was given by our practitioner. Even so, it is very frustrating to go through all of those tests and not know the cause of his condition. We are no further than when we started.

## Metachromatic Leukodystrophy

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*Prenatal/Child/Adult/Post-mortem*

In 2002, my husband, my daughter, and I were all tested for metachromatic leukodystrophy along with an amniocentesis of my 30-week old baby. We had a mutation and urine screen for arylsulfatase. We wanted to find out what type of leukodystrophy my daughter had. We gave informed consent and were given the recommendation to get counseling. It took about a month to get the urine results and about three months to get the DNA tests through the phone and in-person appointments. We found out the mutation that caused leukodystrophy in our first daughter and that our second daughter was a carrier. We understood that the results were 98% accurate in all of our cases. The lab that ran our tests was very compassionate, and they prioritized our case. The testing was done very professionally and as quickly as they could. In 2005, we had DNA tested so that we could commence with an embryo biopsy technique in Australia through in vitro fertilization.

Our daughter passed away at age four. Her DNA was screened one and a half years after her death. This was an excruciating experience as I found the people doing the testing treated her like a bunch of numbers. A genetic counselor delivered her results to us. She was very understanding and offered counseling and a hug, which is about all there is in that situation.

When you are on a journey such as we were, you become desperate for any information and a correct diagnosis so that you can move forward, in whichever direction that may be. I think the most important thing for anyone that is new to the experience is for staff members to understand that the parents are desperate for knowledge, hope for a diagnosis and hope for treatment or a cure for their child. To be given empathy and patience as a newly diagnosed family is truly a gift because when something so devastating occurs, you tend to not speak with your normal support group of family and friends. You are also anxious to have your results as soon as possible because when you are awaiting a diagnosis time is so precious. I don't think there is any action you can take to help parents awaiting a diagnosis, as the waiting consumes your every moment and hour. I feel the best gifts you can give to them are empathy, sympathy, and patience, as parents become guilt ridden, anxious, and desperate, which creates the possibility that they will be short with the testers.

Closure is an important thing. I know of parents whose children were undiagnosed, and for them I know they will always wonder. Fortunately, we had a ‘good’ experience with the lab we worked with and were treated with respect. Testing was done quickly, although it never helped with a cure for our daughter as there is no cure for metachromatic leukodystrophy. I think having a ‘spiel,’ as you could call it, to teach your staff what to expect of newly diagnosed families would help in making their traumatic experience just a little bit easier. Maybe even offer a brochure on places of help or a prayer of hope, not that I am religious.

*Editor’s note: An embryo biopsy is part of a technique called preimplantation genetic diagnosis, when the embryo is tested for a known mutation(s) before being implanted into the mother’s womb.*

## **Mosaic Down Syndrome**

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### *Newborn*

In 1999, when my son was one day old, he was tested to confirm trisomy 21 or Down syndrome (DS). We met with a geneticist to discuss the test and then filled out informed consent. A karyotype was performed, and after four days, we got a telephone call with the results. We were upset because we were supposed to hear on Wednesday, and we didn’t hear until Thursday. They had tested 30 cells and 16 had the extra chromosome while 14 were normal. This was a diagnosis of mosaic Down syndrome (MDS), and he was considered 50% affected. We were given a letter that described common traits of DS and also a list of suggested books. The letter never stated that MDS can be very different. It just told all about what things we could possibly expect because of the DS. In our case, we bought a book on medical conditions that are possible with DS children. After reading about half of it, I threw it out.

At the hospital the geneticist did tell us she was 95% sure he had DS but that usually she is 100% after seeing the baby. MDS was not even brought up until it was confirmed so that was completely new to us. She explained what it was to my husband on the phone. There was literally no information on MDS to be found, other than just a sentence here and there about mosaicism. I founded the MDS group online when my son was four months old. A neighbor, who is a doctor, talked to a colleague who actually knew of another doctor who was conducting research related to MDS, so I contacted her and was able to speak to her. My son was supposed to be part of the research they were doing, but I was later told their funding went away after not hearing back from them for some time. I did receive a booklet on what research they had done on MDS already. My son is seven now, and I know they are once again doing research. He has not really exhibited any of the typical DS medical conditions. My pediatrician gave us the best advice of anyone. She said it will basically be a “wait and see” game all of his life and was very supportive, saying he could do very well and be higher functioning.

## Mosaic Down Syndrome

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*Child*

Back in 1988, my son was tested for mosaic Down syndrome (MDS). We suspected mosaic Down syndrome due to a speech delay and small physical characteristics that were present, so we scheduled a cytogenetic test. I spoke with the geneticist on the same day of testing. The geneticist suggested testing and then we tested. I think I had to sign a paper for consent.

After six very long weeks, I received a phone call to set up an appointment to come in. They explained MDS and showed me a paper with chromosomes on it. It was clear that my son had MDS, but they had no information about the disease. There were no materials at the time that they could offer, which is why I started my own advocacy organization. In fact, outside of one research study written by a university, the only materials on MDS are the ones that I write myself! It was beneficial to get the diagnosis, but the doctors were very negative. They were not helpful at all and suggested that my son would be virtually a vegetable and that I should institutionalize him. Although this happened many years ago, most of our members tell me the same thing is happening to them today. They are not getting any information on MDS, and they hear very negative comments from the physicians who give them the results.

*Editor's note: Cytogenetics refers to the study of chromosomes and a cytogenetic test is a test that examines the chromosomes of a person.*

## Mosaic Down Syndrome

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*Child*

When my son was six months of age in 2004, he was tested for Down syndrome because he showed slight symptoms such as low muscle tone, sizable tongue, epicanthal eye folds, sparse hair, and a gap between his first two toes. Adam's pediatrician expressed her concerns to me and his blood was extracted and sent to a lab where they examined his chromosomes. During the two week wait, I did research on the Internet, as well as looked for information at libraries, in textbooks, etc. When the results came back, I was asked to come into the pediatrician's office, and she informed me of their findings. Out of 20 chromosomes tested, six came back positive for trisomy (myelodysplastic syndrome). However, she was a new doctor and was not too familiar with MDS, so she referred me to a geneticist.

It was very helpful to get the results so we could start Adam in therapy at a very young age. The experience of testing was very scary and surreal. I went from my initial natural response of anger, to fear, to being unsure, to acceptance.

*Editor's note: While all the chromosomes in a cell are examined, only a representative portion of the cells are looked at in a sample. It is likely that all 23 pairs of chromosomes were examined in 20 cells and 6 of the 20 cells were found to have trisomy 21.*

## Mosaic Down Syndrome

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### *Child*

In 2003, my son Dan was diagnosed with a heart defect for which 50% of children also have Down syndrome (DS). A fluorescence in situ hybridization test was performed to look at his chromosomes to see if he had DS. There was no preparation for the test; our pediatrician just told us where to go to get the blood drawn. After two weeks, we went back to the doctor's office and got the results back that my son had mosaic Down syndrome (MDS). We did not know what DS was, let alone MDS, so we were unsure about the diagnosis. Once we did our own research, we were able to find out more about it.

We were referred to a genetic clinic at the children's hospital. This was a horrible experience. The genetic specialist we spoke to told us that our son was not going to walk until he was 36 months old; he walked at 11 months. She told us he would not talk until he was five; he is three and talks non-stop. She told us not to expect much. She said that he would not advance past the age of seven mentally, never live alone, never drive a car. I was alone with my son at the appointment because I did not think talking to a genetic counselor was that big of a deal. I was crying my eyes out in the parking garage and had to pull myself together before I could drive off. She even told me at the end of the meeting that I could put my son up for adoption. I understand that these things may have been told to people with children with DS 40 years ago, but our understanding of DS has changed and so have the expectations for the kids that have it. They can do anything a regular child can do, it just may take a little longer. Kids with MDS, like my son, are even less affected. It is sad that people like this genetic specialist even have the job that they do.

## Ocular Albinism

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### *Child*

Our son was tested for ocular albinism about four years ago. The test was performed to determine which type of ocular albinism he had. There was no preparation for the test; I read about it and decided that I wanted it done.

The results came a month later. They were forwarded to me from our local lab. The results clearly showed that our son had tested negative for the genetic defect. We did not receive any support after the test. An advocacy organization that works with individuals with albinism requested patient history and physical findings, but we still don't have a definite diagnosis. We don't have answers, which is very disappointing.

Our youngest son also has the same clinical characteristics, but I assume that he has the same genetic defect as his brother. I have not had him tested.

## Ocular Albinism

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### *Child*

Because we were interested in knowing whether my son fit into the ocular albinism (OA) 1 gene group due to its current consideration for gene therapy, he underwent a mutation detection test for the OA1 gene in late 2000 or early 2001. He had already been previously diagnosed with OA. We found the preparation for the test to be very easy, as we had one discussion and gave informed consent to participate.

Approximately three weeks later, we received a written response from the university where the testing was conducted, which I then discussed with our local ophthalmologist. The results were inconclusive, and my son's results did not match the OA1 gene. Although the lab thought the results were clear, I did not find them to be quite so clear. Since this testing occurred, I have learned that a researcher thinks she knows a reason why the test may have resulted in a false negative for the OA1 gene. Recently, we have sent another sample off to her lab.

It has been very informative and helpful to start this process. Since Paul is just four years old now, we are hoping that by the time he is much older gene therapy might be a real possibility for him. My local ophthalmologist has been very helpful and accessible as well. We did receive some printed materials, but I do not remember any specifics. Because of the potential advances in upcoming medicine, I strongly recommend genetic testing. I'm hopeful that it will create some choices for us down the road. It has already given us hope for the future and clarification for the present.

## Oculocutaneous Albinism

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### *Child*

Our daughter has been tested by two different hospitals through two different agencies in 2003 and 2006. Both tests have been mutation detection tests for oculocutaneous albinism because she has abnormal pigmentation and a visual impairment. Prior to the test, we were asked to give our informed consent and had a discussion where we received some printed materials.

We received the results from the testing done in Boston after waiting about six months. The results were inconclusive, which told us nothing. We would like to have a more definitive result that tells us exactly what she has or what has happened in her case. This testing was of no use to us. As part of the follow up procedure, counseling and some additional printed information were provided to us. We are still awaiting the results of the second test. We are hoping that these tests will lead to more research and eventually prevention. We live with a verbal analysis of what the doctors believe she has. We would love to have something more accurate.

## Oculocutaneous Albinism

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### *Child*

My son was tested in 2002 and my daughter was tested in 2005 for oculocutaneous albinism. My children underwent molecular genetic testing in an effort to detect the mutation they had, to try to provide additional information, and to get a concrete diagnosis. We requested the test and agreed to it after discussing the option with the geneticist. Other than the standard consent for the blood draw, we signed no additional consents. We were given no literature regarding the testing, nor were we told that it was possible to have albinism without a known mutation.

My son's result took about two months to get back, and my daughter's result came back about four months after the test because of a delay caused by problems with her registration. The results were faxed to my pediatrician's office. The results were that both of my children have OCA1B with known mutations at P406L and P431T. Our pediatrician had no recent genetics background, but we were fortunate enough to be able to speak with another doctor after we received my son's result. He was able to explain the results in much better detail, and I didn't need to discuss my daughter's results with anyone because her results were the same as my son's.

I truly appreciated knowing my son's diagnosis and what it meant for him in the long run. The process was difficult because there are only two places in the U.S. that do this type of testing for albinism, and the one we used required a lot of paperwork. The place also required an outside institution to pay up front for the testing and to guarantee payment of the difference between insurance partial payment and the complete billable cost of the test. For us, no local facility (hospital or doctor's office) would do this, but we were finally able to get the testing hospital to pay. However, this took many phone calls and a lot of energy on my part, and even then, the test results were delayed.

*Editor's note: OCA1B refers to oculocutaneous albinism, type 1B. Mutations can be named after the specific amino acid change(s) they cause in the protein produced by the gene P406L and P431T.*

## Oculocutaneous Albinism/HPV

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### *Child*

In 2005, my daughter Joanna was tested for oculocutaneous albinism (OCA) and human papilloma virus. We had the test completed out of curiosity and to know our options for future child bearing. We discussed the test with a geneticist and then had our daughter's blood drawn during our second visit. I think it took about two months to get the results both in person and by mail. It was clear that Joanna had OCA type 1 B. There was no real support offered, but we did get materials that followed up on the type of albinism our daughter has.

I was actually expecting the results to take longer than they did. The expectations were set with that in mind. We also were told perhaps we might never find out what type she has, so we were pleasantly surprised. The most beneficial part of the whole testing process was just knowing the specific type.

## Oculocutaneous Albinism

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*Adult/Child*

Although we knew my son had oculocutaneous albinism (lack of pigment in hair, eyes, and skin), he, my husband, and I were tested in order to try and learn more specifics about my son's condition and whether he was ty-positive or ty-negative. Additionally, a doctor suggested that we have the test completed. The tyrosinase test or "hair bulb pigmentation test" was administered in 1993 or 1994. It was a rather simple test, as we just went to the doctor's office and they plucked a few hairs out of each of our heads. Though we did give consent, we did not have any meetings to discuss the testing procedure or receive any sort of printed materials.

At the time, I had high hopes that I would have more insight into my son's condition once I received the results of the test. After a few months went by, I called to find out the results. No information was available. For months and months this went on, until a few years went by and I gave up relying on the test for answers. By now my son was old enough that I knew what to expect regarding his albinism.

By the time the results were delivered by mail, we were so frustrated with the test, and we really did not care anymore. Ironically, the results were not very clear and said that my son was in the middle of the continuum between ty-positive and ty-negative. In other words, they still didn't even know what type he was! We were not provided with any support or informational material, just very vague, indefinite results. We did not benefit from the experience at all. I think it was a waste of time and gave me a false sense of hope. The results were so ambiguous that it would not have helped us if they came sooner.

## Osteogenesis Imperfecta

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*Prenatal/Newborn/Adult*

I have osteogenesis imperfecta (OI) and we wanted to know whether or not our children had it. We did not want to know for the purposes of pregnancy termination, only for our own knowledge and information. We've had all three of our children tested—the first just after birth and the second two through amniocentesis. Our first child underwent collagen testing, while the tests for our second and third children involved mutation detection. These tests were performed in 1999, 2003, and 2005. In addition, my husband and I were tested to locate the defective mutation in our family history.

We discussed the implications of each test in-person and over the phone with medical professionals. The genetic counselor was very good at walking us through all three procedures. We were also required to provide our informed consent before testing. The results for all of the tests took four to six weeks and were delivered over the phone. Our first child was positive for OI, but our second and third children were negative. We didn't need any support from our genetic counselor because we already had connections with doctors and knew a great deal about OI from my experiences. Overall, the testing was very beneficial for my family.

## **Osteogenesis Imperfecta**

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### *Child*

I had my foster daughter tested for osteogenesis imperfecta (OI) in either 1987 or 1988. I wanted to find out what type of OI my daughter had. A skin biopsy was done at a government funded institution. Before the test, I signed an informed consent and had a discussion about the test. I think it took close to two years to get the results back. I received the results verbally, but I wish I had gotten them in a printed form. I asked for a printed copy, but I never received it.

The results said that my daughter had type IV OI, which I understood fairly well. The problem with the results was that they did not reflect what could be expected as she grew. I received a lot of information from a related advocacy organization, which has even more information available now than it did 18 years ago. Support was offered in the form of follow-up research, with my daughter returning to the same place every three to six months. She may still return on a yearly basis if she desires. Various tests and physical exams were performed each time she was there.

## **Osteogenesis Imperfecta**

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### *Child*

My daughter was tested in 1992 to confirm a clinical diagnosis of osteogenesis imperfecta (OI) by seeing if she had a mutation for it. Before the test, we met with a team of people at a local genetics clinic to discuss our family history and look for genetic links within the family. This team included our daughter's pediatrician (who is also a geneticist), a nurse (who took a detailed family history), and another geneticist. After the meeting, we took our daughter to a pediatric dermatologist who took a skin biopsy from her upper arm. I don't remember if it involved informed consent.

We got the results about six months later, first via phone and then in a letter. The results were inconclusive because a recognized specific mutation could not be found. We met with a pediatrician to discuss the results. We were disappointed because we already knew the clinical diagnosis and the genetic test didn't offer any real new information other than that our daughter has an unidentifiable mutation. It was also difficult to fight with the insurance company to get them to pay for the test.

The most beneficial part of the experience was hearing from the geneticist that the likelihood of having another child with OI was only three to five percent, since our daughter's mutation was considered to have been spontaneous at conception.

The test seemed really important at the time, but 14 years later, I now realize that even if a specific mutation had been identified, it wouldn't have really told us how our daughter's diagnosis would affect her specifically, since there's so much variability among those with OI. It would have just perhaps allowed us to lump her in a group of others with the same mutation, but it wouldn't have helped with her treatment.



## Osteogenesis Imperfecta

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*Adult/Child*

My daughter and I were tested in 2005 to give us more information about the presence of a detectable deletion or other mutation for mild osteogenesis imperfecta (OI). We were given a clinical physical evaluation, provided information for a genetic family tree, and had a skin biopsy to test for collagen mutations or deficiencies. Additionally, a DEXA scan was completed on both of us, and I had blood chemistry tests done. The DEXA scans showed that both of our bones were significantly less dense than they should be. My blood test showed that, for an undetermined reason, I had no detectable level of Vitamin D. We had a discussion and gave informed consent before the skin biopsy.

The results came back six months after the test, which was three months later than expected, due to a lab error. The doctor gave us the results by phone and also sent a copy of the lab results along with a letter to explain them. The results were inconclusive and unclear because OI could not be ruled out by the tests. We did not receive any support beyond the results and accompanying letter.

I appreciated the direct interaction with the geneticist and the attention she gave us. The least beneficial part was the lab error, which was a very simple error that could have ruined the results. It was surprising because the lab is considered to be one of the top labs in the world, and it is one of only two labs currently doing skin biopsies in the U.S. We discovered the error by accident. Thankfully our doctor was a great liaison to the lab. We were able to work it all out, even though the lab did not make its contact information readily available.

Our geneticist was fantastic and clearly knew what she was talking about. If she had been any less attentive, I shudder to think of what the lab results might have led us to mistakenly believe. As it stands, my diagnosis, as well as my daughter's, is clinical mild OI with probable phenotypic Ehlers-Danlos syndrome overlap.

## Propionic Acidemia

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*Adult/Child*

After my son was diagnosed with Propionic Acidemia, we decided we wanted to determine his gene mutation. In order to do that, they needed blood spots from my husband and myself, in addition to my son. Since Propionic Acidemia is autosomal recessive, we knew my husband and I are carriers, but we didn't know if my son's mutation was homozygous or heterozygous. We also wanted to determine if my other son was a carrier and therefore sent his blood spots as well. The blood spots were obtained through a finger stick in 2000. I don't remember signing consent forms. The results took three to four months and determined my son's mutation and that my other son was a carrier, like my husband and me.

In 2004, my sister decided to check to see if she was a carrier and had her doctor send in her blood spots. Fortunately for my sister and her children, she was not affected at all. All of our results were very clear. The follow-up is the treatment of my son's disorder. We are glad we had the testing done.

## **Trimethylaminuria**

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*Adult*

In 1999, I was tested for mutations that cause trimethylaminuria (TMAU). I requested the test and found a lab that would perform it for free. I had a phone call with the lab and then went to my doctor's office to have my blood drawn. They handled shipping the blood to the testing lab. It took no more than three months to get the test results in the mail. I had tested positive for two alleles attributable to TMAU disorder. The first allele had both a rare and a common mutation, while the second allele had only the common mutation. I felt the results were clear.

After the test, no support was offered, but I did not feel any was necessary. I had a great testing experience. Having clear reasons behind my disorder gave me peace of mind and having a clear diagnosis helped me to form a game plan for treatment. It's also great that I have proof of my disorder in case I need to prove to my employer that I can't help my physical problems. I greatly appreciate the generosity of those who provided the testing and then did it free of charge.

## **Translocated Trisomy 13**

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*Adult/Prenatal*

While pregnant in 2006, abnormal ultrasound results led to a nuchal translucency scan for my baby. The doctors were concerned about the health of my baby, so they ordered a chorionic villi sampling (CVS). The test results confirmed that our baby had translocated trisomy 13. My husband and I then underwent testing to determine whether either of us carried the mutation. We were provided with information regarding the testing by phone.

The fluorescence in situ hybridization test results from my child's CVS took three days, while my husband and I waited seven days for our results. The CVS confirmed our baby's trisomy 13, but the test results were normal for my spouse and me. All of the results were delivered over the phone. The results were not accompanied by any support—only a screening report and a picture of what our chromosomes looked like. I didn't feel like I was informed about what the results and statistics really meant. I had to do my own research online and would have appreciated getting materials that explained the implications of the results. However, it was very beneficial to get the results and find out the information early on in the pregnancy. It didn't change our course of action, but it did help with coming to terms with the loss. Still, I really would have appreciated more information. I would have liked to know what I was looking at when I looked at the picture of our chromosomes.

## **Trisomy 13, Deletion Chromosome 9**

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*Adult/Child*

In 2002, my daughter was born with partial Trisomy 13 and partial deletion of chromosome 9. My daughter was in intensive care after having heart surgery at six weeks old, and she weighed less than six pounds. We could not get her off the vent, and she was very sick. They called me on the phone and told me she had a genetic anomaly, and that she would be severely handicapped for the rest of her life but they would come talk to me about it the next day and hung up. I spent the next few

hours running around the hospital trying to find someone to explain this to me and was very upset. They finally agreed to come talk to me and gave me very bad news, but also said they have never seen her genetics before so they really didn't know what to expect. One of the people kept telling me "I know how you feel." I just wanted to scream at her that there is no way she could have known how I felt. After a few meetings with them, I could not go to anymore, and my husband went instead. They finally said, "We could be wrong—we just really don't know." The geneticists could not give us any new information but wanted us to continue seeing them on a regular basis. When I told them I can only keep up with so much and was seeing four to five doctors a week already, and that we would not go to another appointment unless they had new information for us, they put in their notes that I was a difficult mother who was not doing what was in my daughter's best interest.

Since our daughter had such a unique genetic makeup, my husband and I were tested for trisomy 13 and trisomy 9 using the fluorescence in situ hybridization test. We did have to fill out an informed consent, but there was absolutely no other information provided. It took about six weeks to get back the results. Then we had a short meeting during which we found out that my husband carries a balanced translocation. However, the results were not clearly communicated to us. We were only given a copy of the results with no additional explanation or materials. It was the most horrific experience I have ever had, and I will never see another geneticist again if possible.

*Editor's note: Individuals with trisomy 13 (three copies of chromosome 13) or a deletion of chromosome 9 are expected to have significant medical problems. It is therefore unlikely that this man or woman has either condition. Most likely these parents were tested to see if either had a balanced rearrangement involving chromosomes 9 and 13 rather than an extra or missing chromosome 9 or 13.*

## Undiagnosed

### *Newborn*

Our daughter was tested in 2005 or 2006 because she had congenital heart defects and several other slight abnormalities as well as respiratory difficulties. The test was administered to look for a condition or cause that would explain her symptoms. Chromosome studies, fluorescence in situ hybridization and skin cell tests were administered. Preparation for the test involved one short meeting with a geneticist.

We received the results six weeks later. I had to ask for them over the phone. The tests detected nothing and the results were clear. The chromosome studies and fluorescence in situ hybridization worked, and although they found no genetic conditions, the geneticists explained that she probably still had a condition that they were unable to see. The skin cell test was unsuccessful as the cells did not grow. We had a follow-up visit with the geneticist, which was the most beneficial part of the process. The geneticists offered useful information and were willing to answer questions. The least beneficial aspect was not getting a copy of the results, though we didn't ask for one.

Our daughter passed away February 22, 2006. This changed our experience with the geneticists. They were planning on following her through her development despite the fact that no diagnosis of a genetic condition was found.

## Undiagnosed

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### *Child*

Our nephew underwent genetic testing in 2002. He had multiple symptoms of different syndromes. I don't know any details of the test, but it was performed to provide an explanation for his speech difficulties and to plan ahead for his education.

The decision to go ahead with the testing was well informed. Our nephew's parents had multiple meetings and discussions with experts. I'm not sure how long it took to get the results, but a doctor delivered them. Our nephew was missing a strand of DNA, which explained his symptoms. After testing, additional doctor's appointments provided a continuation of care. The results also helped when it came time to design our nephew's individualized education program.

*Editor's note: A missing strand of DNA likely refers to a deletion or loss of one of the two copies of a gene that a person has.*

## Undiagnosed

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### *Child*

In an effort to understand whether my child's defects were related to a genetic defect or syndrome, he was tested for multiple defects in 2005. In order to accomplish this, a series of blood samples were taken by my physician's lab assistant and sent to a genetic testing laboratory, which performed a high-resolution chromosome analysis at the 610-band level, a fluorescence in situ hybridization (subtelomere panel), and a fluorescence in situ hybridization (3'TUPLE1 probe) to rule out the common 22q11.2 microdeletion.

After signing an informed consent form the test was administered and the results were delivered to my doctor two weeks later. The clearly delineated results of these tests came back normal, indicating that my child had no known syndrome or other chromosomal or genetic defects. The end results were very worthwhile even though they were not accompanied by any support or materials, which I did not feel would have been applicable. Like any test, awaiting the results was the worst part of my experience.

## Velo-Cardio-Facial Syndrome

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### *Child*

My daughter had global delays, learning disabilities, and physical problems that could have been caused by either velo-cardio-facial syndrome (VCFS) or Prader-Willi syndrome. In 2002, we had her tested using a fluorescence in situ hybridization test to determine if she had the mutations for either of these conditions. I don't remember if we filled out an informed consent, but we did have a discussion and an exam the day we met with the geneticist.

About a month or so later, we had a meeting to find out the results. She tested positive for VCFS. I clearly understood the result, but I wish that I had been given resources like a group to contact. We did receive yearly follow-ups and the doctor said that I had covered all my bases. She even offered to test me to see if I had the syndrome too.

I had faith in the doctor when I met her. I got the impression that she knew what the results would be just by meeting with my daughter. The process went very smoothly, and I was glad to be able to meet with a geneticist who was intuitive and warm.

## **Velo-Cardio-Facial Syndrome/Cleft Palate**

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### *Child*

Our 13-month-old daughter Elizabeth was tested at the age of seven months. She had a cleft of the soft palate. Since I am a speech pathologist, I wanted her tested to see if the cleft was part of a syndrome. In order to do so, a fluorescence in situ hybridization (FISH) chromosome testing of the 22nd chromosome specifically was completed in 2005. Our preparation for the testing was probably atypical. I read a book on cleft palate and saw signs in my daughter of velo-cardio-facial syndrome (VCFS). When we went to her surgical pre-op for her cleft repair, we saw the genetic counselor as part of the craniofacial team. I asked the counselor at that time if we could have her blood drawn for the FISH testing while she was already anesthetized during the surgery, so that she wouldn't have to endure another needle stick while conscious. They agreed to draw blood while she was under general anesthesia. We signed a consent form for the testing beforehand.

Because the genetic counselor works at the university, and because we had traveled three hours from home in order to have her surgery, they agreed to tell us the results over the phone so that we wouldn't have to come all the way back. However, the genetic counselor did spend a lot of time with me on the phone that day. She helped me understand what the next steps would be in care and testing for my daughter, as she had tested positive for VCFS. This conversation took place about three or four weeks after my daughter had her blood drawn.

The genetic counselor directed me to a children's hospital where they have an entire center for VCFS and chromosome 22 deletion disorders. She sent me an entire chapter of information in the mail and also gave me phone numbers. From the people I called at the hospital, I was able to get a list of the necessary medical diagnostic procedures that were prudent to be run on my daughter since she did indeed have the syndrome. In just two weeks time, from the date we first knew about her diagnosis, I had spoken to the hospital, gotten prescriptions for the procedures from my doctors, and had already had an EKG, echocardiogram, renal ultrasound, and thoracic x-ray done for my daughter. Therefore, in just two weeks, we were able to tell if her heart, kidneys, and other major organs were affected. They weren't, miraculously, but it was nice to be able to get so much done in such a small amount of time. It helped propel us into a state of advocacy and not just feeling helpless.

I think the thing that helped us the most was that we had access to a knowledgeable medical facility and a genetic counselor to speak with. I think for any family, going to a geneticist and genetic counselor is key. Just getting the blood test would have been scary if my pediatrician had given me the results. My pediatrician knows nothing about her syndrome and we would not have known what to do next. By going to a real geneticist, it ensures that the person will understand the ramifications of a positive diagnosis, and will likely help put you in contact with who you need to talk with next. As is the case with many genetic syndromes, they are not always common enough for a local doctor to have seen any cases previously and know what to do. Therefore, I would say that seeing an actual

geneticist and genetic counselor is key. Beyond that, I think that the genetic counselor is the most important because they have the time to deliver the news and actually “counsel” you. I have called the counselor back on several occasions to ask questions, and she has always been a great point person for me as a mom.

The genetic report itself was hard to understand. It would be nice if maybe the lab could send an info sheet along with the diagnosis that I could have provided to physicians along with the lab paperwork. For instance, it would have been nice to have the International Classification of Diseases code (ICD) for the diagnosis of the syndrome. What is also tough is that when I initially spoke with my pediatrician about having my daughter tested, he disagreed with me stating that she didn't have any signs of the syndrome. In general, the medical community could benefit from ongoing communication regarding typical childhood syndromes. VCFS is the second most common genetic syndrome; Down syndrome is first. However, it does not present as a syndrome where the children “look” any different, so it is not readily noticeable unless you are looking for clinical signs. Therefore, the genetics community would be doing a real service to the rest of the medical community (and patients) by providing ongoing educational materials to pediatricians, who are the first line of defense in seeing children multiple times a year, and who would be the most likely to refer the child for testing. I am a persistent parent who looks at my doctor as simply a consultant but not the end-all-be-all on my daughter's care. I am aware that doctors are human and can be unaware or even wrong at times. Other parents may not be as persistent. Therefore, helping doctors learn about syndromes or at least learn to refer to geneticists if nothing else, will really help. If I had not referred my daughter for testing myself, we would not have known that she is affected. We would therefore not have known how to treat her. So, early diagnosis and the subsequent need for education of the medical community is key.

## **Von Hippel-Lindau**

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### *Child*

In 2006, my child was tested for von Hippel-Lindau (VHL). The test was performed to determine whether a mutation had been passed down to our child, since the disease is present in our family. The results of the mutation detection test were delivered over the phone, approximately three weeks after testing. I don't think that it is normal for information to be given over the phone, but the genetic counselor and I had many discussions prior to the results, and since I am a mosaic, we really were expecting the results to be negative. The results clearly showed that our child had inherited the mutated VHL gene.

After the results were delivered, we were offered information and resources for support. I had also received materials and information with my own diagnosis. Despite the long wait at the doctor's office during testing, my overall impression of the experience was positive.

## Von Hippel-Lindau

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*Adult*

Through a mutation detection test in 2001, I was tested for von Hippel-Lindau (VHL). Because I had started to exhibit signs of VHL, as well as having a family history of the condition, it was decided that the testing was necessary.

Many of the details surrounding the preparation for the test, as well as the follow up, are no longer very clear in my memory. Prior to the test, I met with a genetic counselor who explained the disease to me. I also met with her after I had received the results for more information about the diagnosis. Approximately one month after I was tested, I received the results, which indicated that I had an A to G mutation at the VHL locus. The results were presented in a clear and easy to understand manner.

My genetic counselor was very helpful and offered a lot of support to both my parents and me. I was sad when we moved not long afterwards and weren't able to see her anymore, as she was so helpful and understanding. I have found it really helpful to be able to share what I have learned about the direct mutation with other family members, as this allows them to be tested for the specific mutation.

## Von Hippel-Lindau

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*Adult*

I underwent DNA testing for von Hippel-Lindau (VHL) in 2004. The tests were administered because I had two manifestations that indicated the possibility of VHL. I didn't know that the tests were being performed until my doctor called and said that a blood sample was being tested for VHL and MENS. I did not have a chance to discuss the tests beforehand and didn't really know what the results might mean.

The results were delivered over the phone six weeks later. I had tested positive for the mutation. I was not offered support after I received the results; all that I got was some outdated information from the Internet. While peace of mind from "knowing" was very beneficial, my geneticists and physicians knew next to nothing about the disease. I had to find out what type of medical care and ongoing screening I would need on my own.

I received a clinical diagnosis from an ophthalmologist before the DNA results came back. I did research and had contacted an advocacy organization that was familiar with VHL. They immediately sent me information before I met with the genetics doctors. By the time I met with both my endocrinologist and the geneticists, I think I knew a lot more about VHL than they did since the information they gave me was about five years out of date as far as things like screening recommendations.

*Editor's note: MEN(S) refers to Multiple Endocrine Neoplasia, of which there are types 1 and 2.*

# Gene Mapping

## **Albinism**

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*Adult*

In 1980, my parents, my siblings (four sisters and one brother), and I were tested for albinism. All of us were tested, both albino and non-albino, because they were looking for the albinism gene. The testing was done via a hair pull. Two of my sisters and I also had our hearing tested because we were albino. Since my sister lived near the researcher, she had multiple meetings with him prior to the test. There were consent forms and printed material provided to all of us. My other sister and I were flown out for the hearing test part, and they paid for all of our expenses.

We never received the results from the test. However, more than a year later, we were provided with a reference to the journal in which the research was published. The study found that albinism affects human stereo vision, and that those pathways in the brains of albinos are different because of the lack of 3-D vision. Apparently, people have pigment in their ears, and the same thing is true of human stereo hearing; however, normal human stereo hearing is so poorly directional that the effect is not really noticeable. I was able to easily understand these results. I do not recall if we were given any more materials as a follow up to the test, but the researcher did tell us about an advocacy organization related to albinism.

The best part of the experience for me was that I got to meet other albinos on the trip. However, the researcher made the mistake of having me and my two sisters wait in a room with another test subject before starting. We were in our early 20's and he was in his teens and did not have any albino siblings. The researcher couldn't figure out why this kid was too nervous for the electroencephalogram (EEG) tests after that. In 1989, I also mailed a blood sample to a genetic researcher in Pennsylvania. The costs of the blood draw and shipping were covered in this case.

## **Idiopathic Pulmonary Fibrosis**

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*Adult*

I enrolled in a university study so my DNA could be tested for idiopathic pulmonary fibrosis (IPF). My father died of this disease at 59 years of age, and I was diagnosed with it in 2001 at the age of 57. I sent genealogic information to the university, and they told me who should be tested. I called everyone on the list and got their permission for the university to contact them. Anyone interested in participating could do so at no cost; the university would pay all expenses and testing would be done close to home. All test results would then be sent to the single university. We completed forms, signed permissions, and had a phone call discussion.

My husband, children, brothers, sister, and some nieces and nephews (over 40 years of age) were the desired subjects. Testing is currently being conducted at several sites, and then forwarded to the university. We have no results yet. The most beneficial result will be if a genetic linkage to IPF is discovered. This may help future generations who are predisposed already. More people should be willing to participate in genetic testing.



### Rett Syndrome

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*Child*

My daughter was diagnosed with Rett syndrome based on clinical symptoms before the culprit gene for this condition had been identified. In 1999, I chose to send a blood sample to a researcher who was looking for the gene. Because I chose to send the sample, there was no discussion or informed consent before the test. After the gene was identified, we received the results within weeks. We were first notified of the results via e-mail followed up with a written report.

The results were clear. It turned out that my daughter was positive for a point mutation that later was discovered to be the most common mutation. There was no support offered, but we didn't feel that any support was necessary. I was relieved to finally have an answer.

### Van der Woude Syndrome

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*Adult/Child*

In 2003, a university requested blood samples from my family, as they were trying to discover the gene marker for van der Woude syndrome. As a result, my husband, affected son, and I participated in a blood test. As I remember, we signed papers giving our informed consent. After approximately three months had passed, we received the news by mail that the team had been able to locate the marker for van der Woude syndrome. The results presented to us were very clear. We were offered support upon receiving the results, but I am not sure if any printed materials were offered.

From this process, we now know that my son definitely does have van der Woude and popliteal pterygium syndrome as well as the exact gene marker. We still do not understand why he has this as my husband and I do not and this is an inherited syndrome.

# Predictive

## **Breast Cancer (BRCA)**

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*Adult*

A strong family history of breast cancer caused me to be tested for the BRCA1 gene. My mom died at age 26 from breast cancer, when I was six months old. My maternal aunt (who had been diagnosed with breast cancer in 2002) tested positive for the BRCA1 gene a month prior to me being tested. She was tested at a major university as part of a genetic/familial study. When I found out that she was participating in a genetic study, and our family was being studied, I wanted to be part of it in hopes of helping to find a cure for breast cancer. I had known about the test for years but hesitated to have it done, as I had already been extremely diligent about getting my mammograms and doing self-breast exams.

Coincidentally, I was diagnosed with breast cancer a week after I had scheduled an appointment to have my gene test. I paid to have the results expedited, as it was imperative to help determine my treatment (surgery, chemo, radiation, post treatment). With my diagnosis in 2005 at the age of 32, I became the sixth woman on my mom's side of the family to be affected with breast cancer. The fraternal twin of my aunt that was diagnosed in 2002 tested negative for BRCA1. One of my female cousins also tested negative.

While my entire experience was stressful due to the nature of the test, the genetic counselor that I met with before and after the test has been exceptionally helpful. Before the test, she provided me with the necessary consent forms to sign as well as materials to read. She did an excellent job of answering all of the questions I had. Two weeks later, I learned during a meeting with the genetic counselor that I had tested positive for the BRCA1 gene. The results were presented in a very clear fashion, and I was offered materials and support in many forms; from brochures to DVDs, in addition to further consultation with my genetic counselor. She still calls me every few months to see how I am doing. By having the test completed, it has been very helpful in assisting my doctors and myself to decide my treatment options.

## **Ovarian Cancer**

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*Adult*

I am a 63-year-old ovarian cancer survivor with several other family members who have suffered from the disease. Because I have two granddaughters, I wanted to discover if the mutation ran in my family. In 2005, I had blood drawn to test for the mutation. Before the test I had two meetings, was provided with comprehensive reading material, and had a detailed discussion with a genetic counselor. The time between the meetings allowed me to think and process the information I had received about testing. I scheduled an appointment about two months after the test to receive the results. The result was positive.

I had a very positive experience in that it was very professional and yet very caring. The results were clearly articulated and I received a lot of support. I had a telephone follow-up where I could discuss any questions that I had at that time. I'm glad to have the information that I have the gene so that future family members will know and can be tested if they choose. My granddaughters are only two and four years old, but I also have three sons. The responsibility for knowing affects you more than you think it will. I am still of the opinion that knowledge is good.

### **Von Hippel-Lindau**

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*Adult/Child*

My spouse was definitively diagnosed with von Hippel-Lindau (VHL) syndrome in 1992 and passed away from complications in 2002. Upon the initial diagnosis in 1992, our children were screened for the VHL mutation. It was a blood test, but I don't know the medical terminology well enough to describe it beyond that. The tests required informed consent and the procedures were discussed beforehand. Several weeks went by before we received the results at our genetic counselor's office.

We learned that our son had inherited the VHL mutation, while our daughter had not. My spouse's brother was also tested at that time, and found that he did not possess the VHL mutation. The results for the test in 1992 were supposed to be 95% accurate. My daughter later underwent additional testing in 1997 or 1998, the results of which were supposed to be more than 99% accurate. Her results from the second test were again negative.

We were offered further contact with our genetic counselor following the results of the tests. The tests were very beneficial, in that they let our family know what we were dealing with. My son's health has been monitored regularly since his diagnosis. Although little information was offered in 1992, my son has received enormously useful information and support from a VHL support organization. He subsequently had his two sons screened, both of whom tested negative. In appreciation for all the support organization has offered to my family and other VHL families, I continue to support the organization financially and through service.

# Prognostic

## **Albinism**

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### *Child*

My first child, who is now nine years old, had genetic testing done six years ago. We were hoping to determine whether my son was tyrosinase positive or negative in order to get a better idea of his ultimate visual limitations. At the time of the testing, I was pregnant with our third child, so we were also interested in determining whether that child would be affected. However, we were told that the only way to determine whether our unborn child was affected was to locate the defective genes in our child with albinism and then to do an amniocentesis on the unborn child to look for the known defects. We chose not to do the amniocentesis because of the risk, but we proceeded with the testing of my first child in order to determine tyrosinase positive or negative.

My child with albinism was tested via a blood test but my spouse and I had not been tested. They found one of the known defects but did not find the other. They had told us before testing that this could happen. As a result of him having one of the defects and the characteristics of albinism, he was given a diagnosis of albinism. When my third child was born, her cord blood was tested. Although they found the same defect in her blood, she does not display any symptoms of albinism and is presumed to be a carrier. If we had chosen to do an amniocentesis, it would have been inconclusive because they would have found the known defect meaning that she was at least a carrier.

If my second child were tested today, the result could very likely be inconclusive unless he possessed the same defective gene. However, it is my understanding that there have been more of the defects identified over the past several years and that they may be able to locate the other defect if the testing was repeated today.

## **Breast Cancer (BRCA)/Ovarian Cancer**

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### *Adult*

My mother and aunt died of breast cancer and I have ovarian cancer, so I wanted to see if I had a BRCA1 mutation. I was tested in 2002 by a lab based in the UK. I had to fill out an informed consent, had a discussion about the test and also received printed materials. The results came by mail about six months later, as it takes a long time to get the results in the UK. The letter was very comprehensive. With the technology we have today and about 60% of the BRCA1 gene tested, there was no mutation found.

I was interested to find out where my cancer came from, especially whether it was genetically based. I was very happy to find out the females in my family that had cancer did not have it as a result of a gene mutation. It was quite a powerful experience.

## Lynch Syndrome/Breast Cancer

---

*Adult*

Over the past five years, I have undergone three genetic tests. Throughout 2001 and 2002, I was tested for mutations related to BRCA 1 and 2 as well as for Lynch syndrome. Testing was conducted for these disorders due to previous family cancer histories and diagnosis. Two sisters and I were all diagnosed with different Lynch syndrome cancers within a 2-year period.

In preparation for the testing, I signed the informed consent form. Prior to the actual testing, I networked extensively with others who had undergone genetic testing and also spent much time doing research on the Internet. My sole purpose of genetic testing was to move research forward. To supplement this information, I had multiple meetings with healthcare providers, including my own oncologist.

With testing completed, I waited six months for the BRCA results and over a year for the results for Lynch syndrome (HNPCC). I was informed of the outcome of the BRCA tests through an in-person meeting, whereas I received the results of the Lynch syndrome test by phone and fax per my request. I was found to be negative for both BRCA mutations, but the Lynch syndrome test was inconclusive. Even though the test for Lynch syndrome yielded inconclusive results, all of the test outcomes were presented in a clear manner.

Even though the results were presented in an understandable manner, I received limited and outdated materials. As a matter of fact, an international conference took place in Europe in 2005 and I was interested to know the outcome of the meeting, as they might directly apply to any new mechanisms for screening. What I was really asking was if there was anything new, at all, which might be of interest to me. I was quite annoyed recently to find a synopsis of the conference (online) and that the conference did in fact discuss not only screening, but fascinating information as to how Lynch syndrome is clinically dealt with, depending on the country. In addition, further professional research has determined some new and interesting aspects as it relates to Lynch syndrome. I find the lack of communication surrounding new research results to be unsatisfactory. Significant international studies are being discussed in research circles, but not translated or shared with those who are most intimately involved—the patients. While I understand that this research will not make an impact today and even may not be widely accepted within the research community, it is significant and it is of value to me as an informed patient/consumer. In the absence of comprehensive guidelines, it was helpful to have the ability to determine, on my own, what follow-up I needed to have with my own oncologist. My plan, which my oncologist has agreed to, gives me a sense of control over my own situation. In reality my plan is not necessarily a good plan of surveillance, but it is the best that I can do, given the status of the research at the moment.

I believe that what is not understood, or maybe not verbalized, by the medical community, is that for many, confirmation of a positive genetic test may lead to not only the risk of a particular cancer, but also a second or third cancer. At one point I tried to express this to an oncologist and was dismissed by the response that it only affected very few people. “The risk is only low if it is not you,” was my response at the time.

I can understand and appreciate the failings of science and where we are at the moment in relation to genetic testing. At the same time, however, I needed it to be well understood that I live in the 'today'. I do have to say though that I am quite impressed with the recent attention that Lynch syndrome has received from multi-disciplinarian teams within research/teaching cancer centers. However, I still am required to educate other medical professionals because most often it is a syndrome that garners little or no attention. I am particularly concerned that further education is required at the family physician and nursing levels.

Despite my observations, my overall impression of the experience was fine, but I do and must insist, that new developments are adequately communicated with patients. Cancer patients themselves, in general terms, are simply not aware that they may have an increased risk of a second cancer, or in fact, that they are at risk at all. I find it intellectually and emotionally draining to continually witness over years the distress of others when communication/education could go a long way to reduce these issues.

*Editor's note: HNPCC stands for Hereditary Non-Polyposis Colorectal Cancer.*

## **Nail Patella Syndrome**

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### *Adult*

In 1969, my grandfather, mom, brother, sister, aunt and her six kids, and I underwent testing. We had previously been diagnosed with Nail Patella syndrome (NPS). A pediatric geneticist ordered the tests. At that time, they were looking at the ABO locus on chromosome 9 using a blood test. I was diagnosed with NPS while I was a student nurse at a hospital. I drew my grandfather and my family's blood, and hospital staff drew blood from my aunt's family. In 1969, there was no informed consent. They called my aunt to make an appointment. I just brought tubes home and drew blood from my family. We never got any official results. They were hoping to see if one particular blood type was common. When I called the pediatric geneticist, he said everyone was O positive except me (B plus after my dad), so the results were inconclusive.

Then in 1996, I found out about a glaucoma study at a local university that was looking into families with glaucoma. I was contacted immediately after the doctor got my letter. I noticed that everyone in my family with glaucoma also had NPS but only one article about NPS had mentioned the connection. Everyone in our family participated in the study at various times. The university even flew my cousins in from Hawaii. We did sign consents and discussed the procedure with a genetic counselor. My grandfather and mom were dead but I was able to get some samples of my mom's autopsy slides. The testing included eye exams, pictures, and blood draws.

Three of four generations of our family participated. Our family and a family in Ohio showed the connection of glaucoma and Nail Patella syndrome and we are included in a related journal article. In 1998, at our first NPS conference, I got a copy of our DNA results and specifics on our mutation. A genetic researcher was doing research on NPS and was obtaining blood samples. He passed out the results to everyone who had sent in blood at this conference, as well as mine which was obtained from the previous university study I participated in. Our family's mutation is a frame shift deletion of a C and A, which originally was called 232 Del TG.

I went up to the local university where I had participated in the previous study to get a better explanation of the results to enable me to be able to explain them to my family in lay terms. I talked with the genetic counselor that I had previously seen while there. I typed up the explanation, made a copy of the mutation information I received, and gave a copy to all of my affected family members, both for the information and their records (my aunt's family isn't "into" NPS). This was before 2001. Shortly after this, I found out another university was doing research on NPS with X-rays, eye exams, blood tests, etc. I went in April 2004. They drew more blood for DNA testing. There were lots of papers to sign then. At that time, I also learned about the new mutation No. PL101FS. Over 134 mutations have been discovered now. We were told we would not get the results of most of the tests as they were for research—only abnormal labs. We got results for the kidney and bone density lab tests. I got a slip detailing the results of my sugar test, but I am diabetic so that was not new information.

Additionally, in 1999 I sent blood and urine samples for a study on nurses conducted by a well-respected medical school. The blood is on ice, so to speak, to be used for research on different diseases they are studying. It is to be used for DNA markers, if, for example, they want to check breast cancer rates in nurses. I had to sign a consent form and fill out a questionnaire. I was provided with a number to call if I have questions. We get a newsletter periodically with findings from the questionnaires we fill out and specimens we send. They provide the blood and urine collection materials, shipping box, and labels to send it back.

In November 2004, I was diagnosed with chronic lymphocytic leukemia (CLL). My bone marrow specimen was sent for cytogenetics on the fluorescence in situ hybridization scale. I had to sign a consent form for the procedure, which was done on an outpatient basis. I was given an informational pamphlet on bone marrow aspirations. I got the results in December and was given copies. They found that I have a deletion of chromosome 13q14, which has a fairly good prognosis of the three that can cause CLL. I was negative for the other two mutations.

None of my experiences were bad. Being a nurse, I hope it helps others. I just was shocked about having the leukemia. I just retired from nursing, as I'm legally blind from the NPS glaucoma, and that is why I do not mind participating in these studies.

*Editor's note: "B plus" refers to the blood type known as B positive.*

A frame shift deletion of a "C and A" likely refers to the deletion of two DNA bases, (a "C" base and an "A" base) from the sequence of DNA bases of the gene. Mutations (such as No. PL101FS) are usually named by the person that first identifies them and sometimes the same mutation can be named differently by different people. To compare mutations between individuals it is important to know how the mutation was given its name because the same mutation can have different names.

The study of chromosomes is called cytogenetics. Each individual has 23 pairs of chromosomes numbered 1 through 22, with the 23rd pair representing the sex chromosomes, X and Y. Each chromosome is made up of two sides or arms, called p and q and each chromosome arm is made up of multiple regions or bands that are numbered. This individual's bone marrow cells carried a deletion of chromosome 13 on the q arm at band 14, described as a 13q14 deletion.

## Russell Silver Syndrome

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*Newborn*

In 2004, my daughter was tested because she was suspected to have Russell Silver syndrome (RSS). She was tested for the mutation UPD7 at my request. There is only a 10% chance of testing positive for UPD7 when diagnosed with RSS but it helps confirm a diagnosis. Unfortunately, there is not a diagnostic test specifically for RSS at this time. The test looked for maternal disomy on chromosome 7. Six weeks later, my doctor delivered the news that the test results were negative. She was diagnosed as small for gestational age with characteristics of RSS. I did not receive any materials or additional support, but I already have support. The most beneficial part of the testing process was knowing the result.

*Editors note: "UPD7" stands for uniparental disomy of chromosome 7. Such conditions are the result of having at least one copy of a gene from only one parent rather than a copy from each parent. Maternal disomy refers to the presence of two copies of the mother's gene, rather than one copy from the mother and one copy from the father.*



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