CMDB Bootcamp

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# 1 Homepage

This is the course homepage and digital textbook for CMDB Bootcamp.

#### 1.0.0.1 Instructors

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#### 1.0.0.2 Schedule & Logistics

Class is **Tuesdays from 3-3:50PM**, in **UTL G89**.

Please bring your laptop with you to every class.

| Session | Content |
| --- | --- |
| **Session 1:** | The reference genome & genome browsers |
| **Session 2:** | *De novo* mutations |
| **Session 3:** | Linkage disequilibrium |
| **Session 4:** | Simulating evolution |
| **Session 5:** | Population structure – part I |
| **Session 6:** | Population structure – part II |
| **Session 7:** | Genome-wide association studies – part I |
| **Session 8:** | Genome-wide association studies – part II |
| **Session 9:** | Scans for selection – part I |
| **Session 10:** | Scans for selection – part II |
| **Session 11:** | Archaic admixture |
| **Session 12:** | Gene expression |
| **Session 13:** | Coronavirus phylogenetics |

# 2 Discovering mutations

In this module, we’ll use DNA sequencing data from human families to explore the relationship between parental age and *de novo* mutations in their children.

#### 2.0.0.1 Learning objectives

After completing this chapter, you’ll be able to:

1. Create plots to visualize the relationship between two variables.
2. Interpret the results of a linear model.
3. Compare the impact of maternal vs. paternal age on *de novo* mutation counts.
4. Explain what a confidence interval is and why it’s useful.

## 2.1 *De novo* mutations

Mutation and recombination are two biological processes that generate genetic variation. When these phenomena occur during gametogenesis, the changes that they make to DNA are passed down to the next generation through germline cells (i.e., sperm and oocyte).

***De novo* mutations (DNMs)** arise from errors in DNA replication or repair. These mutations can be single-nucleotide polymorphisms (SNPs) or insertions and deletions of DNA. Every individual typically carries around 70 *de novo* SNPs that were not present in either of their parents.

## 2.2 Recombination

**Crossovers**, or meiotic **recombination**, occur during prophase of meiosis I, when homologous chromosomes pair with each other. Double-strand breaks are deliberately generated in the DNA, and are then cut back and repaired based on the sequence of the homologous chromosome. These repairs can sometimes resolve in a crossover event, where sections of DNA are swapped between chromosomes.

Because the sequences of homologous chromosomes differ at sites where they carry different alleles, recombination generates genetic diversity by creating new haplotypes, or combinations of alleles.

Crossovers are required for meiosis in most organisms because they ensure proper homologous chromosome pairing and segregation. Humans experience 1-4 crossover events per chromosome, with longer chromosomes having more crossovers.

# Authors

| Credits | Names |
| --- | --- |
| **Pedagogy** |  |
| Instructor | [Rajiv McCoy](https://mccoy-lab.org/) |
| Content Author | [Stephanie Yan](https://stephaniemyan.github.io/) |
| Content Author | [Kate Weaver](https://kweav.github.io/) |
| **Website** |  |
| Template | [Jeff Leek](https://jtleek.com/) & [The Johns Hopkins Data Science Lab](https://jhudatascience.org/index.html) |
| Design Inspiration | [Ali Madooei](https://engineering.jhu.edu/faculty/ali-madooei/) & [JHU Data Structures](https://cs226sp22.github.io/) |
| **Funding** |  |
| JHU Center for Educational Resources | Techology Fellowship Grant |

## ─ Session info ───────────────────────────────────────────────────────────────  
## setting value   
## version R version 4.0.2 (2020-06-22)  
## os Ubuntu 20.04.5 LTS   
## system x86\_64, linux-gnu   
## ui X11   
## language (EN)   
## collate en\_US.UTF-8   
## ctype en\_US.UTF-8   
## tz Etc/UTC   
## date 2023-07-14   
##   
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## cachem 1.0.7 2023-02-24 [1] CRAN (R 4.0.2)   
## callr 3.5.0 2020-10-08 [1] RSPM (R 4.0.2)   
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## desc 1.2.0 2018-05-01 [1] RSPM (R 4.0.3)   
## devtools 2.3.2 2020-09-18 [1] RSPM (R 4.0.3)   
## digest 0.6.25 2020-02-23 [1] RSPM (R 4.0.0)   
## ellipsis 0.3.1 2020-05-15 [1] RSPM (R 4.0.3)   
## evaluate 0.20 2023-01-17 [1] CRAN (R 4.0.2)   
## fansi 0.4.1 2020-01-08 [1] RSPM (R 4.0.0)   
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## htmltools 0.5.5 2023-03-23 [1] CRAN (R 4.0.2)   
## knitr 1.33 2023-03-28 [1] Github (yihui/knitr@a1052d1)   
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## memoise 2.0.1 2021-11-26 [1] CRAN (R 4.0.2)   
## ottrpal 1.0.1 2023-03-28 [1] Github (jhudsl/ottrpal@151e412)   
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## rlang 1.1.0 2023-03-14 [1] CRAN (R 4.0.2)   
## rmarkdown 2.10 2023-03-28 [1] Github (rstudio/rmarkdown@02d3c25)  
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## withr 2.3.0 2020-09-22 [1] RSPM (R 4.0.2)   
## xfun 0.26 2023-03-28 [1] Github (yihui/xfun@74c2a66)   
## yaml 2.2.1 2020-02-01 [1] RSPM (R 4.0.3)   
##   
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