

A comparison of drug-relevant genetic raw data from direct-to-consumer genetic test providers and evaluation services with conventional laboratory tests

Submission date 05/07/2023	Recruitment status No longer recruiting	<input checked="" type="checkbox"/> Prospectively registered <input type="checkbox"/> Protocol
Registration date 10/08/2023	Overall study status Completed	<input type="checkbox"/> Statistical analysis plan <input type="checkbox"/> Results
Last Edited 17/10/2023	Condition category Other	<input type="checkbox"/> Individual participant data <input type="checkbox"/> Record updated in last year

Plain English summary of protocol

Background and study aims

Exploratory, descriptive analysis to compare the results of pharmacogenomics between free available direct-to-consumer genetic testing companies (DTC-GT), i.e. 23andme or MyHeritage, and controlled laboratory methods, as a proof of concept. Biological samples (cheek swabs, venous blood) will be sent to 3-4 DTC-GT and to a Swiss laboratory. The results will be compared descriptively for CYP2D6, 2C19, 2C9 and UGT1A1 regarding the question if the data provided by DTC-GT is trustworthy. This study has no aim of validating a method, but only to give a first insight into the quality of the data that patients could explore by themselves and confront their GP with.

Who can participate?

Healthy volunteers between 18 and 75 years old with no risk for bleeding or infections

What does the study involve?

The study involves a comparison between pharmacogenomics results of free available DTC-GT with conventional laboratory testing for drug-relevant metabolic enzymes.

What are the possible benefits and risks of participating?

The benefit is that the participants will know their metabolic panel for several drug-relevant metabolic enzymes (CYP2C9, 2C19, 2D6 and UGT1A1). The only risk is a local reaction after the blood sampling.

Where is the study run from?

University Hospital of Zürich in Switzerland

When is the study starting and how long is it expected to run for?

February 2023 to February 2024

Who is funding the study?

Theodor und Eva Herzog-Egli Stiftung is funding a part of the costs of the study. Third party funds will cover the rest of the costs

Who is the main contact?

Dr. med. Jérôme Bonzon, jerome.bonzon@usz.ch

Contact information

Type(s)

Principal investigator

Contact name

Dr Jérôme Bonzon

ORCID ID

<https://orcid.org/0000-0003-0195-0115>

Contact details

Rämistrasse 100

Zürich

Switzerland

8091

+41 (0)442554074

jerome.bonzon@usz.ch

Additional identifiers

Clinical Trials Information System (CTIS)

Nil known

Protocol serial number

Nil known

Study information

Scientific Title

A comparison of pharmacogenomic raw genotyping data from direct-to-consumer genetic testing services and third-party interpretation websites with conventional laboratory testing

Acronym

CoPharmDL

Study objectives

There are differences in the results between pharmacogenomic raw genotyping data from direct-to-consumer genetic testing (DTC-GT) services and third-party interpretation websites and conventional laboratory testing

Ethics approval required

Ethics approval required

Ethics approval(s)

approved 11/07/2023, Cantonal Ethics Committee Zurich (Stampfenbachstrasse 121, Zürich, 8090, Switzerland; +41 (0)2432597970; info.kek@kek.zh.ch), ref: 2023-00730

Study design

Monocentric exploratory descriptive study

Primary study design

Observational

Study type(s)

Diagnostic

Health condition(s) or problem(s) studied

Descriptive analysis of pharmacogenomic results of from DTC-GT services and third-party interpretation websites compared to conventional laboratory testing

Interventions

Comparison of pharmacogenomic results of from DTC-GT services and third-party interpretation websites with conventional laboratory testing regarding CYP2D6, CYP2C19, CYP2C9 and UGT1A1

Intervention Type

Genetic

Primary outcome(s)

Gain an insight into the quality of personal PGx information freely accessible by patients via DTC-GT, based on the raw genotyping data provided by DTC-GT, the evaluated raw genotyping data by TPI services and by comparison to results obtained through conventional laboratory testing. Completeness and correctness of the results will be assessed descriptively for each PGx enzyme tested (CYP2D6, CYP2C19, CYP2C9 and UGT1A1).

Key secondary outcome(s)

There are no secondary outcome measures

Completion date

29/02/2024

Eligibility

Key inclusion criteria

1. Signed informed consent
2. Ability to understand and follow study procedures and understand informed consent
3. Age 18-75 years

Participant type(s)

Healthy volunteer

Healthy volunteers allowed

No

Age group

Adult

Lower age limit

18 years

Upper age limit

75 years

Sex

All

Key exclusion criteria

1. People at higher risk for infection: age over 75 years, people taking immunosuppressive drugs or with immunodeficiencies
2. People at higher risk for blood loss: people taking anticoagulation and or antiplatelet drugs, people with coagulation disorders

Date of first enrolment

01/09/2023

Date of final enrolment

30/11/2023

Locations**Countries of recruitment**

Switzerland

Study participating centre

USZ

Klinik für Klinische Pharmakologie & Toxikologie

Rämistrasse 100

Zürich

Switzerland

8091

Sponsor information**Organisation**

University Hospital of Zurich

ROR

<https://ror.org/01462r250>

Funder(s)

Funder type

Charity

Funder Name

Theodor und Ida Herzog-Egli Stiftung

Results and Publications

Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are not expected to be made available due to the Swiss law on human genetic testing (GUMG) art. 13, 31 & 33.

IPD sharing plan summary

Not expected to be made available