A Pharmacist’s Journey into Genomic Medicine

by Cathelijne van der Wouden
Pharmacists-in-training don’t usually get involved in genomics research. As graduate students in a PharmD program at Utrecht University in The Netherlands, my peers and I were on the way to becoming pharmacists. Our program only covered basic genomics, since — at least traditionally — genomics is not considered essential knowledge for pharmacists. But I was intrigued to learn why certain patients respond differently to the same drug regimen, and the most fundamental way to describe human variation is through genomics.

Driven by this curiosity, I reached out to the Genomes2People Research Program (G2P) in the U.S. to inquire about working with them as a trainee. Although my background as a pharmacist in training was unusual for a researcher in the field of genomic medicine, I felt that the most interesting discoveries are done at the intersection of multidisciplinary fields, a notion deeply embedded in G2P.

At the time, the G2P team was, among other projects, working to analyze the PGen Project dataset, recruiting patients in the MedSeq Project, and preparing the launch of the BabySeq Project. I was ecstatic to learn I would be given the opportunity to directly contribute to the field by working on a PGen Project research paper, and to learn more about research design and logistics by
observing meetings of the MedSeq and BabySeq Project teams. As a result, I had the extraordinary opportunity to work with G2P from September 2014 through March 2015.

An early eye-opener for me was the extent to which the finish line of the trial has to be anticipated in great detail at the beginning. By shadowing BabySeq management team meetings I became familiar with the complexities of clinical trial design, management, logistics, and ethical dilemmas from the first stages of complex study design and launch. In a more hands-on project, I was allowed to work on a research paper using the PGen dataset and later drafted the results into a first-author research paper that was later published in *Annals of Internal Medicine*.

After returning to the Netherlands, finalizing my PharmD, and starting a PhD, I began working for the coordinating center of the Ubiquitous Pharmacogenomics Consortium, a large-scale European consortium aiming to implement pharmacogenomics (PGx) across Europe. An integral part of the consortium is the PREPARE Study, a randomized controlled trial recruiting in seven European countries, which seeks to quantify the collective clinical utility of a PGx panel to personalize pharmacotherapy. Here, patients in the intervention arm receive their PGx panel testing results before initiating a new drug. Their pharmacist or physician can use these results by to tailor the prescribed drug choice and drug dose to the patient’s genetic profile to reduce the risk of
adverse drug events. The PGx panel results can also be used to tailor more than 40 drugs that patients may use during their lifetime. To enable access across healthcare systems, patients receive their PGx panel results on a card. This trial is unique in its multi-gene, multi-drug, multi-healthcare center approach.

It was my task to coordinate and contribute to the design, launch, management and analysis of the PREPARE Study. From my experience in the BabySeq meetings, I was prepared for tough discussions on ethical dilemmas and knew the importance of a clear common objective among all stakeholders. I also learned that it is imperative to begin with a clear, collective vision of the final manuscript’s format, so that members of the consortium could work towards that in great detail during the preparation phase. Most importantly, though, I had learned to keep the patient journey in the foreground of our focus at all times.

Now the PREPARE Study is nearing a successful conclusion. Enrollment was completed in July 2020 and we aim to report the findings by the end of 2021. During this process, thousands of patients across Europe have received their PGx results to enable physicians and pharmacists to tailor their drug regimen to their genetic profile. I credit my early career experiences with G2P, as a pharmacy student entering the world of genomics, with guiding me through the process — and with supporting my objective to improve human health and patient care throughout my lifetime.
Cathelijne van der Wouden, PharmD, holds a Ph.D. from Leiden University Medical Center in The Netherlands. She is working with the European Horizon 2020 funded Ubiquitous Pharmacogenomics Consortium (U-PGx) as the coordinating researcher for the PREPARE Study, a randomized controlled trial aiming to implement PGx guided prescribing across seven European countries by quantifying the collective clinical utility of a PGx panel to personalize pharmacotherapy.

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Genomes2People (G2P) is a program of Brigham and Women’s Hospital, the Broad Institute and Harvard Medical School. Visit genomes2people.org for more and follow us on Twitter @Genomes2People.