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SAMPLE CV NARRATIVE
INSTRUCTOR
AoE Inv

I am an investigator in genetics with a long-standing interest in unraveling the molecular mechanisms that underlie hereditary disorders. In particular, I focus on understanding the regulation of mRNA splicing and how its disruption leads to human disease, with the ultimate goal of developing new therapeutic approaches.

My interest in splicing disorders was established during my doctoral studies when I was responsible for a project that aimed at characterizing the pathogenic mechanism underlying facioscapulohumeral muscular dystrophy (FSHD). I characterized the FSHD region gene 1 (FRG1) protein using a disease specific mouse model and unraveled the role of FRG1 in RNA splicing. This work was carried out in the laboratory of Dr. Rossella Tupler at the University of Modena and as a visiting graduate student in the laboratory of Dr. Michael Green at the University of Massachusetts Medical School. I conducted splicing analysis studies in muscles from FSHD mice and patients to demonstrate that FRG1 has direct role in the aberrant splicing of muscle specific transcripts. This work was carried out in the laboratories of Dr. Tupler at the University of Modena and of Dr. Michael Green at the University of Massachusetts Medical School, where I was a visiting graduate student.

My enthusiasm for studying splicing disorders encouraged me to continue my research in this field and to join, at the end of 2012, the laboratory of Dr. Susan Slaugenhaupt in the Center for Genomic Medicine (CGM) at Massachusetts General Hospital (MGH) and Harvard Medical School (HMS). Since joining MGH, I have been working on familial dysautonomia (FD), a genetic sensory and autonomic neuropathy caused by an mRNA splicing defect in the Elongator Complex Protein 1 (ELP1) gene. During my postdoctoral fellowship, I generated the first mouse model for FD that can be used to evaluate the in vivo efficacy of splicing modulation. This new mouse is truly a breakthrough in the field of FD and allowed me to initiate a preclinical trial of the splicing modifier, kinetin, to fully evaluate the benefit of modulating ELP1 splicing on disease phenotype and to set the stage for clinical trials in FD patients. I was able to show that kinetin leads to significantly improved ELP1 splicing and has beneficial effects on the disease phenotype.

In 2017, I was appointed Instructor in the Department of Neurology at HMS and became a junior faculty member at the CGM. I have been working with my mentor Dr. Slaugenhaupt to establish my independent research program on developing treatments targeting splicing alterations across a variety of neurologic disorders. To date, I have presented my research findings at several international meetings including the Bermuda Principles 4th Annual Meeting and the Annual Rotary Foundation meeting as well as at institutional research seminars, retreats, and poster sessions. I have been successful in obtaining funding for this work: I am co-investigator in a R01 that explores the therapeutic potential of a new targeted approach to modulate splicing using modified U1 snRNA molecules and am Co-PI in a R21 grant aimed at mitigating retinal degeneration in FD using a combinatorial approach based on gene

therapy and RNA splicing modulation. Our collaborator at Montana State University, Dr. Frances Lefcort, is the lead PI in this grant. Lastly, I am Co-PI with my mentor Dr. Slaugenhaupt in the HMS Freeman Award. The goal of this proposal is to evaluate the efficacy of our new splicing modulator therapy in restoring sensory neuron development.

I supervise and mentor my team of three research technicians and a post-doctoral fellow, who was awarded a Meritorious Abstract Travel Award from the American Society of Gene & Cell Therapy (ASGCT). My mentees have all produced scholarship under my direction. I have presented locally, nationally, and internationally.

Since being appointed Instructor, I have gained expertise in the generation and characterization of animal models, transcriptome sequencing analysis and interpretation, the identification of novel approaches to modulate splicing, manuscript preparation, and the scientific presentation of data. I have also learned how to manage international collaborations. My goal as investigator is to develop better therapeutic approaches for a group of incurable neurodegenerative disorders that share a similar pathogenic mechanism.