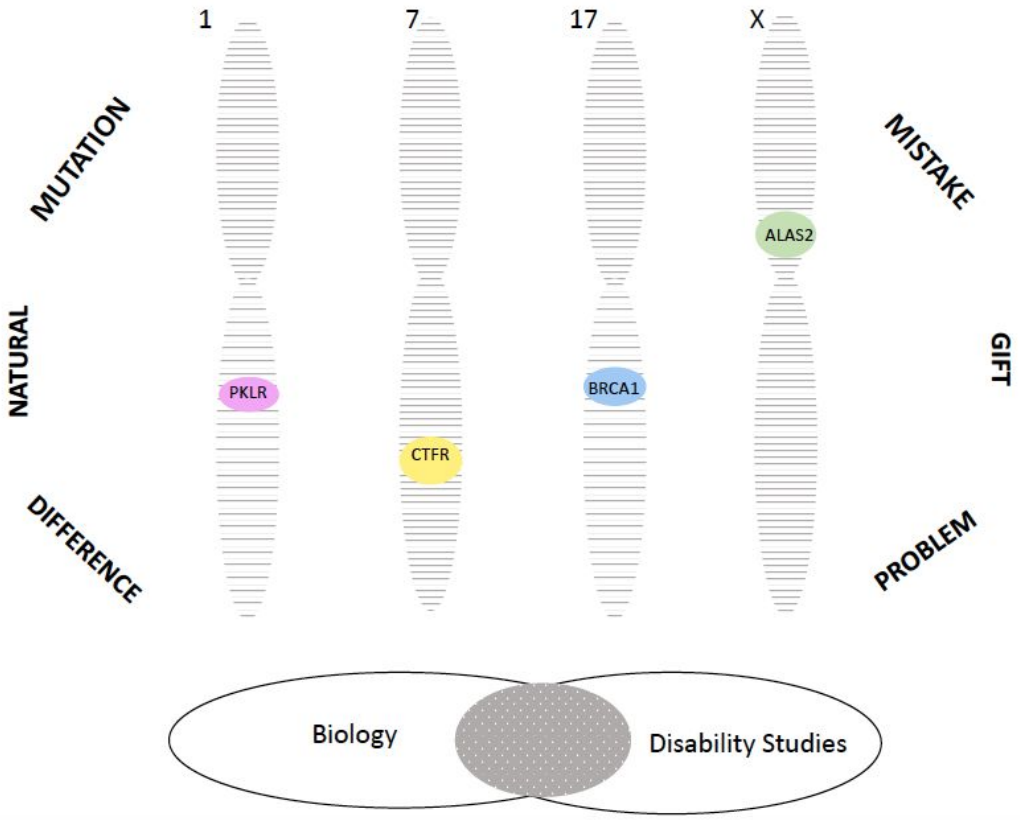
When Genes Speak:

Shifting the Way Biology Students at Haverford Understand Disease and Difference

By Mali Axinn



Introduction

First year biology at Haverford, and most colleges, is a survey course. We are taught how cells function and about the cellular processes that occur inside our bodies. Sometimes we discuss what happens when an essential process isn't functioning "properly". In this context, we are introduced to the idea of disease and disability, learning about gene dysfunction, clinical symptoms, and drug therapies.

The cellular processes leading to disease are discussed in biology class in their scientific form. Diseases are introduced to us by the blips in the cellular process they arise from protein misfolding, red blood cell deficit, and the accumulation of cellular material. In other words, we learn about diseases and disabilities as cellular dysfunctions which negatively impact biological processes. When we discuss specific diseases, we learn to associate Pyruvate Kinase Deficiency with a dysfunction in glycolysis, Cystic Fibrosis with a mutation in the CFTR gene, Porphyria with failure of the heme biosynthesis pathway, and Ovarian Cancer with stem cell formation.

Class lectures explore topics of disease and difference through clinical descriptions and images of dysfunction. For each disease, we review symptoms, "side effects", and medications used to treat them. Sometimes we are shown graphic depictions of symptoms like enlarged fingers, bulbous blisters, and bloody skin. These images often evoke pity, leading to an understanding that life with disease and difference is undesirable and that our job as biology students is to help find a cure. This search for cures shapes biology education, drives biology research, and has resulted in improved treatments for many diseases. However, for those living with diseases and differences, this focus on "cure" can lead to a reductionist belief that the lives of the disabled are lesser than lives of the non-disabled.

The current framework of biology class has the unintended consequence of marginalizing the lives of the disabled. It is not my intention to negate the significant lessons of biology 200/201

and their transformative effect on students' understanding of disease. Instead, it is my hope that my project will enlarge the perspective of Haverford's biology classes to include a disabilities perspective. Within a disability studies context, the voice of the disabled community can be heard in tandem with medical professionals, politicians, and scholars to create an expanded perspective on disease and difference.

In disability studies, we discuss disease and difference in relation to the personal story, transcending symptoms, side-effects, and medications. The knowledge that can be gained from the personal stories provides an enlightened perspective on living with disease.

Over the next year, I will work with the biology department to create a curriculum to complement the existing biology 200/201 coursework. In order to put the lessons from biology in conversation with disability studies, I created an anthology of patient testimonials with accompanying questions and a student assignment. In this anthology you will find patient testimonials written by individuals living with Cystic Fibrosis, Pyruvate Kinase Deficiency, Porphyria Cutanea Tarda, and Ovarian Cancer. The accompanying student writing assignment is based on the guidelines of Professor Robert Fairman's molecular biology assignment where he asks students to write a 350 word report that tells a story about a biochemical process with a relevant application. I adopted this format to provide synchronicity with the existing curriculum and to align the narrative styles.

During a preliminary conversation with Professor Rachel Hoang, the chair of the Biology department, she reviewed my project and provided feedback. We discussed introducing a project for Bio 200 in which students work in groups to create a poster presentation on a specific disease. For this project, students will use lecture materials and patient testimonials to provide a comprehensive overview of the disease that applies a biology and disability studies perspective. The anthology of patient testimonials that I created will serve as a resource for students completing this project. Next year, I look forward to working with Professor Rachel Hoang and Professor Kristen Lindgren to write and implement this curriculum.

Student Handbook:

An Anthology of Patient Testimonials of Living With Disease and Differences for Students of Biology 200/201

In this anthology you will read patient testimonials of those living with Cystic Fibrosis (CF), Pyruvate Kinase Deficiency (PKD), Porphyria Cutanea Tarda (PCT), and Ovarian Cancer. This anthology is intended to parallel the coursework in Biology 200/201 on genetic dysfunction and disease in hopes of constructing a humanistic narrative that can complement and expand the genetic and clinical models already introduced by the course. The creation of the anthology is informed by a disabilities perspective which privileges the individual experiences of those with differences, believing that there is no single experience or perspective which defines who a person is or how they live their life.

Here are some of the foundational principles that can be learned from a disability studies perspective:

- Disability as a construct
    - Disability is an evolving concept that is shaped by societal forces and use of conventional labels. It is the spaces we inhabit that create disability, not the people themselves that are disabled.
- Disability pride
    - There is a diverse community made up of people with disabilities that is loud, proud, and politically engaged. Rosemarie Garland-Thomson explains, "Becoming disabled means moving from isolation to community, from ignorance to knowledge about who we are, from exclusion to access, and from shame to pride".(*Opinion | Becoming Disabled - The New York Times*)
- "Cure me? No thanks."
    - Within the disability community there is a constant debate surrounding the politics of cure. While it is generally acknowledged that advances in medicine and technology are beneficial to the disabled community, the emphasis on cure can undermine disability pride. As Ben Mattlin writes, "not all of us in the target

market are actually seeking this solution. I can't help feeling there's a contradiction between taking pride in one's disability and hankering for a cure. You don't try to cure something you like about yourself".(Mattlin) When considering cure, it is important to consider the implications of changing something that is essential to an individual's identity.

- "I'm not your inspiration". (TED)
    - There is a tendency in our culture to use the accomplishments of people with disabilities to show that "anything" is possible. This tendency to tokenize people with disabilities is problematic because it objectifies them in order to inspire able-bodied people.

While there is a premium on time and space in biology class, it is my belief that engaging with patient testimonials will improve the understanding and treatment of disease.

Guiding Questions

Please consider the following questions when reading the anthology:

1. Based on your study of genetic conditions in Biology 200/201, what are your prior associations with the conditions described in the narratives?
2. How do the patient testimonials describe living with the conditions? Are there common experiences across the narratives? What experiences are unique to specific conditions?
3. How do the patient testimonials confirm or challenge your preconceptions of living with genetic differences?

Biology 200/201 Assignment

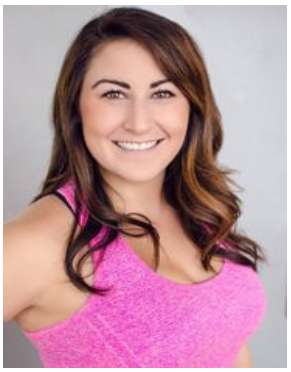
Choose 1 of the conditions: CF, PKD, PCT, Ovarian Cancer

Write a one paragraph essay not to exceed 350 words.In a cohesive and coherent narrative, tell a story about the condition. Include specific references to the patient testimonials and lecture material. Be sure to introduce the topic with a single sentence which includes the perspective gained by reading the patient testimonials and end your paragraph with a closing sentence that discusses your understanding of genetic difference.

Patient Testimonials

Porphyria Cutanea Tarda (PCT)

- Murphy's Story



"It was confirmed that I had Porphyria Cutanea Tardae (PCT) caused by a mutation in the UROD gene. Because I was predisposed genetically for PCT, it could've laid dormant my entire without ever rearing its ugly head. Though we don't know for certain what exactly caused it, I believe the birth control in large part was the major player. After discussing all possible options, I determined that I would start my treatment with phlebotomies. I decided against the drug route initially because it was clear my liver wasn't functioning properly and I didn't want to send it into overdrive.

After two treatments, regular lab work, and maintaining a healthy lifestyle, I am now in remission. I now know what I need to do to manage my PCT moving forward. PCT doesn't define me, but it is a part of me. The scars on my hands are a constant reminder that I am greater than my circumstance. I believe that the Lord bestows some of the biggest challenges to the strongest, most resilient people. While He might test you, it is important to always be steadfast in your faith because you never know what life might throw your way. My diagnosis changed my perception about the world and made me realize how sweet life really is. I no longer take my health for granted. I am grateful, unlike many with PCT, to be able to wake up every day and live a relatively normal life. Now, at 28 years old, I am a bit more lighthearted and know that with the Lord, anything is possible. I am a better person because of my PCT."(*Member Stories - PCT | Porphyria Foundation*)

- Kathryn’s Story

“My Porphyria Cutanea Tarda (PCT) experience began roughly seven years ago. At the time I was living in Irving, Texas. When lesions appeared on my face, forearms and legs, I thought that perhaps it was related to Psoriasis, an autoimmune disorder I had since I was a child. As a result, I resorted to a fairly common approach to Psoriasis which is exposure to UV rays. I spent an hour or so every afternoon in the sun, but more lesions developed and the existing ones grew in size. Repeated visits to my Dermatologist resulted in a variety of diagnoses including eczema, hives and finally a "picking" disorder which basically meant the doctor believes you are picking at your skin causing sores and infections.

It was several months later when I met with Dr. Melissa Costner in Dallas. By that time, most of the lesions and blisters were healed, however, the splotchy dark patches and scarred white areas covered most of my arms, face and large portions of my legs. I described my experiences over the previous three years. Dr. Costner listened closely and then said she wanted to do some blood work. Three weeks later, I returned to her office where she explained I had a familial type of PCT. In one minute, all of the heartaches of the previous four years made sense. She listened to me, something the previous doctors failed to do. She believed me when I said I wasn't "picking" my skin and said that rather than affecting areas where I "could reach," PCT was actually affecting areas being exposed to the sun. By reducing the time I was spending in the sun due to my energy issues, I had actually started the PCT healing process.

Today I still have very dark patches on my skin and very white scars but I have few recurrences of PCT. I play close attention to the medications I take, particularly since I have other autoimmune disorders which require a heavy regimen of treatment. PCT was not something I had ever heard of and was honestly not my biggest health concern at the time. But finding a doctor who listened gave me a sense of empowerment that I continue

rely on today in all aspects of my health and with all my doctors."(*Member Stories - PCT | Porphyria Foundation*)

- Diana's Story

  "Last summer I was babysitting my four grandchildren at my daughter's house while they were on vacation, I noticed a large blister on my hand and thought I had splattered grease on it. After I went home a week later, I had more blisters on my hands of different sizes. I had a really big one on my right hand, and every time I shook hands it would hurt.

  A friend suggested that I should sterilize a needle and drain it. I did so and put Neosporin on it as well. Needless to say it got infected, so I went to the Dr. and he started running tests on me. After several tests, he found my liver enzymes were high, so he sent me to a dermatologist who did a biopsy of a small blister and had me do the 24-hour urine test. That is when I found out I had Porphyria Cutanea Tarda (PCT).

  Next I started the phlebotomies. On Dec. 1st, the 8th time the Dr. did the blood draw, I got what I thought was the flu and was so sick, I could not go anywhere without falling back on the couch, out of breath and I was very weak. My best friend told me to call the Dr. and she would take me as I couldn't drive. I did call and she took me to see my regular doctor. His nurse walked in and took one look at me, and said I was anemic. The doctor checked me and said I needed iron. I told him I couldn't because of my porphyria.

  In January of this year, the doctor took another blood test that was better. Now I am going in every three months for my blood checks. The scars on my hands are fading slowly. My granddaughter says your "owies" are better and I can hold your hands, and my skin is not so fragile as it was, although I am a grandma and most grandmas have fragile skin, it should not be that fragile yet.

  I was noticing some of the other symptoms of PCT, and I just went to the Dr. complaining of pain in my arm, hands, and insomnia. Where I live they don't know much about porphyria. My life is so much better now, but I still stay out of the sun, avoid alcohol and estrogen, and yes my problems could be much worse."(*Member Stories - PCT | Porphyria Foundation*)

- Kelly's Story

  "My name is Kelly Story and I live with my husband, Chad, and two cats in Kissimmee, FL. In July of 1999, one month after our wedding, I was out of town on business, and I noticed tiny little water blisters all over my hands. The blisters didn't itch and were not painful. Although I found them to be very odd, I didn't worry much.

  Over a short period of time, my skin became extremely fragile. It seemed like almost anything would cause a scrape on the skin of my hands. Plus, the blisters got much worse. My hands looked like something out of a horror movie. I was so embarrassed all of the time, and I cried a lot. I went to a dermatologist. After two visits, he told me that he was fairly certain that I had PCT but suggested that I go to a specialist. After several humbling tests and weeks of waiting, I was officially diagnosed with PCT.

  The doctor said that I would have to start phlebotomies. This meant that over a pint of blood would be taken from me at one time. Even after I began the phlebotomy treatments, the blisters moved to my arms. These blisters itched tremendously. My poor husband didn't know what was happening, because, I would wake up in the middle of the night with a frenzy of scratching and crying. The itching was unbearable. I would feel so guilty afterwards. After all, I was receiving treatment for my condition and knew that I did not have a fatal disease. There are other people in the world with so much more serious problems. But, it was still uncomfortable, and my hands and arms were just so ugly.

  I was tired a lot during the six month or so period I was having the phlebotomies. The doctor said that I would most likely be temporarily anemic. This was the result of having the blood taken from me. I used to do aerobic exercises at least four times a week, but that stopped. I also got headaches quite often. Finally, I was in remission. No more cuts, no more blisters, and no more itching.

  I lived for a while with the dark scars on my hands from all of the sores and blisters and several lighter ones on my arms. But now, the scars have faded tremendously. You can't see any on my arms, and the ones on my hands are very pale. It may sound vain, but I never thought I would have pretty hands again, and now I do. To this day, I still cringe if I accidentally knock my hand into something. But, I am always relieved when I look down and see no scrape or cut.

  Since being diagnosed with PCT, I've taken estrogen and alcohol out of my life, and I try my best to stay out of the sun. It's hard living in Florida, but I just keep applying that good old sunscreen."(*Member Stories - PCT | Porphyria Foundation*)

Ovarian Cancer

- Linda's Story

"I'd been having problems during the festive season. My husband and I had gone to stay with friends but I just didn't feel right, I was constantly tired and jaded. Not long after, we went on holiday and I remarked to my husband that I was putting on weight. I felt as if my stomach was getting bigger and bigger. For a few weeks I thought it was just menopausal weight gain or mid-life changes. It was persistent bloating as it wouldn't come and go. I was also having bowel problems and was tired all the time. I decided to go to the GP to talk about all of my symptoms. She thought it was probably thyroid problems and sent me to have blood tests done. Nothing came of it.

Despite the blood test results, my symptoms began to get worse. I started to feel full quickly after beginning meals and my abdomen became very hard. I went back to the GP and asked for more help because I knew something wasn't right. She then ordered a CA125 test. After an ultrasound and CT scan, I was given an appointment with the consultant. I was really pleased that my GP had sent me for tests so quickly. I went to this consultant's appointment on my own, but they brought my husband in. I was told it was ovarian cancer and it had spread. The more she told me, the more I thought, 'this is bad'. I had nodules of cancer in the omentum, liver and all the way up to my diaphragm. The weight gain, what I'd thought was just menopausal weight gain, actually turned out to be ascites. I had three litres of fluid drained to make things a little more comfortable.I started chemotherapy in mid-April – following a 'plan of action' I had two cycles of paclitaxel, carboplatin and two of Avastin. I was also scheduled for surgery.

The operation and recovery was tough. I had an epidural, which was left in for four days, and I was on very strong painkillers. I found the first few days after the surgery very difficult. I felt much better after I was able to have a wash and go to the toilet by myself. Once home, my GP was supportive, which really helped. It was reassuring to have that

extra support. After the surgery and subsequent chemotherapies, I showed no evidence of disease. I held the surgeon's hand and said 'thank you'."(*Linda's Story | Target Ovarian Cancer*)

- Moira's Story

"For months, most of my symptoms were put down to IBS or stress. I'd been going back and forth to the GP with abdominal pain and fatigue. One night I just knew there was something really wrong. I had a feeling, a really terrible feeling. I was finally diagnosed with ovarian cancer after I went back and saw a female doctor.

Eventually, after various scans and blood tests, I was referred to oncology. Finally I got a diagnosis; I had stage III ovarian cancer. My path from seeing the GP to my eventual diagnosis went over the 64-day timescale.

I underwent six rounds of chemotherapy and then a debulking operation. The care I received after my diagnosis was fantastic. My oncologist explained everything in a way I could understand. He had a great way about him. He didn't build my hopes up, but he didn't dash them either. He just inspired confidence. My chemo nurses were also amazing. They were such a jolly bunch and so good in their job.

Before I started treatment I had this horrible picture of chemo in my head. I suppose I imagined lots of people with shaved heads sitting around connected to drips; it wasn't like that at all. In fact, I used to come out of the chemo unit feeling uplifted and happy. There was a lot of support.

After a bit more recovery time, I had another two rounds of chemotherapy, and another scan. My husband died on 31 July – he had dementia – and then I was told on 1 August that the cancer was gone." (*Moira's Story | Target Ovarian Cancer*)

- Pat's Story

"I had been visiting my GP for two years due to frequent urination. I could never seem to get a full night's sleep, and I would be up 10 times during the night. My doctor eventually referred me to a bladder clinic where I was told I'd be given a prescription and physiotherapy. When I didn't hear anything back from the clinic, I forgot about it.

A year later, I received a letter from the clinic giving me another appointment. I phoned them back and told them that I didn't really see the point in going since they had never sent me a prescription or a physiotherapy appointment after the last time. That afternoon my GP phoned to say my prescription was ready to pick up, I asked from whom and was told that it was from the bladder clinic – one whole year later. Apparently the delay was due to a clerical error. To say I was angry was an understatement.

Then, in April, during a trip to Scotland to see a friend, I began experiencing terrible pain in my right side. I could hardly walk and when I passed urine the pain in my side got worse. I got an appointment with my doctor and I explained my symptoms. She felt my stomach and said there was nothing untoward but that she would send me for a scan. I got the appointment for my scan six months later. That was the day I was told I had stage IV high grade serous ovarian cancer.

After a small operation for a biopsy, I was told I had a tumour in my fallopian tube and that I would need a hysterectomy. I went away to wait for the operation but was called back in so they could explain more to me. I was expecting to be given some pre-op tests but instead I was told that I wasn't going to have a hysterectomy at that time, without any

explanation. I thought I was going to get the operation; I had had no idea that it was going to be this news.

I'm currently having chemo – taxol and carboplatin. I've responded really well to treatment so far and of the 10 tumours in my abdomen, five have disappeared, and five have shrunk to the size of a marble. I met some lovely people during my treatment – including women who had a diagnosis of ovarian cancer. That's where I got the idea to play around and get creative with scarves. It got to the point where other women would ask me for advice! It's also where I heard about Ovarian Warriors – a Facebook group for women with ovarian cancer in Northern Ireland. Reading other people's stories and being able to give support – it's all very positive." (*Pat's Story | Target Ovarian Cancer*)

- Renate's Story

"Hello. My name is Renate McGrail. I am 46 years old. I was diagnosed with Stage 3b ovarian cancer on New Year's Day January 2012.

After having an emergency CT scan at the hospital my diagnosis was swift. On January 10th they performed a complete hysterectomy. They removed my uterus, ovaries, Fallopian tubes, omentum and part of my diaphragm. They were able to obtain optimal debulking and were confident that they had removed everything and that the cancer seeds that were remaining would be addressed with chemotherapy.

I underwent 6 rounds of aggressive chemotherapy directly into my abdomen. I had read that this type of chemotherapy had the highest success rate. Unfortunately, my body was resistant to this type chemotherapy and that since I had had the most aggressive therapy there was that there was not much else they could do. Fortunately, my story had not yet ended. I was lucky to get a spot in a clinical drug trial in September 2012. This seemed to work for me and held the disease stable until February 2013 when they had to remove me

from the trial because the tumours had started to grow again. Again I have been fortunate to be able to join another drug trial. I have been on this trial for 8 weeks and will find out on May 6th whether this treatment is helping me.

Our family has been blindsided by all of this. There is no history of ovarian cancer in our family. I had experienced stomach aches and heavier menstrual symptoms. I had an ultrasound in March 2011 and there was no sign of anything. I have 3 daughters that are 17, 13 and 6. I want a test to be found in their lifetime to diagnose this disease early. My two daughters are doing their part to create awareness by participating in a legacy girls study at Princess Margaret Hospital. My thirteen year old daughter is also creating t-shirts for the first world ovarian awareness day with her friends."("Renate McGrail")

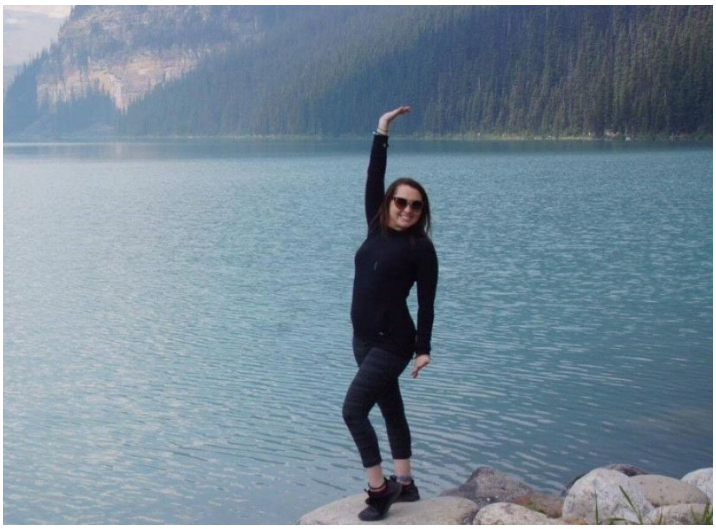
Cystic Fibrosis (CF)

- Karen's Story

"I started experiencing symptoms when I was four months old. That's the first time I had pneumonia (inflammation of the lungs) and after that I had pneumonia a couple of times a year. I played sports in high school and because I grew up in such a small town, the local doctor used to come to my games with epinephrine (adrenaline), oxygen and whatever else I needed because he knew that by the third quarter, I was going to need it. I had pancreatitis (inflammation of the pancreas) for the first time at the age of 11 and I have had it many times since then. During my adult life, I experienced every symptom of cystic fibrosis at one time or another. When I was diagnosed, my doctor went through a list of all the symptoms with me and I had experienced all of them."

When I started on Kalydeco®, it didn't take long for it to start working. Maybe a month at most before I started seeing changes. Today, I've been on Kalydeco® for about a year and I probably feel better at 70 than at 17.I don't become out of breath quickly anymore and I deal with weather changes a lot better, which always used to make me feel unwell and short of breath. I haven't had any gastro problems [stomach issues] since I've been on it.I hadn't mowed my yard in years, I had to hire someone to do it. Now I'm mowing and working in my yard all day long. I play golf now and I go for walks. For 70 I think I'm pretty good. I'm so grateful for this treatment and for the researchers who made it."(*Karen's Story*)

- Alana’s Story



“At school camps I’d have to go up and take medication and had to be wary about swimming or cross country because contact with soil and some types of water can harbour bacteria that can be dangerous for someone with CF. But I refused to let my family coddle me but if someone finds out you have an illness that’s what they tend to do. There were times when I hid my illness.

But I was quite lucky in that I wasn’t too sick during my teenage years. I still managed to have a gap year when I was 18 and go travelling which has always been a big focus for me. There were a lot of people who tried to talk me out of it because I was going away for five months and doing a Contiki which meant a lot of young people living a party lifestyle. I tried to keep up treatment as much as I could, and I did manage, but it wasn’t easy.

When I returned to NZ I got a certificate in nannying, followed by an Early Child Education degree and an Infant and Toddler Mental Health diploma, and started a career in early childhood education.

When I was 25 I embarked on another big trip around Europe and Egypt but this time my health was starting to fail and I got exhausted very quickly.

By the time I was 28 my health was really declining. I would have to have rest breaks while brushing my teeth because I’d lose my breath. But I still didn’t think I needed a lung transplant – I was in denial that I was that bad. Like every CFer we think that we can do it on our own and if we do our nebulisers and we do our physiotherapy we won’t need a hospital admission – that’s how stubborn we are. Then in just five months I declined

very quickly. My lung function dropped by 20 percent, I was on oxygen 24/7, I weighed just 42kgs and my hair was falling out.

I remember by the time it was decided a lung transplant was my best chance for a future, it was almost a relief. I was scared that I wouldn't be accepted on to the lung transplant waiting list because there are a lot of tests you need to pass before you are. The medical team need to be certain that donor lungs go to someone who will care for them.

I was only on the list for six weeks before my transplant which was a surprise because I had a rare blood type. There was a lot of support from Cystic Fibrosis New Zealand from the time of transplant assessment right through to post-transplant.

Looking back since my transplant in 2015, I can't believe how sick I actually was. I had managed to work until a few months beforehand but I would have to spend my lunch break hooked up to an oxygen machine.

After my transplant I vowed to continue my love of travelling, and this time I was able to do it easily and all with the awareness that the gift from my lung donor made the journey possible.

I celebrated my 30th birthday with 28 friends and relatives in Las Vegas, went to the Grand Canyon, Niagara Falls, New York and went ziplining at Whistler. I want to go back to Europe and go to all the places I struggled to do before."(*Alana, Adult from Christchurch » Cystic Fibrosis NZ*)

- Taylor's Story

  "I am currently 24. I was diagnosed with CF right before my 16th birthday in 2010. I was tested because for my entire life I had what they thought were terrible allergies and "faked" stomach pains — every doctor I had seen was adamant that I was faking to skip school.

  My dad, who is in the U.S. Army, got stationed in Hawaii, and the military hospital just so happened to be a CF center. After a few visits for the same [stomach] issues with my new doctor there, he believed my symptoms and was determined to help me. He put together all the pieces, met with the CF team and decided to sweat test me. The first [test] was borderline, as was the second. So, we did genetic testing and it came back to show I carried the deltaF508 genes — two of them. Once I was diagnosed, they tested my entire family and my mom was diagnosed in her mid-30s. We were all really shocked; we had never even heard of CF before!"("3 Cystic Fibrosis Diagnosis Stories from Our Community")

- Sabrina's Story

"My life journey with cystic fibrosis (CF) began over 25 years ago, when I was diagnosed with the double F508del mutation at the age of four years. I am one-quarter Tlingit(KLIN-KIT) Indian (Alaska Native) and live in Anchorage, Alaska. Because CF is not usually found among Native American/Alaska Native people, it took a lot of time before a sweat test was suggested.

My running journey started when I was 12 years old. My mother wisely believed that running could be used as a form of airway clearance. She would take me to a local track in Anchorage and set a goal for me to run 10 minutes straight. She always ran with me. We learned that running helped me to loosen the thick mucus from my lungs and cough it out.

When I was 18 years old, doctors found a cancerous tumor on my spine, and I was diagnosed with non-Hodgkin's lymphoma. This was a brutal time. The chemotherapy and radiation treatments made it difficult to continue my running routine and I eventually had to stop altogether.

My running revival came in 2010 after being in remission for five years. I wanted to become more proactive in regards to my cystic fibrosis and my overall health. Once I started running again, I realized how much mucus I was bringing up and how clear my lungs were feeling after each run. I started out by setting goals like running a 5k race. After I accomplished that, my new goal was a 10k, then a half marathon, then a 16-mile mountain run, then a full marathon!

My lung function will sometimes drop and indicate that I need antibiotics or hospitalizations, but that always reminds me how important running is in my life and to my health.

My main motivation for running has been to outrun cystic fibrosis and to prevent further lung deterioration. I am determined to run and exercise for my health. I now have a wonderful son, Leo, and It's not just about me anymore. I have a child who needs me; I want Leo to grow up with his mom."(*Running the CF Road – Cystic Fibrosis Research, Inc.*)

Pyruvate Kinase Deficiency (PKD)

- Unnamed Woman

"I was born in 1994. My mother's pregnancy went fine. When I was born, someone realized something was wrong. I was born with no color. My dad says I was grey. Doctors say I had barely any blood running through my body, so the moment I was pushed out I was rushed out to get a transfusion. I got 1 blood transfusion. I was in the hospital for about 2 weeks and then I was able to go home. No one had any idea what was wrong with me so I just went on being a kid.

When I turned 10, I started to notice my eyes turning yellow. I was told it was just jaundice and I found out I was anemic. I expected to be tired and weak. Still, I went on with life as a normal teenager. One night, I was sleeping and I felt a horrible pain in my side. I was 15 at this time and figured it was cramps. But the pain got worse. My mom rushed me to the hospital and turned out to be an ovarian cyst. But, the doctor who was working with me wasn't too worried about my cyst but the coloring of my eyes. He said it's more than just me being anemic. He ran some test and my Hb was extremely low. If I´m not mistaken it was a 6 g/dl. He did some research and that's how I found out I had Pyruvate Kinase Deficiency. I met up with a blood specialist and went over everything that I was going to go through. I got 2 blood transfusions and it made me feel better. I felt normal, well, normal from feeling tired my whole life.

About a year later I was looking into joining the military. When I was told I couldn't because I have a blood disorder and I don't have a spleen, my dreams felt like they were just stolen from me. I was angry, I blamed my parents for giving me this. I hated it. I was extremely depressed for the longest time because it stopped me from doing what I wanted to do.

By 18, I noticed I was starting to feel how I did before I found out I had Pyruvate Kinase Deficiency. The doctors were confused because they said I shouldn't feel like this. I should feel better. They decided to see if my spleen had grown back. 3 years laters and my spleen had grown back. So I got ready for yet another surgery. March 2013, I went in yet another time to have my spleen removed. As I lay in the bed, I just thought about my life. How much stuff I've been through and as I looked around I saw that it could be worse.

That's when I realized what I wanted to do. My goal now is to be a nurse. I see what these wonderful people do for me, and for me to say 'Thank you' isn't enough. I want to do what they do. It's been an year since my last surgery and I feel amazing. I'm about to graduate high school and go to college. I dance a lot and walk to maintain my exercise. I am just happy to be alive. I'm still going to go to school to be a nurse. I'm still on antibiotics and I still have to watch out for any flus or colds. Getting sick comes a normal thing. I'm not letting it stop me though. I'm now at peace with having PKD. My doctor has been a Dr. for 20 years and I'm the 2nd person he's met with Pyruvate Kinase Deficiency. Overall, I'm just happy to be living this life." (*Personal Stories | Pyruvate Kinase Deficiency*)

- Unnamed woman

"My 3 years older sister has Pyruvate Kinase Deficiency. When she was born in 1984 having severe jaundice and a low hemoglobin, the doctors had to run a lot of tests to make a diagnose. My mother read the medical file afterwards and was shocked by all the terrible diseases they were investigating. After some months, the diagnosis of PKD was made. My sister has a quite mild type of PKD and she didn't need to be transfused often.

When my sister was 2 years old, my parents wanted to have another child. They went to a genetic counselor, who told them they had 50% chance a second child would also be affected, but the disease could be 10 times more severe. He was quite pessimistic, but luckily my parents didn't listen to him and conceived a second child. When I was born, in 1986, I was as yellow as a banana. My parents immediately knew I had PKD. I had an exchange transfusion and was put under the UV-lamp.

Until the age of 6, I had a blood transfusion every 6 weeks. My parents learned to read the signs of low hemoglobin. Sometimes my Hb dropped to 3 g/dl and I happily ran around without noticing I was running in a zigzag line. My parents would then rush me to

the hospital for a transfusion. When I entered the pediatric service, the nurses called me "snow white", and when I left the service after the transfusion, they called me "little red riding hood". As a kid, I really liked the transfusions, because in the hospital room I could watch Disney movies on the tv.

When I was 6, my spleen and gallbladder were removed. I don't remember much from the surgery, except for the annoying tube in my nose. I had a lot of visits in the hospital from my friends and family and got a lot of attention (and presents), which was nice at that age. My sister had her spleen, gallbladder and appendix removed a year earlier. After the spleen removal, my hemoglobin went from an average of 5 g/dl to 8 g/dl. Both my sister and I didn't need transfusions anymore. We took antibiotics for about a year and from that moment, we only went to the hospital once a year for a checkup with our hematologist.

I lived a very active life: our parents wanted us to do a lot of activities, so our weeks were filled with music classes, singing in the opera choir, horse riding, theater, ballet, jazz dance, … Every day after school, I had another activity to attend. Even though I'm not the most sportive girl, I was perfectly able to live a normal life. Having Pyruvate Kinase Deficiency was something I could easily forget, if it wasn't for the yellow skin and eyes. Being a teenager, it sometimes was annoying having to answer the same question over and over again: "why are your eyes so yellow?". But luckily, I wasn't bullied for it at school nor anywhere else.

After high school, I went to university to become a pharmacist. I had a lot fun, partied a lot and had no difficulties with studying. I graduated with great distinction and started to work as a pharmacist. I am now 29 years old, and with my partner we enjoy travelling, camping, hiking and backpacking. We did some serious treks in the mountains and I did even climb Half Dome (!). My hematologist always warned me for high altitude, so I try to avoid heavy exercise above 3500m (11.500 feet), but I like pushing my limits." (*Personal Stories | Pyruvate Kinase Deficiency*)

Acknowledgements

I would like to thank Professor Kristen Lindgren for providing me with direction for this project and supporting me throughout the process. I would also like to thank Professor Rachel Hoang from the biology for her time and consideration of how this curriculum might be implemented at Haverford.

Works Cited

"3 Cystic Fibrosis Diagnosis Stories from Our Community." *Cystic Fibrosis News Today*, 14 Sept. 2018, https://cysticfibrosisnewstoday.com/2018/09/14/cystic-fibrosis-diagnosis-stories-community/.

Alana, Adult from Christchurch » Cystic Fibrosis NZ. https://www.cfnz.org.nz/life-with-cf/stories/alana/. Accessed 15 May 2019.

Karen's Story: 70 with Cystic Fibrosis. https://raremark.com/cystic-fibrosis/articles/karens-story-70-with-cystic-fibrosis--534. Accessed 14 May 2019.

Linda's Story | Target Ovarian Cancer. https://www.targetovariancancer.org.uk/stories/symptoms-ovarian-cancer-awareness-month-awareness/lindas-story-0. Accessed 15 May 2019.

Mattlin, Ben. "Opinion | 'Cure' Me? No, Thanks." *The New York Times*, 20 Jan. 2018. *NYTimes.com*, https://www.nytimes.com/2017/03/22/opinion/cure-me-no-thanks.html.

Member Stories - PCT | Porphyria Foundation. https://porphyriafoundation.org/member-stories/porphyria-cutanea-tarda. Accessed 14 May 2019.

Moira's Story | Target Ovarian Cancer. https://www.targetovariancancer.org.uk/stories/symptoms-ovarian-cancer-awareness-month-awareness/moiras-story. Accessed 15 May 2019.

Opinion | Becoming Disabled - The New York Times. https://www.nytimes.com/2016/08/21/opinion/sunday/becoming-disabled.html. Accessed 15 May 2019.

Pat's Story | Target Ovarian Cancer. https://www.targetovariancancer.org.uk/stories/symptoms-awareness/pats-story. Accessed 15 May 2019.

Personal Stories | Pyruvate Kinase Deficiency. https://pyruvatekinasedeficiency.com/personal-testimonials/. Accessed 6 Mar. 2019.

"Renate McGrail." *World Ovarian Cancer Day*, https://ovariancancerday.org/renate-mcgrail/. Accessed 14 May 2019.

Running the CF Road – Cystic Fibrosis Research, Inc. http://cfri.org/running-the-cf-road/. Accessed 6 Mar. 2019.

TED. *I'm Not Your Inspiration, Thank You Very Much | Stella Young*. *YouTube*, https://www.youtube.com/watch?v=8K9Gg164Bsw. Accessed 15 May 2019.