Genetics of Vitiligo Susceptibility

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Our goals for this research project are to expand our collection of vitiligo patient and family DNA samples, begin screening candidate susceptibility genes for association with vitiligo, and perform pilot studies of gene expression in melanocytes. We have made good progress on our first goal, with the help of many members of the National Vitiligo Foundation and their families. We have collected 414 samples in all, including 217 vitiligo patients and 197 unaffected family members. The patient group consists of 132 females and 85 males, with 159 Caucasians, 20 Hispanic, 10 African-Americans, 6 Indians, and 6 Asians. Within the next few months, we hope to increase the numver of samples and obtain 200 Caucasian patient samples, and as many as possible for all other groups. The average age of vitiligo onset is our patient group is 19 years old. As reported for other vitiligo patient groups, our group appears to have a higher incidence of thyroid disease and some other autoimmune conditions than the general population.

We have been collecting samples from affected and unaffected family members of vitiligo patients for family-based genetic association analyses. We have obtained samples from71 vitiligo patients without family members, 64 patients with only one or two first-degree relatives, and 37 patients with three or more relatives. It is the last group that is the most useful for family-based studies, and our emphasis is now to continue collecting family samples consisting of patient, both parents, and at least on sibling. We are also attempting to collect families with multiple affected members and/or multiple generations affected by vitiligo.

Our second goal was to begin case/control association studies of candidate vitiligo susceptibility genes. The time it has taken to collect enough numbers of samples to begin these studies was unexpectedly long. However, we have begun the testing of 21 candidate genes, including nine genes important in melanocytes and twelve genes involved in regulating the immune system. Based on our preliminary results, six of the genes tested demonstrate no significant association with vitiligo in the Caucasian patients tested so far. The results for four candidate genes are more promising, but more patients need to be tested. Genetic markers are also being identified for a number of additional candidate genes.

The third goal of these studies is to perform pilot studies of gene expression in melanocytes grown in culture from skin biopsies. We are currently establishing the culture conditions for growing human melanocytes, and we will begin comparing patterns of gene expression in melanocytes grown from the skin of vitiligo patients and their unaffected relatives later this year. The aim of these experiments is to identify genes that are expressed abnormally in patient melanocytes compared to control melanocytes using molecular biology techniques that do not rely on previous knowledge about melanocye function.

We are continuing to collect DNA samples from vitiligo patients and family members, especially where both parents and at least one sibling are available (regardless of whether they have vitiligo). Families with multiple affected members will be especially helpful. Please note that there will be no direct benefit to participants in this study. This is not a diagnostic test, and the research results will not be useful to you in deciding how to treat your vitligo. These studies should, however, help us to understand why some people develop vitiligo, and this may eventually benefit others at risk of developing vitiligo or may lead to finding better treatments.

For more information, please see the website at: www.ned.ufl.edu/path/faculty/mccormac'particip.html.