

НИС ДЗ - Фармакогеномика

<https://github.com/PharmGKB/PharmCAT-tutorial>

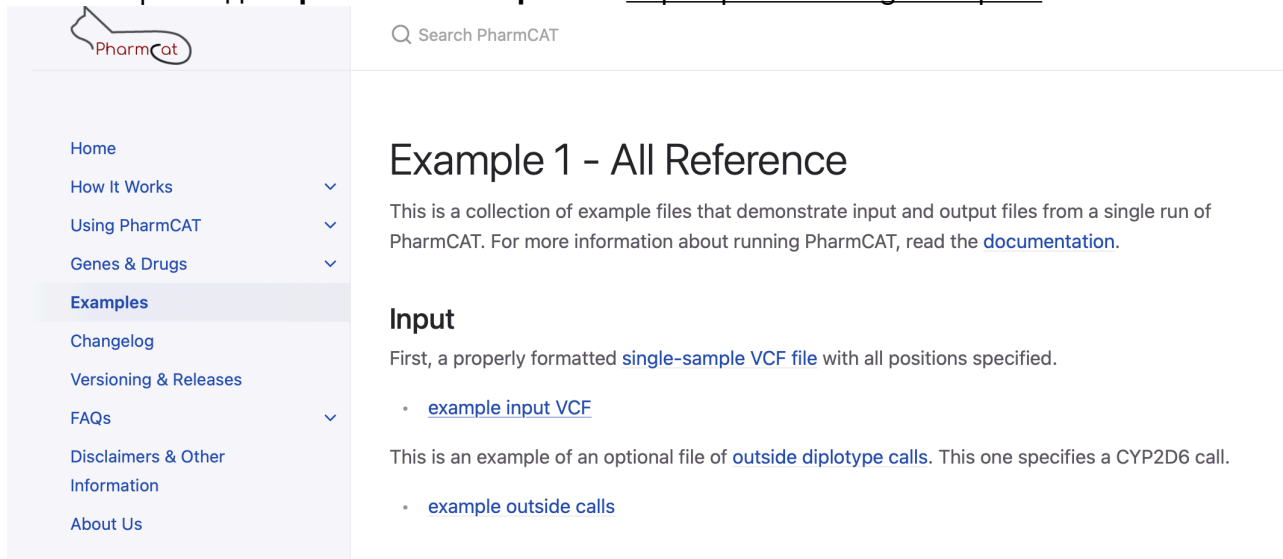
1. Установите фармкат локально или на сервере сами (не используя докер).

Подсказка: тул **pharmcat** имеет расширение **.jar**

Скачала файл **pharmcat-2.15.5-all.jar** отсюда: <https://github.com/PharmGKB/PharmCAT/releases/tag/v2.15.5>.

2. Запустите pharmcat на данных pharmcat.example.vcf по tutorialу приведенному сверху

Скачала файл здесь **pharmcat.example.vcf**: <https://pharmcat.org/examples/>



Образец кода:

```
java -jar <path_to_the_latest_pharmcat_jar> -vcf <sample_file>
```

Моя команда:

```
java -jar pharmcat-2.15.5-all.jar -vcf pharmcat.example.vcf
```

```
(base) jupyter-gorovenko-e@hse-students:~/common/K/pharmcat$ java -jar pharmcat-2.15.5-all.jar -vcf pharmcat.example.vcf
Saving named allele matcher JSON results to /srv/common/K/pharmcat/pharmcat.example.match.json
Saving phenotyper JSON results to /srv/common/K/pharmcat/pharmcat.example.phenotype.json
Saving reporter HTML results to /srv/common/K/pharmcat/pharmcat.example.report.html
Done.
```

3. Получите отчет Reporter в формате .html

Получила отчет в файле **pharmcat.example.report.html**.

Сказала себе этот файл с сервера.
Открыла файл в браузере.

file:///Users/rivikta/Downloads/pharmcat.example.report.html

PharmC...

PharmCAT Report

pharmcat.example

Date created December 13, 2024

PharmCAT Version 2.15.5

Data Version 2024-11-06-02-36

Sections

I. [Genotype Summary](#)

II. [Prescribing Recommendations](#)

III. [Allele Matching Details](#)

IV. [Disclaimers](#)

Disclaimer: PharmCAT is only able to generate recommendations based on the information provided to the software. The gene and variant information for all reported sections are interpreted directly from user-supplied data. The user recognizes they are using PharmCAT at their own risk. For a full list of disclaimers and limitations see [Section IV](#).

Section I: Genotype Summary

Genotypes called: 18 / 19

Drugs	Gene	Genotypes Genotype	Allele Functionality	Phenotype
allopurinol rosuvastatin	ABCG2 [†]	rs2231142 reference (G)/rs2231142 reference (G)	Two Normal function alleles	Normal Function

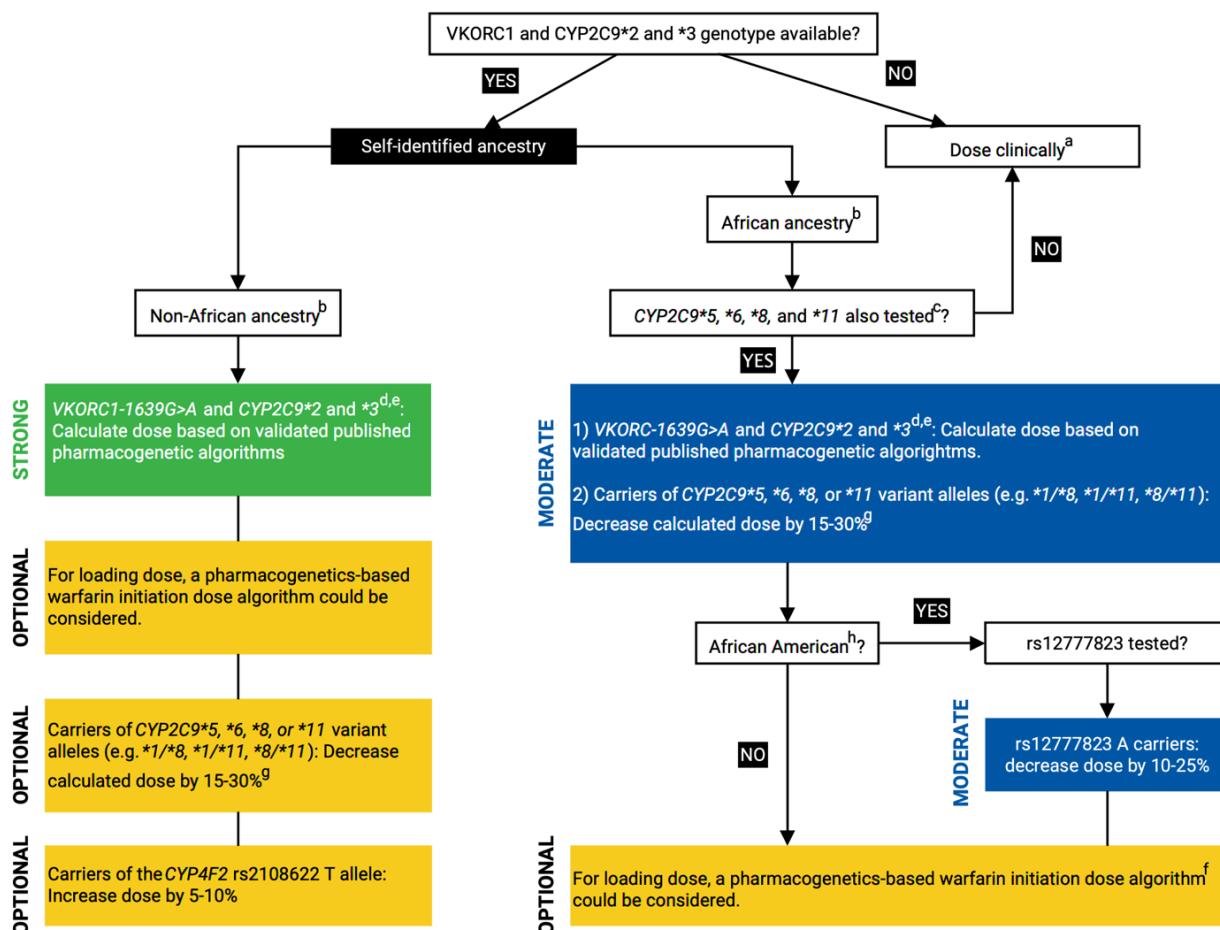
Задания:

1. Напишите полученные генотипы для генов *VKORC1* и *CYP2C9*

VKORC1
rs9923231 reference (C)/rs9923231 reference (C)

CYP2C9
*1/*1

2. Напишите фенотип метаболизатора варфарина



Фенотип: Dose clinically (a)

3. Какая доза варфарина рекомендована гайдлайном CPIC для этого пациента?

- (a) "Dose clinically" means to dose without genetic information, which may include use of a clinical dosing algorithm or standard dose approach
- (b) Data strongest for European and East Asian ancestry populations and consistent in other populations.
- (c) 45-50% of individuals with self-reported African ancestry carry CYP2C9*5, *6, *8, *11, or rs12777823. IF CYP2C9*5, *6, *8, and *11 WERE NOT TESTED, DOSE WARFARIN CLINICALLY. Note: these data derive primarily from African Americans, who are largely from West Africa. It is unknown the same associations are present for those from other parts of Africa.
- (d) Most algorithms are developed for the target INR 2-3.
- (e) Consider an alternative agent in individuals with genotypes associated with CYP2C9 poor metabolism (e.g., CYP2C9 *2/*3, *3/*3) or both increased sensitivity (VKORC1 A/G or A/A) and CYP2C9 poor metabolism.
- (f) See the EU-PACT trial for pharmacogenetics-based warfarin initiation (loading) dose algorithm (33) with the caveat that the loading dose PG algorithm has not been specifically tested or validated in populations of African ancestry.
- (g) Larger dose reduction might be needed in variant homozygotes (i.e. 20-40%).
- (h) African American refers to individuals mainly originating from West Africa.

Рекомендовано назначение дозировки препарата без генетической информации, то есть пациенту назначат стандартную дозу препарата.