Activity 2

A. Working plan

1. Create a new gene panel including variants related Charcot-Marie-Tooth disease.
2. Upload data for 10 patients to evaluate possible candidate variants (VCF files): (this step is ready for you)

- Create a new project + study (Family): CMT.
- Upload all VCF files.
- Add a suspected diagnostic.
- When the status is “ready” and the suspected diagnostic is added we can start the analysis.

3. Analyze each sample from Run Diagnosis:

- Select your sample and the panel
- Choose a name and run!

B. Questions

B.1. Overview

1. How many variants does the sample include?
2. Are there any diagnostic variants?
3. Download “Diagnostic file” and “Secondary Findings” and describe its contents.
4. What is the gene which presents more variants?
5. Download the plot: “Number of variants by gene”.
6. What is the consequence type more frequent?
7. Download the plot: “Number of variants by Consequence Type”

B.2. Diagnostic

1. Comment information about diagnostic variants.
2. Interpret the meaning of the plot down.
3. Check diagnostic variants in the Genome Viewer.

B.3. Secondary Findings

1. How many secondary findings did you find?
2. What is the meaning of the filters? Check static and custom filters.
3. What about “Frequencies” for these variants?
4. Go to the different links for obtain more information about variants. (ENSEMBLE, BEACON, OMIM...)

B.4. Report

1. Include patient data, find confirmed diagnostic and save for a future review.
2. Could you generate a nice report? Save it as a .pdf.