

Scholarship Report

My name is Laura Gonzalez Trueba and I am a Spanish first year PhD student at the School of Cardiovascular & Metabolic Health at the University of Glasgow. I was awarded the David Fleming-Brown Travel Scholarship (£400) so I could go on a two-month research trip to the Centre for Genomic Medicine at Massachusetts General Hospital (MGH), Boston. For this visit to take place and for myself to be able to afford this experience I had to apply to multiple grants. Being awarded this grant made it possible for me to go to Boston, learn first-hand new techniques that I will apply in my PhD studies and take part in this amazing opportunity.



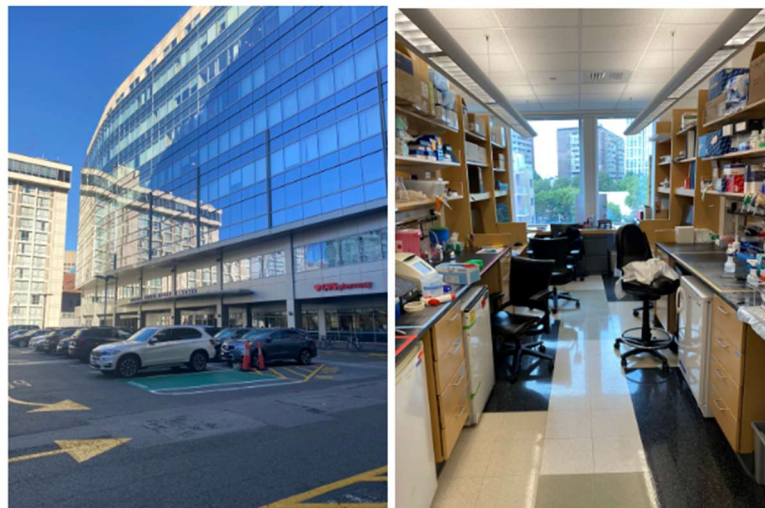
Me and the John Harvard Statue at the Harvard main campus. Rubbing his left foot is meant to bring you good luck. And a picture of the campus.

My PhD thesis is entitled: *Gene therapy approaches for collagen IV related vascular disease*. My project focuses on mutations in the genes COL4A1 and COL4A2 which cause COL4A1 syndrome. This is a hereditary disorder that encompasses a wide range of symptoms that vary from haemorrhagic stroke, kidney, eye, and muscle defects. Gene therapy to correct the mutations in the genes of the patients that have these diseases hold the promise of a cure negating the need of lifelong treatments. I focus on applying base editing using modified CRISPR-Cas9 technology as a gene therapy approach. This technology requires the use of enzymes called “base editors” most of which have been developed at the Center for Genomic Medicine by Dr. Kleinstiver. My

lab at the University of Glasgow collaborates with Drs. Musolino and Kleinstiver at the Center for Genomic Medicine. Going for a two-month stay allowed me to learn first-hand how these enzymes work are generated and the way the therapeutic approach is designed. Furthermore, it helped me establish connections that will be able to help me as I develop these approaches now that I am back in Glasgow.

Details of My Visit

My visit started the 4th of August when I travelled from Glasgow to Logan Airport in Boston, the next day I was already in the lab and I worked there for two months until the 30th of September when I flew to Madrid to visit my family for a few days before heading back to Glasgow. The aim of my visit was for me to learn the base editing technique from the people that developed the enzyme that I was using as well as learn assays that they carried out in their labs that I didn't do in Glasgow that I could learn and bring back. In summary I was there to learn as much as I could from everyone I met, an amazing opportunity.



Simches Research Center where the Centre for Genomic Medicine is based and the bench where I worked

I was based in Dr Musolino's lab where I was able to work with fibroblasts derived from patients that she treats in the hospital that have the condition that I study. I characterized these cells and I tried to cure them using a treatment that they had found to work in another cerebrovascular condition that they focused on. While in Dr

Musolino's lab I attended weekly meetings with their collaborators within Harvard University, this showed me first-hand the process to get a therapy to the clinic as they were patenting a gene therapy and allowed me to network with people from other departments. As Dr Musolino is a clinician and works in the paediatric Stroke and Cerebrovascular Service in MGH I was able to shadow her in the clinic and meet the patients that we are trying to cure. This was an amazing experience that gave me a clearer view of what I am trying to do with my research and the importance of it, I am really grateful that I could take part in it. I also treated the cells that I work with at Glasgow that have a mutation in COL4A1 with the base editor therapy and learned how the therapy approach is designed for the beginning.

While I was there, I tried to do as much as I could throughout the afternoons and weekends and to explore the surroundings. I went camping while the weather was good, I also went to New York for Labor Day weekend and explored all that Boston can offer.



From left to right. Salisbury Beach State Reservation. World Trade Centre, New York. Massachusetts State House, Boston. Boston skyline.

Impact of the Travel Scholarship

My research visit to the Centre for Genomic Medicine in MGH has been an incredible experience. It is amazing to have the opportunity to take part in this experience in the first place, but it wouldn't be possible for me to go if it wasn't for the money awarded to me by this travel scholarship.

This experience has allowed me to generate data for my thesis and advance on the genetic therapy for my research. I was able to characterise for the first time cells

from these patients resulting in novel data that can help advance in the research of COL4A1 syndrome.

As I have mentioned this experience has helped me see the importance of my research through a new perspective by seeing first-hand how patient's lives depend on it. Overall, I come back from this experience more motivated in my research. I am truly grateful for this experience and the funding bodies that have made it possible.