

Genomics & Bioinformatics

Gen711/811

<http://gen711.readthedocs.org>

ANNOUNCEMENTS

- Email: Matthew.MacManes@unh.edu
- Twitter: @MacManes #Gen711
- Office hours: Tuesday/Friday 8-10am in 189 Rudman, or by appt. Schedule at <http://genomebio.org/office-hours/>

ANNOUNCEMENTS

- Lecture: (you've figured this out)
- Lab: Friday 310-500pm Hewitt 301
- Blackboard: Grades and other unimportant things
- Website: <http://gen711.readthedocs.org>

GOALS OF THE CLASS

- Cloud Computing
- UNIX
- Don't be afraid of the command line
- Assemble genomes and transcriptomes
- Map billions of sequence reads
- Annotate genomes
- Learn some biology
- Have fun!

SYLLABUS

- <http://gen711.readthedocs.org/>





Intro to Bioinformatics

**Lecture 1
Aug 31, 2015**

GENOMICS

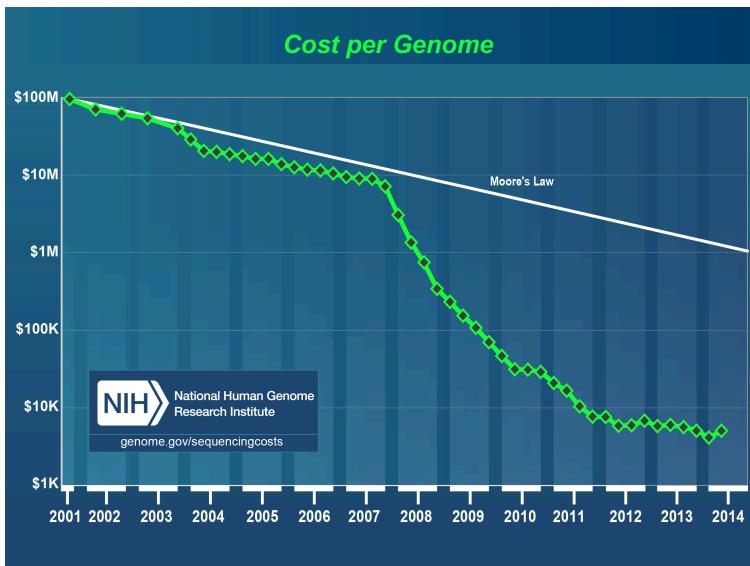
- What is genomics?

GENOMICS

- What is bioinformatics?

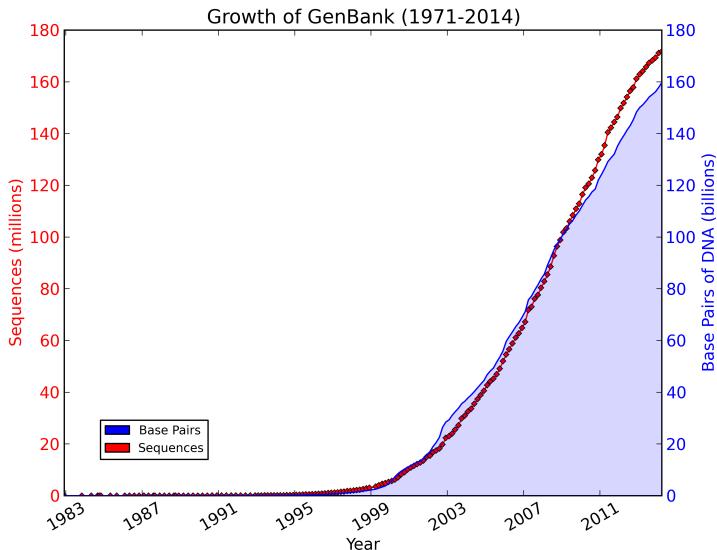
BIOINFORMATICS

- Why now??



BIOINFORMATICS

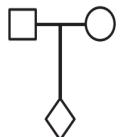
- Why now??



DO OLD FATHERS PASS ON MORE MUTATIONS

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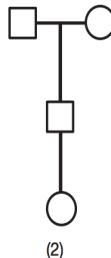
a 57 simple trios



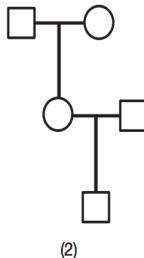
b 6 sib-pairs



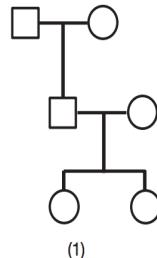
c 5 three-generation families



(2)



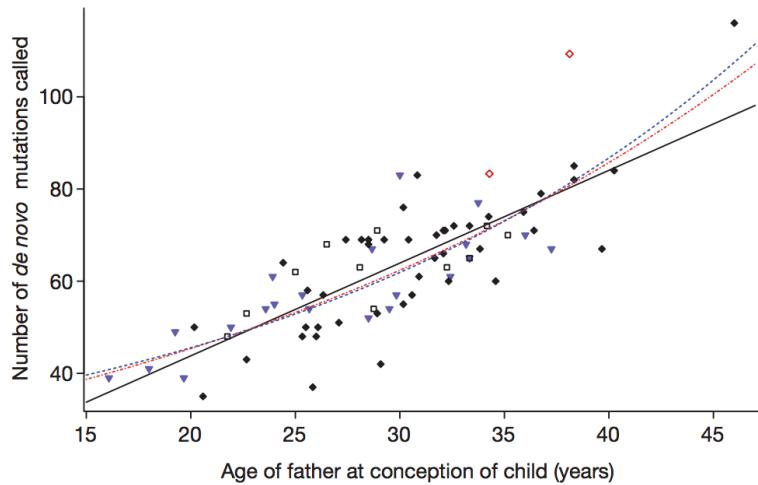
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(1)

Kong et al 2013 Rate of DeNovo Mutations and the Importance of Father's Age to Disease Risk Nature 488

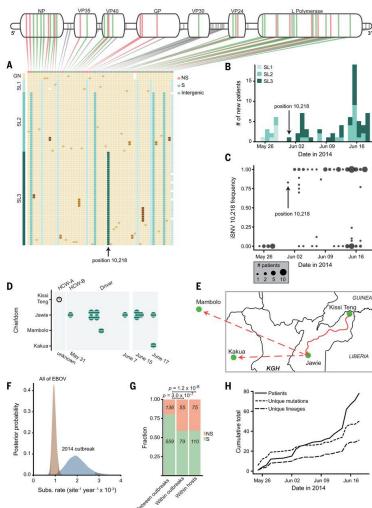
DO OLD FATHERS PASS ON MORE MUTATIONS



Kong et al 2013 Rate of De Novo Mutations and the Importance of Father's Age to Disease Risk Nature 488

WHY IS 2014 EBOLA SO DEADLY?

WHY IS 2014 EBOLA SO DEADLY?



Gire et al 2014 Genomic Surveillance elucidates Ebola Origin *Science*

- Discover other stuff, too
 - Genes
 - Expression levels
 - Repetitive regions
 - Community composition
 - Methylation
 - ...

SEQUENCING PLATFORMS

- Illumina (@illumina)

SEQUENCING PLATFORMS

- Pacific Biosystems (@PacBio)

SEQUENCING PLATFORMS

- Oxford Nanopore (@nanopore)



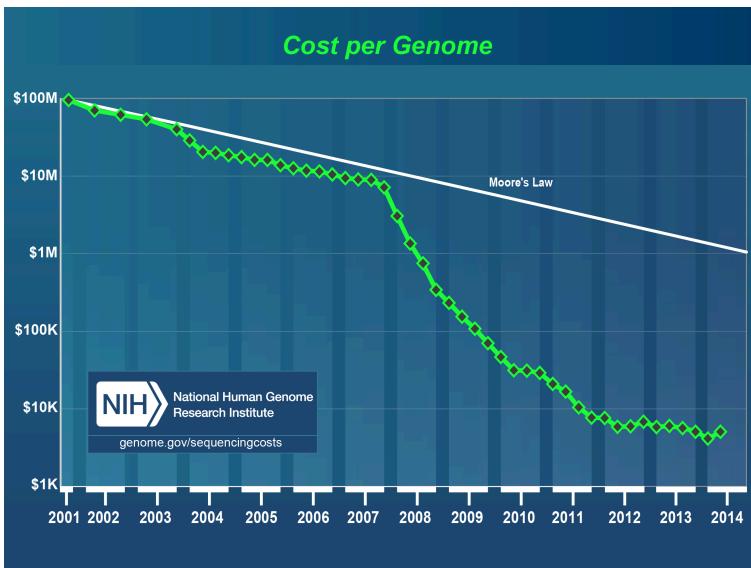
SEQUENCING PLATFORMS

- 454 (@LifeTech)

SEQUENCING PLATFORMS

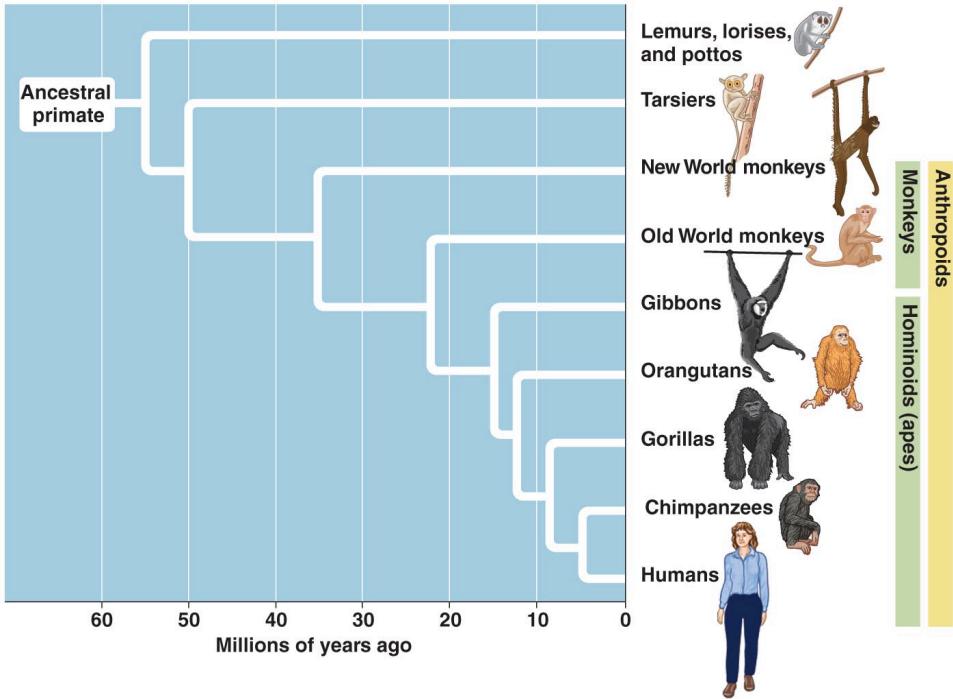
- Sanger

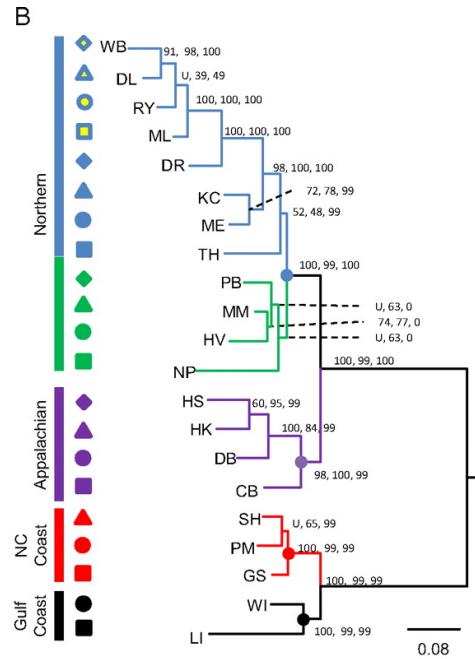
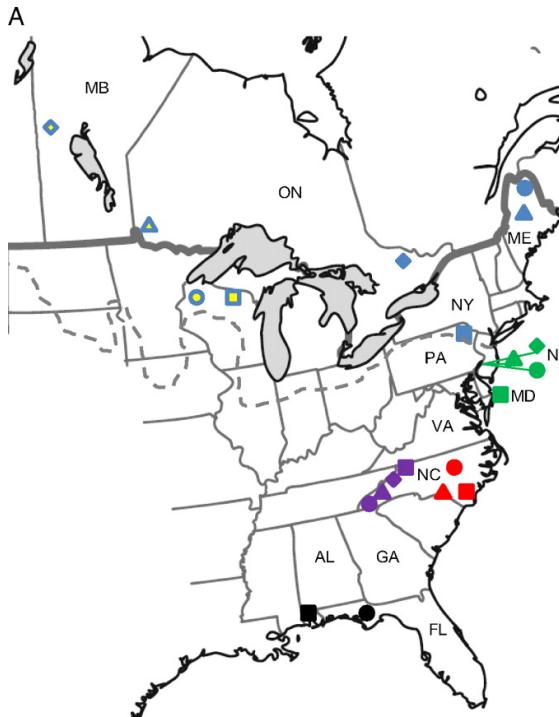
SEQUENCING PLATFORMS



CONTEXT IS EVOLUTION

- Evolution





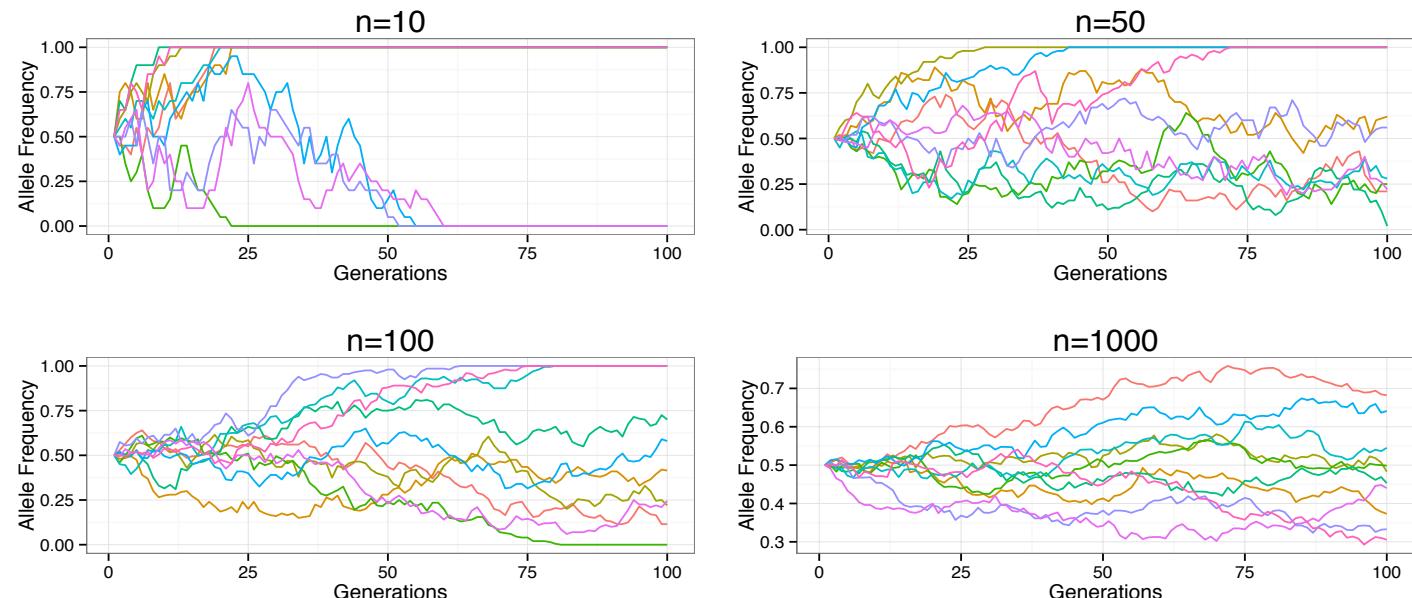
MECHANISMS OF EVOLUTION

- Gene Flow

MECHANISMS OF EVOLUTION

- Genetic Drift

MECHANISMS OF EVOLUTION



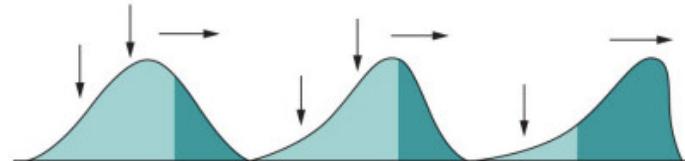
MECHANISMS OF EVOLUTION

- Mutation

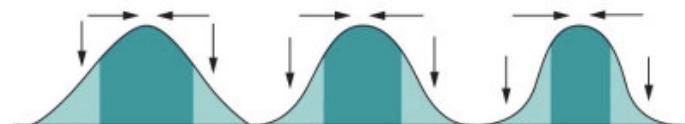
MECHANISMS OF EVOLUTION

- Natural Selection

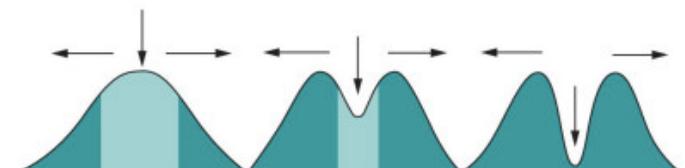
SELECTION



(a) Directional selection



(b) Stabilizing selection



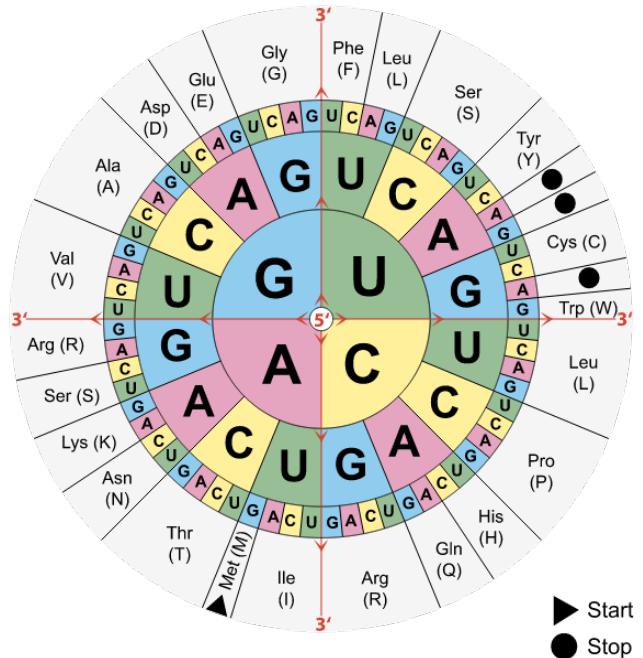
(c) Disruptive selection

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SELECTION

- Synonymous change
- Non-synonymous

SELECTION



SELECTION

- dN/dS

Finding Data & Pairwise Alignment

Lecture 2
Sept 9, 2014

FINDING DATA

- Genomics has embraced Open Science & Open Data

Genbank	ENA
Uniprot	dbSNP
Refseq	ClinVar
Swiss-prot	Rdp
Pfam	OMIM
Ensembl	UCSC GB
SRA	

FINDING CODE

- Genomics has embraced Open Science & Open Data

Github
[Biostars](#)

[Stackoverflow](#)

PAIRWISE ALIGNMENT

- Definition

PAIRWISE ALIGNMENT

- Homologous
 - Paralogous
 - Orthologus

PAIRWISE ALIGNMENT

- Alignment demo:
 - [mafft](#)

PAIRWISE ALIGNMENT

- What is aligner actually doing?

PAIRWISE ALIGNMENT

- Scoring Matrices