For Risks and Side Effects Consult Your Smartphone

Developing a User-Validated, Comprehensible Application for the Personalized Communication of Pharmacogenomic Information

Even in modern medicine, finding the right drug for a patient's personalized treatment sometimes follows a trial-and-error process: some medications work better than others or can cause serious side-effects. The decision of which drug to choose often depends on a physician's experiences and personal preferences. Simultaneously, sinking costs of genomic testing make genomic data available more broadly. This improves researchers' understanding on how the DNA influences treatment and disease, including on how the DNA affects a person's response to drugs, called pharmacogenomics. In this project, we want to bring findings from pharmacogenomic research directly to the user, as for example shown in Figure 1.

Figure 1. *Mock-ups of how a user-facing pharmacogenomics app could look like.*

Project

The aim of this project with the life science company Illumina is to follow a user-centric approach to develop a mobile application that helps patients to choose the drugs that work best for them based on their genome without going through cumbersome self-tests.

Together with Illumina, you will touch upon different aspects, building up on initial work that was already done in our research group, focusing on interoperability of genomic data, a pharmacogenomics pipeline and comprehensible user interfaces:

- Develop a **mobile application** that can receive and visualize users' genomic data by connecting to established, global data sources, as Illumina's global, connected analytics platform (ICA), Mount Sinai's BioMe biobank, and others
- Adapt and improve the present **pharmacogenomics processing pipeline**, aiming to integrate it with the Illumina Connected Analytics platform, and evaluate it on thousands of real-world samples
- Develop different **user interfaces using user-centered design** principles to convey pharmacogenomic information in a comprehensible way and develop an approach to **digitally assess users' comprehension** of the interfaces to enable validation and content personalization

To participate in the project you should be interested in working in a multidisciplinary team and learning about genomic medicine and user-centered design. The present project builds upon a Python and Docker backend, FHIR specifications for data interoperability, and a React Native and Expo cross-platform app; but we are open to broadening the spectrum of technology involved.

Are you interested in designing user-centered apps to empower patients? Then join us for our project – *For Risks and Side Effects Consult Your Smartphone*!

Pharmacogenomics

An example for a pharmacogenomic application are nonsteroidal anti-inflammatory drugs (NSAIDs), such as ibuprofen [1]. NSAIDs often are easy to buy over the counter without a prescription. Figure 2 depicts how ibuprofen is broken down – metabolized – into non-active components by the liver enzyme CYP2C9. However, if the enzyme has a decreased or no function due to certain genomic variants, it cannot metabolize ibuprofen as expected. This poor metabolization rate leads to a higher level of ibuprofen in the body over a longer time, which is associated with severe gastrointestinal, renal, and cardiovascular adverse events.
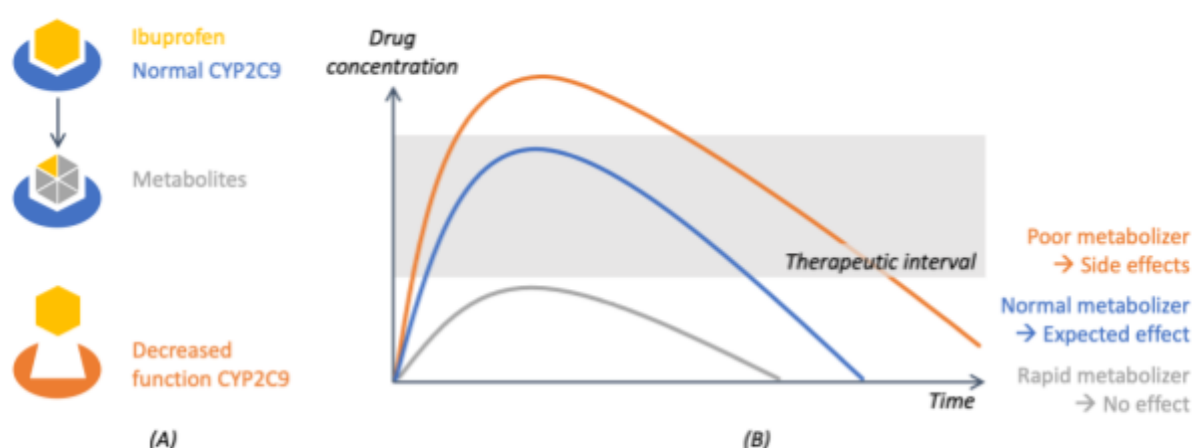

Figure 2. Metabolism of ibuprofen (adapted from [2]).

People that are considered poor metabolizers could reduce their dosage or choose a different drug that is not metabolized by CYP2C9, for example, naproxen. Such

recommendations are published in different guidelines that collect evidence from pharmacogenomic studies, for example CPIC [3] and DPWG [4].

Illumina

Illumina is a leading developer, manufacturer, and marketer of life science tools and integrated systems for large-scale analysis of genomic variation and function. These systems are enabling studies that were not even imaginable just a few years ago, and moving us closer to the realization of personalized medicine. With rapid advances in technology taking place, it is mission-critical to offer solutions that are not only innovative, but flexible, and scalable, with industry-leading support and service. Illumina Connected Analytics (ICA) is a comprehensive cloud-based data management and analysis software platform empowering researchers to manage, analyze, and interpret large volumes of multi-omics data in a secure, scalable, and flexible environment.

References

[1] Theken, Katherine N., et al. "Clinical pharmacogenetics implementation consortium guideline (CPIC) for CYP2C9 and nonsteroidal anti-inflammatory drugs." *Clinical Pharmacology & Therapeutics* 108.2 (2020): 191-200.

[2] Reis, Sara S., Ana S. Carvalho, and Rúben Fernandes. "Pharmacogenomics, CYP2D6, and tamoxifen: a survey of the reasons sustaining European clinical practice paradigms." *Medicina* 55.7 (2019): 344.

[3] https://cpicpgx.org/guidelines/

[4] https://www.knmp.nl/patientenzorg/medicatiebewaking/farmacogenetica/pharmacogenetics-1/pharmacogenetics
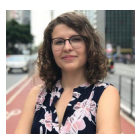
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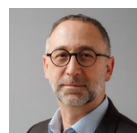Room: G-2.2.34
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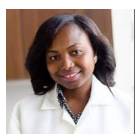Room: G-2.2.30/31
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