



CEITEC

Central European Institute of Technology  
BRNO | CZECH REPUBLIC

**Introduction to Bioinformatics  
(LF:DSIB01)**

**Week 8 : Bayesian modeling**



# Bayes' theorem

## Conditional Probability: Bayes' Theorem

$$P(A|B) = \frac{P(B|A) P(A)}{P(B)}$$

# Bayes' theorem

**LIKELIHOOD**  
the probability of "B"  
being TRUE given that "A" is TRUE

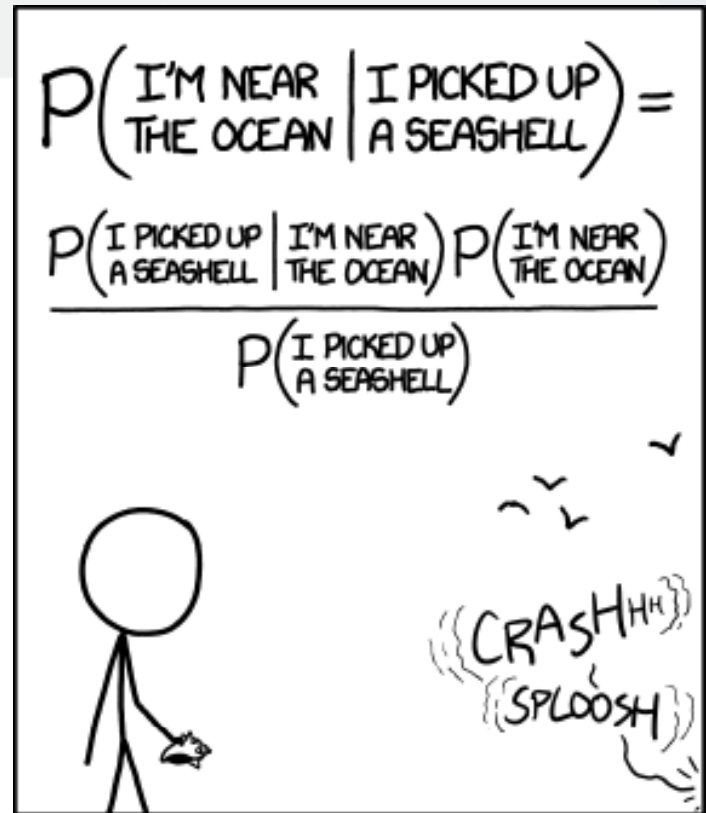
**PRIOR**  
the probability of  
"A" being TRUE

$$P(A|B) = \frac{P(B|A) P(A)}{P(B)}$$

**POSTERIOR**  
the probability of "A"  
being TRUE given that "B" is TRUE

The probability  
of "B" being  
TRUE

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STATISTICALLY SPEAKING, IF YOU PICK UP A SEASHELL AND DON'T HOLD IT TO YOUR EAR, YOU CAN PROBABLY HEAR THE OCEAN.

# Bayes' theorem

$$P(A | B) = \frac{P(B | A)P(A)}{P(B)}$$

Posterior

Likelihood

Prior

Normalizing constant

$$P(B) = \sum_Y P(B | A)P(A)$$

# Bayes' theorem

$$P(A_i|B) = \frac{P(B|A_i)P(A_i)}{P(B)} = \frac{P(B|A_i)P(A_i)}{\sum_{j=1}^n P(B|A_j)P(A_j)}$$

$A_1, \dots, A_n$

When A has 1 to  $n$  cases, and we choose some case  $i$  to find the probability of

The normal Bayes' theorem equation, where we chose a case A (called  $A_i$ )

The normal Bayes' theorem equation, where we chose a case A (called  $A_i$ )

Here we sum over each case, so there is  $n$  cases and we start at  $j$ , that is, we start at case 1, then case 2, all the way to case  $n$

# Bayes' theorem - example

- You might be interested in finding out a patient's probability of having liver disease if they are an alcoholic.
  - Past data tells you that 10% of patients entering your clinic have liver disease.  $P(A) = 0.10$ .
  - Five percent of the clinic's patients are alcoholics.
  - Among those patients diagnosed with liver disease, 7% are alcoholics.

$$P(A | B) = \frac{P(B | A) \cdot P(A)}{P(B)}$$

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$$P(A | B) = \frac{P(B | A) \cdot P(A)}{P(B)}$$

- $P(A|B) = (0.07 * 0.1) / 0.05 = 0.14$
- If the patient is an alcoholic, their chances of having liver disease is 0.14 (14%). This is a large increase from the 10% suggested by past data.

# Bayes' theorem - example

- What is the probability that a woman has cancer if she has a positive mammogram result?
  - One in 1000 of women have breast cancer.
  - 98 percent of women who have breast cancer test positive on mammograms.
  - 1 percent of women without breast cancer have a positive mammogram.

$$P(A | B) = \frac{P(B | A) \cdot P(A)}{P(B | A) \cdot P(A) + P(B | \neg A) \cdot P(\neg A)}$$



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$$P(A | B) = \frac{P(B | A) \cdot P(A)}{P(B | A) \cdot P(A) + P(B | \neg A) \cdot P(\neg A)}$$

- $P(A)=0.001$ ,  $P(\neg A)=0.999$ ,  $P(B|A)=0.98$ ,  $P(B|\neg A)=0.01$
- $(0.98 * 0.001) / ((0.98 * 0.001) + (0.01 * 0.999)) = 0.0893$
- The probability of a woman having cancer, given a positive test result, is ~9%.

# Bayes' theorem - applications

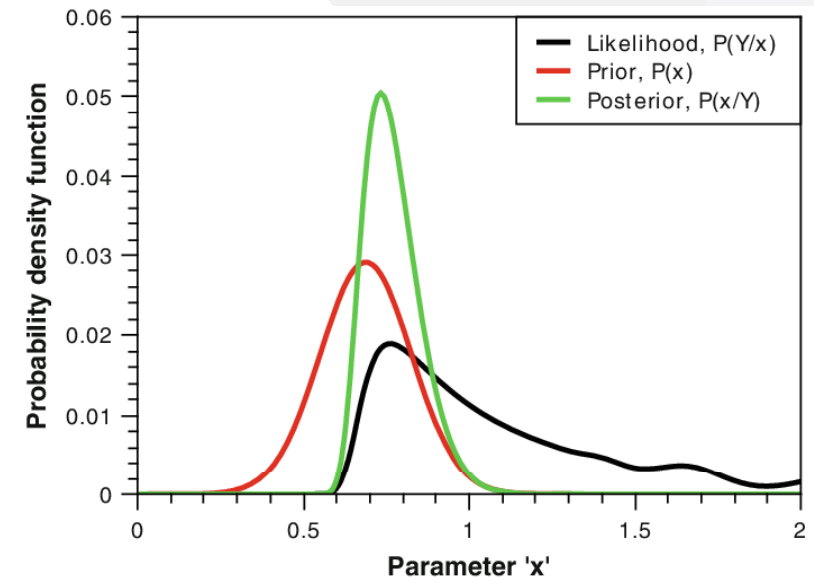
- Bayesian statistics
  - Data modeling
  - Parameter estimation
- Bayesian networks
  - Naïve Bayesian classifier
  - Dynamic Bayesian networks
    - Hidden markov models
- Can be used in many different context
  - Neural networks

# Bayesian statistics

- Bayesian statistical methods use [Bayes' theorem](#) to compute and update probabilities after obtaining new data.

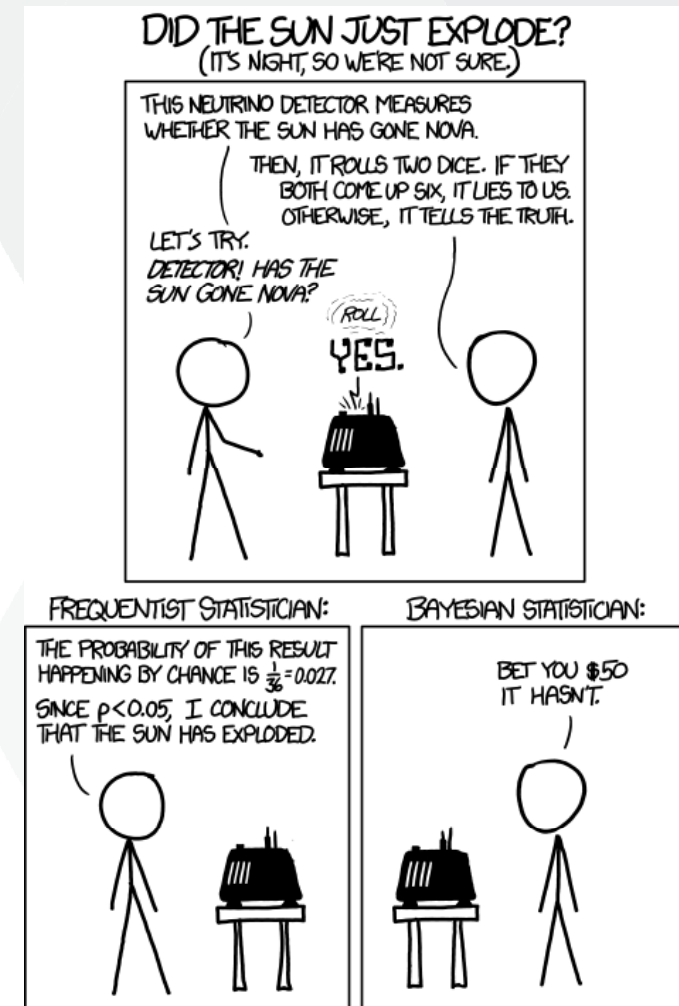
$$P(A|B) = \frac{P(B|A)P(A)}{P(B)}$$

Posterior  $P(A|B)$  is equal to the product of Likelihood  $P(B|A)$  and Prior  $P(A)$ , divided by the Normalizing constant  $P(B)$ .



# Bayesian vs. frequentists statistics

- Bayesian interpretation of probability where probability expresses a degree of belief in an event
- In the Bayesian view, a probability is assigned to a hypothesis, whereas under frequentist inference, a hypothesis is typically tested without being assigned a probability.
- The frequentist interpretation that views probability as the limit of the relative frequency of an event after many trials



# Bayesian vs. frequentists statistics

- Two main sticking points
  - Prior believe
  - Small amount of data situations

DID THE SUN JUST EXPLODE?  
(IT'S NIGHT, SO WE'RE NOT SURE.)

THIS NEUTRINO DETECTOR MEASURES  
WHETHER THE SUN HAS GONE NOVA.

THEN, IT ROLLS TWO DICE. IF THEY  
BOTH COME UP SIX, IT LIES TO US.  
OTHERWISE, IT TELLS THE TRUTH.

LET'S TRY.

DETECTOR! HAS THE  
SUN GONE NOVA?

ROLL!

YES.



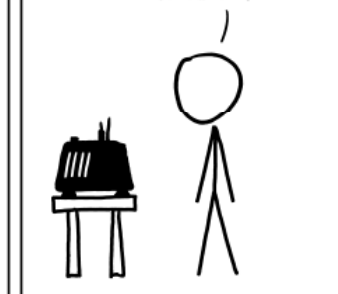
FREQUENTIST STATISTICIAN:

THE PROBABILITY OF THIS RESULT  
HAPPENING BY CHANCE IS  $\frac{1}{36} = 0.027$ .  
SINCE  $p < 0.05$ , I CONCLUDE  
THAT THE SUN HAS EXPLODED.



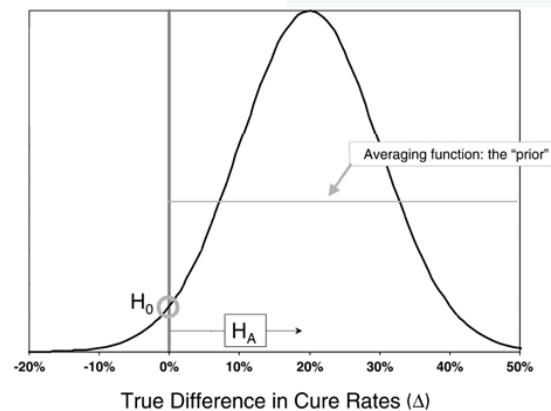
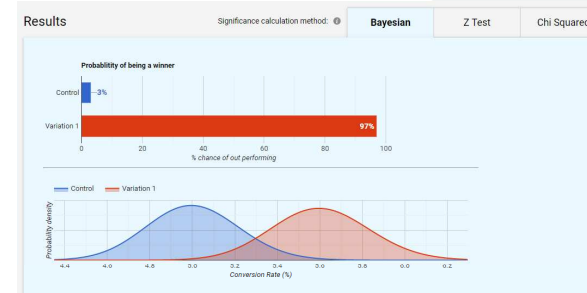
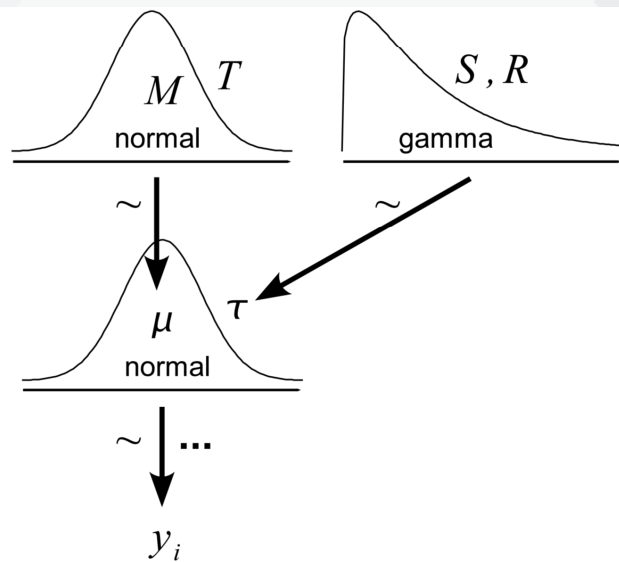
BAYESIAN STATISTICIAN:

BET YOU \$50  
IT HASN'T.



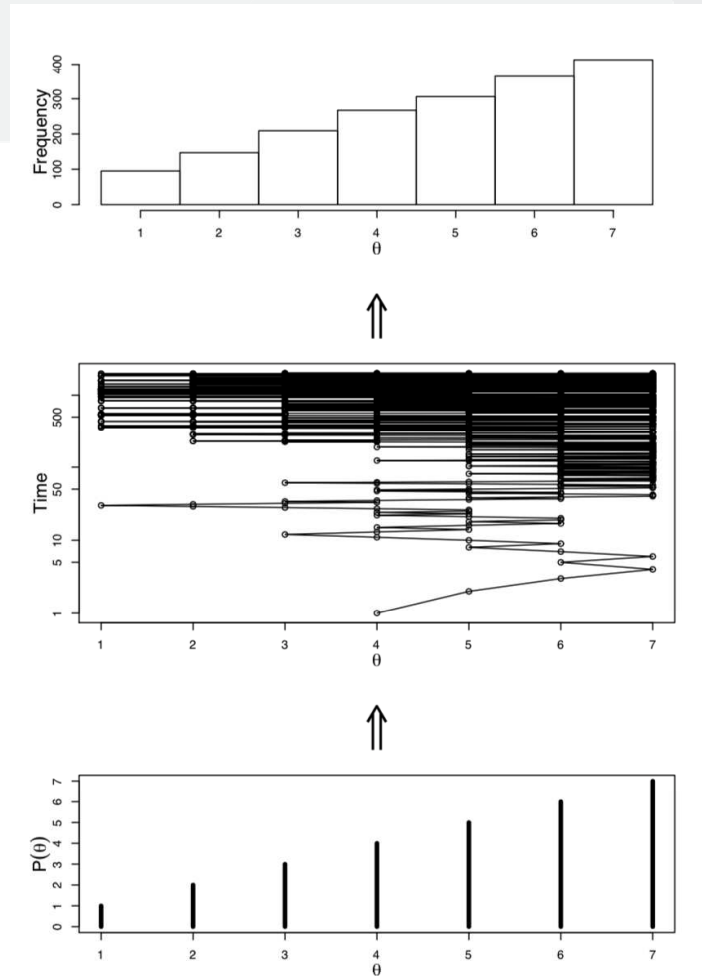
# Bayesian statistics – parameter estimation

- Data from some probability distribution function (PDF)
- The goal is to get PDF of the parameter of this function



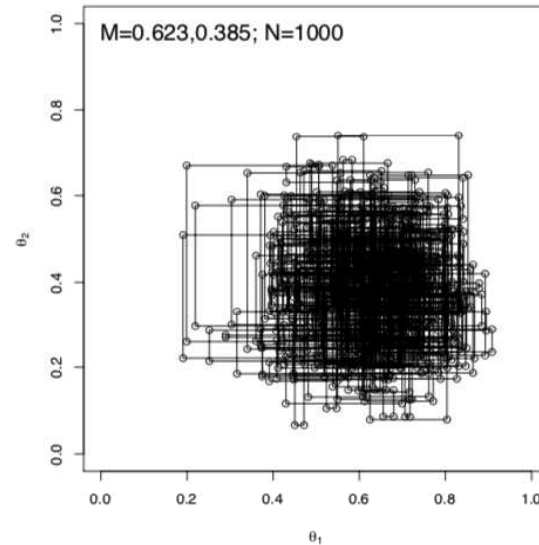
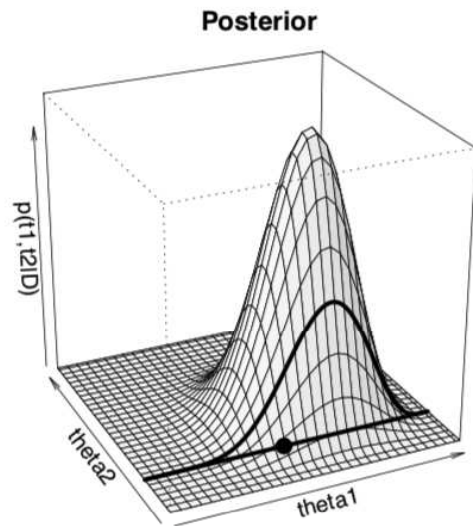
# PDF estimation with sampling

- Impossible (very hard) to compute analytically
- Markov chain Monte Carlo (MCMC)
  - This allowed usage of Bayesian statistics



# PDF estimation with sampling

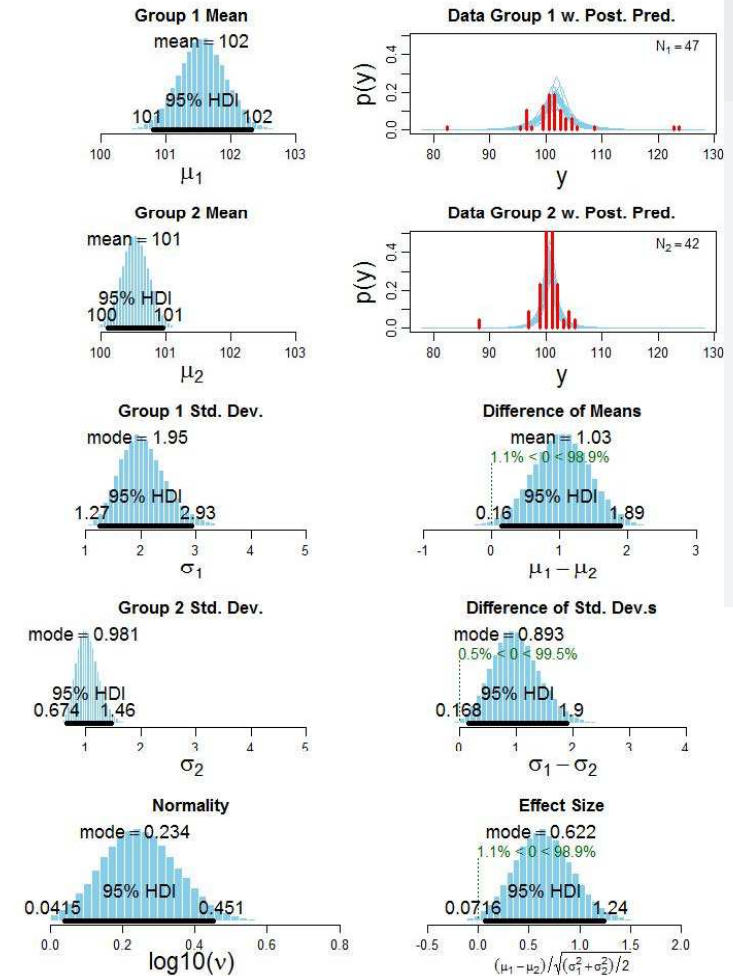
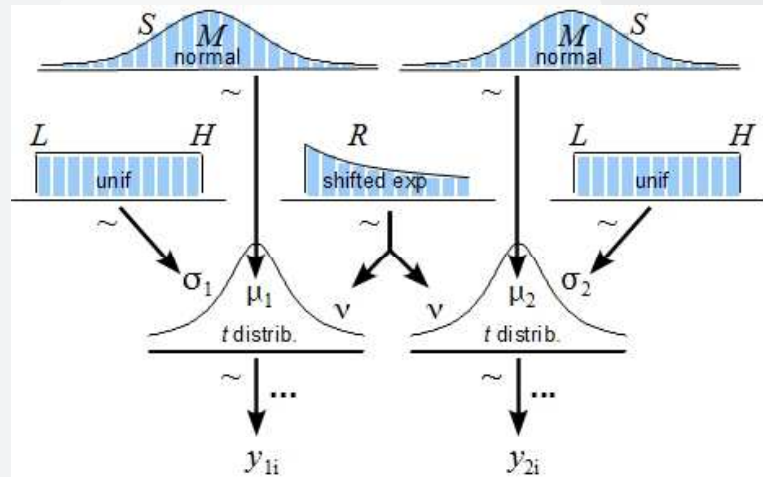
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# Bayesian statistics - example

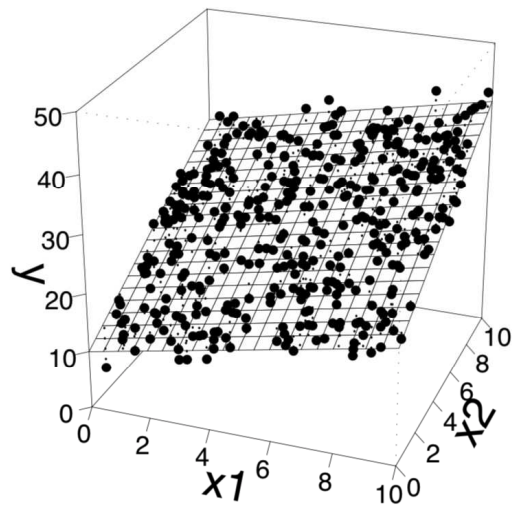
- Student t-test



# Bayesian statistics – Generalized linear model

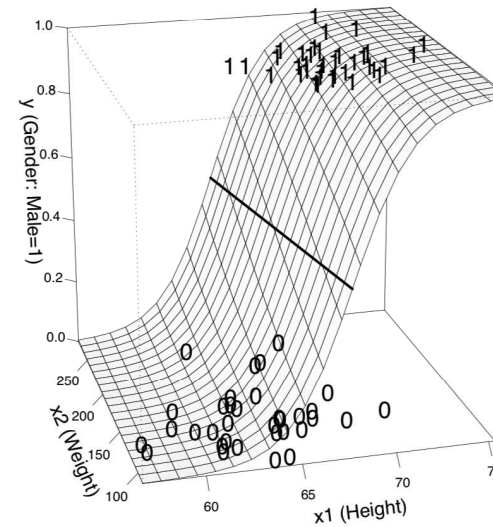
$$y = \beta_0 + \beta_1 x_1 + \beta_2 x_2$$

$$y \sim N(m, sd=2), m = 10 + 1x_1 + 2x_2$$



$$y = \text{sig}(\beta_0 + \beta_1 x_1 + \beta_2 x_2)$$

$$y \sim \text{dbern}(m), m = \text{sig}(1(0.66x_1 + 0.0096x_2 - 45))$$



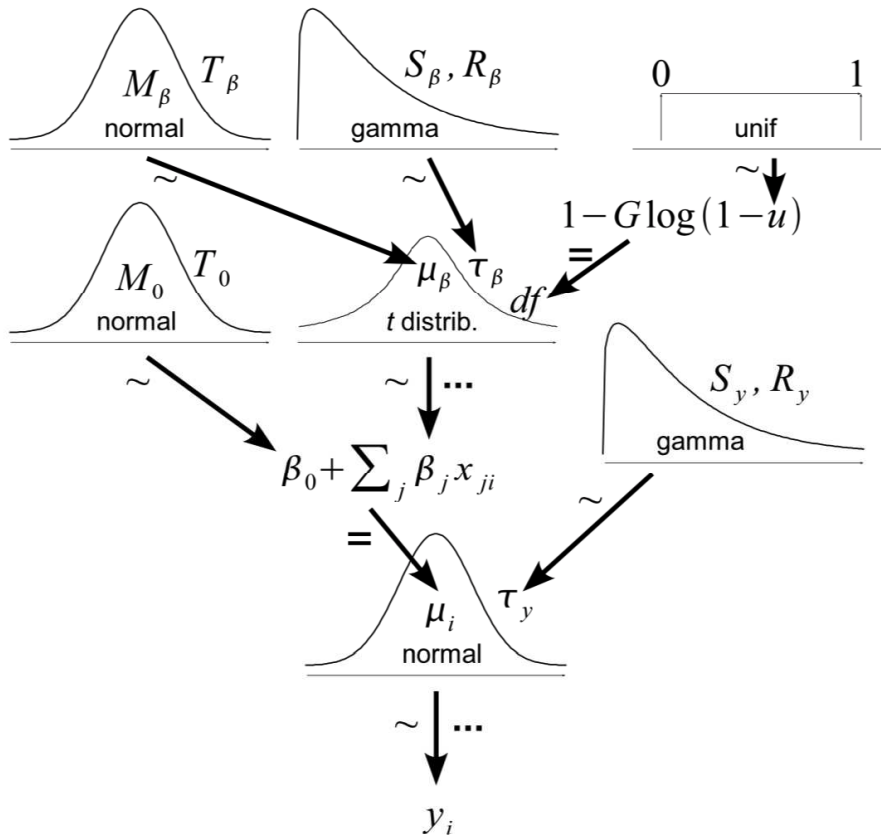
# Bayesian statistics – Generalized linear model

y Scale Type	Link Function	pdf
metric	identity	normal
dichotomous	logistic	Bernoulli
ordinal	thresholded cumulative normal	categorical
count	exponential	Poisson

# Bayesian statistics – Generalized linear model

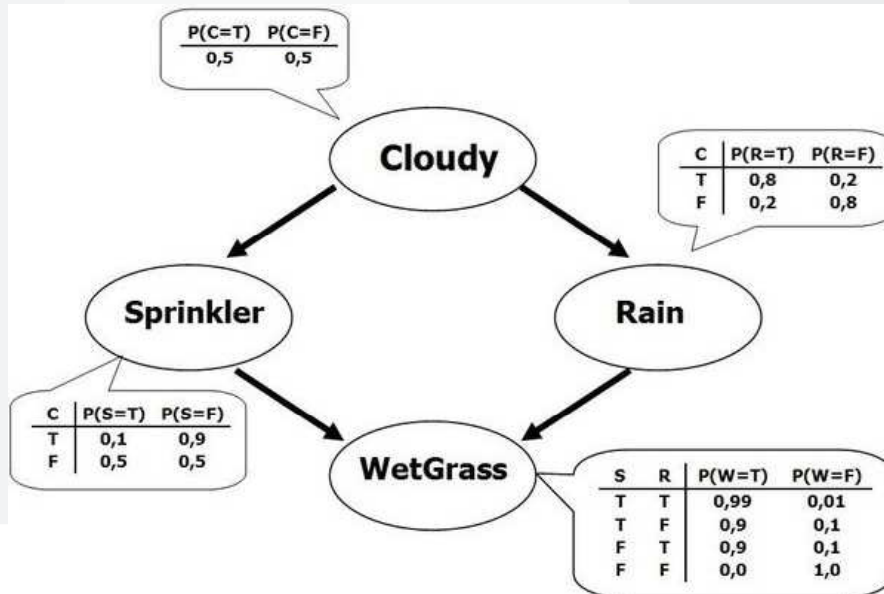
Response variable type	Explanatory variable type	Example test type
Categorical	Categorical	Fisher test
Categorical (two groups)	Continuous	t-test
Categorical (multiple groups)	Continuous	ANOVA
Continuous	Continuous	Linear regression
Continuous	Categorical (two groups)	Logistic regression

# Bayesian statistics – hierarchical models

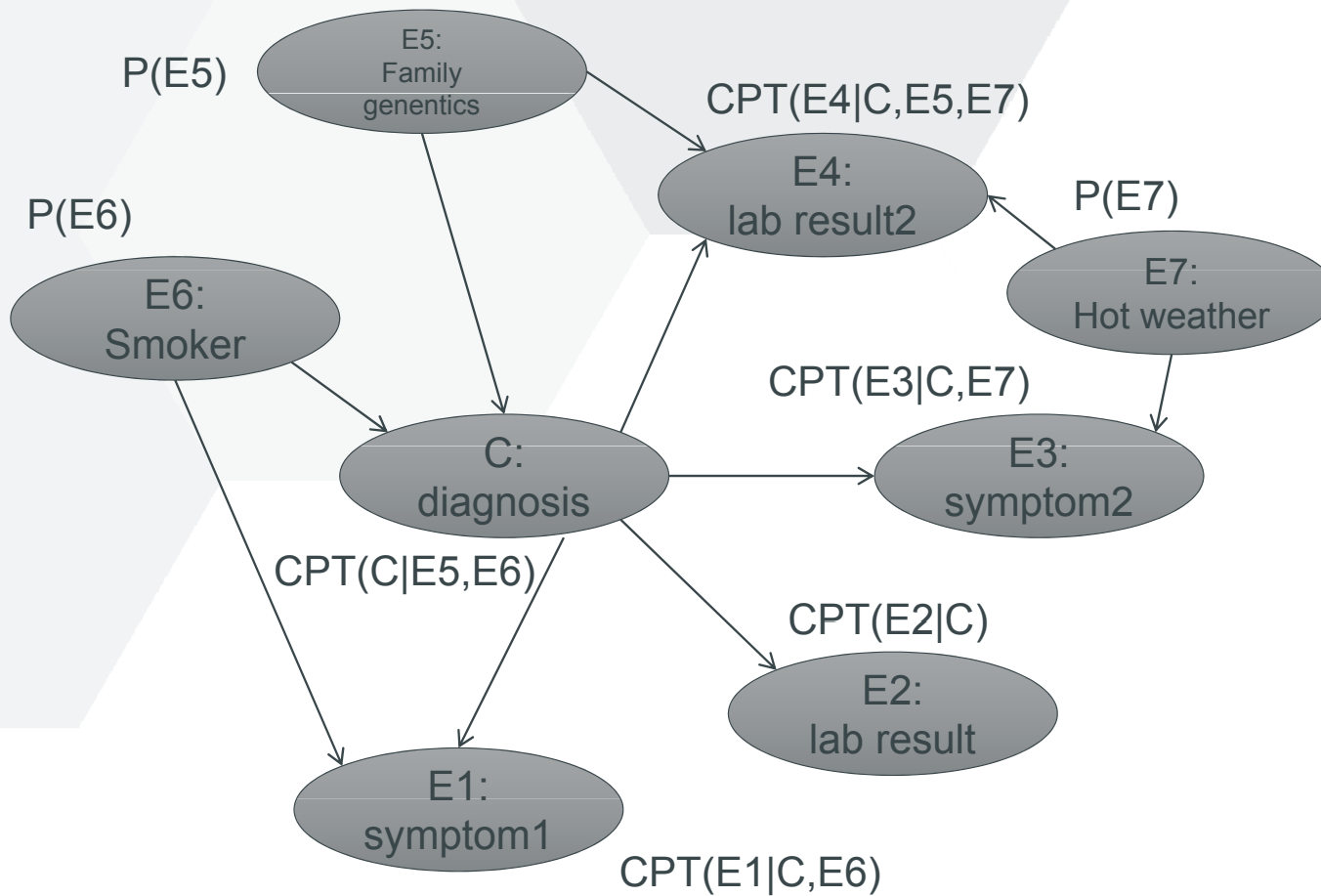


# Bayesian networks

- Directed acyclic probabilistic graph
- Represents a set of variables and their conditional dependencies

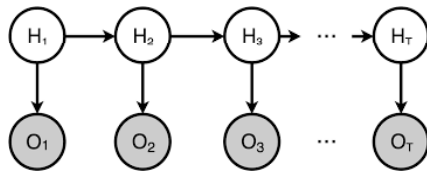
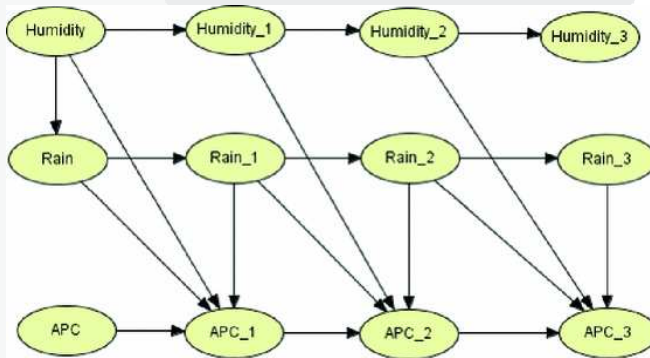


# Bayesian networks - example



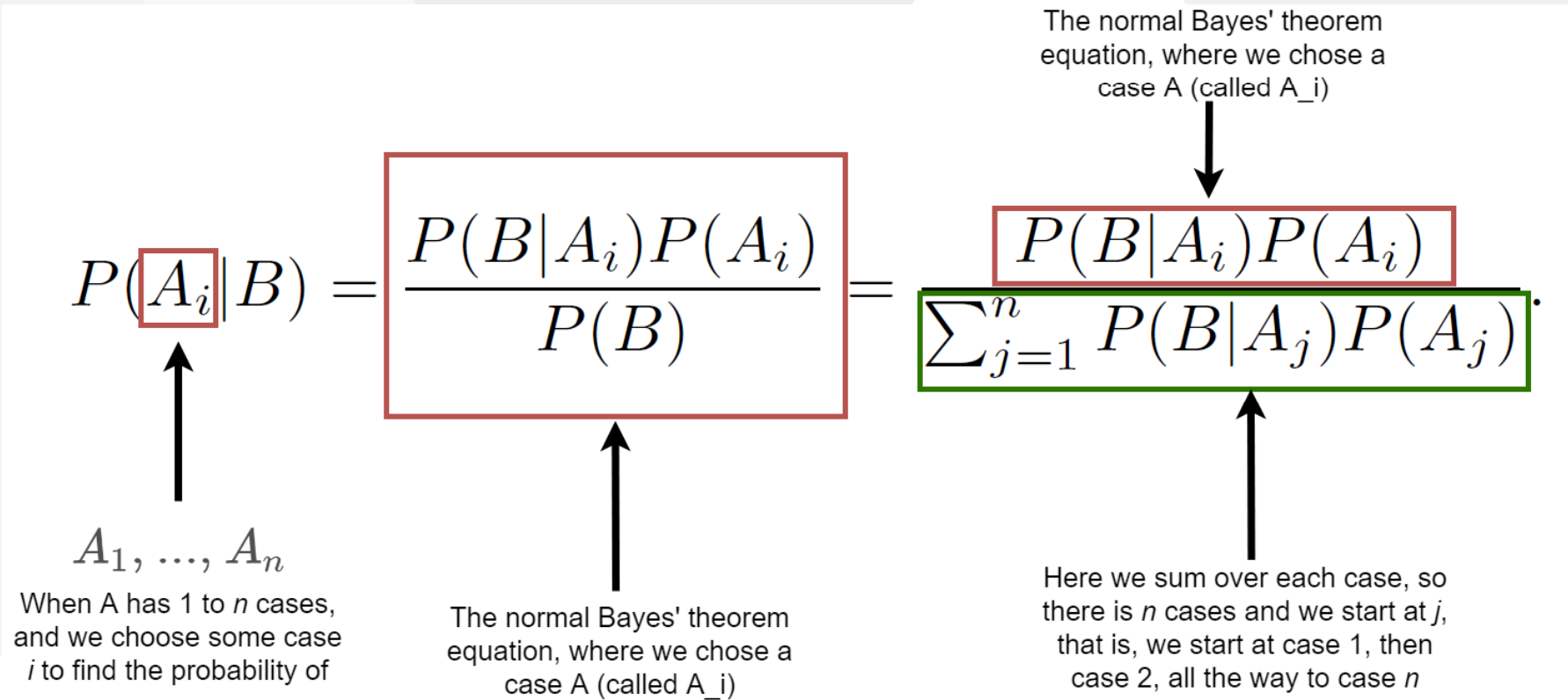
# Dynamic Bayesian networks

- Hidden markov models (hmm) are a (simple) special case of DBN

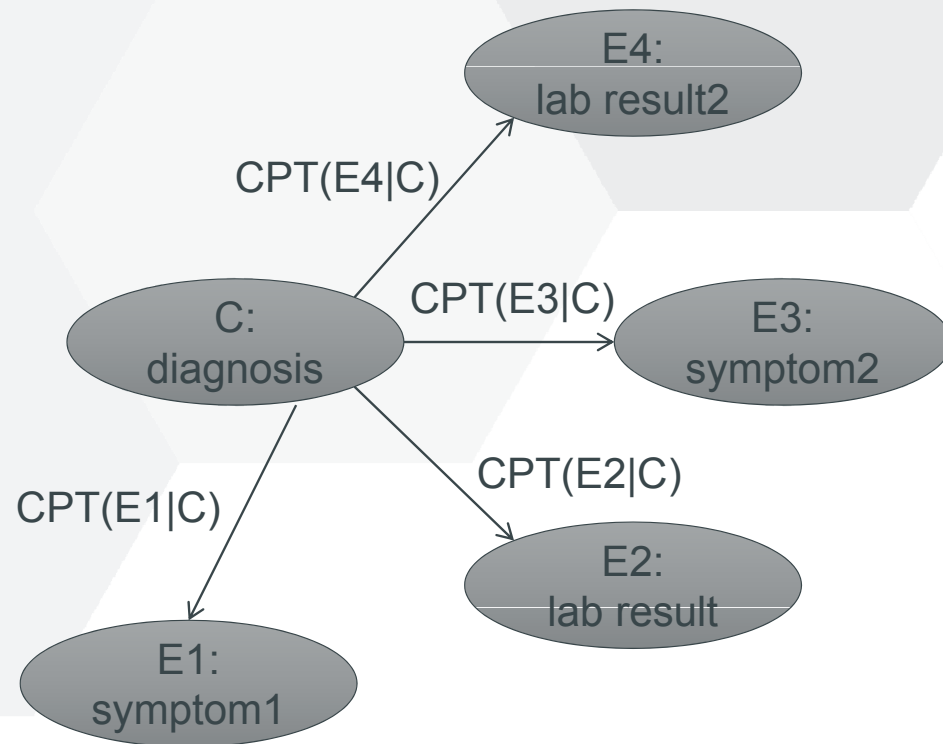




# Naive Bayesian Classifier



# Naive Bayesian Classifier – simple example

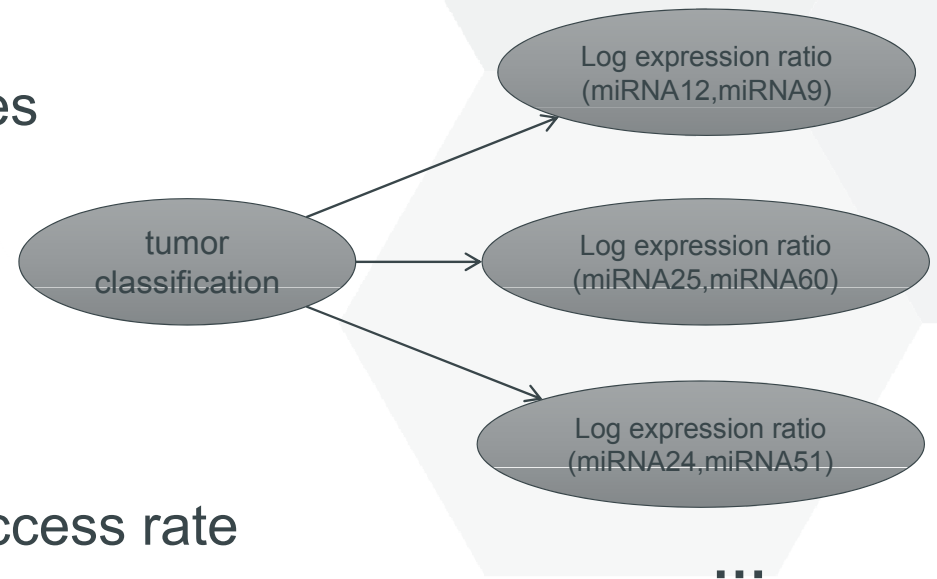


# Naive Bayes classifiers - properties

- highly scalable, requiring number of parameters linear in the number of variables
  - curse of dimensionality
- training can be done by evaluating a closed-form expression which takes linear time
- Assumption of independence
  - Simple model
- Successfully being used
- Best if you have poor understanding of the problem.

# Naive Bayesian Classifier - example

- Pancreatic tumor classification based on miRNA levels
- miRNA sequencing from plasma samples
  - ~300 miRNAs
- Select ~20 miRNAs to classify tumor types
  
- Bayesian model - 24 miRNAs -> 85% success rate





Thank you for your attention!  
60 minutes lunch break.



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