ISRCTN24498451 https://doi.org/10.1186/ISRCTN24498451

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

Registration date
10/08/2023

Overall study status
Completed

Last Edited
17/10/2023

Condition category
Other

[X] Prospectively registered
[_] Protocol
[_] Statistical analysis plan
[_] Results
[_] Individual participant data
[_] 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
http://orcid.org/0000-0003-0195-0115

Contact details
Rämistrasse 100
Zürich
Switzerland
8091
+41 (0)442554074
jerome.bonzon@usz.ch

Additional identifiers

EudraCT/CTIS number
Nil known

IRAS number

ClinicalTrials.gov number
Nil known

Secondary identifying numbers
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

Secondary study design
Exploratory study

Study setting(s)
Hospital, University/medical school/dental school

Study type(s)
Diagnostic

Participant information sheet
Not available in web format, please use contact details to request a participant information sheet

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 measure
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).

Secondary outcome measures
There are no secondary outcome measures

Overall study start date
23/02/2023

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

Age group
Adult

Lower age limit
18 Years

Upper age limit
75 Years

Sex
Both

Target number of participants
20

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

Sponsor details
Rämistrasse 100
Zürich
Switzerland
8091
+41 (0)442554074
jerome.bonzon@usz.ch

Sponsor type
Hospital/treatment centre

Website
http://www.en.usz.ch/Pages/default.aspx

ROR
https://ror.org/01462r250

Funder(s)

Funder type
Charity

Funder Name
Theodor und Ida Herzog-Egli Stiftung

Results and Publications

Publication and dissemination plan
Planned publication in a high-impact peer-reviewed journal.

Intention to publish date
31/12/2024

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