

## **PAF and Coriell Announce Collaborative Project to Further Research on PA**

The Propionic Acidemia Foundation and Coriell Institute for Medical Research in Camden, NJ have initiated a collaboration to provide the NIGMS Human Genetic Cell Repository with samples from affected individuals and their families a thorough set of clinical, molecular and laboratory data. An extensive questionnaire has been developed with the expertise of the Medical Advisory Board of the PAF, which will be administered for each submission.

The Coriell Institute for Medical Research in Camden, NJ has pioneered Biorepositories for the study of human diseases. Most of the repositories at Coriell are sponsored by agencies of the NIH. The samples are used by scientists for gene discovery, functional assays, reference materials in assay development and genomics. The goal of providing the samples is to improve the understanding of human genetics and human diseases.

The Human Genetic Cell Repository, which is sponsored by the National Institute of General Medical Sciences (NIGMS), has cell lines and DNA samples from more than 500 inherited diseases. These cell lines have been an important resource for the identification of genes in which a mutation or chromosomal defect leads to the disorder. Yet, this is often only the first step. For some inherited disorders, it has been possible to perform a “genotype-phenotype” analysis, connecting the patient outcome with the type of symptoms displayed by the patient. For others this connection is not obvious. One possibility is that the outcome of the disease is due to variants in other genes. In order to investigate these possibilities it is necessary to have excellent longitudinal clinical information in addition to the laboratory analyses. To build these resources we have begun establishing interactions with groups which interact directly with the patients on a continuing basis.

As noted above, it is critically important not only to find the gene which is the primary cause of the disorder, but also to understand the genetic and other factors which could lead to an understanding of the way the disease unfolds. To achieve this Coriell and the PAF have developed a protocol whereby subjects who submit samples to the Repository for the preparation of a cell line, will also be recontacted in the future so that the course of the disease can be known to the scientists who obtain the cell lines or DNA from the Human Genetic Cell Repository. To protect the identity of the subjects and their families, Coriell developed a protocol, approved by the Institutional Review Board, where the Propionic Acidemia Foundation would be the only ones who would be able to connect the identity of the subject with the sample in the Repository. The PAF would also undertake to follow-up on the patients every few years to create a database on the status of the patients in addition to information about cells derived from the patients. Our goal is to create a new type of resource for the study of rare diseases. We have already started processing the first samples and hope to have them available to the scientific community in the spring of 2008.

In order to make this a successful endeavor in bringing about more knowledge about PA we need as many people as possible to contact PAF to request a sample kit, which will include a questionnaire. Our hope is that each affected individual and their immediate family members become a part of this pursuit. Please contact PAF at 877-720-2192 or [paf@pafoundation.com](mailto:paf@pafoundation.com) for questions or to request a testing kit.