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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 “Default Study” and wait 4-5 min to indexing tool. http://bioinfo.cipf.es/apps-beta/gda16/team/patients.vcf
3. While the file is indexing we will design all the panels.

Design panels

Cataracts

1. Use the ”+ Panel” menu and create a new panel for “Cataracts”.
2. Select the next phenotypes: “Cataract” and “Cataracts”.
3. Use Panel App and Search 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.

Hyperammonaemia

1. Use the ”+ Panel” menu and create a new panel for “Hyperammonaemia”.
2. Select the next phenotype: “Hyperammonaemia”.
3. Use Panel App and Search 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.

Osteogenesis imperfecta

1. Use the ”+ Panel” menu and create a new panel for “Osteogenesis imperfecta”.
2. Select the next phenotype: “Osteogenesis imperfecta”.
3. Use Panel App and Search 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.

Fanconi anemia

1. Use the ”+ Panel” menu 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.

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?