Practical Day 4: Fine Mapping

In this practical, we will use three different approaches to better characterize association signals within a genomic region, identify independent signals and perform fine-mapping. Details on how to do these have been provided in the demo.

LDLR -LD-Assoc-tools

1. Upload data - text file provided (**LDLR.txt**) ; SNP to test : **rs112552009**
2. Select one super-population at a time and study LD between significant SNPs in the region and the lead signal?
3. Export images for AFR, SAS, and EAS?
4. Which of these populations show fewer SNPs in high LD with the lead SNP?

Casual-DB

1. Search for Cholesterol LDL in Causal-DB (please search it in the correct category!)
2. Locate Prins et al. 2017 study
3. Navigate to the first peak on chromosome 19
4. Which genes does the peak in this genomic region correspond to?
5. Check how many SNPs are there in the 95% credible set predicted by the 3 programs?
6. Check how many SNPs are there in the 95% credible set predicted by the 3 programs?
7. Sort SNPs by PAINTOR scores
8. Compare the top 6 variants to see which two SNPs have the strongest functional evidence?
9. Sort SNPs by FINE-MAP scores.
10. Compare the top 6 variants to see which two SNPs have the strongest functional evidence? Do FINEMAP and PAINTOR identify the same two best SNPs?

COJO

Run GCTA COJO on file provided (**ldlr.ma**)

```
gcta64  --bfile san.chr19  --cojo-file ldlr.ma   --cojo-slct --out san.chr19
```

How many independent associations can you see in the SAN population?