Slide #1: The personal genomics revolution
- More than 1M people now have access to their own genomes, for just around $100
- Compare that to sample sizes in academic studies of 1000s of thousands
- Power of personal genomics: in exchange for your data, empower consumers to analyze their own genome
- Genomics so cheap, now a commodity

Slide #2: The power of commercial genome databases
- With a database of 1M+ users, can simply ask users to fill out surveys, correlate with the genome
- E.g., dress, survey assK-ppl in week!
- or morning vs. night person? messy data, perhaps, but can reveal real genetic hints related to circadian rhythms
- In academia, want grant to fund, in the approval, collect forms, call samples, ... CPS, later, and participants may never see results.

Slide #3: I have longstanding interest in genetics
- I first got interested in genetics when I learned as a kid that twins share 99% of their genomes!
- Later tested if I was a twin.
- Which one is me?

Slide #4: Outline
- Why would someone want to analyze their genome?
- Course overview: what to expect
- Whitewashed history of genetics
- Basic bio/humgen
- Work on PSI

Slide #5: Why analyze your genome?
- In most cases, the answer is because it's fun!
- Spoiler alert: you are unlikely to learn anything hugely important health wise, or even that much new about your family, etc...
- But will learn about what you can realistically expect, and how sequencing your own genome can end up helping other people

Slide #6: Mutations have implications for human health
- Perhaps most obvious reason to look at your genome is to learn about your health. For most people, you are much healthier.
- But there are many highly informative mutations
- Example, one of 1st diseases mapped: CF
- A copy of mutation in CFTR
- Quite common in Europeans, low life expectancy
- Can now be prevented by carrier testing to inform reproductive options

Slide #7: Our genomes contain a record of human history
- Mutations in the genome act like a clock, across a range of time scales
- Another popular, recent, sequence: nuclear genome, more reliably than medical info
- Close relatives: e.g., parents, siblings, cousins
- Ethnicity: e.g., European genome recapitulates map, see who you fall
- Ancient history, migrations, Neanderthals

Slide #8: Your genome is uniquely identifying
- Thus, it is highly useful in forensics
- We'll talk about what that means for privacy implications
Slide #10: Course overview
- Course objectives
- Course structure, schedule, modules
- Outline of sessions
- Grading, assignments
- If you want to drop out...

Slide #15: A whirlwind, history of human genetics
- This course won't give comprehensive human genetics course.
- Instead, we're going to squish an entire semester of human genetics into one class. Important to have some basic background, so please ask questions!

Slide #16: Mendel establishes heredity as a principle (1865)
- We've known about heredity for thousands of years: kids tend to look like their parents.
- Mendel studied pea plants.
- If you cross F1, always get yellow! (F1)
- But if you cross F1, green reappear!
- Mendel reared 0.5 mm peas into reproductive ratios.
- Can explain a “unit” called genes.
- Allows heir to “skip” generations.

Slide #17: mid-1900s - DNA as genetic material
- Won't go over in detail, but fill in our timeline.
- Griffith: back to transfer genetic info.
- AMC: this is DNA.
- Hers: DNA indeed transfers genetic info.
- Watson & Crick: DNA structure

Slide #18: First disease gene mapped (1983)
- HTT, huntington's disease, abnormal movements, tumours, families.
- Only been described for 100 years.
- Especially present in large pedigrees in Venezuela.
- Sequenced others this painstaking methodical effort.
- “HTT” is called “Huntington’s” disease.
- Many other diseases followed in early 90s.

Slide #19: The human genome is sequenced (2000)
- Huge public effort over 10 years, 3 billion.
- Competing project from Venter/Colo.
- By 2000 had sequenced, mostly from man.
- Sequenced complete, but huge landscape.

Slide #20: Toward the $1000 Genome
- You're likely seeing this slide before, better than Hitt's law in 10 years.
- 2015: $1 and $1 per mark for entire WGS.

Slide #21: The personal genomics revolution
- Direct-to-consumer companies offer
- SNPs, i.e. single nucleotide positions.
- Note: not whole genome, but on a subset of 1.5 M positions, we'll learn about different technologies.
- Pundits say, “only” how many companies.
- Give users health, beauty, ancestry.
- Hand’s mine as an example.

Slide #22: Biology Intro
- Go over some basic biology I'll assume we all know for rest of the course.

Slide #23: Bird's eye view of the human genome
- Body made up of cells, each cell has nucleus.
- DNA lives in the nucleus.
- Normal human has 46 chromosomes.
- 23 from mom, 23 from dad.
- XY sex chromosomes.
- XX, XY.
- 1-22 autosomes.

Slide #24: DNA Structure
- Components: 4 bases, sugar, phosphate.
- Bases pair according to strict rules.
- Watson & Crick.
- Among four phosphates, sugar (backbone).
- Always reads in 5' to 3' direction.
- 5'-Thymine G-C
- Reverse complement: 5'-A-T.
Slide #36: Modes of inheritance - dominant
- 1 copy of dominant gene => affected
- example: pedigree
  - example disease: Marfan's syndrome

Slide #37: Modes of inheritance - recessive
- 2 copies of recessive gene => affected
- example: CF

Slide #38: Modes of inheritance - X-linked
- Need at least one unattacked copy of X
- Usually males affected, females "carriers"
- e.g. Hemophilia A

Slide #39: Example recessive trait - red hair
  (I'm heterozygous)

Slide #40: Example recessive trait - eye color
- 65% of control most of eye color
- blue eyes primarily controlled by 1
  SNP: regularly change in T R E Q

Slide #41: Beyond Mendelian - complex traits
- Many (most?) traits controlled by many
genes, not just 1
- Fisher's: many genes contribute small
  additive effects toward phenotype
- e.g. height: 1000s of genes

Slide #42: Example complex traits - SCZ
- 80% h², only recently discovered
  true genetic association

Slide #43: PS1

Slide #44: Getting started