# CLINICAL TRIAL REVIEW

Clinical Trial Review is a JDD department designed to provide physicians with information on drugs and devices undergoing clinical testing. It is our goal to inform the reader of the status of select drug and device studies relevant to the practice of dermatology before this information is available through standard channels. To participate in or learn more about these and additional trials, visit www.clinicaltrials.gov.

## SKIN CANCER

### Cancer Risk in Carriers of the Gene for Xeroderma Pigmentosum

This study will determine if family members of patients with xeroderma pigmentosum (XP) have various abnormalities including: skin abnormalities; nervous system abnormalities, such as hearing problems; skin, eye, or internal cancers; or other changes. XP is a rare inherited disease that involves an inability to repair damage to cell DNA (genetic material). It can affect several organ systems, including the skin, eye, nervous system, and bones. Patients have a more than thousand-fold increase in frequency in all major skin cancers.

Parents of patients with XP are carriers of the abnormal XP gene. Other family members may also be carriers of the abnormal XP gene. Carriers do not develop the disease themselves; symptoms develop only in children who have inherited the faulty gene from both parents. This study will try to clarify the genetic basis for XP and to understand the increased frequency of cancer in the disease.

Both XP patients who have been evaluated at the National Institutes of Health Clinical Center and their relatives are eligible for this study. Newly diagnosed XP patients are also eligible. Spouses of relatives will also be included as control subjects.

| Condition | Study Type |
|---|---|
| **Xeroderma pigmentosum; melanoma; squamous cell carcinoma; basal cell carcinoma; skin cancer.** | Observational |
| **Sponsor: National Cancer Institute**<br>**Study ID Numbers:** 020313 02-C-0313<br>**ClinicalTrials.gov Identifier:** NCT00046189 | |

## VITILIGO

### Open-label Pilot Study of Abatacept for the Treatment of Vitiligo

Vitiligo is a chronic autoimmune disease with evidence of CTLA-4 involvement. This pilot study of the treatment of new onset or actively progressing vitiligo with abatacept is to determine if weekly self-injections of medication lead to clinical improvement in vitiligo lesions.

Abatacept has been shown to decrease T-cell activity and reduce symptoms associated with rheumatoid arthritis. Similar pathways have been shown to be involved in vitiligo. Therefore, 10 adult patients with active vitiligo who meet specific inclusion and exclusion criteria will be recruited to receive self-administered injections of abatacept weekly, starting at week 0 and continuing until week 24. A week 32 follow-up visit will also be performed to evaluate secondary endpoints. Patients will be monitored to see if skin lesions of vitiligo stop spreading and start to repigment with continued treatment.

| Condition | Drug |
|---|---|
| **Vitiligo** | Abatacept |
| **Sponsor: Brigham and Women's Hospital**<br>**Collaborator:** Bristol-Myers Squibb<br>**Study ID Numbers:** 2014P000699<br>**ClinicalTrials.gov Identifier:** NCT02281058 | |

## PROTOPORPHYRIAS

### Erythropoietic Protoporphyrias: Studies of the Natural History, Genotype-Phenotype Correlations, and Psychosocial Impact

The initial objective of this protocol is to assemble a well-documented group of patients with confirmed diagnoses of the erythropoietic protoporphyrias for clinical, biochemical, and genetic studies. The long-term objectives are (1) to conduct a longitudinal investigation of the natural history, complications, and therapeutic outcomes in people with erythropoietic protoporphyria; (2) to systematically investigate the psychological effects of the erythropoietic protoporphyrias on children and adults; and (3) to investigate the correlation between the identified genotypes and the resulting clinical presentation, also determining the possible interaction of other genetic markers.

The porphyrias are a group of rare metabolic diseases that may present in childhood or adult life and are due to deficiencies of enzymes in the heme biosynthetic pathway. The most common manifestations are related to accumulation of intermediates in the pathway and usually occur as acute neurological attacks (as in the acute or hepatic porphyrias), or cutaneous photosensitivity (as in the cutaneous porphyrias, including the erythropoietic protoporphyrias).

The risk of disability or death from these disorders is significant, in part because diagnosis is often delayed due to lack of

adoption of diagnostic testing in clinical practice. Moreover, the natural history of these disorders is not well described and it is not known what determines differences in outcomes. New therapies are needed. For existing therapies, high-quality evidence on short- and long-term efficacy and safety is generally lacking. Therefore, the purpose of this study is to provide a better understanding of the natural history of these disorders, as affected by available therapies, and to aid in developing new forms of treatment.

| Condition | Study Type |
|---|---|
| **Erythropoietic protoporphyria** | Observational |
| **Sponsor: Icahn School of Medicine at Mount Sinai**<br>**Collaborators:** Rare Diseases Clinical Research Network; Office of Rare Diseases; National Institute of Diabetes and Digestive and Kidney Diseases<br>**Study ID Numbers:** GCO 08-0959-04; HSM12-00307; U54DK083909<br>**ClinicalTrials.gov Identifier:** NCT01688895 | |