- Zimmer V1

	Homozygous	Heterozygous
Pre-filter	22	64
After-filter	13	42

- 1KG P3, see here: https://public.tableau.com/profile/yali.xue#!/vizhome/1000G_phase3_per_individual_count_all/Sheet1

After filtering, ~ 10 homo & 36 ~ 40 heterozygous

- In Zimmer genome, all 4 potentially deleterious LoFs are heterozygous and predicted to be recessive, meaning not causing any phenotype

- We used machine-learning method to train our model on disease-causing and benign LoF variants with various features, e.g. evolution, biological networks and functional impact.
- Our model is able to distinguish deleterious LoFs from benign ones and also give a prediction whether the LoF will cause disease in a recessive or dominant pattern.
- The deleterious scores produced by the model will be the probability that a particular LoF is causing disease in recessive/dominant pattern or totally benign.
- Usually we took a cut-off 0.5 to make a decision whether a variant is deleterious or not.
- The prediction score is unrelated to the homozygous or heterozygous genotypes of the individual, which needs to be combined with the individual's genotype to determine which LoFs are disease-causing for this individual