

The Smith Family Kidney Adventure

October 21, 2007

Dear Family and Friends,

Many of you have been asking for an update on our kidney situation. It has been a busy summer!

As you may know, Katie, Samuel, and Esther were diagnosed with juvenile nephronophthisis back in March and April. Juvenile nephronophthisis is an autosomal recessive kidney disease that is a subset of the larger family of cystic kidney diseases. There are 5 types of nephronophthisis labeled NPHP1 – NPHP5 according to the specific gene that is affected. The incidence of NPHP is 9 cases in 8.3 million live births in the United States. NPHP type 1 occurs during childhood and typically progresses to renal failure before the age of 20, with the median age being 13.

The Gleeson Lab at the University of California San Diego School of Medicine has been doing some genetic testing on our family. The test results show that our family has a mutation in the NPHP1 gene which is located at chromosome 2q12-q13. This confirms that the children have juvenile nephronophthisis. The fact that we have children with this disease means that Tom and I are both carriers. Statistically, each of our children would have only a 25% chance of receiving a recessive gene from both of us, thereby having the disease. Somehow we both managed to pass the gene to 50% of our children. My engineer husband says “we overachieved”! We have another meeting with the geneticist at the hospital on November 5th. We are hoping she will have the formal report from the lab and be able to answer our many questions. Hannah, Grace, and Lydia appear to be unaffected, but we’d like to know whether they are carriers or not.

NPHP is sometimes associated with other disorders such as liver disease, bone abnormalities, and mental retardation. Retinitis pigmentosa (RP) is associated with 10% of cases of NPHP1, NPHP3, and NPHP4, all cases of NPHP5 and, until recently, thought not to occur in cases of NPHP2. This association of nephronophthisis and retinitis pigmentosa is known as Senior-Loken syndrome (SLS), named after two doctors who first identified the connection in 1961. Katie, Samuel, and Esther are among the 10% of NPHP1 patients who also have RP. The term retinitis pigmentosa is used to describe a large group of degenerative eye diseases that affect the light gathering rod cells in the retina. Rod cells are responsible for peripheral and night vision. The progression of RP is varied and difficult to predict. Some people are mildly affected only having trouble seeing at night; many people eventually become legally blind but retain some eyesight all their life; and other people lose their eyesight completely. Because Senior-Loken syndrome is so rare, there is a limited amount of information available. We have no idea, at this time, how RP will impact the children’s lives. They have some additional testing scheduled (electroretinograms) and will be seen by a retina specialist in November.

My dear friend Elisabeth Lipponer in Germany emailed me recently with an amazing story. She sings in a choir that meets once a month for practice. After a long summer break, they gathered once again to rehearse. One of the choir members is a young, talented doctor who does research in retinal transplants. Elisabeth told him a little bit about us and immediately he knew who/what she was talking about – an American family with six children and three with this condition. He didn’t know our name, only the unusualness of the case and the ages of the children. Isn’t that incredible?!?...not only that he knew of us, but that Elisabeth discovered it? My prayer is that our “notoriety” will connect us with someone who will be able to use our family’s genetic information to advance the understanding of this disease.

Well, that’s enough of the background medical information! On to the day-to-day details.....

Esther is doing well with 50% kidney function. She is being closely monitored and we are hopeful that it will be a few years before she needs a transplant. She takes several medications including iron supplements and weekly shots of erythropoietin for anemia. Esther is a good candidate for a preemptive transplant, meaning at the time when she no longer has sufficient kidney function, she would go directly to transplant and avoid dialysis altogether. This plan would, of course, be dependent on having a living donor arranged and waiting in the wings. Esther has grown quite a bit this summer and is doing well with her studies.

Katie has had her share of challenges this summer. When she was first diagnosed in April, she had 25% kidney function. Two months later, she was down to 15% function and needed to begin dialysis. She chose to do peritoneal dialysis because it is done at home during the night and it wouldn’t interfere with her school schedule (ha, ha!).

On June 29th, Katie had surgery to have a special dialysis catheter placed in her peritoneal cavity. To keep this letter from becoming a novel, I will omit all the gory details of her ordeal. The short version of the story is that Katie ended up having four surgeries in 3 ½ months. She had to abandon peritoneal dialysis after two consecutive catheters became clogged with granulation tissue. The surgeon who put in and took out the two catheters says he should write a paper about Katie’s granulation tissue for publication in a medical journal. He says it is “unheard of”. Katie feels very special! ☺

Katie was feeling very ill through these months of complications and, in September, had to drop three of her five college courses. The workload was too much for her after missing classes because of the surgeries. This was a big disappointment, but the lighter class load seems to be working well for her. She now comes with Samuel and me to the hospital three days a week for dialysis. We are quite the little group! Hemodialysis seems to agree better with her. We are hopeful that Katie's hemodialysis will proceed smoothly and be terribly boring. This business of being special and unusual is getting old fast!

We would ask for your prayers regarding a kidney donor for Katie. Our original plan was that I would donate to Katie, but the Lord seems to have someone else in mind. I made it part way through the screening process, but was eliminated as a donor when my EKG showed that I have had a heart attack. I wasn't altogether surprised by the news as there was a time 2 years ago when I thought I was having a heart attack. I was only 43 at the time, I was away from home, and it seemed too impossible that it really could be a heart attack. I didn't want to be a hypochondriac and it was just plain inconvenient at the time; consequently, I talked myself out of going to the ER. I am grateful that the Lord preserved my life and that I didn't die that day on the New Jersey Turnpike (now that would be a terrible fate! ☺). I am scheduled for a cardiac stress test this week.

So far, seven incredibly generous people have offered to donate a kidney to Katie. Unfortunately, five of the seven have been turned down by the transplant team. Statistically, only 25% of the people who present themselves as donors actually pass the screening process and end up being able to donate. Two of the seven would-be donors are still under consideration. We will have to wait patiently through the process. We are very grateful to all of you who have offered such a gift!

Samuel has also had a busy summer with his share of difficulties and complications. In addition to going to the hospital three days a week for dialysis, we made multiple trips to the ER in August and September as Samuel was having trouble with migraines. His blood pressure would skyrocket and he would become nauseated. I wasn't able to give him anything to lower his blood pressure or ease the pain as he would just throw it up. His BP would get dangerously high and we would have to go to the ER to get medicated intravenously. It was thought that the migraines were caused by his vascular system responding to fluid shifts in his body and that the best way to help him would be to remove one of his kidneys.

So....on September 13th, Samuel had a nephrectomy. He was in the hospital for 6 days. After losing quite a bit of weight and taking 4 weeks to recover, he is doing much better. Thankfully, the frequent migraines and trips to the emergency room have stopped. Samuel now no longer needs to drink 6 liters of water a day just to stay hydrated. His urine output has significantly decreased and we are hoping that his bladder will return to a more normal size. Typical bladder capacity is about 400 milliliters (just over 1 ½ cups). Samuel's bladder was so stretched out, it could hold 1550 milliliters (over 6 ½ cups)!

One morning two weeks ago, Samuel complained of chest pain. Dr. Guillot, his pediatric nephrologist, asked me to bring him in to the hospital to be evaluated. The chest pain that Samuel described didn't quite fit with a heart problem, but our very thorough doctor ordered a chest x-ray, EKG, and blood work to determine if something was going on. Dr. Guillot didn't expect to find anything and sent us home before the test results were available. A couple hours later, I received a phone call with instructions to bring Samuel back to the hospital immediately. The blood work showed that his potassium level was dangerously high and the EKG showed that his T waves were spiking. He was at risk of having a heart attack. I brought him back to the hospital to be dialyzed right away. The doctor is still not sure that the chest pain was actually connected to the heart issue and called it "fortuitous". We believe that the chest pain was "providential" and what the Lord used to bring the issue to the doctor's attention so it could be addressed. Praise God!

So....Samuel's newest challenge is restricting his potassium intake. Even if he were to only eat foods that are considered to contain low levels of potassium, by the time he ate the number of calories that he needs in a day, he would exceed his potassium restriction. The recommendation is to eat lots of sugary, empty calories – not something I can get excited about.

Dr. Guillot told us recently that Samuel can have his transplant whenever it is convenient for our family (is there ever such a time? ☺). We had planned to wait until early 2008 to give Samuel as much growing time as possible, but he has had the good "pop" in his growth velocity that the doctor was wanting and there doesn't seem to be any benefit in waiting. I called the transplant team last week to see what they had for available dates over the next couple months. They only do living donor transplants on Wednesdays, so after eliminating the options of the day before Thanksgiving and the week before Christmas, the only two choices left were October 31st and January 16th. Given the new potassium challenge, we have scheduled Samuel's transplant for October 31st! It is a bit of a scramble for both us and the medical team, but it

seems to be coming together. Tom will be the donor. He had a CT scan of his kidneys on Friday. They will use the scan to make a computerized 3-D model of his left kidney and all its plumbing. This will give the surgeons a clear picture of what they will find when they go to remove Tom's kidney. Assuming everything looks good with the CT scan, the last hurdle to clear will be one more crossmatch of Tom and Samuel's blood on the 29th. The crossmatch must be negative, meaning Samuel's blood does not react to Tom's. Next week will be busy with dialysis, preparatory meetings, pre-op appointments, more labs, and a grand tour of the ICU. The "big day" will start very early in the morning on the 31st. We are expecting Tom to be in the hospital for 2 or 3 days and Samuel to be there for a week. It is both an exciting and scary time!

Our prayer for Samuel through all of this comes from Luke 2:40. It is a verse that speaks about Christ as a child. Please pray with us that Samuel would grow and become strong in spirit, that he would be filled with wisdom; and that the grace of God would be upon him. There are sure to be many difficult days ahead. A kidney transplant is a treatment, not a cure. The immunosuppressive drugs that Samuel (and eventually Katie and Esther) will be taking after a transplant come with a whole new set of challenges and potential complications. A transplant is not the end, but rather the beginning of a new phase in our kidney adventure.

The strain of the last seven months is wearing on all of us, particularly some of the children. Samuel will have had 98 dialysis sessions by the time he has his transplant on the 31st. Between dialysis, doctor's appointments, surgeries, and one medical test after another, I am very often away from home. Please pray for Hannah and Grace as they are carrying the bulk of the burden at home these days. It has been a difficult time for all of us. Nevertheless, we are experiencing God's grace and many blessings in the midst of it.

Our "thankful list" is very long. Here are just a few of the many blessings....

- Dr. Guillot, our pediatric nephrologist - It is a privilege to be working with her. She makes things happen! She has been so patient to answer all my questions and has made herself very available to us. We look forward to working with her in the years ahead.
- The terrific team of caring dialysis nurses - They have taken good care of Samuel, and now Katie. They threw quite a surprise party for Samuel's 14th birthday! He still talks about it! They have made those long hours in dialysis much more bearable.
- A friend of Samuel's who has given up many hours to spend time with him in dialysis - It is a special treat for Samuel and makes the long dialysis session go by much quicker.
- The many meals that have been delivered to our door - Our church family and folks from the community (many of whom we don't even know!) have provided us with many delicious meals. This has been a HUGE blessing to us!

We are grateful for the many kindnesses – large and small – that have been shared with us. We are humbled to know that people, literally around the world, are praying for us. Our immediate request would be for an uncomplicated transplant for Samuel and specifically that his body would not reject Tom's kidney. Please pray that both Samuel and Tom would recover quickly and completely. The next couple of months will continue to be a bit intense as the doctor monitors Samuel closely for any signs of rejection.

Don't be shy about calling or emailing. It seems many people have been worried about "bothering" us and have hesitated to contact us. We are feeling a bit lonely! ☺ I'll email again after the transplant to let you all know how it went.

God is good!

Nancy

"Therefore, having been justified by faith, we have peace with God through our Lord Jesus Christ, through whom also we have access by faith into this grace in which we stand, and rejoice in hope of the glory of God. And not only that, but we also glory in tribulations, knowing that tribulation produces perseverance; and perseverance, character; and character, hope. Now hope does not disappoint, because the love of God has been poured out in our hearts by the Holy Spirit who was given to us." Romans 5:1- 5