Before signing on for a clinical trial, considerable thought before a consent form, indicating willingness to participate, is signed. Establishing an understanding about what the study involves may require several discussions and also an opportunity to ask questions about the study. This process where you learn about and consent process that provides details about the study and includes important information themselves or other family members, either now or in the future. They may have hope for a newer or better therapy, or even a cure for their disease. They may just want to contribute to the body of information about their health condition. How does a person decide whether a particular clinical trial is right for them? All clinical trials include a required informed consent process that provides details about the study and includes important information (often in the form of questions) to help you decide whether you want to participate. There is also an opportunity to ask questions about the study. This process where you learn about and establish an understanding about what the study involves may require several discussions and considerable thought before a consent form, indicating willingness to participate, is signed. This article highlights some of the questions that we suggest asking yourself and the researcher before signing on for a clinical trial:

1) Will my child benefit from the research? Will there be possible benefits for others with the same disease or condition?

2) Can my child be harmed by the research? It is important to consider potential risks of procedures and side effects of medication. How likely is it that they will occur? Is it

3) Will my child possibly get placebo (a “sugar pill”) instead of the medication?

4) Who is paying? Is the drug investigational and provided by the study sponsor? Are there required procedures and, if so, is the cost covered by the study or my insurance? Are there extra costs (i.e., gas, parking, childcare for other children) if I get hurt during the study, who pays for my medical care?

5) Why is the research being done? If a new drug is being tested, will I have the option of opting out of this investigational medication at the end of the study?

6) How much of my time will the study take? How long is the study?

7) What if I choose not to be in the study? What are the alternatives to being in the study?

8) What if I choose not to be in the study? What are the alternatives to being in the study? If it start the study and change my mind?

9) Who is in charge of my care while I am in the study? My regular doctor?

10) If I have questions during the study, who do I call? The coordinator? The investigator?

Patient participation in research trials is necessary for the advancement of science and the development of new therapies. Participation involves a real commitment on the part of the individual and their family. If a particular trial means you, it is important to participate actively in the informed consent process where you have ample time to develop a clear understanding about what the trial involves, review the consent form and have your questions answered to determine if the trial is right for you.

Enzyme replacement therapy had been suggested to me in 1991 by Dr. John Barranger who had been involved in the research and development of treatment for Gaucher disease. How fortunate I was to live only 50 miles from a major treatment center with an amazing doctor! There were some insurance issues to resolve and once that was complete, I started enzyme replacement in June 1992.

Bone damage had already been done and I found a right total knee replacement soon after. 1993. Post-op prayer seemed so slow, but that Spring I was able to attend my daughter’s First Holy Communion and her son’s high school and college graduations. Insurance again became an issue and I missed a few infusions before being assisted by the Charitable Access Program. Eventually I obtained an insurance policy with no lifetime max.

Looking back at a Clinical Trial; why did I participate? by Irene Marshall

“I want a normal mother!”. This is a statement from my 7 year old daughter 16 years ago, one that made me pause to get well and stay well, to be a pioneer. To a seven year old, I wasn’t normal. She could see me baring pain and using crutches for what must have seemed to her like an eternity. It’s kind of ironic that I was actually diagnosed with Type Gene therapy trial involved injections of GCFF and leukopheresis before receiving any cells that had been exposed to a retroviral vector carrying an uncharged copy of the Gaucher (glucocerebrosidase) gene. I understood that the vector could potentially activate an unwanted oncogene which could possibly cause cancer.

I received four gene transplants from July, 1995 to August, 1996. The plan was simple. If the corrected gene was not in my blood, my dose of enzyme would be reduced by half several times until I was eventually weaned from the drug. My infusions were discontinued in October, 1997. I felt great and had no health issues but in January, 2000, the corrected gene was no longer seen in my labs which meant counting enzyme therapy. Mine was the only trial out of four total patients in which the transplant was temporarily successful.

I was optimistic that as we learn more through present & future clinical trials, there will be new and improved medications and ultimately, a cure for Gaucher disease... Hopefully in my lifetime.
that the sound was delivered efficiently to the structures that process sound. Drum is intact, and if the ear drum is able to transfer sound. That is important for hearing ear. The audiologist can determine if the ear canal is plugged with wax or debris, if the ear Tympanometry determines if sound is being transmitted through the outer and middle order to be aware of a sound, the sound must be effectively transmitted to the inner ear. The described below are evaluating whether the anatomical structures are functioning normally. For young children, hearing is assessed by demonstrating to the child that a head turn in auditory information in children. Most people are familiar with routine hearing testing in which earphones are used and the individual diseases and with each individual. Auditory symptoms typically do not cause a patient with Fabry disease to seek medical treatment. Whether prospective diagnosis or newly discovered, audiologists can help individuals navigate the myriad issues that come with a diagnosis of Gaucher disease. Individuals requiring a Gaucher Mentor may be referred to the Program through Gaucher treatment centers, the NGF at 800-GAUCHER, the NGF website or call Genzyme Treatment Support Services (GTS) at 800-745-4447. Interested persons may also call or email the NGF at ngf@gaucherdisease.org with a Mentor request. Clinical Trials: Is Research for Me? Dawn Laney, MS, CGC, CCRC and Karen Grinzaid, MS, CGC, CCRC Emory Lysosomal Storage Disease Center Medical genetics, genetic counselors, and other health care professionals involved in the care of people with lysosomal storage diseases (LSD) are always looking for better hearing! or in the second decade of life. Most people with high frequency hearing loss function well, of hearing loss is common in aging individuals. But in Fabry disease, this loss occurs much more quickly. Fabry disease is not typically characterized by childhood hearing loss; however, hearing loss in adults with Fabry disease is characterized by a progressive high frequency sensorimotor hearing loss. High frequency hearing loss is characterized by the loss of hair cells that are responsible for high frequency sensitivity. The amount of hearing loss in common in aging individuals. But in Fabry disease, this loss occurs much more quickly. The hearing loss identified in Fabry disease is often identified in the late teens or in the second decade of life. Most people with high frequency hearing loss function well, with no difficulty, in quiet situations. All of the hearing loss examples described above may benefit from amplification or in the use of a cochlear implant. All of these cochlear implants are described below are evaluating whether the anatomical structures are functioning normally. A normally functioning system allows sound to move effectively through the system. In order to be aware of a sound, the sound must be effectively transmitted to the inner ear. Tympanometry Although part of the audiologic test battery, tympanometry is not a hearing test. Tympanometry determines if sound is being transmitted through the outer and middle ear. The audiologist can determine if the ear canal is plugged with wax or debris, if the ear drum is intact, and if the ear drum is able to transfer sound. That is important for hearing to occur, but does not determine if hearing has occurred. Normal tympanometry suggests that the sound was delivered efficiently to the structures that process sound