

ARC Newsletter



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Dear Friends and Supporters of ARC,

The last year has been an extraordinarily busy one on many fronts, which distracted us from putting together our regular newsletters. We have been hard at work on our existing studies, and also initiating a number of new research projects, some of which are briefly described below. The pace of scientific progress in the field of ALS has been steadily increasing. We feel privileged to be contributing to this effort and wish to thank you for your ongoing contribution to the research enterprise. Only through research is there hope for a future without ALS.

With best wishes, Michael Benatar

Interested in ALS Clinical Research?

Visit our website and use our newly added interactive map to locate ALS clinical research studies currently taking place in the United States. The map shows the population density (by county) as well as the location of and details for ongoing studies. Follow the instructions on the website to tailor your search to your interest.

Website: www.als-research.org

Research Updates

Pre-fALS

As many of you know, we have long believed in the importance of studying pre-symptomatic disease in ALS. The essential idea is that the disease process begins well in advance of the appearance of clinical symptoms (e.g. weakness) and, by the time symptoms do appear, the disease process is already quite far advanced. This is problematic because treatments are expected to be less useful if started later in the course of disease. Our long-term goal has always been a disease-prevention (or very early stage) treatment study. In order to develop such a study, we need a better understanding of the earliest manifestations of disease – which was the motivation for initiating **Pre-fALS**, in which we study people who may be at risk for ALS (but who are not yet affected). Since the only people known to be at risk for ALS are relatives of those with familial ALS, we have focused our attention on familial ALS in which the underlying genetic cause of disease in the family can be determined. The **Pre-fALS** study continues to gain momentum. So far, 226 people have been consented and screened for participation, and 85 enrolled in longitudinal follow-up. We appreciate the continued participation of so many of you who have been a part of this study since its early days, and welcome the participation of many new family members. To learn more about **Pre-fALS**, please contact us using the email or phone number on the next page.

Pre-Symptomatic Studies in ALS (PRESS-ALS) – NEW STUDY

More recently, we have developed a *novel* approach to studying pre-symptomatic disease not only in pre-symptomatic individuals, but also in people who have been diagnosed with ALS but are still relatively early in the course of disease (Benatar et al., *Muscle Nerve*, 47(5):629-31, 2013). The idea is elegant in its simplicity, and it builds upon the recognition that the disease process in ALS is typically focal when it begins. As many of you know, the first sign of disease may be weakness in an arm or a leg (so-called “limb” onset), or difficulty talking and swallowing (so-called “bulbar” onset). When weakness first appears for example in the right arm, other body regions (e.g. left arm, both legs, the bulbar region) may still be clinically unaffected. But since we know that, as the disease progresses, these initially unaffected regions will become affected, these regions are essentially “pre-symptomatic” at this early point in the course of disease. This realization offers us an opportunity to broaden our study of pre-symptomatic disease to also include people who are affected with (familial or sporadic) ALS, and it serves as the basis for the “**Pre-Symptomatic Studies in ALS**” (**PRESS-ALS**) study that we launched about a year ago. For the **PRESS-ALS** study, we are looking to recruit ALS patients, as well as healthy individuals who may serve as controls. **All PRESS-ALS participants will be recruited locally, from the South Florida Area.** To learn more about the **PRESS-ALS** study, please contact our research team using the email or phone number on the next page.

Clinical Trial of Arimoclomol in SOD1+ Familial ALS

Our clinical trial of **arimoclomol** in people with SOD1+ familial ALS is wrapping up. This study is closed to enrollment and the last few patients who were on study drug have just completed their last follow-up. We will be busy analyzing the data in the coming months and look forward to sharing the results in the early part of 2015.



Happy Holidays



Family Pedigrees

We have 521 families enrolled in our research program so far. Maintaining up-to-date family pedigree information is crucial for the work we do. Please continue to help us update our records by contacting us if any members of your family receive a diagnosis of ALS (or a related neurological disease), or if a loved one passes away. Additionally, please contact us if you obtain access to medical records of family members affected with ALS (or a related disorder).

CONTACT INFORMATION

Pre-fALS & pedigree updates : fals@med.miami.edu

PRESS-ALS: alsresearch@med.miami.edu

CReATe: www.rarediseasesnetwork/CReATe

Phone: 1-888-413-9315

Visit our website: www.als-research.org

New Additions to the Research Team



Danielle Dauphin

Danielle has a B.A. in psychology. She has previous clinical research experience with large multisite pediatric cardiomyopathy and HIV studies and also worked for the National Cancer Institute's Cancer Information Service. Danielle joins our team as a study coordinator for the *Pre-fALS* study.



Corinne Barbato

Corinne has a B.S. in Health Science. She worked in clinical research at the Joslin Diabetes Center in Boston, where she was involved in trials looking at pharmacogenetics of anti-diabetic medications, and inflammation in diabetes and cardiovascular disease. She joins our team as a Research Assistant.

Newsroom

Clinical Research in ALS and Related Disorders for Therapeutic Development (*CReATe*)

Dr. Michael Benatar was awarded a large NIH grant to establish a Rare Diseases Clinical Research Consortium called "**Clinical Research in ALS and Related Disorders for Therapeutic Development**" (*CReATe*). This multi-center consortium includes collaborators at the University of Miami*, St. Jude Children's Hospital in Memphis, Mayo Clinic in Jacksonville*, University of Kansas Medical Center*, University of California San Diego*, University of California San Francisco*, Duke University, the Medical University of South Carolina, and the University of Tübingen in Germany*. (Asterisks denote the 6 recruitment sites.)

As we have showed in our *IBMPFD-ALS* study (Benatar et al, *Neurology*, 80(20):1874-80, 2013), there is great value in studying ALS and related disorders together because, due to the overlap of the underlying biology and/or clinical manifestations between these diseases, what we learn from one can help our understanding of another. During the five-year grant period, the *CReATe* consortium will carefully evaluate the relationship between phenotype and genotype in ALS and a group of related disorders (e.g. PLS, HSP, FTD, ...); search for novel genetic causes of disease; identify genetic modifiers; and advance the development of biomarkers that may aid therapy development. In addition, as part of the consortium's mission, we will train and mentor fellows and junior faculty in clinical research related to ALS and related disorders. Although *CReATe* will not be conducting any clinical trials at this time, all aspects of the consortium's activities are devoted to filling some of our knowledge gap about these diseases for the advancement of therapy development and future clinical trials.

Dr. Benatar is "particularly excited about the translational nature of this consortium, in which neurologists, geneticists and other scientists have joined forces to advance scientific progress toward effective therapies for this devastating group of diseases." In addition, a special and important aspect of *CReATe* is its partnership with a number of organizations that represent patients afflicted with this group of rare diseases: The ALS Association, the Muscular Dystrophy Association, the ALS Recovery Fund, the Spastic Paraplegia Foundation, the Association for Frontotemporal Degeneration, the National ALS Registry, and PatientsLikeMe. As Dr. Benatar said, "Clinical and translational research is motivated by the needs of patients and can only succeed if predicated upon genuine partnerships with patients and patient advocacy groups. Herein lies a unique underpinning of the *CReATe* consortium."

For more information about the *CReATe* consortium, please visit www.rarediseasesnetwork.org/CReATe or our website (als-research.org). An entry about this study will soon also appear on clinicaltrials.gov.



Happy Holidays

