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FAROE GENOME PROJECT

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Why study the Faroese population?

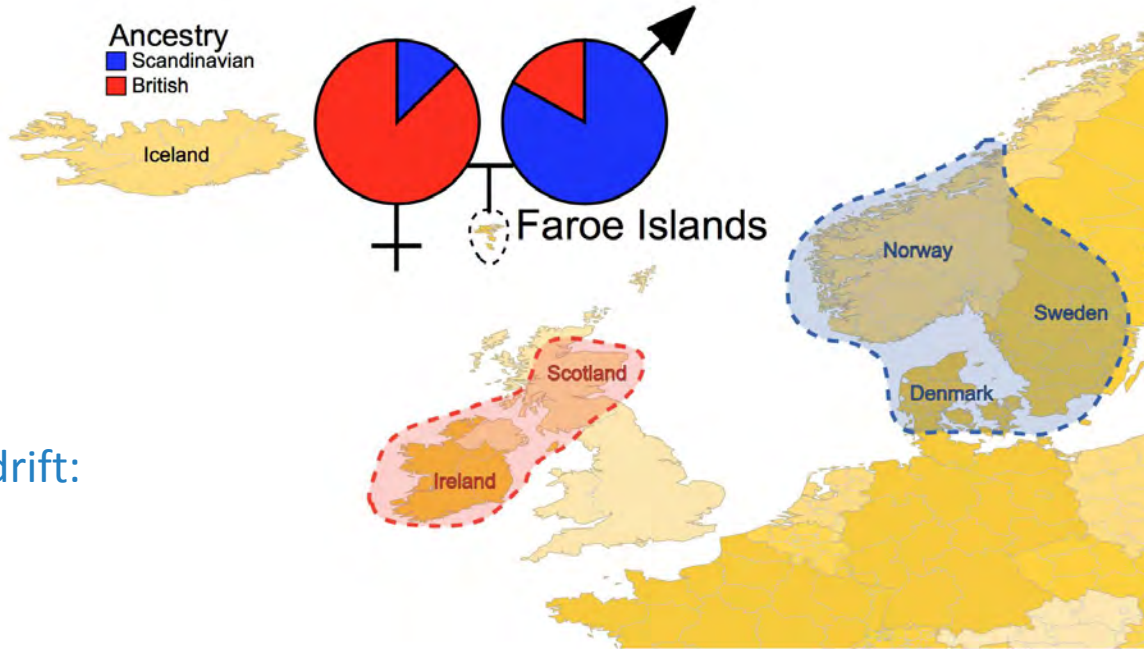
- Current population: ~50 000

Demographic history:

- Few founders
- Little immigration for centuries
- Recent population expansion

High founder effect and genetic drift:

- Homogeneous population
- Enrichment of rare variants
- Novel variants



Jørgensen et al. 2002, Als et al. 2006

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Genetic disorder	Prevalence		Gene	Number of genetic variants (clinical)	
	Whole world	Faroe Islands		Whole world	Faroe Islands
Mono-genetic			Genotype		
Cystic fibrosis	1 in 3.000	1 in 1.775	*CFTR* (recessiv)	1289	1
Carnitine transporter deficiency	1 in 100.000	1 in 300	*SLC22A5* (recessiv)	148	4
Phenylketonuria (PKU)	1 in 12.000	1 in 10.000	*PAH* (recessiv)	635	NA
Glycogen storage disease (GSDIIIA)	1 in 50.000	1 in 2.700	*AGL* (recessiv)	43	NA
Genetic complex			**Haplotype**		
Glaucoma	1 in 10.000	1 in 70	*MYOC* og *CYP1B1*	36, 46	NA
Inflamatory Bowel Disease	1 in 1.000	1 in 75	*ATG16L1* , *IRGM* , *NOD2* og *IL23R*	19, 6, 121, 14	NA
Panic disorder	1 in 100	1 in 100	*TMEM132E, COMT, DGKH*	3, 172, 28	3, 8, 19
Parkinson's disease	1 in 1.000	1 in 500	*LRRK2, PARK2, PARK7, PINK1* , og *SNCA*	138, 98, 32, 31, 19	NA

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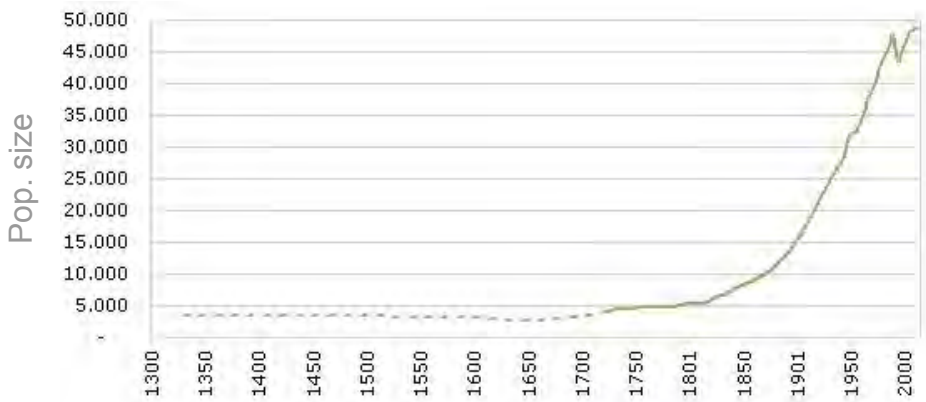
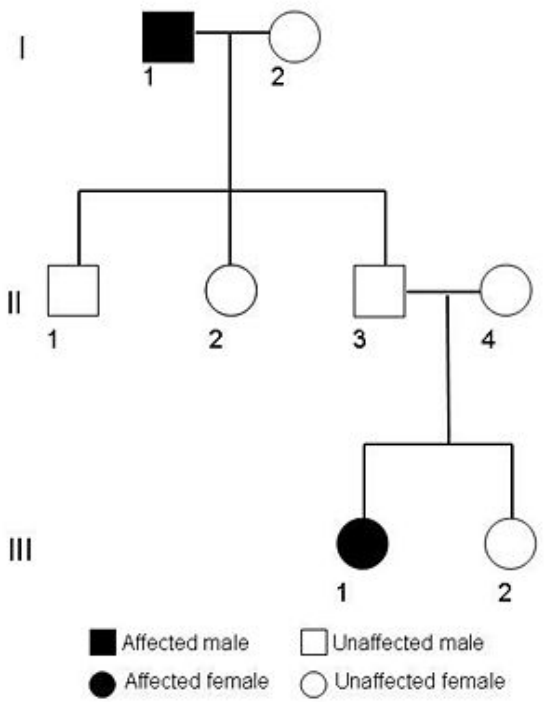
Genealogy registry

- Detailed information from about year 1650
- Uses cases:
  - Gathering cohorts for association studies
  - Population consanguinity





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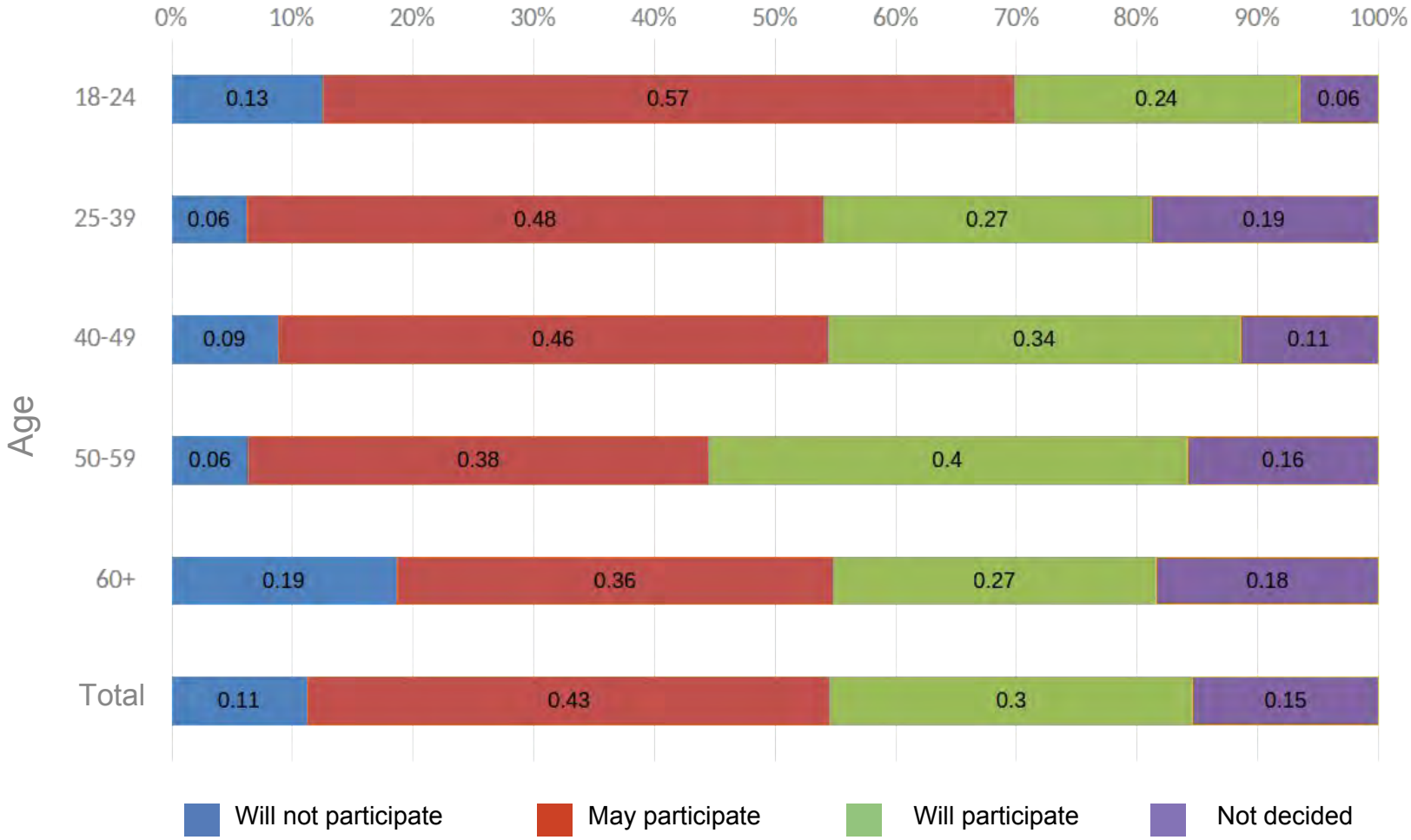
Do people want to participate in FarGen?



Gallup, 2015.

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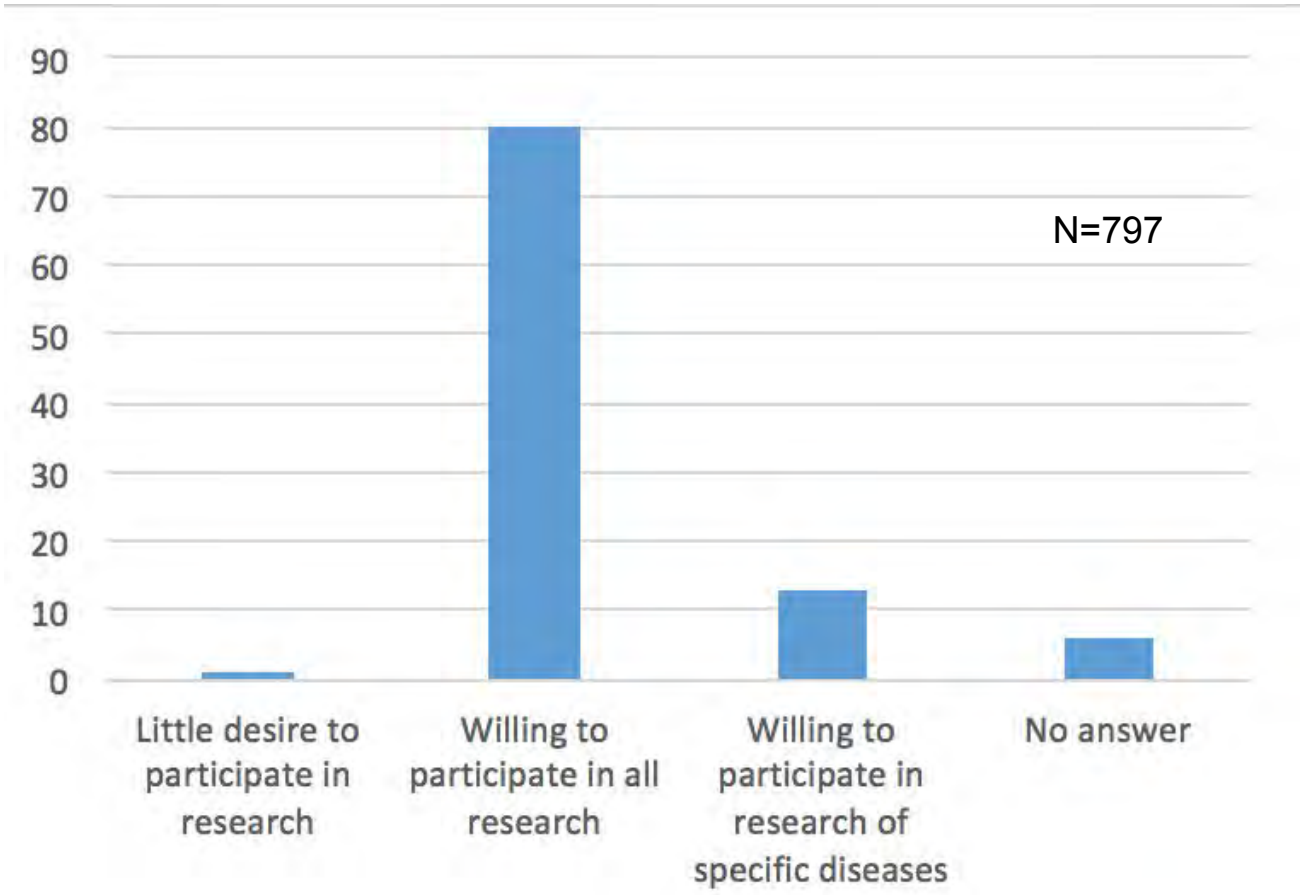
Do people want their data to be used in research?



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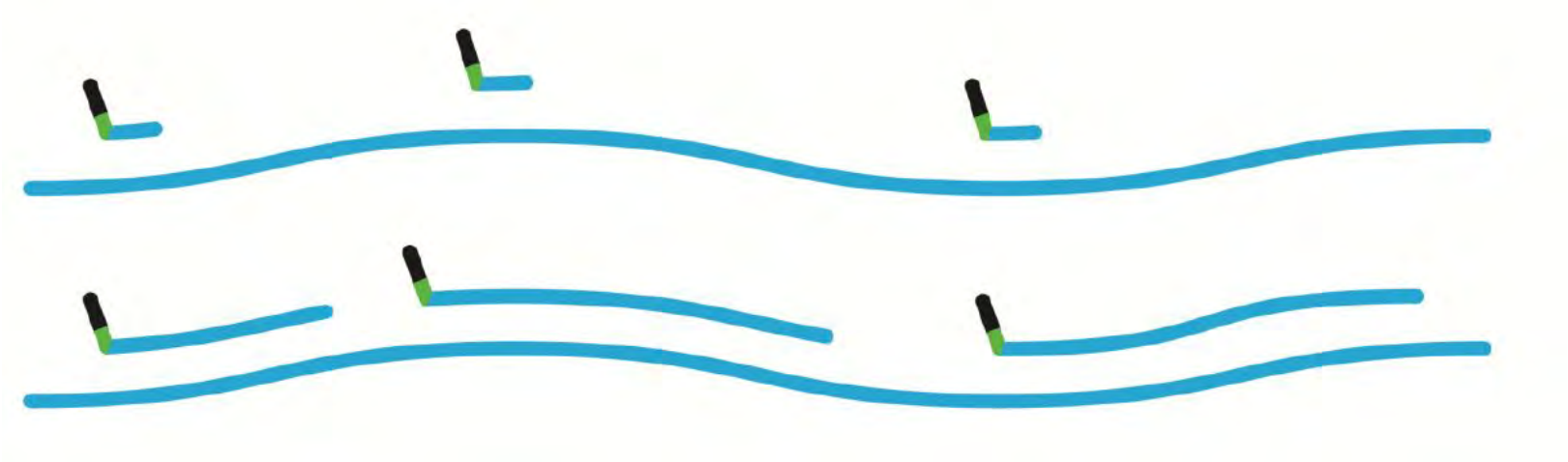
10x Genomics

"Long range information from short-read sequencing"

- Increased contig size (> 100 kb)
  - Accurate *de novo* assembly
  - Structural variants
- Phase blocks sizes > 2.5 Mb
  - Accurate phasing
  - "True diploid assembly"



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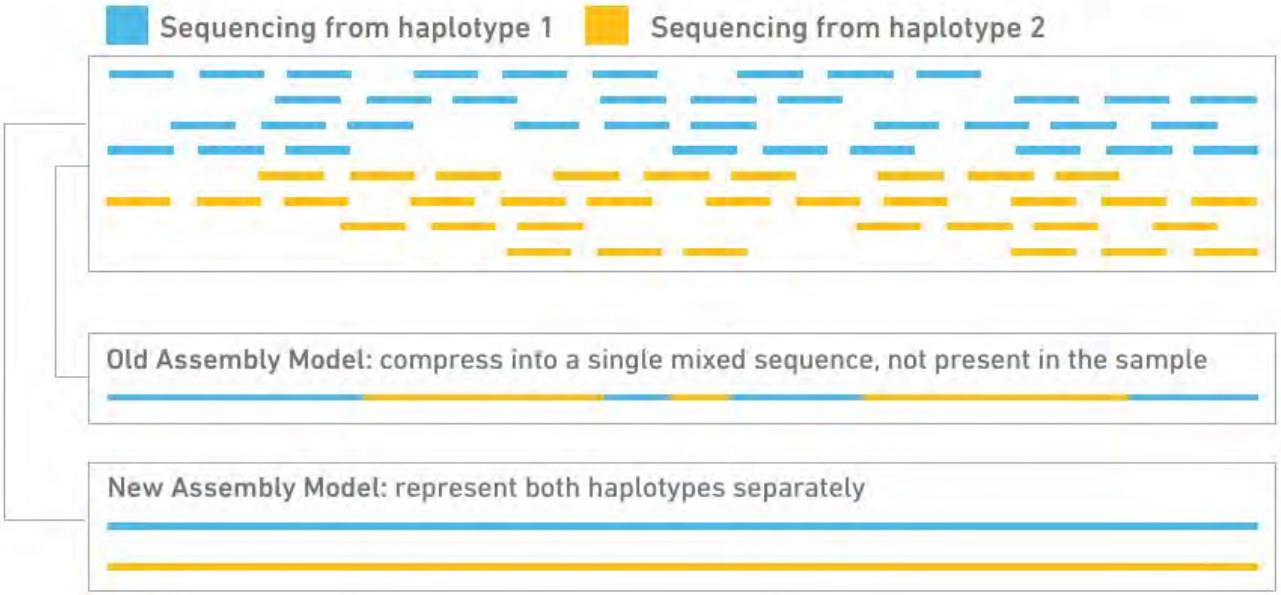

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Plans

- Whole-genomes, whole-exomes, trios
- Catalogue of genetic variation
  - SNPs, indels and CNVs
  - Structural variants
  - *De novo* mutations
  - Novel variants
- Demographic inference
  - Population structure
- Population specific resource for whole-genome imputation based on whole-exome sequencing

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Population stratification

- What is the structure?
- How significant is the stratification?
- Should the structure be considered when sampling from the population?

- Hypothesis driven (as in image)
- Clustering
- Isolation by distance
- Use genealogy registry to generate hypothesis?

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Where are we now?

- About 1700 people have registered for the FarGen project
- About 1100 blood samples have been taken
- Sequencing has begun

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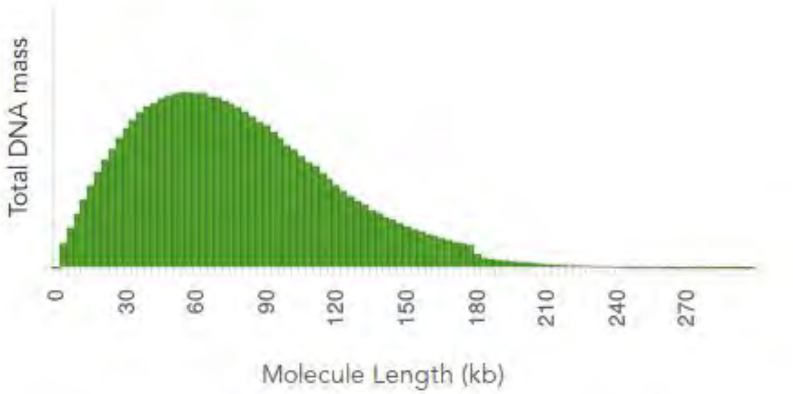
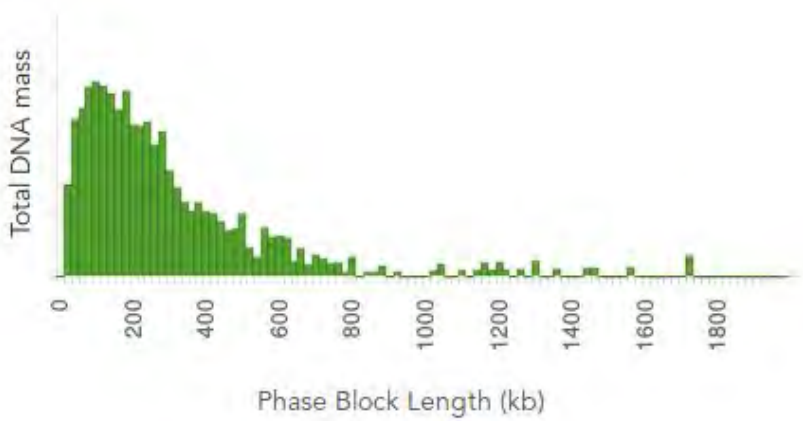
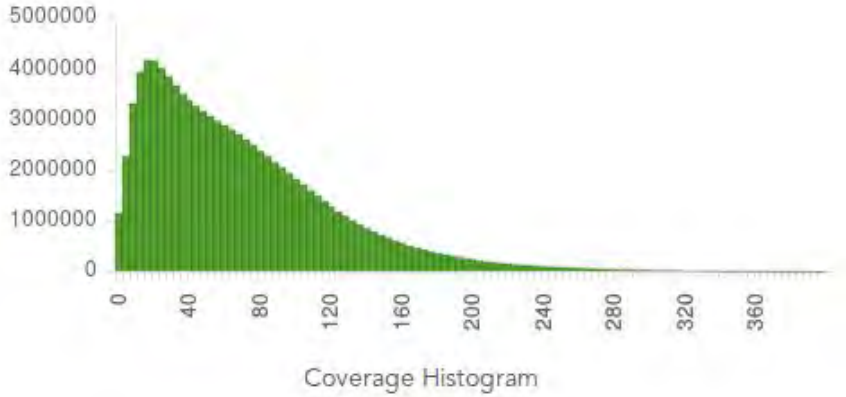
Where are we now?

- Mean molecule length: ~85 kb
- Mean coverage: 75.5x
- 96% of genes phased







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Thank you

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