Elizabeth is a beautiful brown hair, brown-eyed girl. She sits with poise and dignity in her hospital chair every Monday. During the last year, I have come briefly into her life with the intention of understanding more about the patient in the context of her illness. However, my experience instead was with a young girl who refused to let her illness place her into any type of context. She is strong. She is in control. She has been handed a situation of circumstances that would be hard for most to handle. Yet, without a complaint, she tackles the obstacles while studying for 7th grade trigonometry and planning her summer vacations. How are some people so strong in the face of a chronic, life-long illness? How can some stand so strong, beautiful and proud in the face of so much adversity and personal angst?

In 2008, at age 5, Elizabeth presented to the ER with diarrhea, weight loss, swelling of her lower extremities and puffiness in her eyes. Histology showed lymphangiectasia, a pathologic dilation of lymph vessels, in the second portion of the duodenum. The dilation of the lymph vessels and obstruction of the lymphatic drainage of the intestine can progress to a rupture of the intestinal lacteals. This can lead to leakage of lymph into the lumen of the bowel. The common clinical findings with intestinal lymphangiectasia include diarrhea, abdominal distention, peripheral edema, chylous effusions, and repeated infections (Hoffenberg). Since the chylous effusions represent the direct absorption of fat from the small intestine lacteals, the effusions are rich in fat, vitamins, immunoglobulins, and calories (Evans). Laboratory testing of individuals with lymphangiectasia often show decreased immunoglobulin and serum albumin levels, lymphocytopenia, anemia. In addition, the patient's serum calcium and magnesium levels

are often decreased because the cations are lost in the diarrhea with the unabsorbed fatty acids (Hoffenberg).

Elizabeth's symptoms in 2008 resolved but after 5 stable years, Elizabeth presented again to the hospital with puffiness in her eyes and swelling of lower extremities. Her laboratory studies indicated low lymphocyte count and serum albumin level. Elizabeth was placed on bactrim for prophylaxis to decrease the risk of a bacterial infection. A follow-up CT scan and endoscopy revealed lymphangiectasia in most of her small intestine. The results of Elizabeth's lymphangiogram showed complete obstruction of the thoracic duct, the largest lymphatic vessel in the human body. In April of 2014, Elizabeth was hospitalized to receive albumin, Lasix infusions, and nutritional evaluation. Elizabeth was started on a high-protein, low-fat diet in the form of nutritional protein shakes.

After Elizabeth's hospitalization, a treatment plan was put into place for her to receive weekly albumin infusions, followed by Lasix infusion to help with her edema. It is more like a maintenance plan than treatment plan, hoping to keep Elizabeth's body as healthy as possible. Elizabeth knew this was the case and these treatments were not curative. Elizabeth has continued to struggle with hypocalcemia, hypomagnesmia, and hypo vitamin D, despite being place on maximum doses. From the low ionized calcium levels, Elizabeth experiences sporadic episodes of tetany, or cramping in her hands, that could last up to 30 minutes and was hospitalized multiple times for hypocalcemia with fear it could lead to a heart arrhythmia.

I first met Elizabeth during an intense family meeting to discuss whether she will be put on Sirolimus. Sirolimus, an FDA approved immuno-suppressive medication,

inhibits mTOR. mTOR is involved in the activation of protein synthesis, which results in activation of cellular processes including angiogenesis and cell proliferation. The thought is that mTOR may play a role in the vascular and lymphatic abnormalities present in diseases. By blocking mTOR, you are preventing the activation of the protein synthesis and downstream cell proliferation and increased angiogenesis (Lackner).

Elizabeth had bright and piercing brown eyes, and was a plump young girl with radiant skin and a beautiful smile. Her appearance was quite different from the fragile and sickly looking child I had imaged her being. She greeted her physician warmly and agreed quickly to have a medical student follow her case. There was a heaviness in the room, though, as the family discussed the serious side effects of Sirolimus, such as decreased cancer surveillance in the body.

Over the next seven months, I met with Elizabeth as she received her infusions and learned both about the obstacles and joys in her life. Elizabeth is in a program that takes students from low-income areas and places them into suburban schools. The bus to Elizabeth's school leaves around 7:30 a.m. In order to finish her morning protein shake and supplements, Elizabeth wakes up between 5 and 5:30 a.m. This process takes multiple hours and she often misses the bus. Since her school is not close to where she lives, her father often drives her to school. Without traffic, the driving distance is 30 minutes. Her father works in the other direction, and is frequently late for work. I had the impression that it was a problem with the father's employer that he was frequently late for work. I can understand why taking the bus for Elizabeth would be stressful. There is no bathroom on the bus and she has just consumed a large amount of liquid.

Elizabeth can no longer eat solid food, except for high-protein lunches at school to avoid questions from her peers. Elizabeth told me on her last visit that she did not mind the diarrhea and that she would choose to have diarrhea every day if that meant she was able to eat what she wanted to. Elizabeth's experiences highlighted the obstacles individuals face who are suffering from a serious medical illness, but don't have many outward physical manifestations of the disease. While I was visiting with Elizabeth in January, she mentioned to me that her teachers often do not let her use the bathroom more than once or twice a week in a certain class. She often has to remember in which classes she has used the bathroom that week and try to spread out which classes that she asks to use the restroom. This is especially hard she said when she "really has to go" or has diarrhea. I let Elizabeth known that I would mention this to her physician, because it did not seem fair that with her medical conditions, the teachers would not be understanding about her having to use the restroom frequently. Elizabeth's quality of life during school is very important, especially as she holds on to normalcy.

When I asked Elizabeth if she tells her friends about her disease, she said when her face is puffy, she tells them she has allergies. The name of Elizabeth's disease, lymphangiectasia, is hard to pronounce and a very rare disease. Would the teachers in Elizabeth's school or her friends be more understanding if Elizabeth had a disease such as diabetes or cancer? Does the foreignness and lack of comprehension about the disease make it harder for people to empathize with Elizabeth? I think this is a real possibility. Even if Elizabeth tried to explain the disease to her friends, it would be difficult to try to figure out what words to use to explain it. Lymphangiectasia is a dilation of lymph

vessels. How do you explain what a lymph vessel is? A lymph vessel drains lymph. What is lymph? Why are they dilated?

So instead, Elizabeth has told people that she has allergies if asked about her eye puffiness or why she is eating a certain diet. This might create a barrier to Elizabeth's friends understanding what Elizabeth is going through. On the other hand, it may also help Elizabeth maintain normalcy in her life. She spends the school day under the guise of normalcy. No one in her class looks at her and see's someone who is sick. They cannot see the intestines that are not working, or her diminished immune system. They do not know that she is on a powerful immunosuppressive drug that puts her at higher risk of infection or even developing cancer later on in life. When they do not wash their hands after the bathroom and touch the door handle, they do not know the impact they could be having on Elizabeth's life. I believe it is our role as physicians to share knowledge with our patients so they can build bridges of understanding between them and their friends and acquaintances about the rare disease. We need to help them prepare how to explain their illness and empower them to speak up about their condition.

One time I went to visit Elizabeth during her infusion in January or February. When I entered into the curtained off room of the Cat-CR, she had the usual IV line placed in her arm. The blue chair had a natural recline to it, but she was sitting up over the push up tray that most people eat off of. She had a large binder opened and she was reading through math notes. The room was loud but brightly lit. The nurses frequently come into to check on Elizabeth and change the infusion medication when the system starts to beep. In the middle of this organized chaos was Elizabeth trying to study for her math exam the next day. She had been performing poorly on her previous math exams. I

wished that her teachers could see her in this moment, trying hard to preserve through the adversity and perform well in school. They might see the girl who is struggling in class in a different light.

After my conversations with Elizabeth, I thought about the factors that can limit our success as a physician in helping our patients. Even if the physician provides the best medical care possible, there are many outside factors that can influence the health status of individuals. I feel that it is our responsibility, along with the other members of the health team such as social workers, to identify and work with these other entities in order to provide the best quality of life for the patient in the face of their illness.

Elizabeth's infusions usually last from 2 p.m. to around 6 p.m. on Monday's. The times when I have arrived at the Cat-CR to visit with Elizabeth before 4 p.m., her mother is often asleep in the chair. She is in her work uniform, and leaves Elizabeth around 3:45 to go to work. At that time, Elizabeth's grandmother arrives to sit with Elizabeth and bring her home from the infusion. Elizabeth's grandmother arrives wearing a maroon polo shirt with the name of a nursing home facility on it. She speaks very little English, and is always pleasant. Within a few minutes of arriving, she is often nodding off against Elizabeth's chair or with her head resting against the wall. She looks young for a grandmother. I would think she is in her early 60s but she looks tired. Both Elizabeth's grandmother and mother look very tired to me, with dark bags underneath their eyes. So at any time you go into Elizabeth's room, there is a guardian who is usually resting against her hospital chair or against the wall in her hospital room. Elizabeth is never there alone.

Elizabeth's family does not have the luxury for her illness to disrupt the inner workings of the family. According to her physician, Elizabeth is very aware of the burden her illness has on the family. She understands the implications that missing the bus might have on her father, and works to make sure that she is on the bus at 7:30 a.m. Elizabeth understands what foods she is able to eat and not eat with her illness. She is not policed by her parents, saying "oh you can not eat that" or attend an event that might serve food that would make her sick, because her parents are working and are not monitoring Elizabeth's every moment. Elizabeth has a great deal of autonomy and ownership of her illness, much more than I would ever expect for a 12 year old. She makes her own decisions of which food to eat, and knows the consequences of what will happen when she eats something that has fat in it. Elizabeth has extensive knowledge about which foods contain fats and what she is able to eat. Elizabeth also understands the financial burden her illness has on the family. She mentioned at one of our last meetings that parking was expensive at the hospital, and that she is planning on taking the public bus to the hospital for infusions when in high school.

When I met with her physician in May to discuss how the experience was following Elizabeth, she made an observation that in the beginning of the year I was asking a lot of questions about Elizabeth's medical illness and as the year progressed, I began asking the majority of questions about the impact of Elizabeth's illness on her life. Through this experience, I learned the importance of learning about the patient as a person and how the social history of the patient can shine much light on to the effects of an illness that cannot be detected by a medical test.

I wrote in the end of my medical school application, "As a physician, I hope to embody the characteristics of a compassionate healer, innovative scientist, and relentless advocate for the vulnerable, speaking for those who cannot be heard. In doing so, I hope to improve the quality of life for those suffering and help others cope with the fear associated with illness. For me, being a physician is not simply a career, but a way of life filled with charity, compassion and the guiding principle of service to others." My experience with Elizabeth has shown me the importance of having a physician who embodies all three characteristics.

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