

# Clustering of the human genome

Menghor THUO, Matthieu DESPREZ, Skander GORGI

## Description of the data

The objective of this project is to apply clustering algorithms to genotype data derived from a large-scale genomic study, specifically focusing on a dataset from the 1000 Genomes Project. By clustering genotypes, we aim to:

1. Identify subpopulations within the dataset.
2. Examine the relationship between genetic variation and phenotypic traits.

The genotype array has the following characteristics:

- **Shape:** (1103547, 2504, 2)
  - **1103547:** The number of variants (rows), which corresponds to the number of different genetic variants the VCF file.
  - **2504:** The number of samples (columns), indicating how many individual samples are represented in the dataset.
  - **2:** The number of alleles (for diploid organisms, each individual has two alleles at each variant site).

There is a list of fields to be extracted from a VCF (Variant Call Format) file when using the `allel` library in Python. Each field corresponds to a specific type of information stored in the VCF file format. Here's a breakdown of each field in the list:

### 1 variants/CHROM:

- **Description:** This field contains the chromosome identifier where the variant is located.
- **Example:** It has values like `chr1`, `chr2`, `chrX`, etc. This indicates which chromosome the variant appears on.

## 2 variants/POS:

- **Description:** This field represents the position of the variant on the specified chromosome.
- **Example:** It is a numeric value (e.g., 123456) that indicates the exact base pair position on the chromosome where the variant occurs.

## 3 variants/ID:

- **Description:** This field contains an identifier for the variant, if one exists.
- **Example:** Commonly, this will be an rsID from dbSNP (e.g., rs123456). If there is no identifier for a particular variant, this field might be represented as a dot (.).

## 4 variants/REF:

- **Description:** This field shows the reference allele at the specified position.
- **Example:** It will contain the nucleotide(s) that are present in the reference genome (e.g., A, C, G, T).

## 5 variants/ALT:

- **Description:** This field lists the alternate alleles present at the position, which represent the variant(s) that differ from the reference.
- **Example:** This could be a single nucleotide like G (for a SNP) or multiple alleles in case of multi-allelic variants (e.g., G, T for a variant that can be either G or T).

## 6 calldata/GT:

- **Description:** This field provides genotype information for each sample at the variant position. It indicates the alleles present in the samples.
- **Example:** Genotype representations include:
  - 0/0: Homozygous reference (both alleles are the reference allele)
  - 0/1: Heterozygous (one reference and one alternate allele)
  - 1/1: Homozygous alternate (both alleles are the alternate allele)

The dataset was found on <http://www.internationalgenome.org/> .