# Activity 1

## Create new user and upload the data

1. Please create your own user and login with this new user.
2. Upload the required VCF file in "My Data" → "Default Study" and wait 4-5 min to indexing tool. patients.vcf
3. While the file is indexing we will design all the panels.

## Design panels

### Cataracts

1. Use the "Panels" menu → "+ New Panel" and create a new panel for "Cataracts".
2. Select the next phenotypes: "Cataract" and "Cataracts".
3. Use "Import from external App" and Search in "PanelApp" if we can get more genes for this panel (select only genes with "HighEvidence").
4. With this panel search 1 or more samples with the disease using "Run Diagnosis".

### Hyperammonaemia

1. Use the "Panels" menu → "+ New Panel" and create a new panel for "Hyperammonaemia".
2. Select the next phenotype: "Hyperammonaemia".
3. Use "Import from external App" and Search in "PanelApp" if we can get more genes for this panel (select only genes with "HighEvidence").
4. With this panel search 1 or more samples with the disease using "Run Diagnosis".

### Osteogenesis imperfecta

1. Use the "Panels" menu → "+ New Panel" and create a new panel for "Osteogenesis imperfecta".
2. Select the next phenotype: "Osteogenesis imperfecta".
3. Use "Import from external App" and Search in "PanelApp" if we can get more genes for this panel (select only genes with "HighEvidence").
4. With this panel search 1 or more samples with the disease using "Run Diagnosis".

### Fanconi anemia

1. Use the "Panels" menu → "+ New Panel" and create a new panel for "Fanconi anemia".
2. Select the next phenotype: "Fanconi anemia".
3. With this panel search 1 or more samples with the disease using "Run Diagnosis".

### My disease

1. We will create a panel for a new disease: "my disease".
2. Copy genes from this file: genes.txt
3. Import mutations from this CSV file: mutations.txt (NOTE: this is a TXT file but you should import it using "import CSV")
4. These genes and these mutations are from a real disease. Do you know the name of this disease?