

## **NIH Natural History Study Article**

When our daughter Leah was diagnosed in 2007 with Neurofibromatosis Type 2 we were completely overwhelmed. Any patient or parent of someone with NF2 has experienced that original shock and feeling of “WOW” this is serious. After educating ourselves on NF2 by reading, visiting doctors in our home town of Buffalo, NY, getting expert opinions from the NF Clinic at Children’s Hospital in Boston and attending a couple of NF symposiums we began to grasp the complexities of having and living with NF2. We came to the conclusion that finding a cure or course of treatment would be very difficult and it would take many different entities to accomplish this goal. These entities consist of scientists, doctors, the US Government (politics), pharmaceutical companies and of course money (fundraising). The often forgotten piece to this puzzle is the patient. If a cure is to be found it will take brave patients to enter clinical trials and possible inherent risks so that a drug or treatment can be explored. Due to our daughter’s age she was not a candidate for any of the drug studies like Lapatinib and Avastin. One day while looking at clinicaltrials.gov, a government website that lists all the current and ongoing clinical trials, we came across the NIH Natural History Study for NF2. We asked Leah how she felt about being part of a study and she thought it would be fun it gave us a sense of doing something, when at the time there was not a lot being done in NF2. We applied and were accepted into the study in June of 2008.

A natural history study tries to gain insight about a particular disease/diagnosis, to learn about it from the beginning in the hopes of appropriately steering further studies into effective treatments. The objective of the NIH sponsored NF natural history study is to “...gain clinical and molecular insight into the effects of this tumor suppressor syndrome on tumor development and progression and to identify factors linked to symptom evolution.” The scientists hope to learn how fast the tumors grow, if certain factors might affect their growth and also examine effects of tumors on patient’s activities of daily living. There are different specialists that each patient sees, some yearly some twice a year. Rovella Hyman schedules all the appointments based on who you are expected to see that visit. Research nurse specialists Hetty DeVroom and Rene Smith oversee that every appointment is completed, labs are drawn and paperwork signed (along with many behind the scenes activities). The study is headed up by Dr. Ashok Asthagiri who meets with you at the end of every visit. Visits are every 6 months, but more often if needed. Labs and MRI’s are done every visit, then oncologists, ophthalmologists, audiologists, speech therapists, ENT’s, neurologists, vestibular (and more if needed) testing are all completed if scheduled. Some of our visits have been 1-1 ½ days, while others are 4-5 days. All expenses are paid for as part of the study. One plane ticket for adults, two for a minor and an adult are paid for or reimbursement for mileage based on federal guidelines is provided if you drive. \$30 in meals per person per day is also provided for study participants, which while not covering everything, certainly helps. Hotels are also paid for, or in our case we stay on campus for free in the wonderful Children’s Inn, which is truly an oasis, a home-away-from-home. Every detail in the facility has been thought of and is beautifully carried out. Volunteers pour in and provide meals some nights, music, massages, and special surprises in your family’s mail boxes. You name it they think of it. There is also a Safra Family Lodge for

adults receiving medical care at NIH. If you fly shuttle busses pick you up at the airport and drive you to NIH. Once on campus the metro has a stop in the NIH campus so if time allows you can go into DC for a quick tour/visit.

One thing is clear when you are at NIH, your tax dollars are at work. You are surrounded by incredibly bright people from every nationality you can imagine. There is state-of-the-art equipment and personnel and top notch researchers, who in our case are genuinely interested in NF2 and want to figure it out. We have met wonderful people from every walk of life at both the Children's Inn and in the various waiting rooms. Some have NF2, some NF1, some have diseases we have never heard of. One thing we have also come to appreciate is no matter how bad we may think we have it, a visit to NIH puts that all in perspective, someone always has it worse. We are just so grateful there is a place like NIH where they are truly trying to find some answers. Again we cannot reiterate enough that we want to be part of the solution with NF2. While we do not want to expose our daughter to unnecessary risks, we need to have as good of an understanding as possible as lay people with the complexities of NF, so that when a trial comes up we can make an informed decision about participation.

If this sounds like something you may be interested in, it is easy to begin the process. For more information you can go to [clinicaltrials.gov](http://clinicaltrials.gov) and look up the NF2 study NCT00598351, or simply contact patient recruitment at 1-800-411-1222 or [prpl.mail.cc.nih.gov](mailto:prpl.mail.cc.nih.gov), or by TTY 1-866-411-1010. After answering some simple eligibility questions someone will contact you from there. After that you give them some dates to work with and your first visit will be scheduled. You have to be part of the solution if you want an answer to a problem, if you are affected by NF2, this is one possible solution.