# CSC 120: Applied Data Analytics

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- Instead of proposing a definition, I will draw some contrasts with classical statistics.

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Question: Which sleep-inducing drug works better?

### Data:

0.7
1.6
0.2
1.2
1.9
0.8
1.1
0.1

Method: Student's paired *t*-test

Results:

```
##
## Paired t-test
##
## data: dat$'Increase in Sleep'[dat$Drug == 1] and dat$':
## t = -4.0621, df = 9, p-value = 0.002833
## alternative hypothesis: true difference in means is not
## 95 percent confidence interval:
## -2.4598858 -0.7001142
## sample estimates:
## mean of the differences
##
                     -1.58
```

Answer: The difference in sleep times is unlikely to be due to chance. We can be reasonably confident that Drug 2 leads to more sleep.

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Question: How are individuals *genetically* predisposed towards asthma?

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- ➤ This sequence is nearly identical for each person, but there are sites in the DNA that vary among individuals. So individual A might have ATTCC{T}ATCGAA at a certain location while individual B has ATTCC{C}ATCGAA at the same location.

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- This kind of variation is known as a single nucleotide polymorphism or SNP.
- ▶ There are millions of SNPs in the human genome.

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- ▶ Data consists of the full genome for a number of individuals.
- Datasets can be enormous: typical studies include hundreds of thousands of SNPs for hundreds or thousands of individuals, and there have been studies including over a million individuals.

- ► The idea is to find the SNPs that are linked to the trait we're studying.
- Some traits are relatively simple, but a trait like asthma can have *hundreds* of causal SNPs.

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- ➤ Sequencing technology is never 100% accurate, so there needs to be quality control.
- Biased datasets have led to inaccurate conclusions: most GWASs have been over-represented by individuals of European ancestry, and results are not automatically generalizable to, say, individuals of African ancestry.

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- But there are problems that require new methods.
- ▶ For example, selecting SNPs that have p < .05 doesn't work when you are testing millions of them. This is the problem of **multiple testing**.

# Genome-wide association studies (GWAS)

▶ **Fine mapping** is another new and interesting problem: SNPs that are near one another on the genome are *correlated*, so it's tricky to find the ones that are truly causal:

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#### Data Science

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- ► All required software is freely available online.
- ► A (reliable) laptop is required.

#### Course Format

There will be a regular weekly routine:

- ► M/Tu: Lecture-based, with slides
- ▶ W: Discussion-based, with live code demonstrations
- ► F: Lab (collaborative problem solving)

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- Be familiar, at a conceptual level, with basic statistical principles of data modelling, including ethical issues and common misunderstandings.
- Have an in-depth understanding of the uses and abuses of one modelling technique (regression).

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- Have technical mastery in R (this is not a programming course).
- Have a detailed mathematical understanding of hypothesis testing, linear regression, etc. (this is not a statistics course).
- Be exposed to a panoply of machine learning techniques such as clustering, support vector machines, and neural networks. These techniques are very powerful, but they require a good statistical foundation if they are to be used responsibly.

#### Course Schedule

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# Assignments for This Week

- 1. Read through the syllabus on Canvas/GitHub. We can discuss on Friday if there are questions or concerns.
- Sign up for a GitHub account and complete the software setup detailed in Assignment 1. Troubleshooting on Friday.
- 3. Request your personal data (Assignment 0). Due Tuesday.