

Bioinformatics

CS300

Chapter 1:

Genetic disorders and Data

Spring 2021
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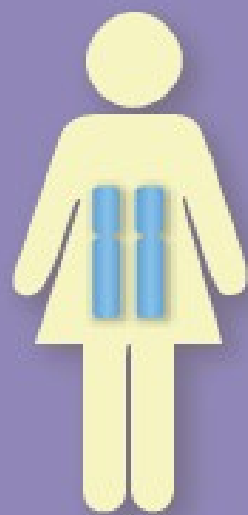


ALLEGHENY
COLLEGE

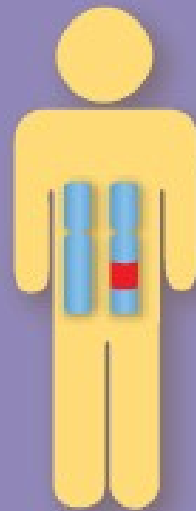
Where Do Some of These Mutations Come From?



Autosomal Dominant Inheritance



MOM DAD



Normal

Affected

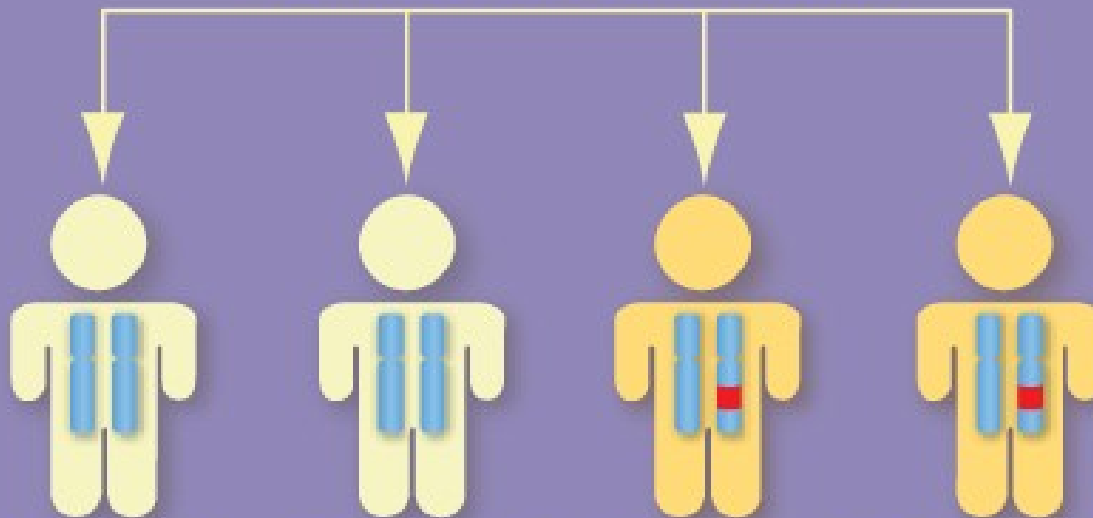


Chromosome with
normal copy of gene



Chromosome with
defective copy of gene

Possible combinations:



Normal

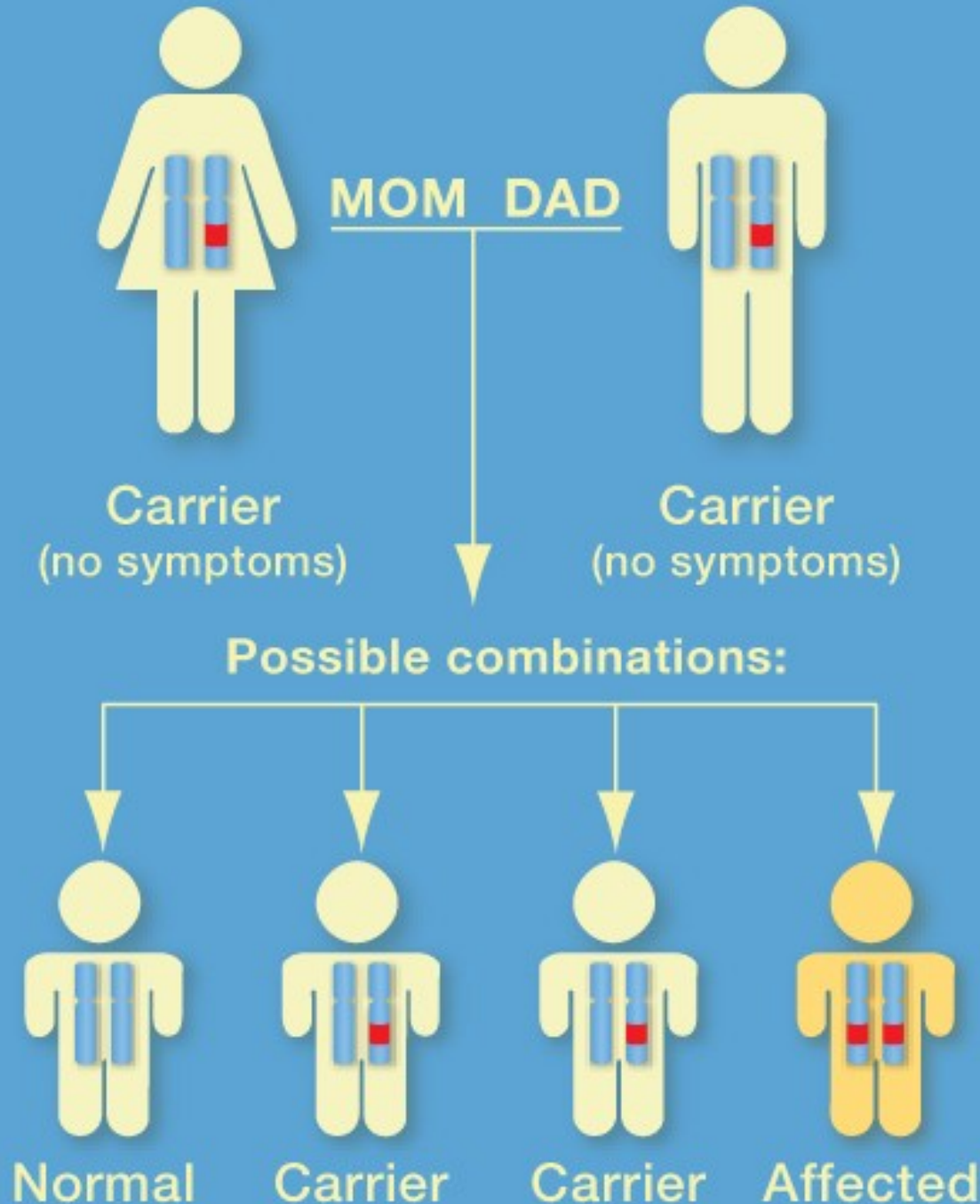
Normal

Affected

Affected

Each child inherits a normal copy from Mom and either a normal or a defective copy from Dad.

Autosomal Recessive Inheritance



- Chromosome with normal copy of gene
- Chromosome with defective copy of gene

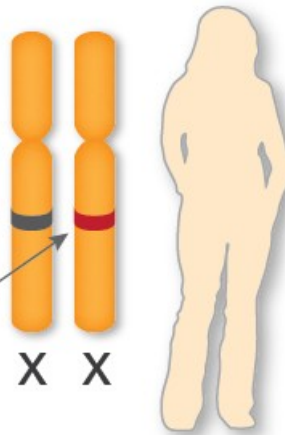
Each child inherits one copy of the gene from each parent.

X-Linked Inheritance

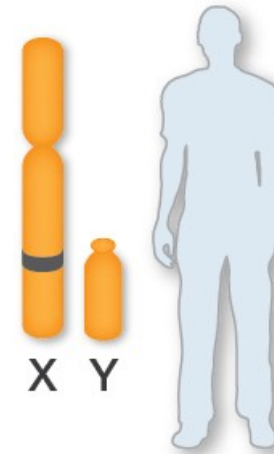
Parents:

Color vision gene

- Normal allele →
- Defective allele →

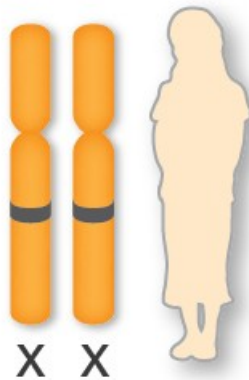


Normal vision
(*Colorblindness carrier*)

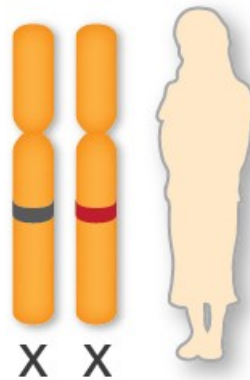


Normal vision

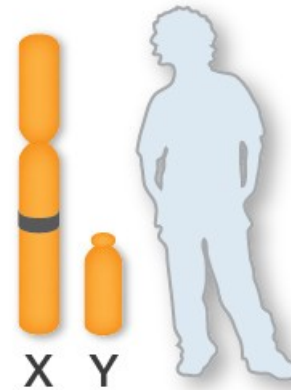
Possible offspring:



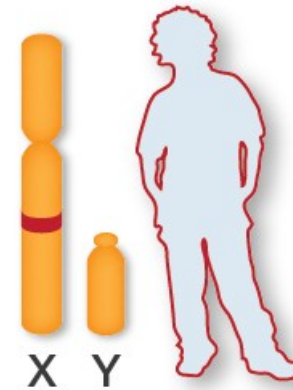
Normal vision



Normal vision
(*Colorblindness carrier*)



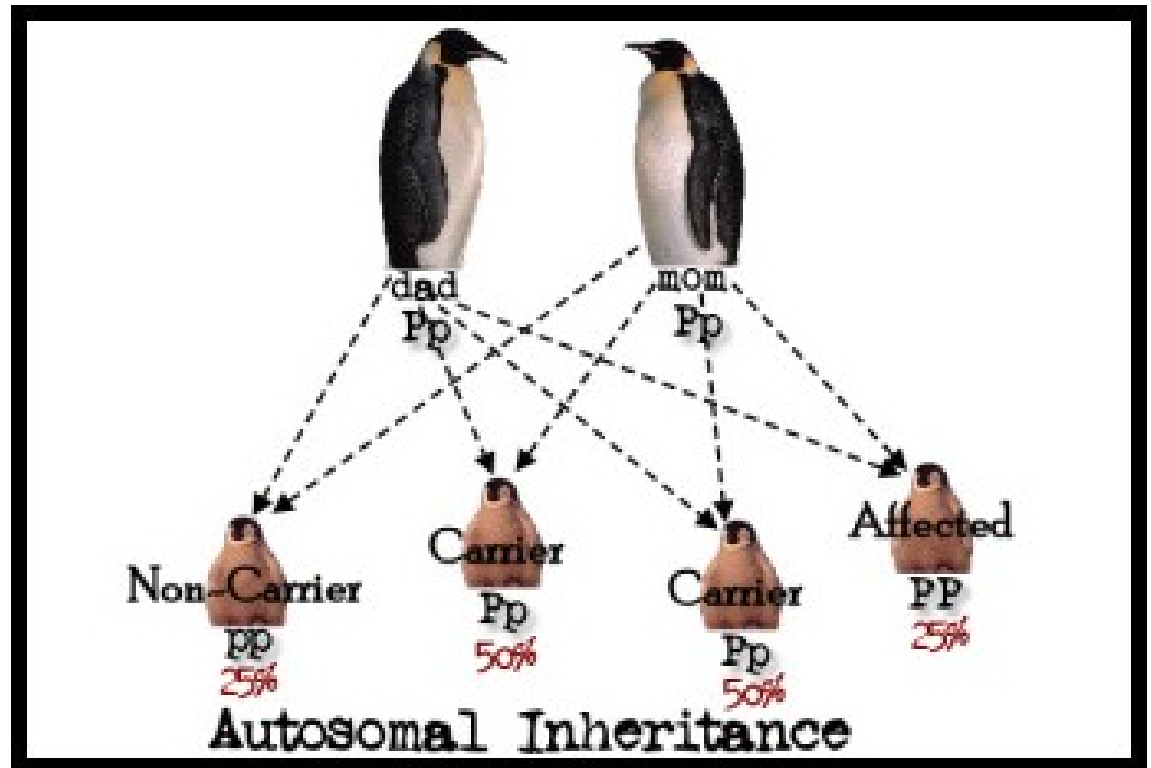
Normal vision



Colorblind

Single Gene Disorders

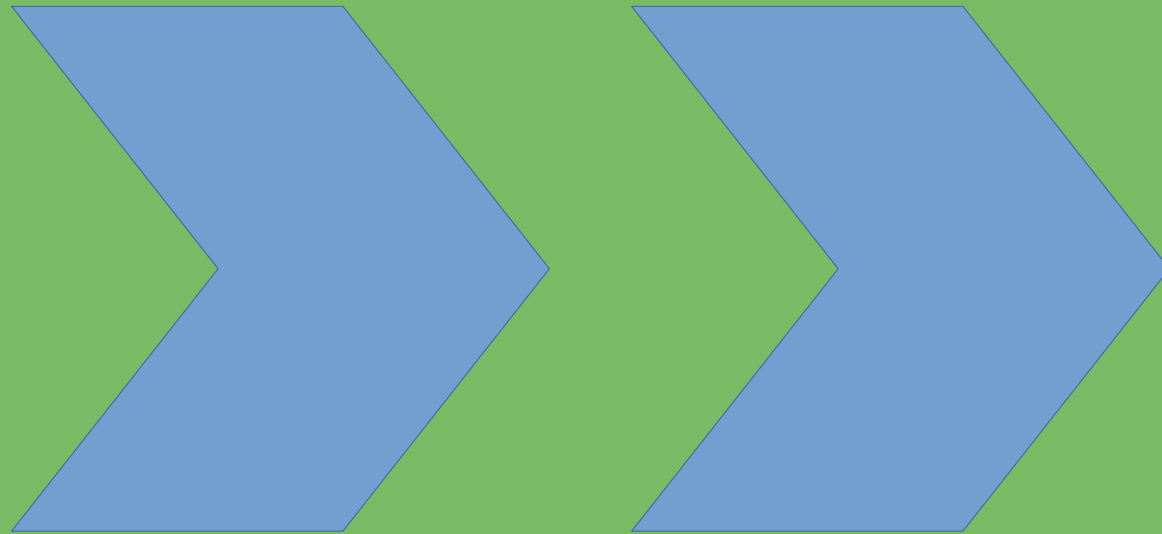
- Inheritance patterns are relatively simple
- Chances of inheritance in the text generation can be predicted by studying patterns in past generations.





ALLEGHENY
COLLEGE

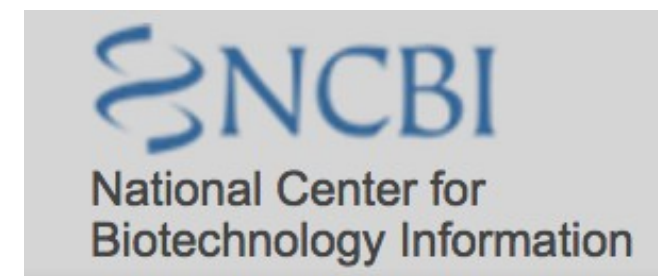
Bring the Data!



Up Next!

Sources of Data

- Typically Protein: **Uniprot**
 - <http://www.uniprot.org/>
 - Search: Pink1 (protein)
-
- Typically DNA and Genes: **National Center for Biotechnology Informatics (NCBI)**
 - <https://www.ncbi.nlm.nih.gov/>
 - Search: “orchid” in Nucleotide database
 - (https://www.ncbi.nlm.nih.gov/nuccore/NC_030915.1)





Let's Talk About It!

- Go to *Links to bioinformatics resources* at link
https://www.cs.alleggheny.edu/sites/bonhamcarter/bioinfo_i.html
- Find and pick an online database or analysis platform to investigate with your group
- Discuss the resources.
 - What use(s) does the resource have?

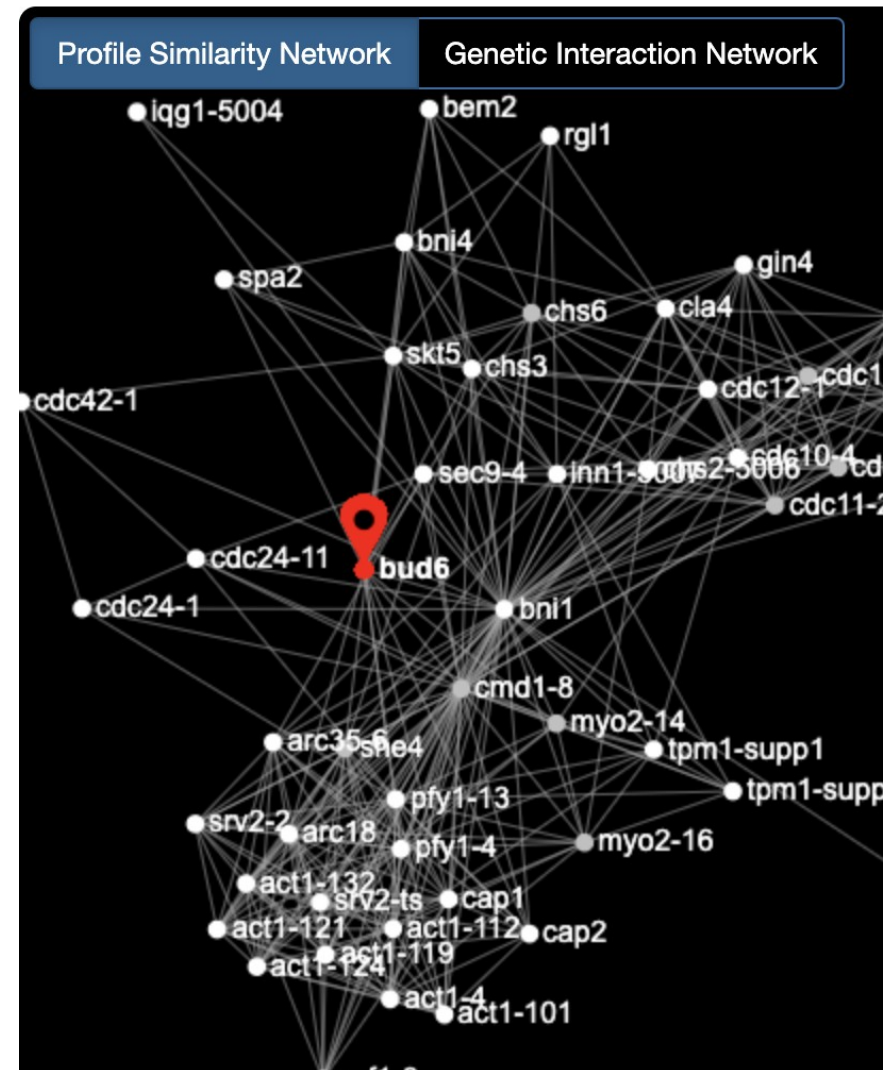


Links to bioinformatics resources

Let's Talk About It!

- Is data offered? Is it tools to work with data?
- Research interests?
- **Activity 4:** Complete the Google form;
<https://forms.gle/nKcJjcgG9gW9Z7ur6>
- A check mark grade

THINK



Genetic Networks from the *Cell Map*