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Successful Relocation of Adult and Pediatric NF Clinics and a Discussion of Increased Breast Cancer Risk in Women with NF1

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Relocation of Adult and Pediatric NF Clinics

I'd like to provide an update of a development mentioned in a previous blog regarding the NF Clinic's relocation to two distinct sites in the UAB Medical Center District. The relocation, which has recently been completed, originated earlier this year with the reorganization of the NF Clinic into adult and pediatric clinics. The adult clinic is located in the Kirklin Clinic at UAB, while the pediatric clinic is at the downtown Children's Hospital of Alabama location. Both of these facilities provide our patients with more convenient parking than our previous NF Clinic location in the Hugh Kaul Human Genetics building. Also, the new locations will enable improved integration with the range of other medical specialties involved in the multidisciplinary care we provide while allowing our patients to remain in one physical location for blood draws, imaging, or consultations with other specialists. Although our previous clinic location allowed us to see adults and children in the same visit, we now see adults at the Kirklin Clinic location on Mondays and children at the Children's Hospital facility on Thursdays; at the Kirklin location, patients must be 16 or older, while patients in the children's clinic must be 18 or younger. Because we understand that these split clinic days could be an inconvenience for some patients, we can certainly arrange to see members of the same family on the same day if needed, by prior request on a case-by-case basis. Overall, we believe that our patients and families will benefit from the convenience and integration of care that our adult and pediatric clinics provide.

Increased Breast Cancer Risk in Women with NF1

Next, I want to review information concerning the increased risk of breast cancer in women with NF1. In recent years, it has become clear that women with NF1 are at an increased risk for breast cancer, with the risk being two to three time higher in women with NF1 than in those in the general population. Also, these cancers occur at a younger age and tend to be more aggressive in women with NF1 than those that occur in women in the general population. The nature and composition of the cancers, however, are not different.

In many women who have been diagnosed with breast cancer, a genetic panel of tests is performed to detect mutations that might be associated with the cancer. The NF1 gene is now being tested as part of this panel, as well as other genes including BRCA1 and BRCA2. However, it's important to note that the increased risk of breast cancer in women with NF1 is not associated with mutations in the BRCA1 or BRCA2 genes.

The reason for the increased risk of breast cancer in women with NF1 is not completely understood. We know that cancer is the result of the accumulation of genetic alterations that cause cells to behave abnormally. The NF1 gene has been shown to have mutated in many common cancers, which might indicate that the NF1 mutation puts an individual one step closer to developing other cancers.

Some women diagnosed with breast cancer have been referred to our clinic because of an unexpected NF1 mutation detected in the genetic testing panel. There are a few possible explanations for this finding, including that the individual has NF1 and was never diagnosed because the clinical features went unnoticed. Another possibility is that the individual has a mosaic form of NF1 that is detected in the blood but may not be clinically evident. Lastly, genetic variants are sometimes found in testing that are different from the normal gene variations. These are known as variants of unknown significance, and it can be a challenge to know what to do with this information. Often, when these patients are evaluated, they are not found to have NF1.

Breast Cancer Screening Recommendations

The increased risk of breast cancer in women with NF1 raises questions about screening recommendations. The National Comprehensive Cancer Network (NCCN), an organization that issues screening guidelines for various cancers, recommends that women with NF1 should be screened for breast cancer at an earlier age than the general population, beginning at age 30. In addition, the NCCN states that some consideration should be given to the use of breast MRI from age 30 to age 50. After this, the guidelines shift back to that of the general population. We are now recommending these screening standards to the patients we see in our clinic with the goal of achieving an early diagnosis for improved outcomes.

Some patients are concerned that neurofibromas in the breast may be confused with breast tumors during imaging. Although neurofibromas can develop in the skin of the breast, they are clinically distinguishable from tumors in breast tissue. However, it is important for radiologists to know the NF history when reading imaging results for these patients.