ALS Research Collaboration Newsletter

@ The University of Miami

May 2013
Volume 4, Issue 1
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RESEARCH UPDATE

Dear Friends,

Please accept our apologies that it has been so long since our last newsletter. We have been distracted by an extraordinarily busy year – engaging in some very exciting scientific advances and also writing grants to secure funding to maintain our ALS research program. It seems fitting, therefore, to share with you some of our progress on each of these fronts.

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Building on our 2010 discovery of mutations in the VCP gene as a cause of ALS, we have recently completed a study investigating the possible relationship between ALS and IBMPFD, a degenerative disorder that affects the brain, muscle, and bone. This study was prompted by the observation that, though mutations in the VCP gene had long been associated with IBMPFD, there was until our paper little recognition of the potential overlap between IBMPFD and ALS. Indeed, we found that, upon closer examination, patients with IBMPFD often developed evidence of motor neuron degeneration (including ALS) as well, but this additional component of the disease had previously been overlooked. The link between IBMPFD and ALS suggests that, by studying the biology of IBMPFD, we may improve our understanding of ALS, and vice versa. These insights also provide new avenues that we might pursue for therapy development.

In a related study, led by our collaborator, Dr. J. Paul Taylor at St. Jude Children's Research Hospital, we have identified mutations in two genes (hnRNPA2B1 and hnRNPA1), in addition to VCP, which may cause both IBMPFD and ALS. Further investigations by Dr. Taylor and colleagues showed that aberrant protein processing and the formation of abnormal protein aggregates are mechanisms central to the degenerative process in these IBMPFD-ALS families. These observations, combined with the multisystem nature of the disease (affecting muscle, brain, bone, and motor nerves) have led us to propose that the new term "multisystem proteinopathy" (or MSP) be used, instead of IBMPFD, to describe this illness.

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Many of you may know that the Department of Defense (DOD) oversees a congressionally directed research program in ALS that aims to improve treatment and to find a cure for ALS. Last year we submitted an application to DOD in collaboration with colleagues at the University of Miami who are experts in drug development. We are very excited to report that this grant has been recommended for funding and we hope to start work in the fall.

In this project we will employ a laboratory approach known as a "semi-high-throughput screen" to rapidly evaluate a large number of potential therapeutic compounds within a relatively short period of time. We are testing both existing compounds (that might quickly be brought to clinical trials) as well as a set of entirely novel compounds. Our goal is to identify, in laboratory experiments, promising treatments for patients with ALS and/or frontotemporal dementia caused by mutation in the C9ORF72 gene. This work complements our ongoing efforts to identify effective therapies for patients with familial ALS due to mutations in other genes (e.g. our trial of arimoclomol in SOD1 ALS), and we will update you on our progress in future newsletters.

Thank you, as always, for your interest and participation in our research program.

The ARC Team

Vital Statistics:

- 190 subjects enrolled in Pre-fALS, with 70 participating in the longitudinal cohort
- 34 subjects enrolled in Arimoclomol. We expect interim analysis to occur within the next 6 months.
- 465 families in our database of families affected by fALS

Contact us at:
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Visit us at:
als-research.org

Remember to join fALSConnect to register your interest in being contacted about new fALS research opportunities.

www.falsconnect.org

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New Additions to the Research Team

Sumaira Hussain, BSc, CCRP
Sumaira has a BSc degree in Neuroscience and Psychology. She was previously involved in pediatric neurotrauma research in the ICU, and joined the ARC team in Nov 2012 as a Clinical Research Coordinator for the Pre-fALS study. She has recently received her CCRP designation from the Society of Clinical Research Associates.

Sara-Claude Michon, PhD candidate
Sara is a PhD candidate in Neuroscience, soon to graduate after presenting her doctoral thesis. After several years of conducting pre-clinical (i.e. laboratory) research, her interest in clinical research on neurodegenerative disorders led her to join the ARC in Jan 2013 as a Clinical Research Coordinator. She is involved in the coordination of various ALS research studies, and will aid in the Pre-fALS study as well.

(Left to right: *Sumaira Hussain, Sara-Claude Michon, Anita Blenke*)

Anita Blenke, PA-C, MS, CCRC
Anita is a Physician Assistant with an MS degree in Clinical Medical Science & Physician Assistant and a CCRC designation from the Association of Clinical Research Professionals. She has worked in the Department of Neurology since 2001, and has coordinated over 60 research studies in the areas of, among others, Parkinson's disease, Huntington's disease, and dystonia. Since joining the ARC team in Jan 2013, she has divided her time between ALS research, notably the Arimoclomol trial, and clinical work in the University of Miami ALS Clinic.

A warm farewell…

Natasha Garcia, BA
Clinical Research Coordinator
Arimoclomol Clinical Trial
& fALS research

Greetings,

I would like to thank each and every person I have had the pleasure of working with through the ALS Research Collaboration. I will be leaving the project at the end of this month to pursue a doctoral degree in neuropsychology. I have learned so much through working with Dr. Benatar and all of you; this experience has shaped my future career path. I plan to acquire more expertise so I may continue working in the ALS field and contribute to the knowledge of this disease. Namely, I would like to evaluate cognition and quality of life in patients with ALS. Additionally, I have also been inspired to seek to better understand stress and quality of life in caregivers, through my many interactions with individuals providing care to those with ALS. I am very proud to be a part of Dr. Benatar's research program and I am very excited about the research we have been conducting. During the time I have been involved in ALS research the knowledge of the disease has grown immensely, although we still have much to learn. Thank you all for your participation and advocacy in aiding ALS research. Please note that Anita Blenke will be taking over the Arimoclomol trial. Anita has 12 years of experience in neurology and she is a great asset to the team!

Warm regards, Natasha Garcia