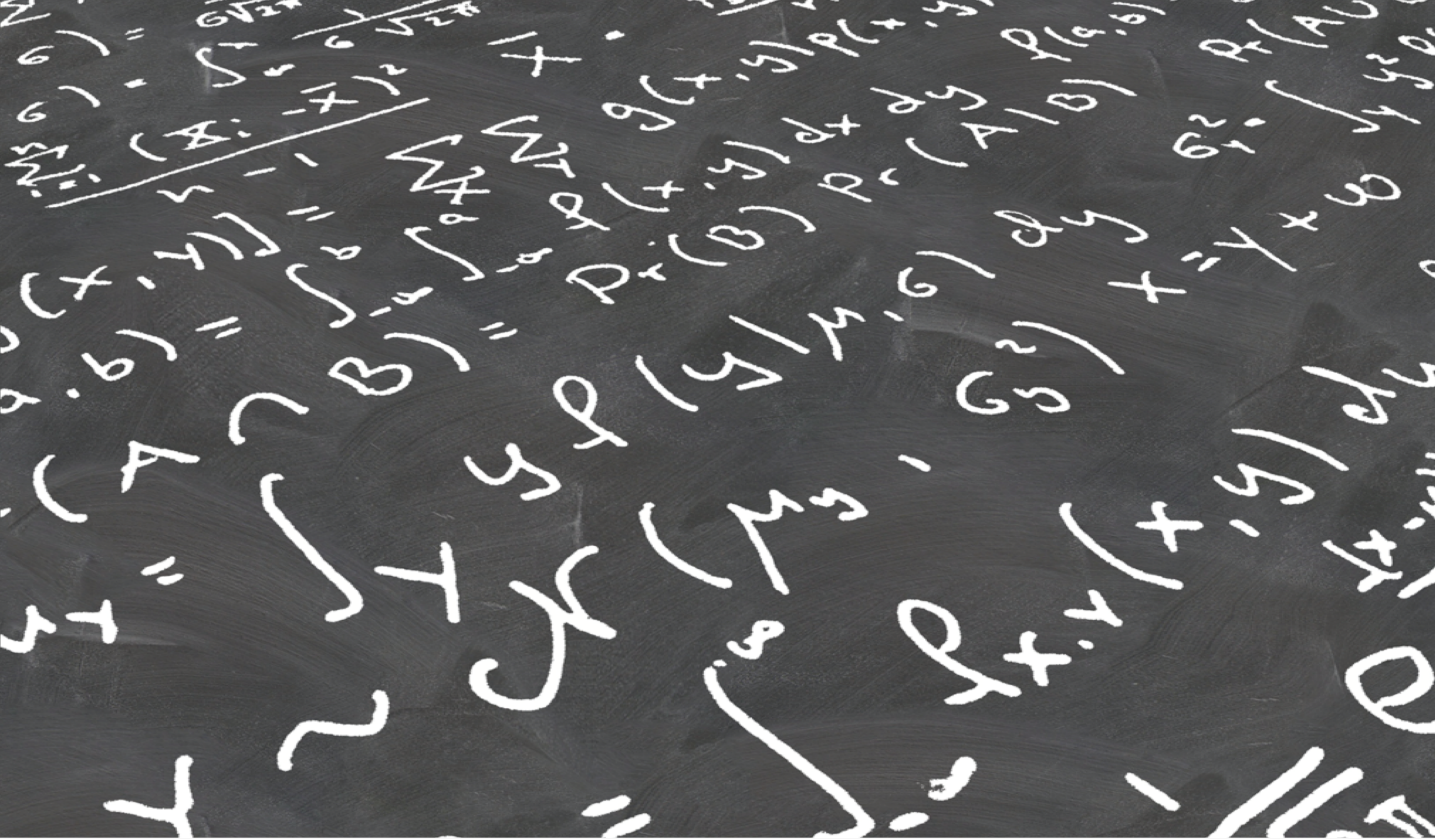

1. Introduction to Probability Theory

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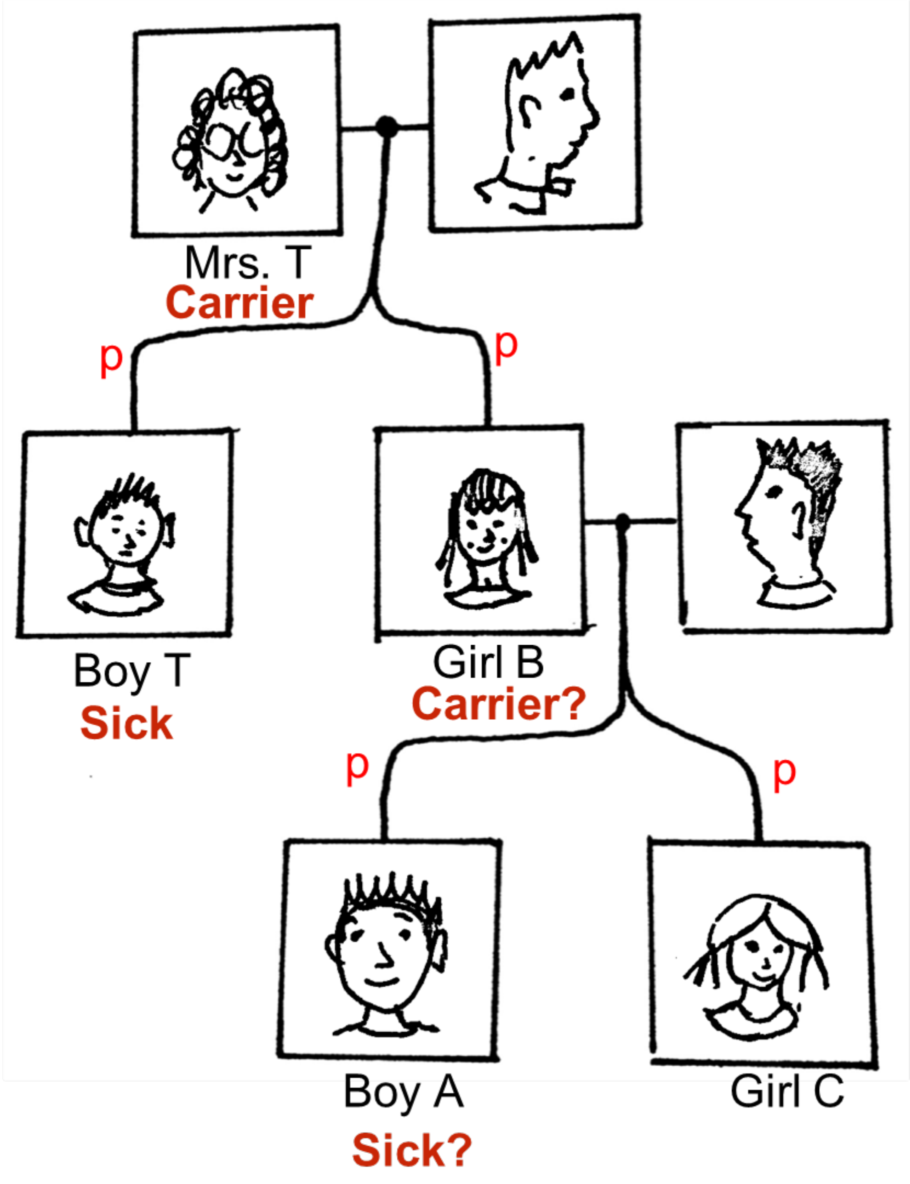
Todays Content

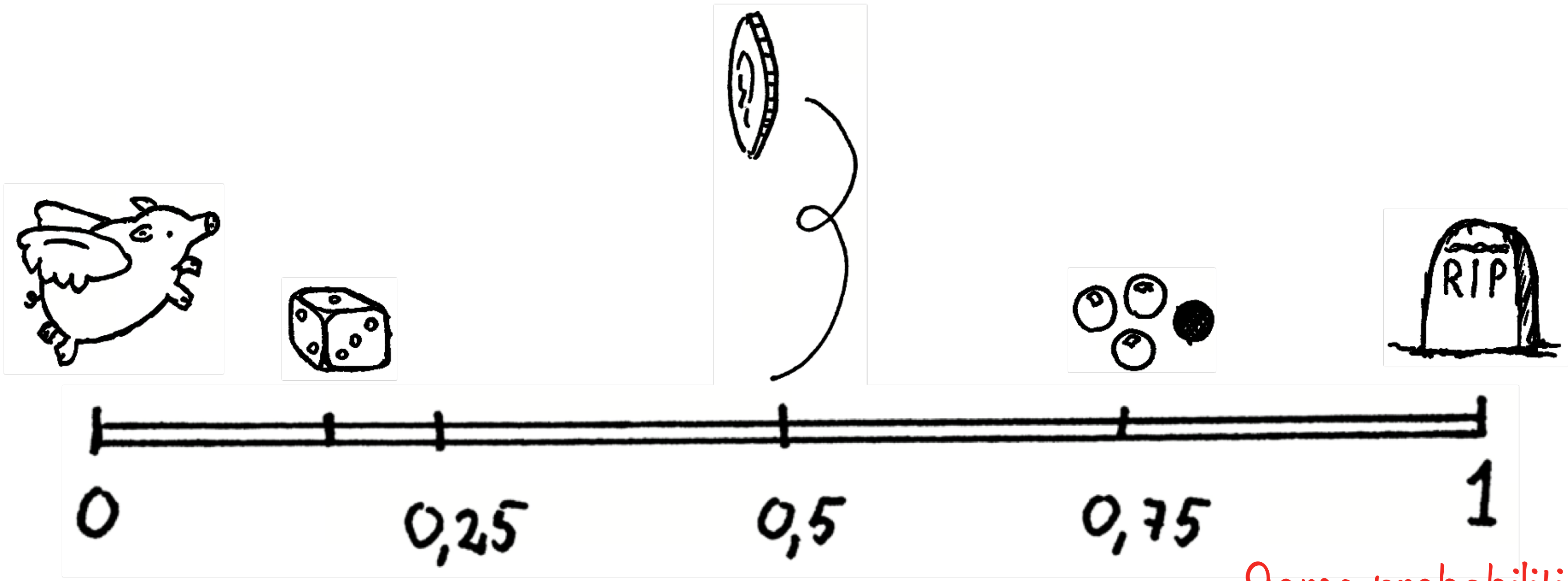
- Introduction to Probability Theory
- Definitions, concepts and notation
- Relative Frequency Approach
- Set theory
- Basic Axions on probabilities

Example - X linked recessive disease

→ Conditional probabilities
→ Don't (only) rely on logic
→ Systematic calculations



Probability Line



Some probabilities you know

- All probabilities are numbers between 0 and 1.
- In percentage, between 0% to 100%.
- We begin with one sample point.

Words to Know

- Experiment/trial (*Forsøg/test*) — Roll a dice
- Sample space (*Udfaldsrum*) — S={1,2,3,4,5,6}
- Sample point (*Bestemt udfald*) — a={4}
- Event (*Hændelse*) — A={2,4,6} (even number)
    - Elementary event — Event that has one possible outcome
    - Joined event — Event that has many possible outcomes
    - Simultaneous event — Event with two or more sub trials

Relative Frequency Approach

- The number of times event A occurs: N_A

- The number of times that all events occur (sample space):

$$N = N_A + N_B + N_C + \cdots$$

- Then we have the relative frequency:

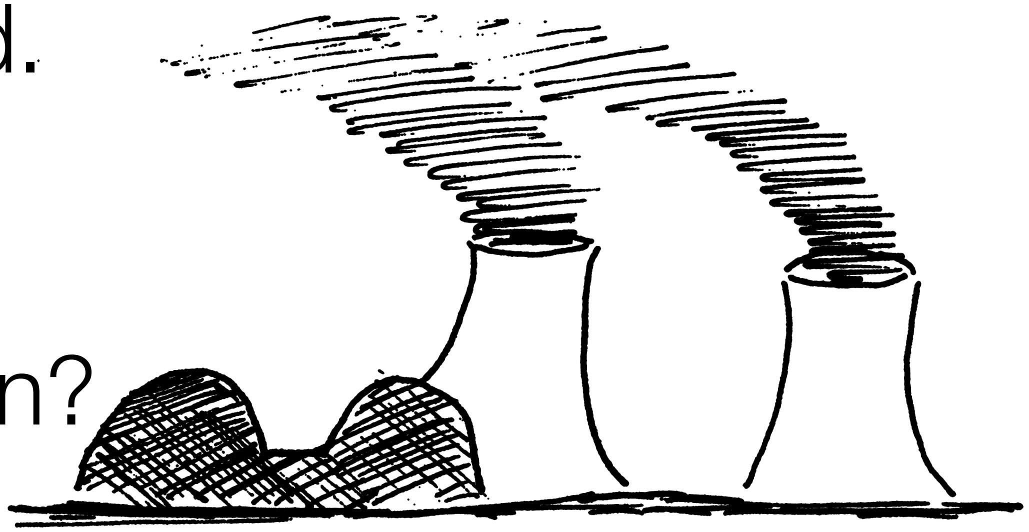
$$\boxed{Pr(A) \sim r(A) = \frac{N_A}{N}}$$

All sample points should have the same a priori probability

- Where: $Pr(A) = \lim_{N \to \infty} r(A)$

Risk of a Meltdown

- There are 437 reactors in the world.
- ~153M operating reactor hours.
- ~Four reactor meltdowns.
- What are the chance of a meltdown?



$$\frac{4}{153M} \text{ pr. reactor pr. hour}$$

$$\sum_{n=1}^{437} \frac{4}{153M} = \frac{1}{87600} \text{ pr. hour}$$

$$\frac{24*365}{87600} = \frac{1}{10} \text{ pr. year}$$

→ Be carefull: Small samples, small probabilities, circumstances, etc.

→ Very uncertain: If number of reactors > 4370 → Pr(Meltdown)>1

Set Theory (*Mængdelære*)

A set:

- A collection of things.
- Elements of sets are not ordered.

Name of set

Some more elements

$$E = \{\alpha_1, \ldots, \alpha_n\}$$

Element

Example:

- The set of all persons in a drug trial group.
- The number of cars i DK.
- All numbers.
- All colours.

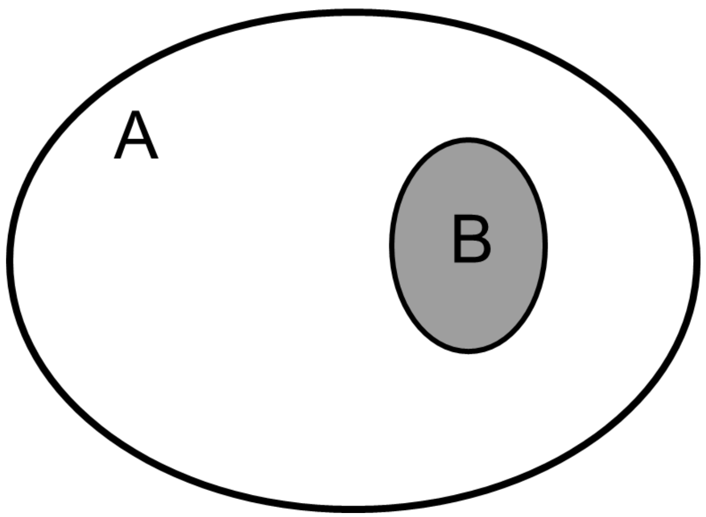
A Subset to a Set *(Delmængder)*

- A subset is any set, where all elements are included in the original set

Notation:

B is a subset to A:

$$B \subset A$$



Example:

For a set $A = \{blue, red, green\}$

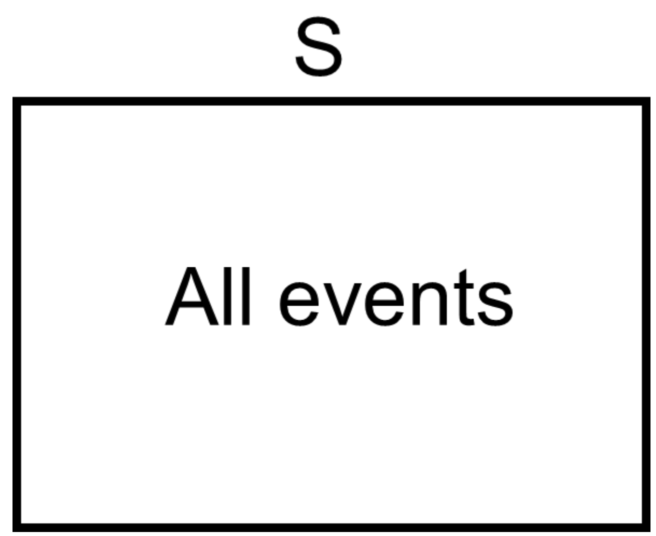
we have a subset $B \subset A$ if B is in A,

e.g. $\{blue, red\}$,$\{blue, red, green\}$,$\{green\}$, $\{\}$

The Sample Space (*Udfaldsrum*)

- The sample space contains all possible events.
- The probability that a sample is from the sample space is 1.

$\Pr(S)=1$

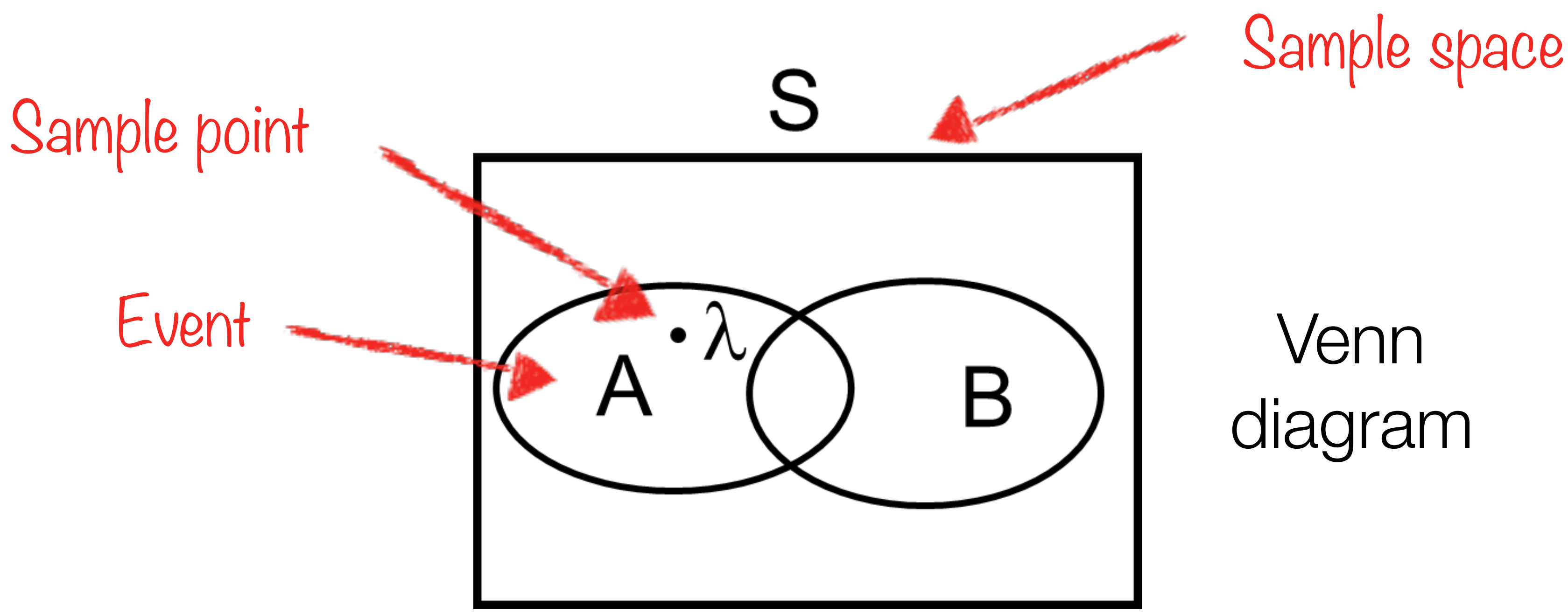


Example:

- A sample space contains 20 people
- 8 has a given disease, 12 is healthy
- Draw a random person.
- What is the chance that a person is a person?
- What is the chance that a person is sick?

A Sample Point *(Udfald)*

- An elementary event.
- Events are collections of sample points.
- Sample space is the collection of all possible sample points.
- Sample points are not ordered.



Venn diagram

Example:

Throw of a dice:

Possible outcomes: 1,2,3,4,5,6 → S={1,2,3,4,5,6}

Events: A={1,2,3} and B={2,4,6}; A⊂S; B⊂S

Basic Axions of Probability

- The probability of a sample point (element of a sample space).
- The probability of a event (E) (collection of sample points).
- All sample points of a probability space (S) sums up to 1.

➢ Basic Axions of Probability:

Axion 1: $0 \leq Pr(E) \leq 1$

Axion 2: $Pr(S) = 1$

The Empty Set (*Den tomme mængde*)

- The empty set is always a subset of any set.
- This corresponds to the impossible event.

$$\varnothing = \{\}$$

The null set

- The probability of the impossible event is 0.

Example:

- The set of boys in an all girlschool.
- The chance of pigs growing wings and fly.
- To get an 8 when rolling a dice.

Summary

The certain event

S is the certain set.

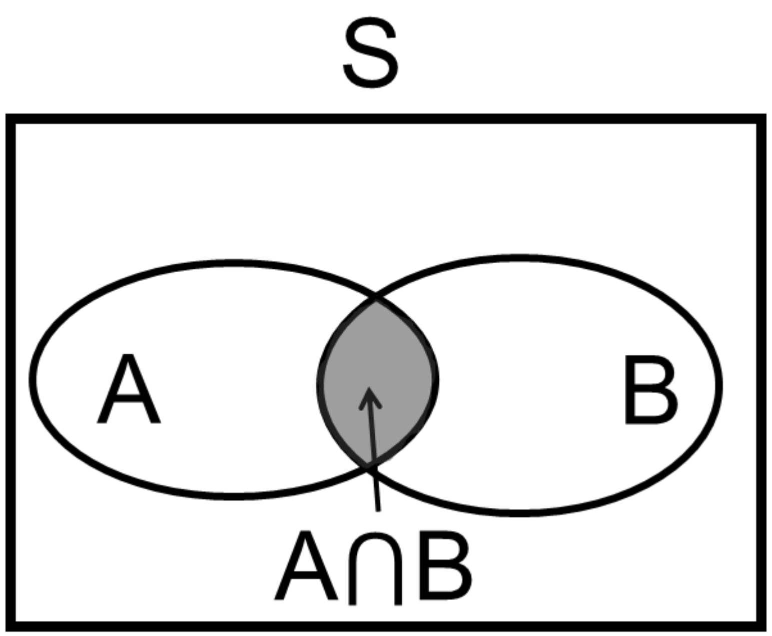
$\varnothing$ is the empty set.

The impossible event

Joint Events (*Fællesmængde*)

- The intersection $A \cap B$ are the common elements of the events A and B
- $A \cap B$ means A <u>and</u> B.

Venn diagram

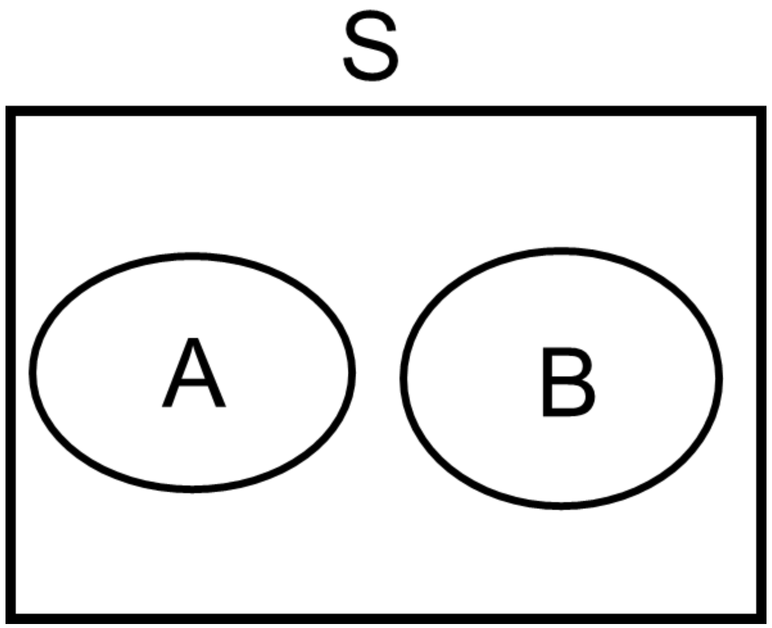


Example:

- A is the event of VW cars i DK
- B is the event of red cars in DK
- The intersection of the events is all red VW in DK.

Mutually Exclusive (Disjoint) Events *(Disjunkte)*

- The sets of A and B are disjoint if: $A \cap B = \emptyset$



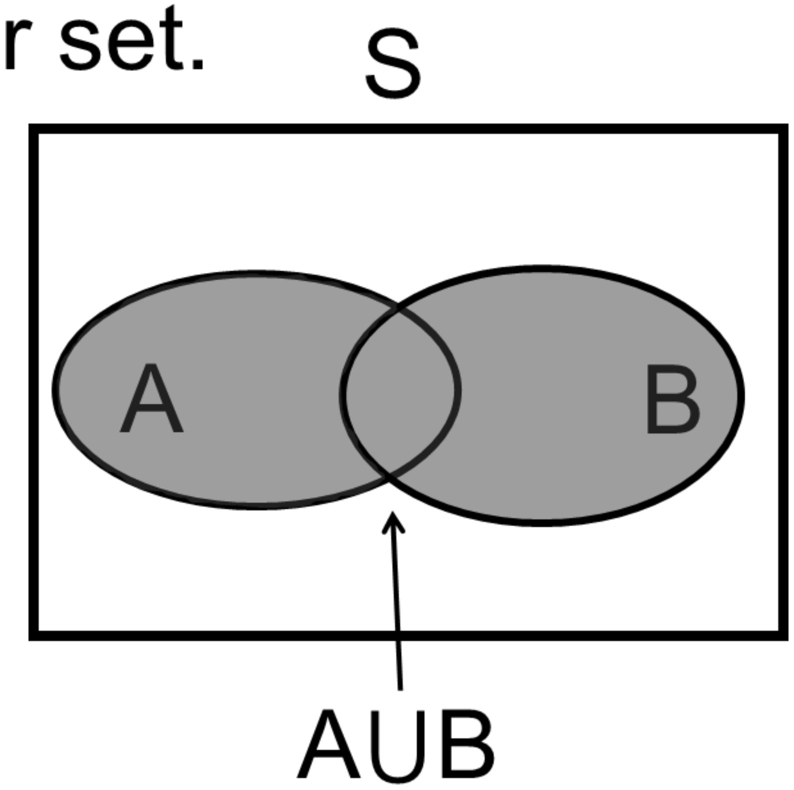
$$A \cap B = \varnothing$$

Example:

- Event A: The child is a girl.
- Event B: The child is a boy.

Union of Events (*Foreningsmængde*)

- The union of events $A \cup B$ are all the events in one set 'plus' the events in the other set.
- $A \cup B$ means A or B.
- $A \cup B = A + B - A \cap B$

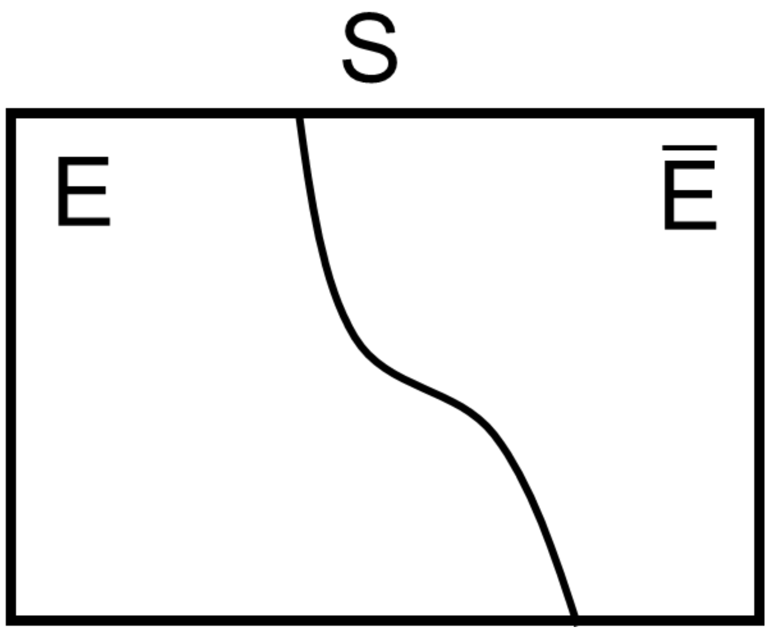


Example:

- I can choose between oatmeal (A) and cornflakes (B) for breakfast.
- The union of the events is that I had breakfast.

The Complement Event (*Komplementær*)

Notation: $S \setminus E = \overline{E} = E^c$ "not-E"



Notice:

$$E \cup \bar{E} = S$$

The certain event

$$E \cap \bar{E} = \emptyset$$

The impossible event

Example:

- The complement of having a disease is not having a disease

Calculation Rules for Set Theory

Commutative law

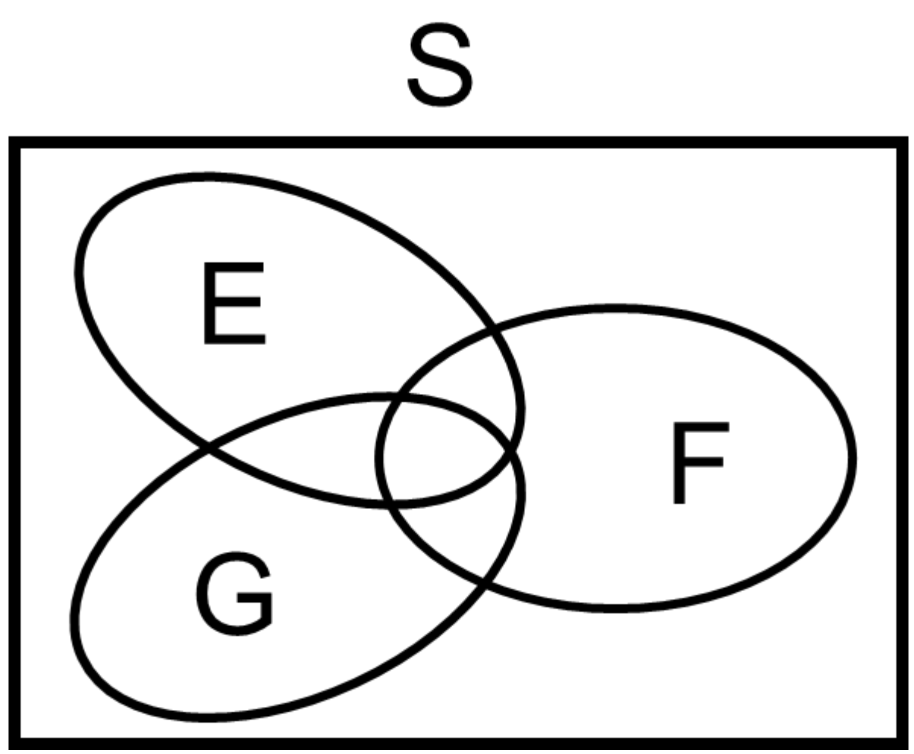
$$E \cup F = F \cup E$$

Associative law

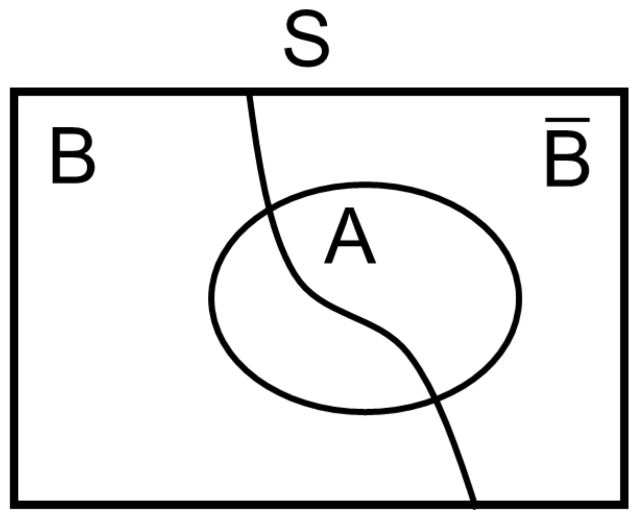
$$E \cup (F \cup G) = (E \cup F) \cup G$$

$$(E \cup F) \cap G = (E \cap G) \cup (F \cap G)$$

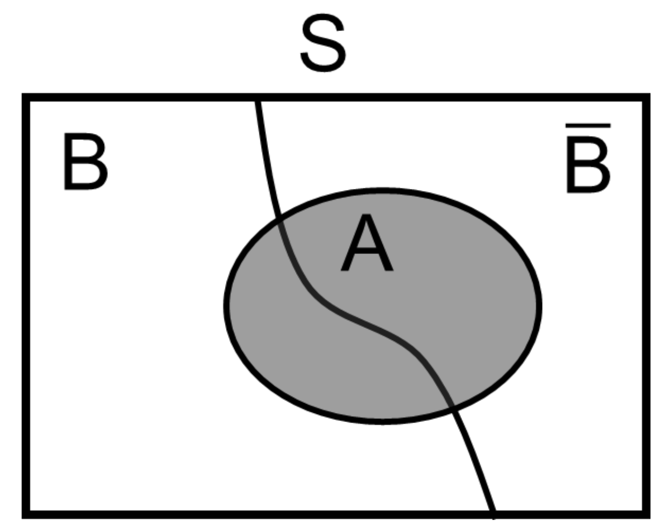
Distributive law



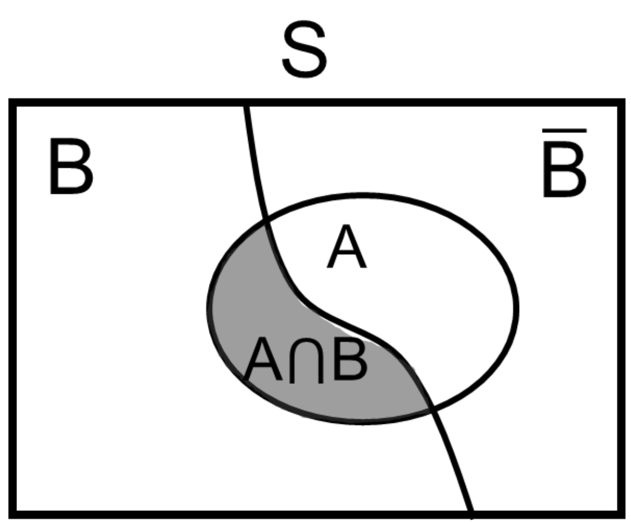
Probability of joint events



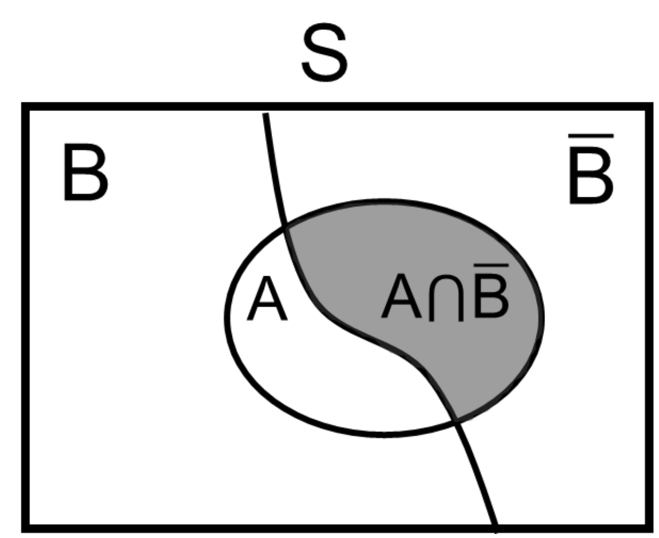
Venn diagram



$$\Pr(A) = \frac{N_A}{N_S}$$



$$\Pr(A \cap B) = \frac{N_{A \cap B}}{N_S}$$



$$\Pr(A \cap \bar{B}) = \frac{N_{A \cap \bar{B}}}{N_S}$$

Independence *(Uafhængighed)*

- We define that two events are independent if and only if:

$$\boxed{Pr(A \cap B) = Pr(A) \cdot Pr(B)}$$

Notice:

- This does not apply if the events A and B are dependent.

Example:

- Two throws with a dice
- The gender of two siblings

Conditional Probability (*Betingede sandsynligheder*)

- We write a conditional probability as:

$$\boxed{Pr(A|B)}$$

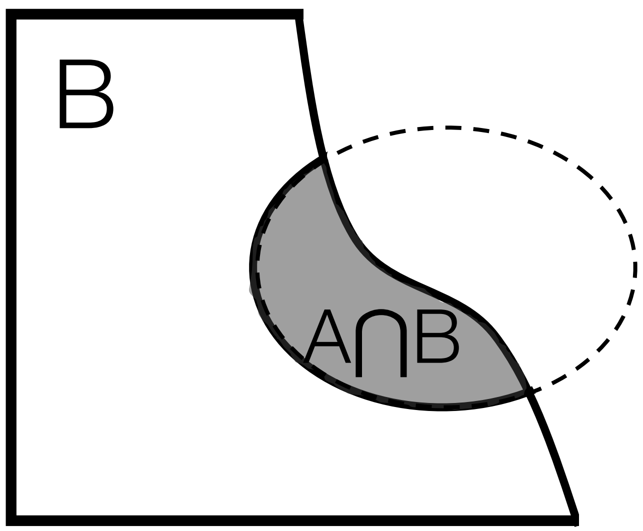
"A given B"

- This means that if the event B has already happened, what is the probability of the event A.
- Reduction of the sample space (possible events) from S to B

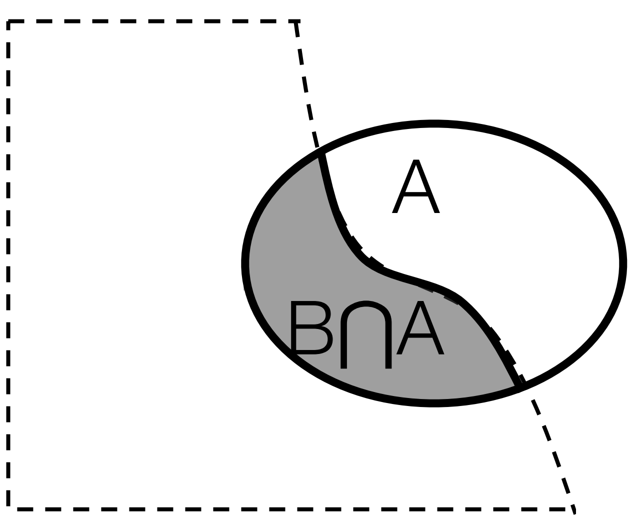
Example:

- From a population, I have selected a female.
- What is the chance that the selected person is below 1.6 m in height?

Conditional Probabilities – Bayes Rule



$$\Pr(A|B) = \frac{N_{A\cap B}}{N_B} = \frac{N_{A\cap B}/N_S}{N_B/N_S} = \frac{\Pr(A \cap B)}{\Pr(B)}$$



$$\Pr(B|A) = \frac{N_{B\cap A}}{N_A} = \frac{N_{B\cap A}/N_S}{N_A/N_S} = \frac{\Pr(B \cap A)}{\Pr(A)}$$

Probabilities of a Joint Event

- We can calculate the probability of a joint event

$$\boxed{Pr(A \cap B) = Pr(A|B) \cdot Pr(B) = Pr(B|A) \cdot Pr(A)}$$

Notice:

- We can extend this rule to multiple events.
- Joint events are not the same as conditional events

- If A and B independent:

$$\boxed{Pr(B|A) = Pr(B) \quad \text{and} \quad Pr(A|B) = Pr(A)}$$

Bayes Rule

Very important!

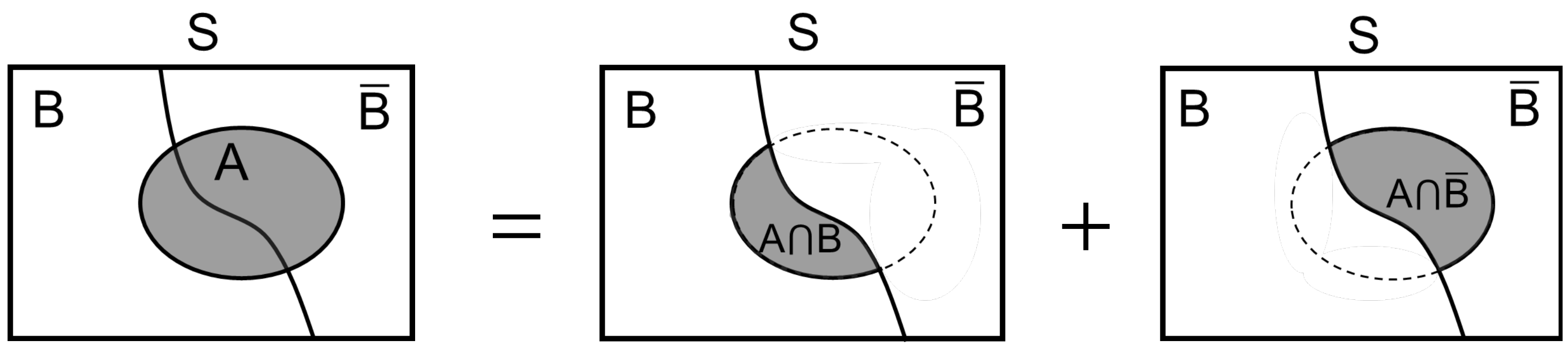
- We can write Bayes rule for two events as:

$$\boxed{Pr(B) \cdot Pr(A|B) = Pr(A) \cdot Pr(B|A)}$$

or

$$\boxed{Pr(A|B) = \frac{Pr(B|A) \cdot Pr(A)}{Pr(B)} = \frac{Pr(A \cap B)}{Pr(B)}}$$

Conditional Probabilities – Total Probability



$$\Pr(A) = \frac{N_A}{N_S} = \frac{N_{A\cap B}}{N_S} + \frac{N_{A\cap \bar{B}}}{N_S} = \frac{N_{A\cap B}}{N_B}\cdot\frac{N_B}{N_S} + \frac{N_{A\cap \bar{B}}}{N_{\bar{B}}}\cdot\frac{N_{\bar{B}}}{N_S}$$

$$\boxed{\begin{aligned}\Pr(A) &= \Pr(A\cap B) + \Pr(A\cap \bar{B}) \\ &= \Pr(A|B)\cdot\Pr(B) + \Pr(A|\bar{B})\cdot\Pr(\bar{B})\end{aligned}}$$

Conditional Probabilities - Example

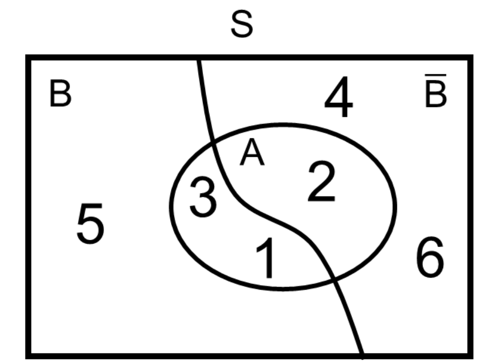
Rolling a dice:

Sample space: S={1,2,3,4,5,6}

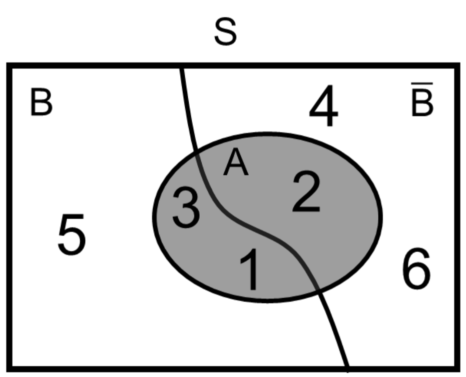
Events: A={1,2,3}

B={1,3,5}

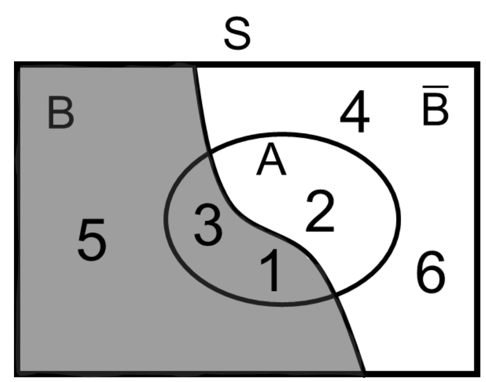
B̄={2,4,6}



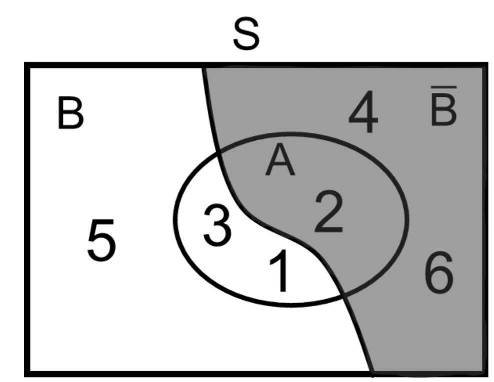
Venn diagram



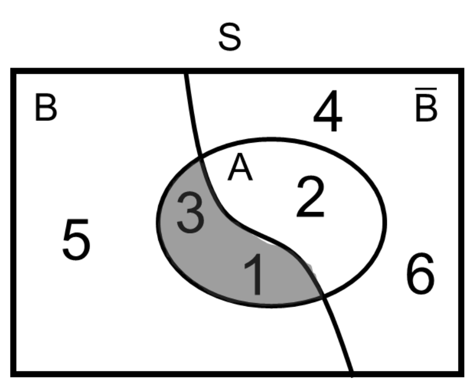
$$\Pr(A) = \frac{N_A}{N_S} = \frac{3}{6} = \frac{1}{2}$$



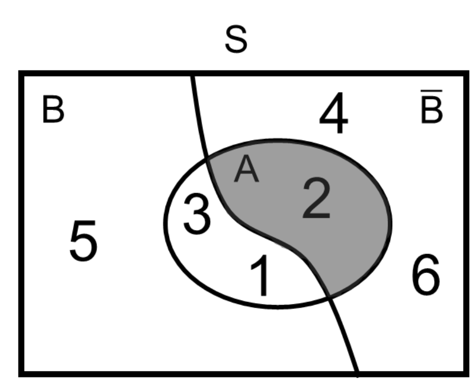
$$\Pr(B) = \frac{N_B}{N_S} = \frac{3}{6} = \frac{1}{2}$$



$$\Pr(\bar{B}) = \frac{N_{\bar{B}}}{N_S} = \frac{3}{6} = \frac{1}{2}$$

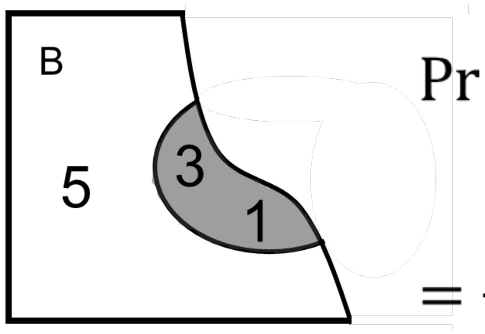


$$\Pr(A \cap B) = \frac{N_{A \cap B}}{N_S} = \frac{2}{6} = \frac{1}{3}$$



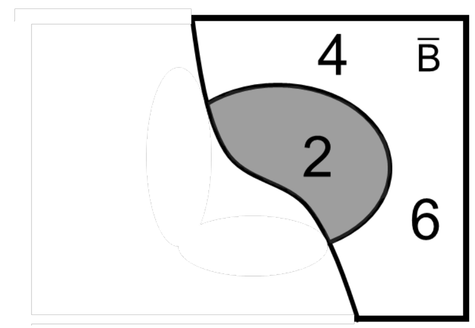
$$\Pr(A \cap \bar{B}) = \frac{N_{A \cap \bar{B}}}{N_S} = \frac{1}{6}$$

Conditional Probabilities - Example



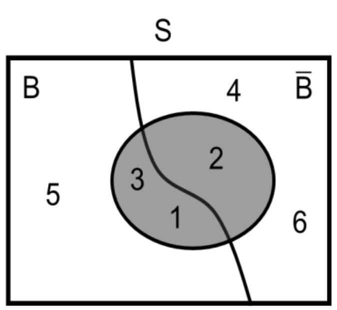
$$\Pr(A|B) = \frac{N_{A\cap B}}{N_B} = \frac{2}{3}$$

$$= \frac{\Pr(A\cap B)}{\Pr(B)} = \frac{1/3}{1/2} = \frac{2}{3}$$



$$\Pr(A|\bar{B}) = \frac{N_{A\cap \bar{B}}}{N_{\bar{B}}} = \frac{1}{3}$$

$$= \frac{\Pr(A\cap \bar{B})}{\Pr(\bar{B})} = \frac{1/6}{1/2} = \frac{2}{6} = \frac{1}{3}$$



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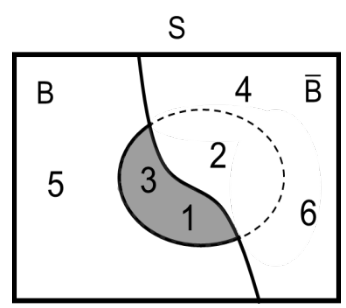

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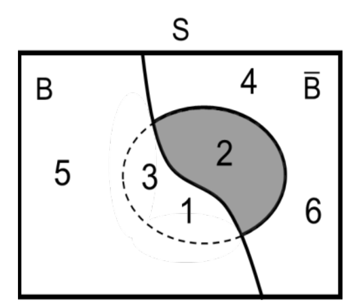


$$\Pr(A) = \frac{N_A}{N_S} = \frac{N_{A\cap B}}{N_S} + \frac{N_{A\cap \bar{B}}}{N_S} = \frac{N_{A\cap B}}{N_B}\cdot\frac{N_B}{N_S} + \frac{N_{A\cap \bar{B}}}{N_{\bar{B}}}\cdot\frac{N_{\bar{B}}}{N_S}$$

$$= \frac{3}{6} = \frac{2}{6} + \frac{1}{6} = \frac{2}{3}\cdot\frac{3}{6} + \frac{2}{3}\cdot\frac{3}{6} = \frac{1}{2}$$

$$\Pr(A) = \Pr(A\cap B) + \Pr(A\cap \bar{B}) = \Pr(A|B)\cdot\Pr(B) + \Pr(A|\bar{B})\cdot\Pr(\bar{B}) = \frac{2}{3}\cdot\frac{1}{2} + \frac{1}{3}\cdot\frac{1}{2} = \frac{3}{6} = \frac{1}{2}$$

Markov Properties

- Bayes rule for tree events:

$$\boxed{Pr(A \cap B \cap C) = Pr(A) \cdot Pr(B|A) \cdot Pr(C|B, A)}$$

- For a Markov chain, it holds that:

$$\boxed{Pr(A \cap B \cap C) = Pr(A) \cdot Pr(B|A) \cdot Pr(C|B)}$$

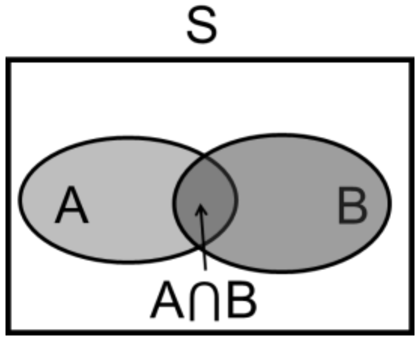
i.e. $$\boxed{Pr(C|B, A) = Pr(C|A \cap B) = Pr(C|B)}$$

(*A* don't give new information)

Probabilities of a Union of Event

- We can calculate the probability of a union of events:

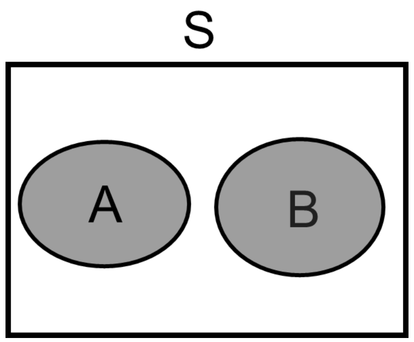
$$Pr(A \cup B) = Pr(A) + Pr(B) - Pr(A \cap B)$$



Notice:

- If the events are mutually exclusive

$$Pr(A \cup B) = Pr(A) + Pr(B)$$



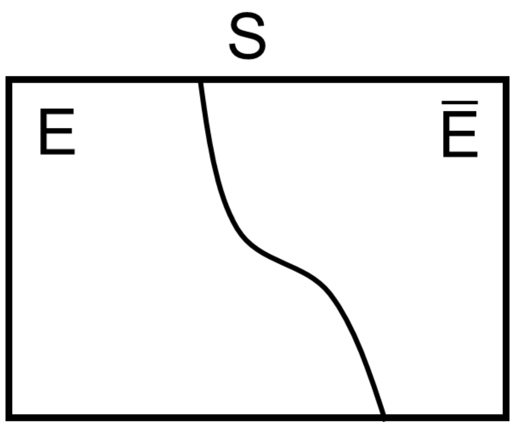
Probabilities of Complement Events

- We can write some rules for the probabilities of a complement event

$$Pr(E \cup \bar{E}) = Pr(S) = 1$$

$$Pr(E) + Pr(\bar{E}) = Pr(S) = 1$$

$$\boxed{Pr(E) = 1 - Pr(\bar{E})}$$



Example:

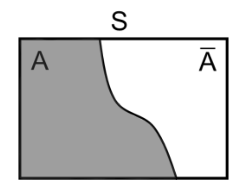
- The probability of not hitting 2 eyes on dice.

$$Pr(\{1,3,4,5,6\}) = 1 - Pr(\{2\}) = 1 - \frac{1}{6} = \frac{5}{6}$$

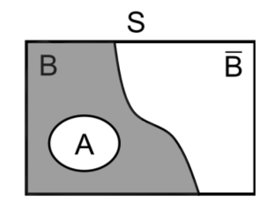
Summary of Probability

Relative frequency: $Pr(A) = \frac{N_A}{N_S}$

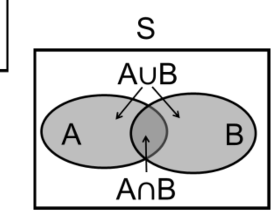
Complement: $Pr(\overline{A}) = 1 - Pr(A)$



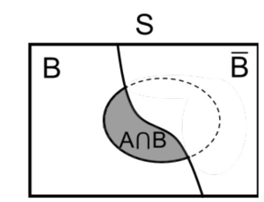
Exclusive: $Pr(\overline{A} \cap B) = Pr(B) - Pr(A)$ if $A \subset B$



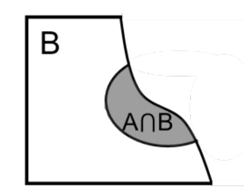
Union: $Pr(A \cup B) = Pr(A) + Pr(B) - Pr(A \cap B)$



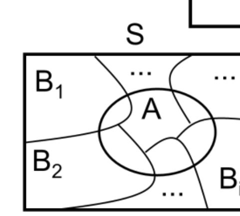
Joint: $Pr(A \cap B) = Pr(A|B) \cdot Pr(B) = Pr(B|A) \cdot Pr(A)$



Conditional: $Pr(A|B) = \frac{Pr(A \cap B)}{Pr(B)}$ if $Pr(B) \neq 0$



Total probability: $Pr(A) = \sum_{i=1}^{n} Pr(A|B_i) \cdot Pr(B_i)$



Bayes rule: $Pr(B|A) = \frac{Pr(A|B) \cdot Pr(B)}{Pr(A)}$

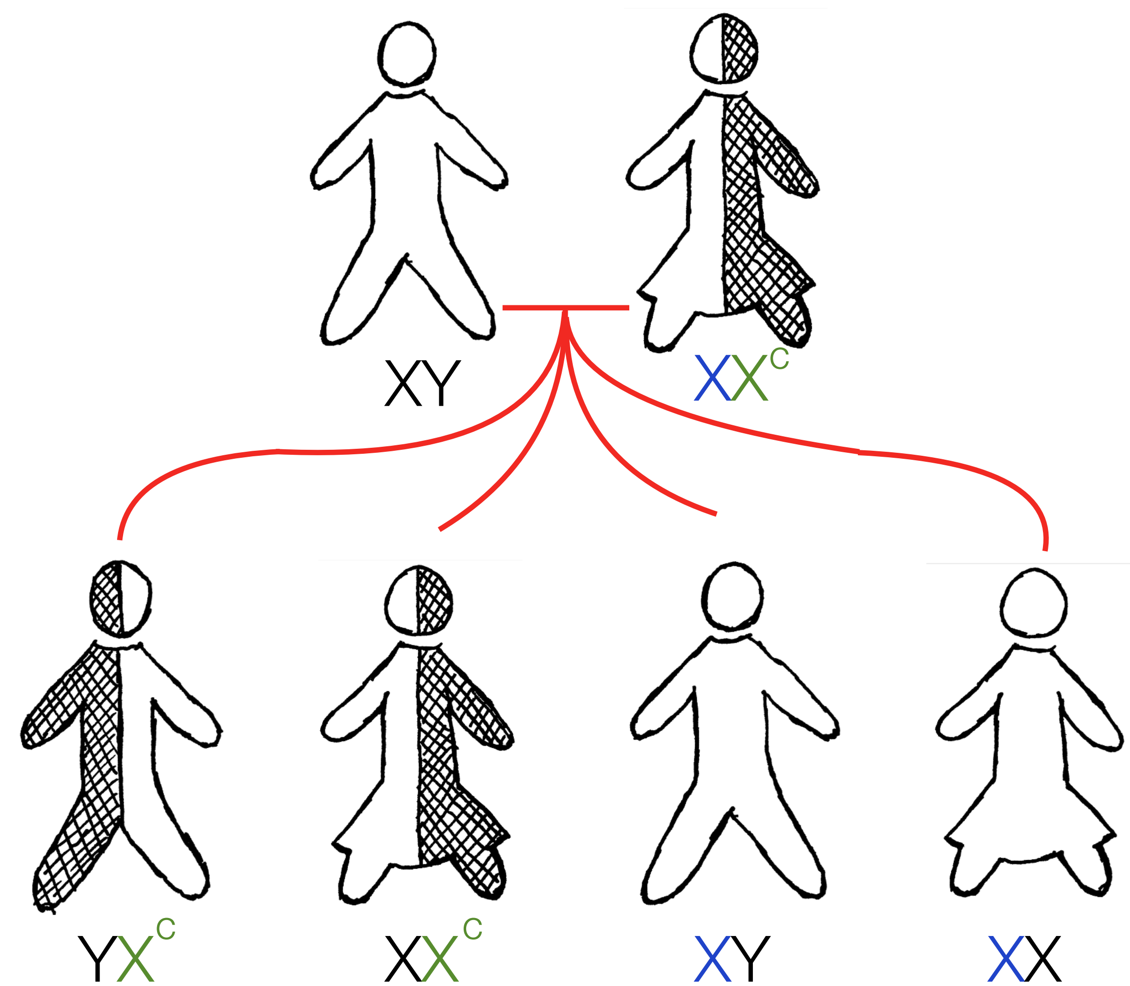
Bayes formula: $Pr(B_i|A) = \frac{Pr(A|B_i) \cdot Pr(B_i)}{\sum_{i=1}^{n} Pr(A|B_i) \cdot Pr(B_i)}$

Independence: $Pr(A \cap B) = Pr(A) \cdot Pr(B)$

Neutralized by a healthy X-gene

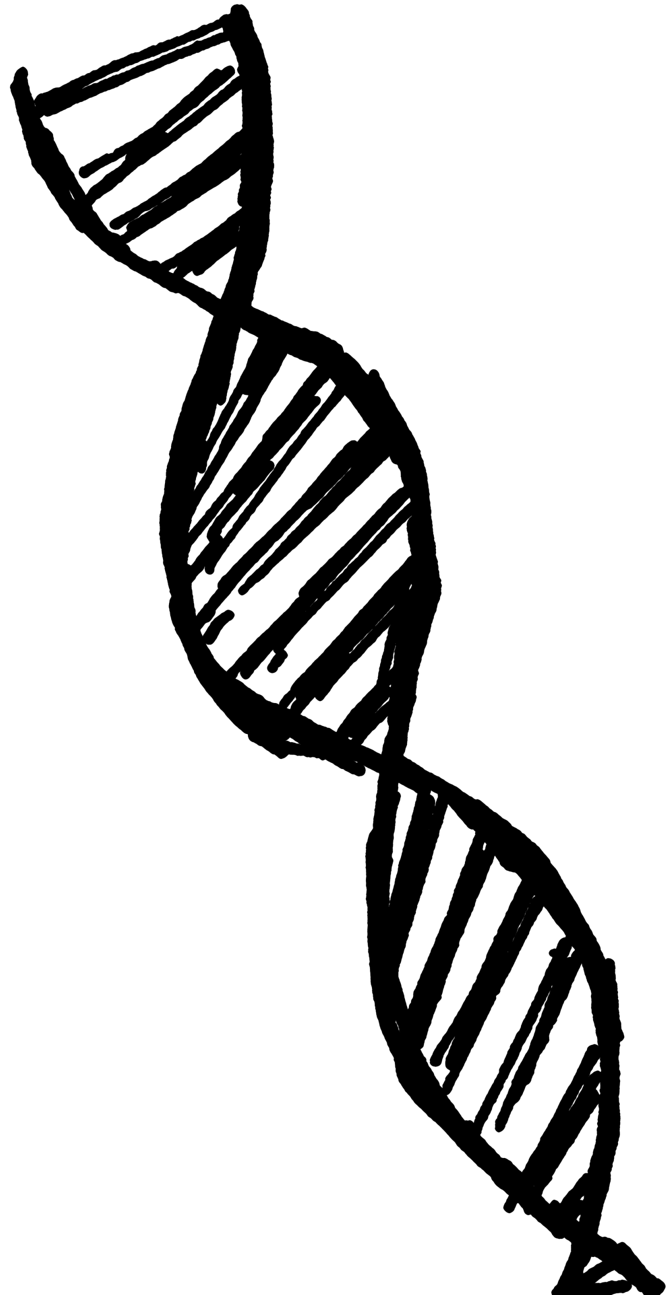
Example - X linked recessive

- A mother has a sick X gene.
- The chance of giving the sick X gene to a child is 50%.
- A boy with the sick X gene has the disease.
- A girl with the sick X gene is a carrier of the disease.
- Of cause the chance of not giving the sick X gene to a child is also 50%.



Hunter Syndrome (MPS II)

- X linked recessive
- 1:130.000 male births
- What are the probability that a boy have Hunter?
- Event A: The boy has Hunter.



$$Pr(A) = \frac{1}{130.000} = 7{,}69 \cdot 10^{-6}$$

Genetic Risk Assessment Hunters Syndrome (MPS II)

Information:

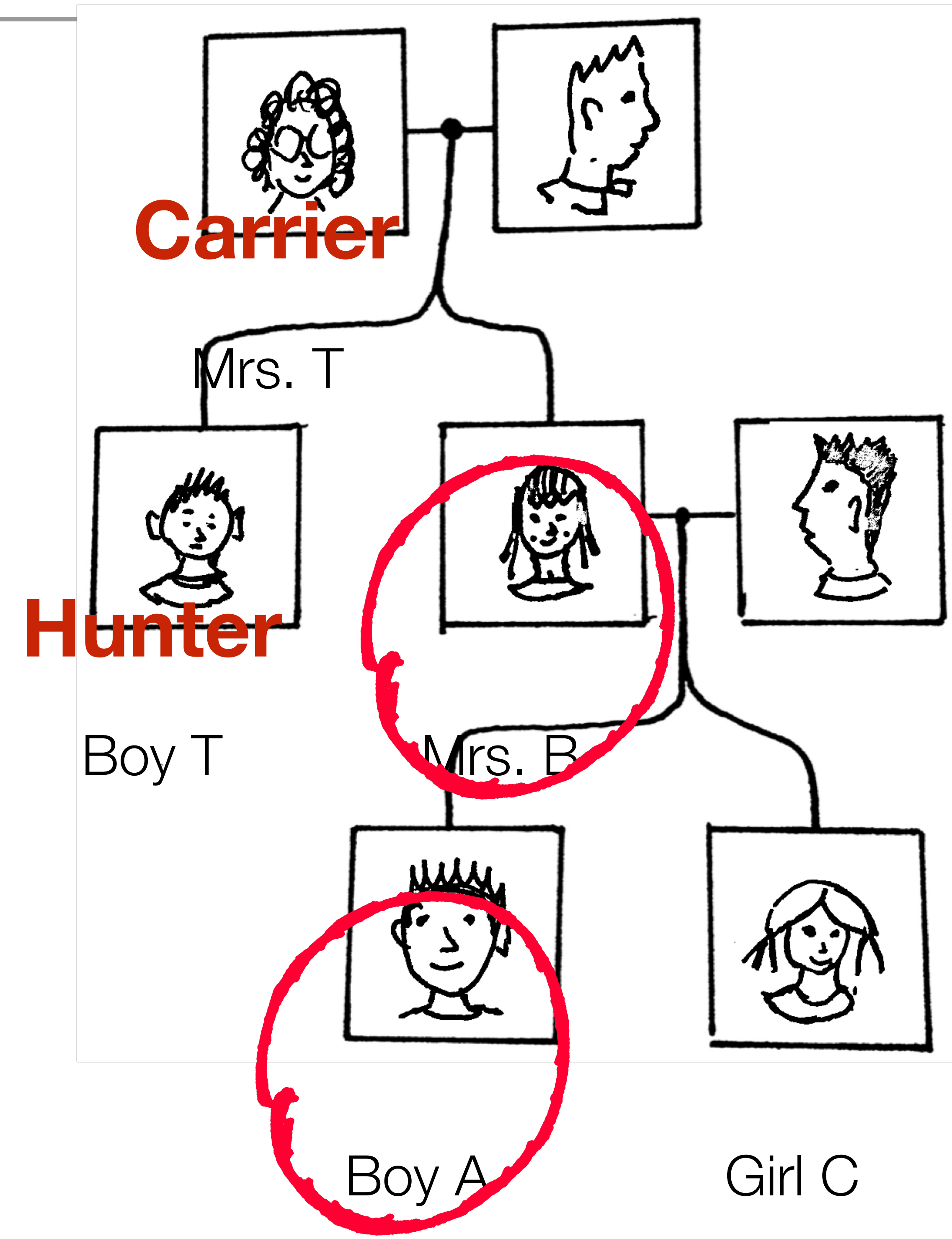
- X linked recessive.
- Boy T has Hunter.

Events:

- Event B: Mrs. B is a carrier.
- Event A: Boy A has Hunter.

Find:

- What is Pr(A)?



$Pr(B)=\frac{1}{2}; \qquad Pr(A|B)=\frac{1}{2}$

$\Downarrow$

Bayes: $Pr(A\cap B)= Pr(A|B)\cdot Pr(B)=\frac{1}{4}$

$Pr(\bar{B})=1-Pr(B)=\frac{1}{2}; \qquad Pr(A|\bar{B})=0$

$\Downarrow$

Bayes: $Pr(A\cap \bar{B})= Pr(A|\bar{B})\cdot Pr(\bar{B})=0$

$Pr(A)=Pr(A\cap B)+Pr(A\cap \bar{B})=\frac{1}{4}+0=\frac{1}{4}$

Genetic Risk Assessment Hunters Syndrome (MPS II)

Information:

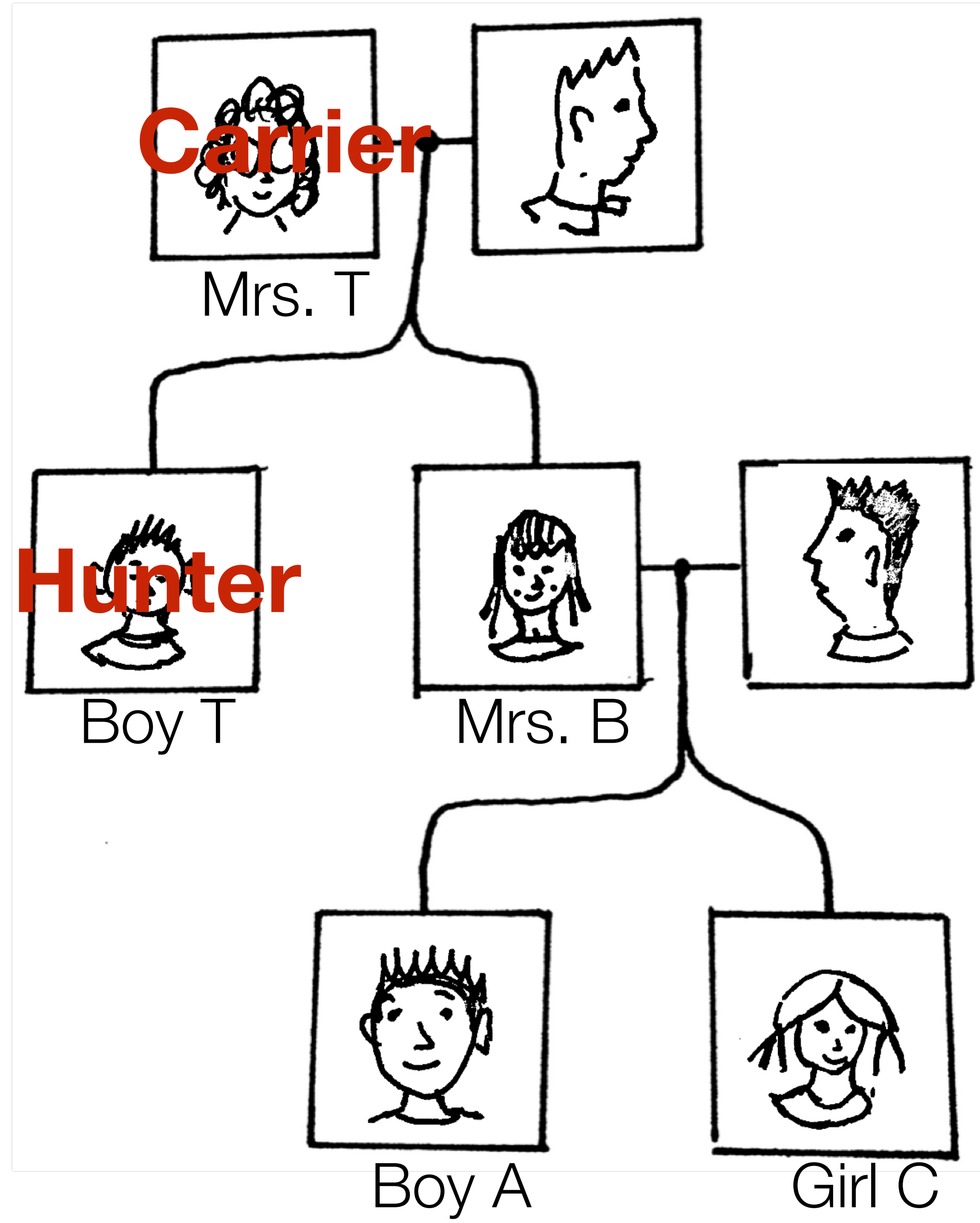
- X linked recessive.
- Boy T has Hunter.

Events:

- Event A: Boy A has Hunter.
- Event B: Mrs. B is a carrier.
- Event C: Girl C is a carrier.

Find:

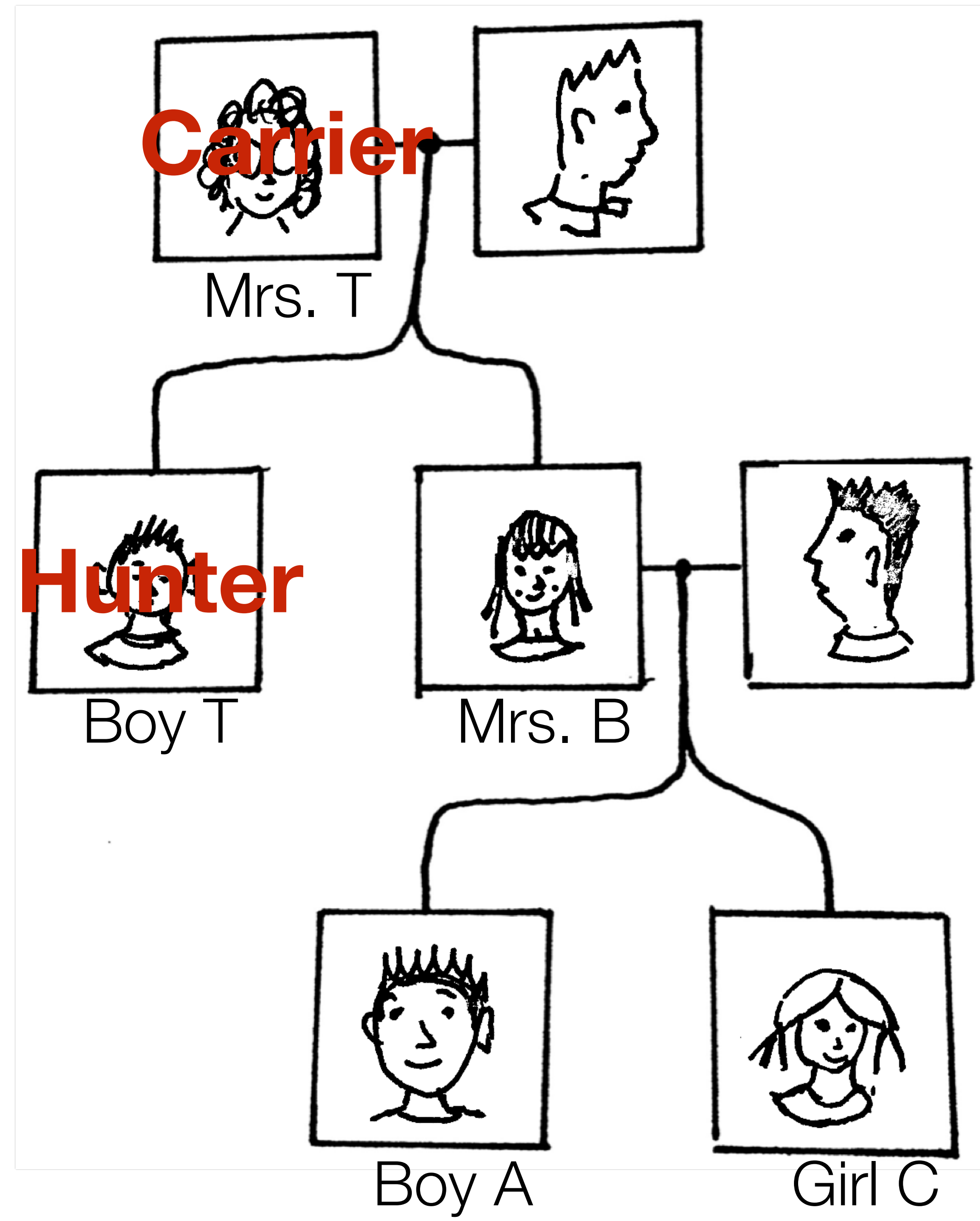
- What is $\Pr(C|\bar{A})$?



Genetic Risk Assessment Hunters Syndrome (MPS II)

Events:

- Event A: Boy A has Hunter.
- Event B: Mrs. B is a carrier.
- Event C: Girl C is a carrier.



$Pr(B)=\frac{1}{2};\quad Pr(A)=\frac{1}{4};\quad Pr(\bar{A})=1-Pr(A)=\frac{3}{4};$

$Pr(A|B)=Pr(\bar{A}|B)=\frac{1}{2};\quad Pr(A|\bar{B})=0:\quad Pr(\bar{A}|\bar{B})=1;$

$Pr(C|B)=Pr(\bar{C}|B)=\frac{1}{2};\quad Pr(C|\bar{B})=0;\quad Pr(\bar{C}|\bar{B})=1;$

$\Downarrow$

$$\text{Bayes: } Pr(B|\bar{A})=\frac{Pr(\bar{A}|B)\cdot Pr(B)}{Pr(\bar{A})}=\frac{\frac{1}{2}\cdot\frac{1}{2}}{\frac{3}{4}}=\frac{1}{3}$$

$$\text{Markov: } Pr(C\cap B|\bar{A})=Pr(C|B)\cdot Pr(B|\bar{A})=\frac{1}{2}\cdot\frac{1}{3}=\frac{1}{6}$$

$$Pr(C\cap\bar{B}|\bar{A})=Pr(C|\bar{B})\cdot Pr(\bar{B}|\bar{A})=0$$

$\Downarrow$

$$Pr(C|\bar{A})=Pr(C\cap B|\bar{A})+Pr(C\cap\bar{B}|\bar{A})=\frac{1}{6}+0=\frac{1}{6}$$

Words and Concepts to Know

Experiment/Trial

Intersection

Markov chain

Sample space

Mutually Exclusive/Disjoint

Event

Sample point

Union

Complement/not

Relative frequency

Independence

Set

Subset

Bayes Rule

Empty set/Null set

Conditional probability

Joint events

Total probability