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Master programmes in Artificial Intelligence 4 Careers in Europe

University of Cyprus

MAI643: Artificial Intelligence in Medicine

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Probabilistic Graphical Models in Medicine

(some material drawn from David HeckerMann slides: A Tutorial On Learning With Bayesian Networks)

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UNIT 3

Probabilistic Graphical Models (PGMs) in Medicine

CONTENTS

1. Importance of causality
2. Probability theory – Bayes rules
3. Bayesian networks
4. Temporal probabilistic graphical models
   a. Dynamic Bayesian Networks
   b. Applications in medicine
6. Naïve Bayes
7. PGMs as machine learning methods
8. Evaluation of clinical-decision support systems

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INTENDED LEARNING OUTCOMES

Upon completion of this unit on Probabilistic Graphical Models, students will be able to:

1. Describe the main parts of a probabilistic graphical model
2. Apply the Baye’s rule for different inference approaches
3. Grasp the importance of PGMs in medical systems
4. Overview the different types of temporal Bayesian networks
5. Explain the difference between a Bayesian network and Dynamic Bayesian network
6. Evaluate a particular medical system by selecting the most appropriate evaluation metrics
7. Appreciate the role of clinicians and medical experts in evaluating a medical system.

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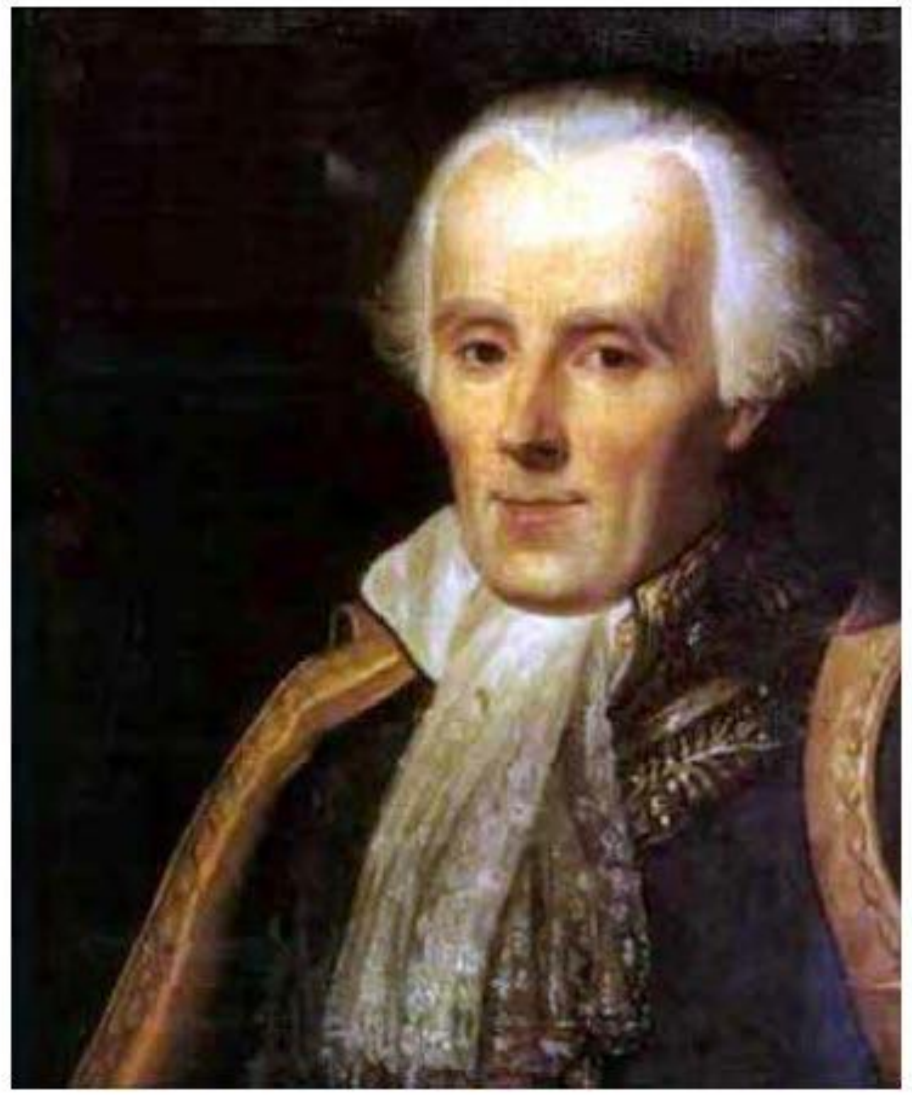
Unit 3

Probability theory is nothing but common sense reduced to calculation ...



Pierre Simon de Laplace (1749-1827), 1812

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Bayesian Probability

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The Bayesian Approach to Probability and Statistics

- **Bayesian Probability :** the degree of belief in that event
- **Classical Probability :** true or physical probability of an an event

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Some Criticisms of Bayesian Probability

- Why degrees of belief satisfy the rules of probability?
- On what scale should probabilities be measured?
- Which probabilities are to be assigned to beliefs that are not in extremes?

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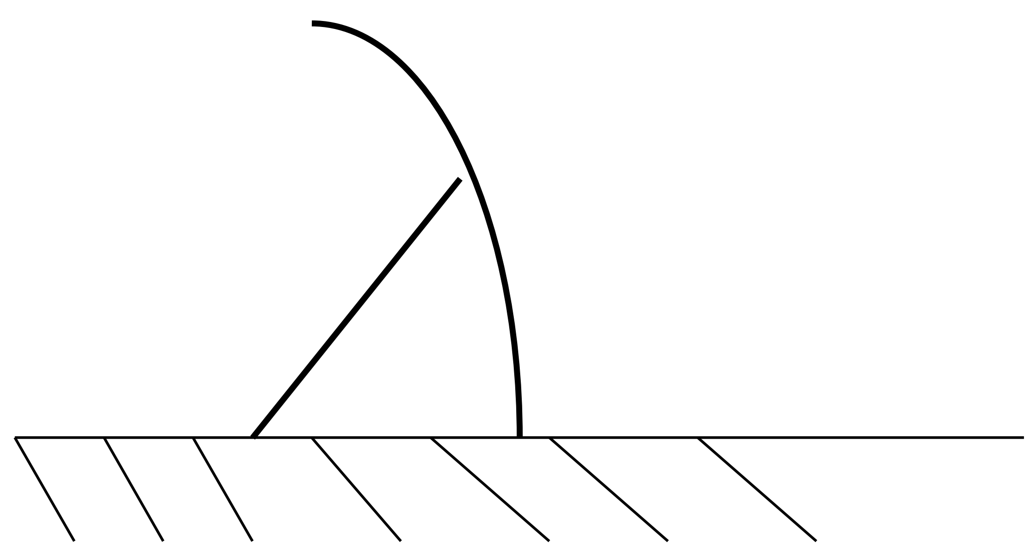
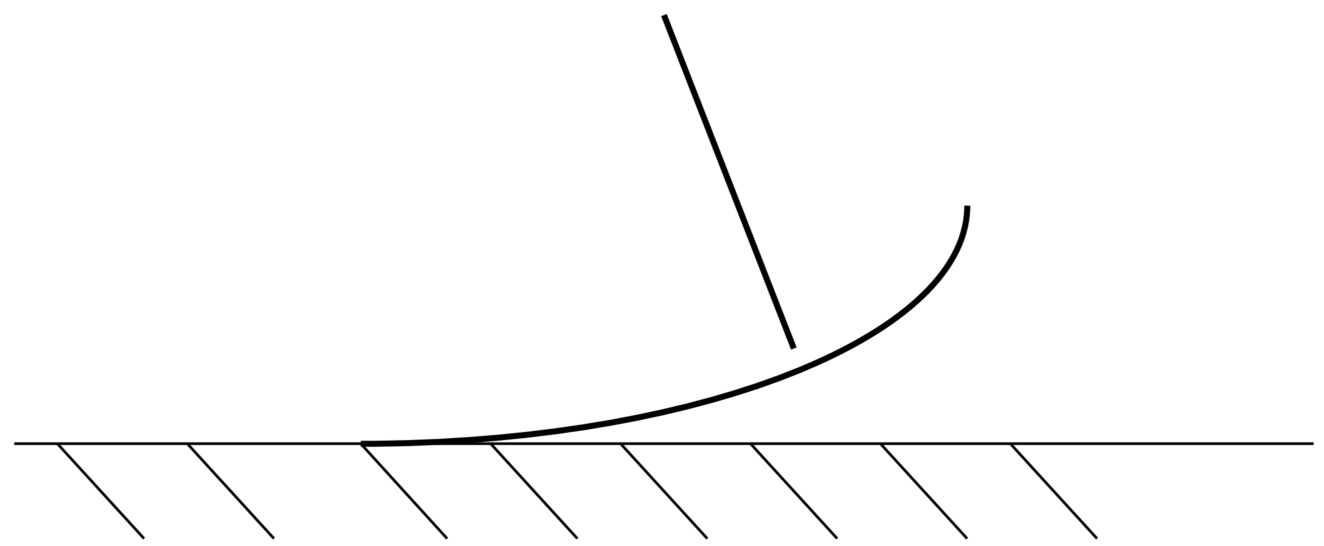
Thumbtack problem

When tossed it can rest on either heads or tails

Heads



Tails



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Problem…

- From N observations we want to determine the probability of heads on the $N+1$th toss.

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Two approaches…

Classical Approach :

- Assert some physical probability of heads (unknown)
- Estimate this physical probability from *N* observations
- Use this estimate as probability for the heads on the *N+1th* toss.

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The other approach

Bayesian Approach

- Assert some physical probability
- Encode the uncertainty about their physical probability using the Bayesian probabilities
- Use the rules of probability to compute the required probability

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Importance of Causality

- Very important in medicine as already seen in early medical systems i.e. **CADUCEUS, CASNET** and **ABEL**
- If we had a complete understanding of how the human body works, we could build models that could predict the response to various conditions and treatments with more accuracy.
- Causality in Bayesian networks is represented as a conditional dependency
  - Risk factors may cause a disease
  - Symptoms are the effects of a disease

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Probabilistic Graphical Models in Medicine

- Bayesian networks
  - Classification
  - Prediction/Prognosis
  - Treatment selection
- Naïve Bayes
  - Classification
- Temporal Bayesian networks
  - Prognosis
  - Treatment selection
  - Patient monitoring

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Bayesian Networks

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Bayesian Networks

- Probabilistic graphical models used as a **knowledge-based** technique
- **Directed acyclic graph**
  - **Nodes:** representation of features/events
  - **Edges:** dependencies between features (causal relationships)
  - **Conditional probability table:**
    - ➢ Quantify the effects of the parents on child nodes:

      P (Xi | Parents (Xi))
    - ➢ Evidences are represented as parents of Xi
- Encodes the assertion of conditional independence

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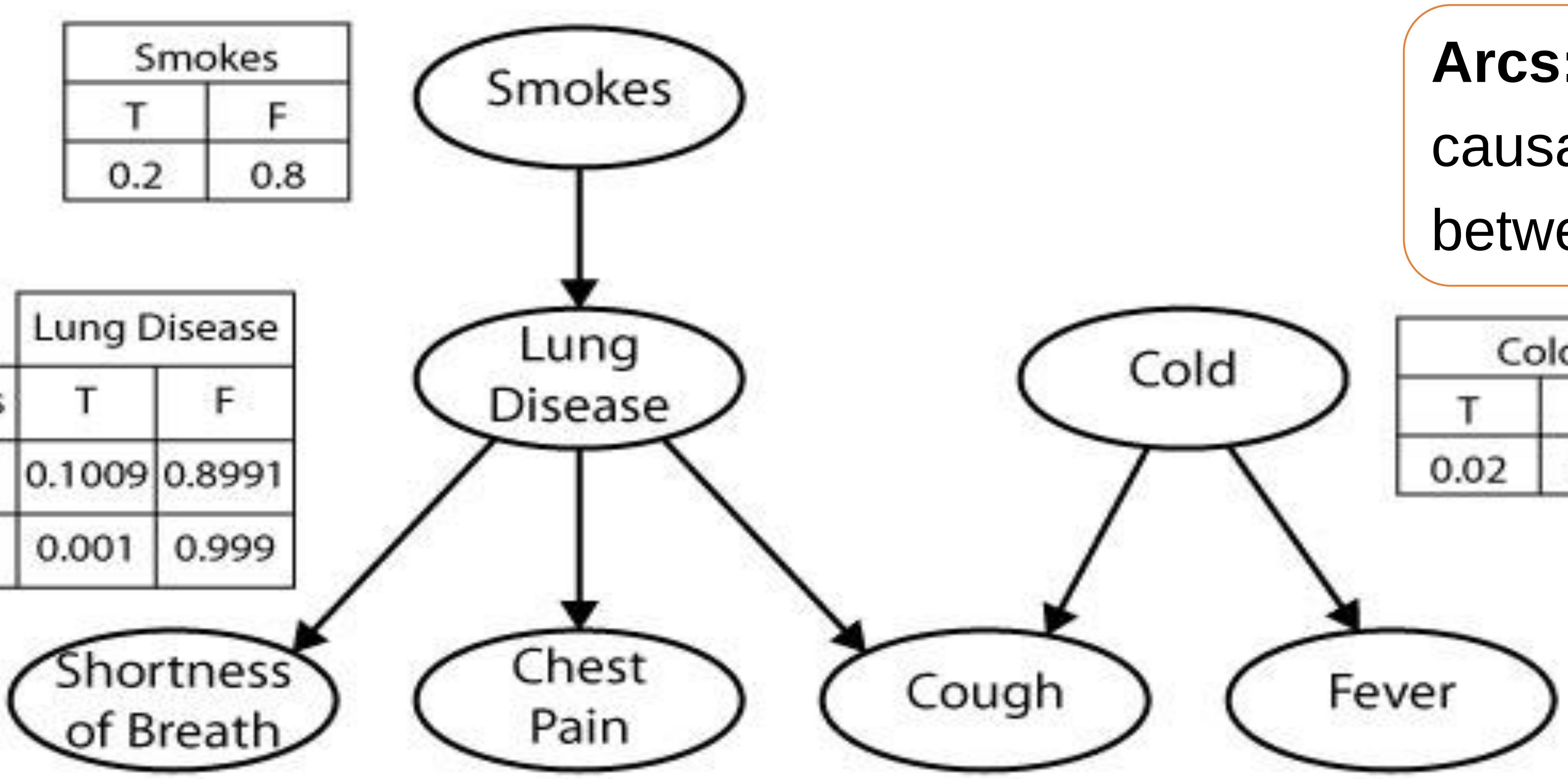
Bayesian Network Structure

Each node has a set of values. (E.g. Smokes has values True, False).

Each node has a **conditional probability table (CPT)** that relates all the values of that node, with all the possible combinations of values of the parent nodes.

Arcs: Independencies or causal-effect relationship between nodes.



Smokes	
T	F
0.2	0.8

	Lung Disease	
Smokes	T	F
T	0.1009	0.8991
F	0.001	0.999

Cold	
T	F
0.02	0.98

	Shortness of Breath	
Lung Disease	T	F
T	0.208	0.792
F	0.01	0.99

	Chest Pain	
Lung Disease	T	F
T	0.208	0.792
F	0.01	0.99

		Cough	
Lung Disease	Cold	T	F
T	T	0.7525	0.2475
T	F	0.505	0.495
F	T	0.505	0.495
F	F	0.01	0.99

	Fever	
Cold	T	F
T	0.307	0.693
F	0.01	0.99

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Lung Cancer Medical Diagnosis - Toy Example

- **Nodes:**
  - Symptoms i.e. Cough,
  - Risk factors i.e. Smoking
- **Class – node:** Disease – values: yes/no
- **Arcs:** Represent the causal-effect dependencies between nodes
  - Smokes can cause Lung Disease
  - Chest Pain is a symptom (can be caused) by Lung Disease

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Constructing a BN

- The approach is based on the following observations:
  - ➢ People can often readily assert **causal relationships** among the variables
  - ➢ Casual relations typically correspond to assertions of **conditional dependence**
  - ➢ To construct a Bayesian network, we simply draw **arcs** for a given set of variables from **the cause variables** to their **immediate effects**.
  - ➢ In the final step we determine the **local probability distributions**.

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Probabilities

- **Prior or unconditional probability:** probability in the absence of any other information

  P(*Cold*= true) = 0.02

  Correspond to belief prior to arrival of any (new) evidence

- **Probability distribution:** gives values for all possible assignments

  P(*Cold*) = <0.02,0.98> (Note: they sum to 1)

  Values of Cold are: *true, false*

- **Conditional** or **posterior probabilities**

  e.g., P(*Lung Disease*| *Chest Pain*) = 0.097

  i.e., probability of lung disease given that *the patient has chest pain* is all I know

Baye's Rule

$$P(H \mid E) = \frac{P(E \mid H)P(H)}{P(E)}$$

where:

P(*H* | *E*): = posterior : the probability that hypothesis H is true given evidence E e.g. P(Lung Disease= True|Chest Pain=True)

P(E | H) likelihood ; the probability that we will observe a new evidence E given that H is true e.g. P(Chest Pain= True|Lung Disease=True)

P(H) = priori; the probability that the hypothesis is true without any additional prior information P(Lung Disease=True)

P(E) = marginal likelihood ; this is the total probability of observing the evidence e.g. P(Chest Pain=True)

Types of Inference

- **Diagnostic Inference**: From effects to causes

For example: Given that the patient has chest pain, what is the probability of lung cancer:

P(Lung Disease=T|ChestPain=T)

- **Causal Inference**: From causes to effects

For example: Given that the patient is smoker what is the probability that the patient has lung cancer:
P(Lung Disease=T|Smoke=T)

- **Mixed Inference:** A combination of the above

P(Lung Disease=T|ChestPain=T, Smoke=T)

Criteria for Model Selection

- Some criteria must be used to determine the degree to which a network structure fits the prior knowledge and data
- Some such criteria include
  - Relative posterior probability
  - Local criteria

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Relative Posterior Probability

- A criterion for model selection is the logarithm of the relative posterior probability given as follows :

$$\text{Log } p(D /Sh) \;=\; \underbrace{\log p(Sh)}_{\text{log prior}} \;+\; \underbrace{\log p(D /Sh)}_{\text{log marginal likelihood}}$$

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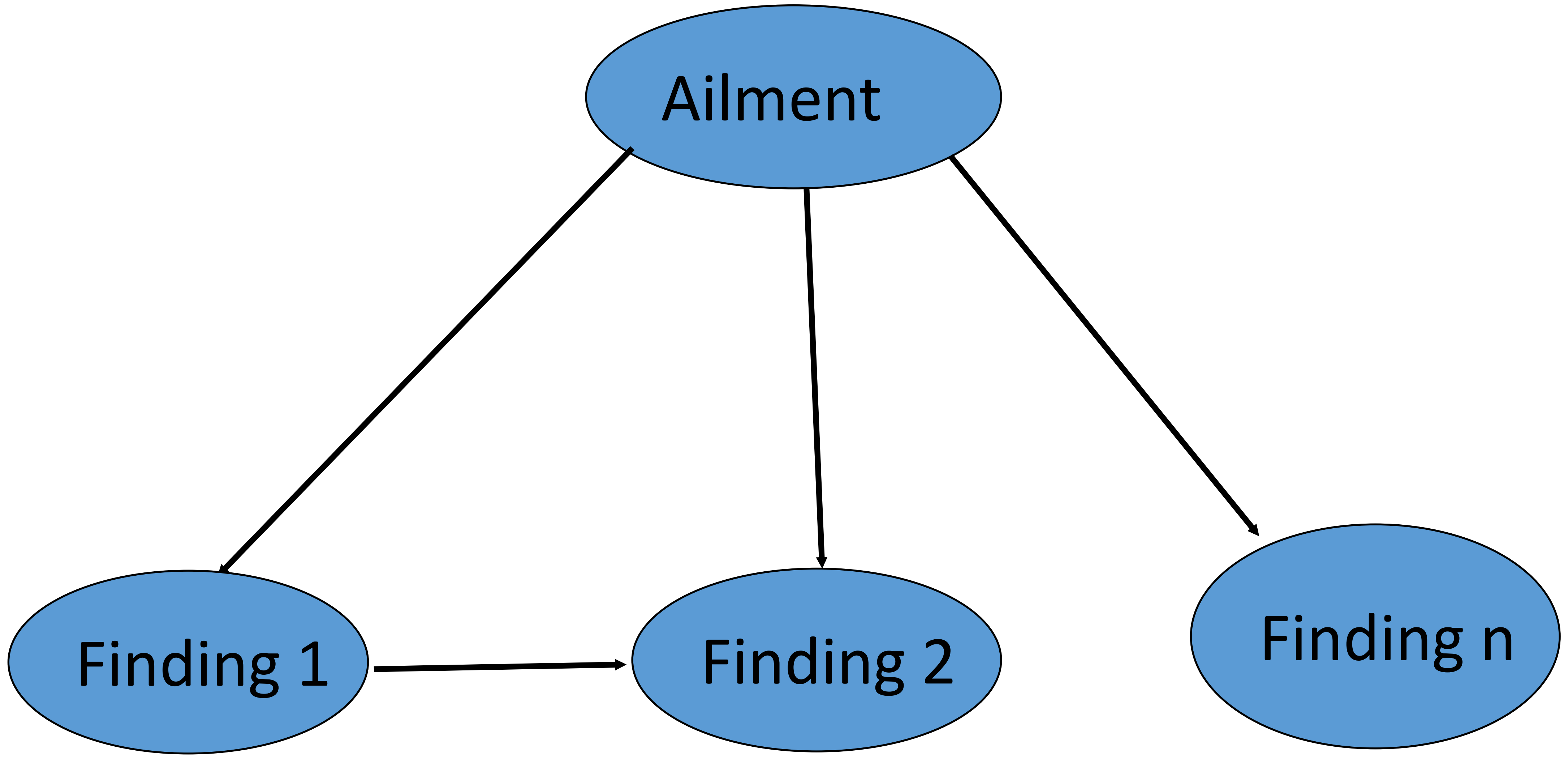
Local Criteria

An Example:



A Bayesian network structure for medical diagnosis

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Applications in Medicine

- Neurosurgical intensive care unit monitoring [Peelen et al., JBI, 2010]
- Forecasting sleep apnea [Dagum et al., UAI, 1993]
- Pittsburg cervical cancer screening model (PCCSM): Prediction of the risk of cervical cancer for patients undergoing cervical screening [Austin et al., Arch. Pathol. Lab. Med., 2010]
- Breast cancer surgery survivability prediction [D.A. Aljawad et al., ICIHT, 2017]
- A multiobjective Bayesian networks approach for joint prediction of tumor local control and radiation pneumonitis in nonsmall-cell lung cancer (NSCLC) for response-adapted radiotherapy [Luo, Y. et al., Med Phys, 45 (8) (2018)]
- Heterogeneous multimodal biomarkers analysis for Alzheimer’s disease via Bayesian network [EURASIP J Bioinform Syst Biol, 2016]
- Medical Image Interpretation [Velikova M. et al., AIME, 2013]

Why Bayesian networks?

- BNs can incorporate the accumulated knowledge of medical experts when the patient data are limited.
- Decision making under uncertainty
- Intuitive knowledge representation
- Transparent ML method

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Temporal Bayesian Networks

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Temporal Extensions of Bayesian Networks (TBNs)

- The initial model (time t0) represents domain knowledge
- Transition model represents temporal dependencies between variables (probabilistic relationships)
- **Representation of time:**
  - **Discrete time (time-slices):** Values of variables occurring at distinct points in time
  - **Interval time:** Values of variables occurring at distinct time intervals
  - **Continuous time:** Variables having a particular value for an infinitesimal (very small) period of time
  - **Irregular time**: Values of variables occurring at irregular time points or time intervals

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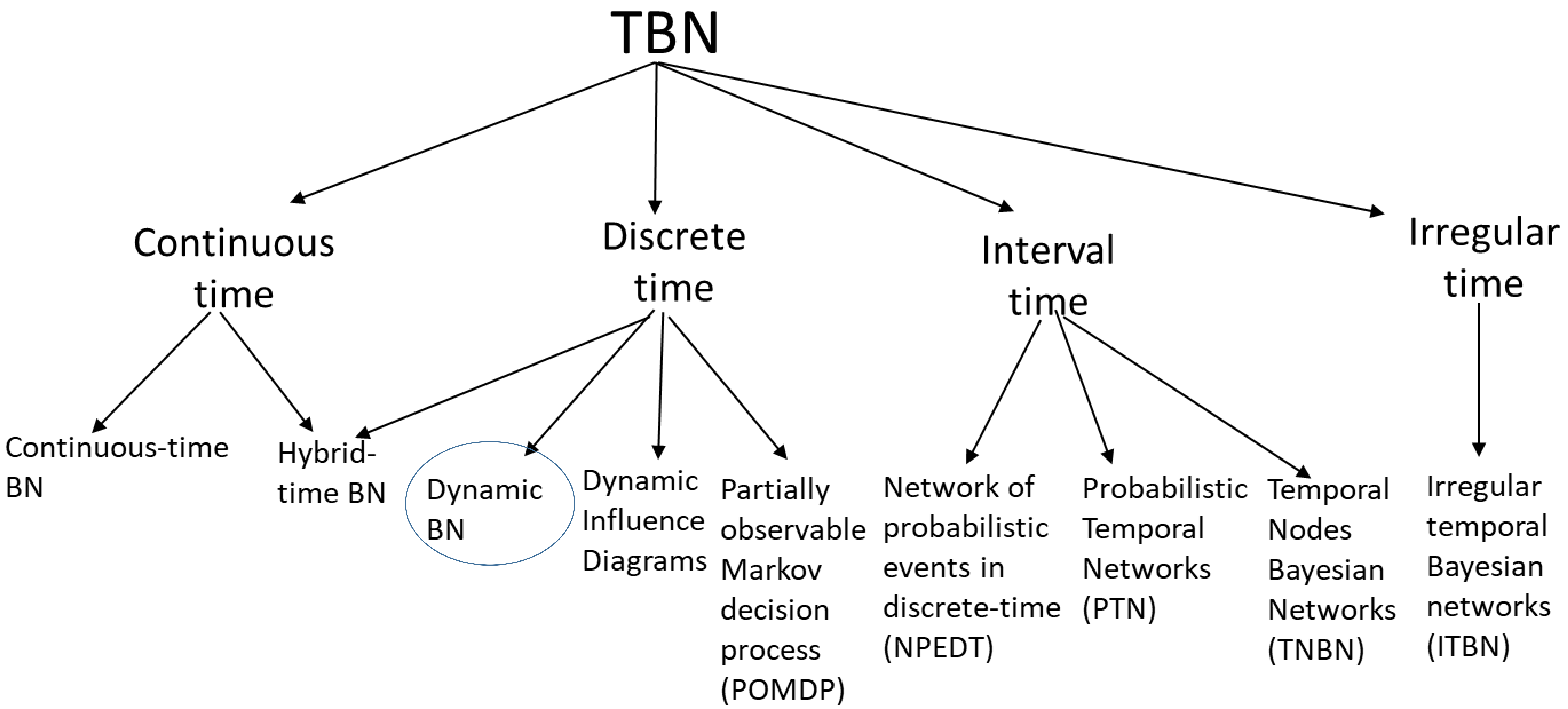
Categories of TBN



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Dynamic Bayesian Networks (DBNs)

- Most widely used temporal extension of BN
- Discrete time
- Repeated structure of BN
  - **Nodes:** Random variables that represent events or facts
  - **Arcs:** Causal and **temporal** relationships between nodes
    - **Intra arcs:** static arcs
    - **Inter arcs**: consider temporal delay

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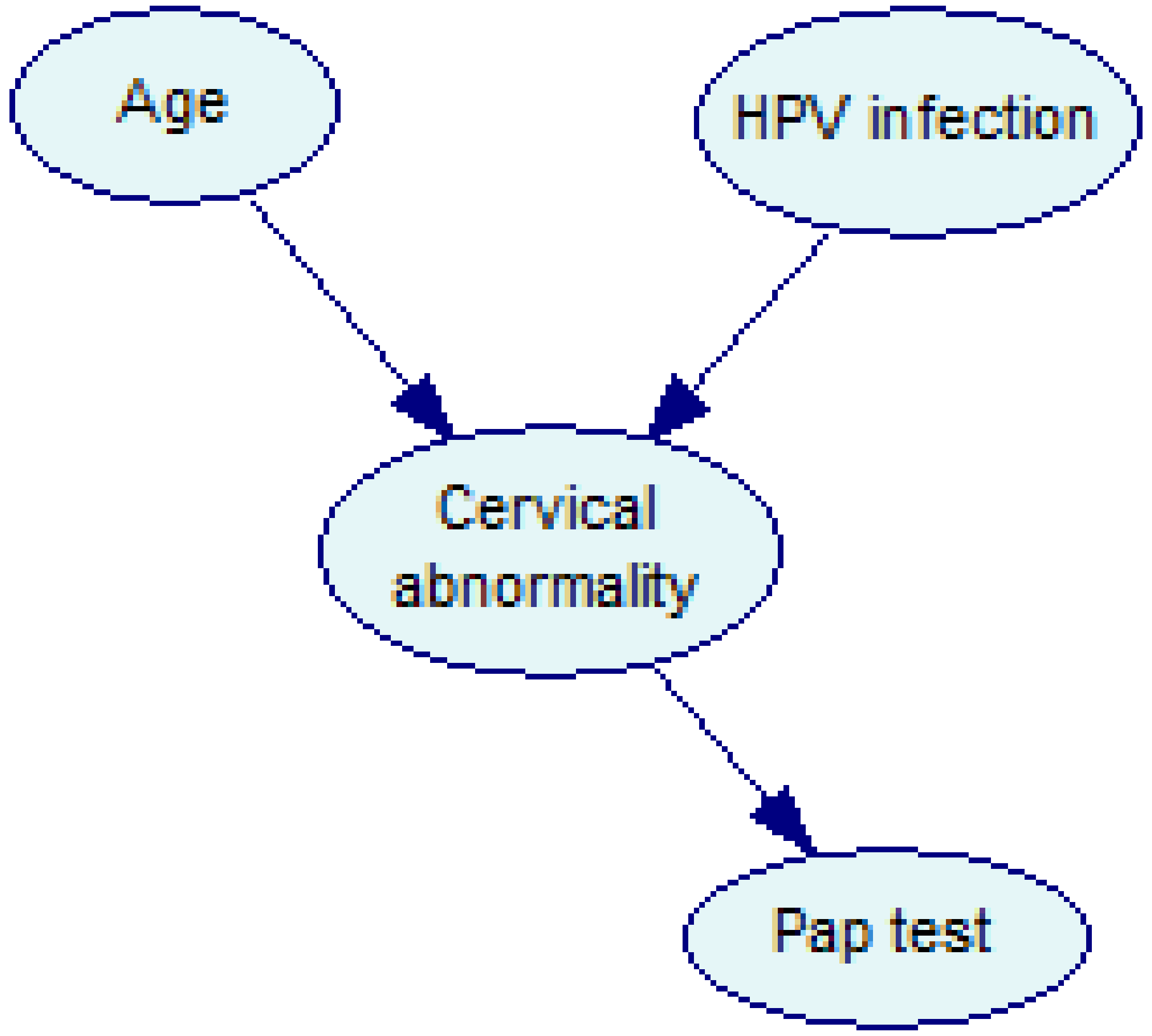
DBNs: Cervical Cancer Screening Example

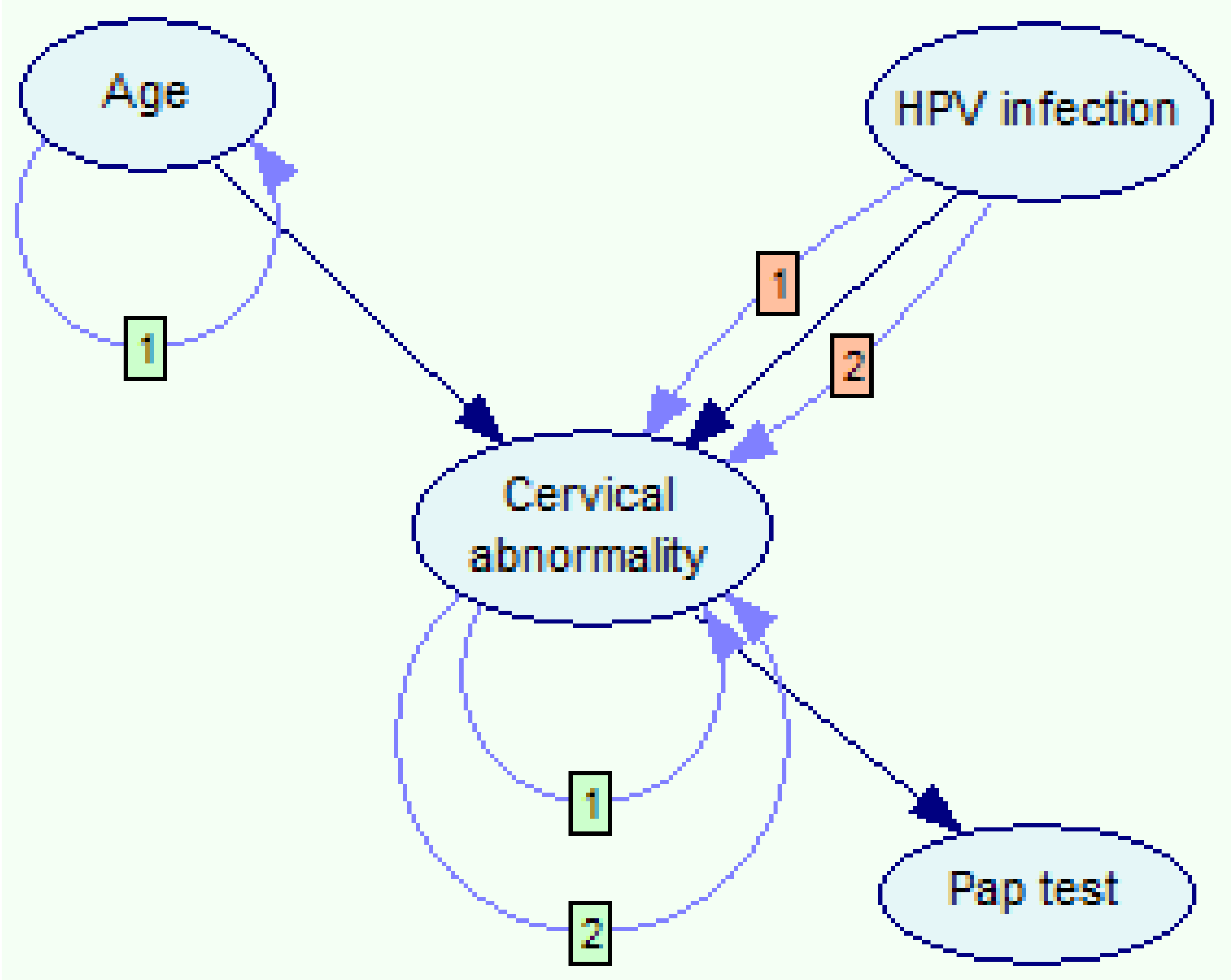


BN model

Consists of:

- random variables
- static arcs

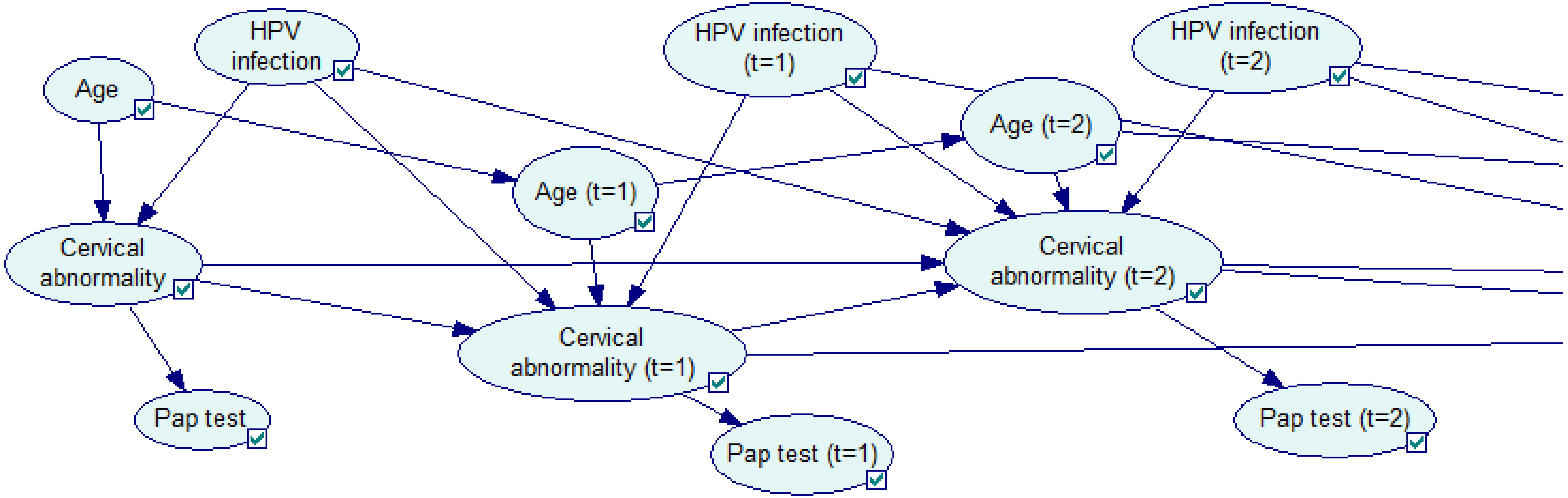
[Austin et al., Arch. Pathol . Lab. Med., 2010]



DBN model

In addition to BN models:
----temporal arcs

DBN – Cervical Cancer: Unrolling the model



Step 0 Step 1 Step 2

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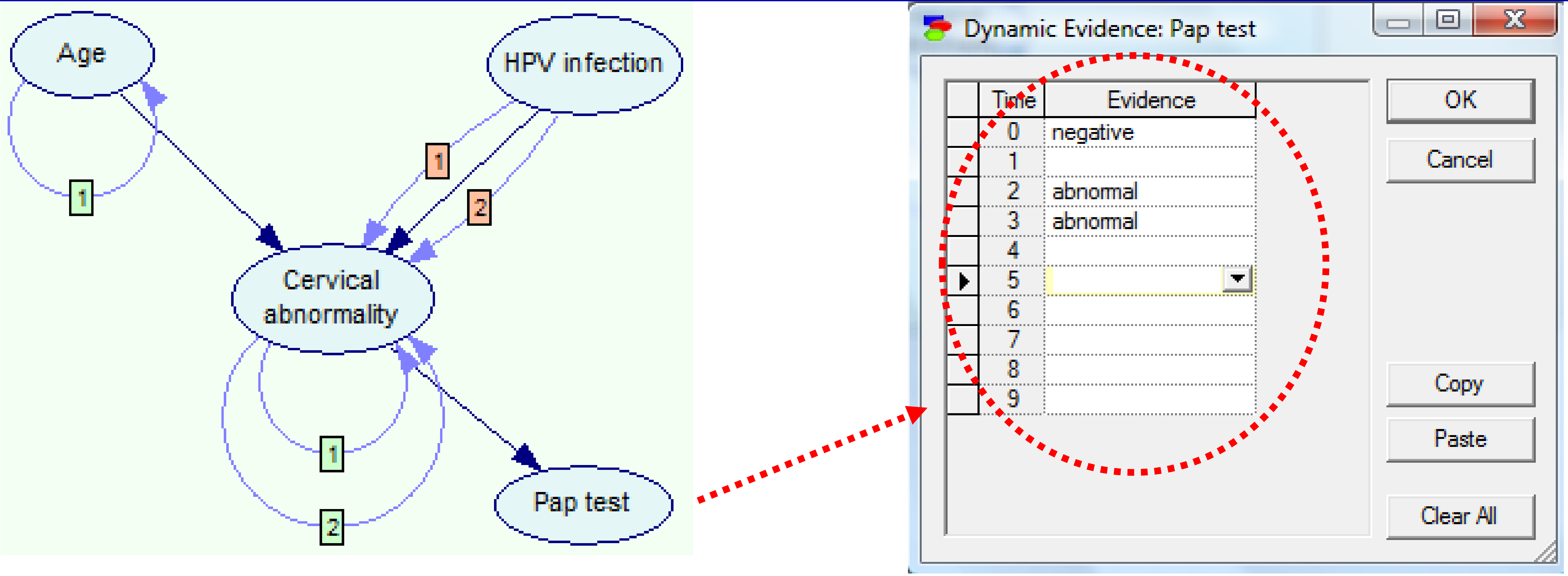
DBN: Temporal evidence



P(Cervix$_t$(abnormal)|Evidence) = ?

[Austin et al., Arch. Pathol . Lab. Med., 2010]

Evidence = Pap$_{t=0}$ (negative), Pap$_{t=2}$(abnormal), Pap$_{t=3}$(abnormal), ...

DBN Applications in Medicine

- Cancer management [Cao et al., IJMI, 1997]
- Neurosurgical intensive care unit monitoring [Peelen et al., JBI, 2010]
- Palate management [Xiang et al, UAI, 1999]
- Forecasting sleep apnea [Dagum et al., UAI, 1993]
- Pittsburg cervical cancer screening model (PCCSM): Prediction of the risk of cervical cancer for patients undergoing cervical screening [Austin et al., Arch. Pathol. Lab. Med., 2010]
- DBN-Extended: A Dynamic Bayesian Network Model Extended With Temporal Abstractions for Coronary Heart Disease Prognosis [Orphanou et al., Journal of Biomedical and Health Informatics, 2016]
- A Dynamic Bayesian Network model for the simulation of Amyotrophic Lateral Sclerosis progression [Zandonà et al, BMC Bioinformatics, 2019]

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Tools to use to construct (temporal) Bayesian networks

- **BayesiaLab** (https://www.bayesia.com/articles/#!bayesialab-knowledge-hub/bayesialab-overview)
- **GeNIe Modeler and SMILE engine** (https://www.bayesfusion.com/genie/)
- **Python packages for Bayesian networks**
  - **Pgmpy:** https://pgmpy.org/models/bayesiannetwork.html
  - **Pomegranate:** https://pomegranate.readthedocs.io/en/latest/BayesianNetwork.html
  - **PyBNesian:** https://pybnesian.readthedocs.io/en/latest/pybnesian.html

Naïve Bayes

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Naïve Bayes

- **Naïve:** Assumes that the occurrence of a particular feature is independent of the occurrence of other features.
- **Bayes:** Based on Baye's rule
- Early medical diagnostic systems (~1960) used Naïve Bayes models to assess the impact of observations about a patient.
- The patient had just one disease and all the manifestations of that disease were **conditionally independent** of each other, depending only on what the actual disease was

→ most appropriate for diagnosis of acute illnesses because newly presenting facts about a patient are likely to be caused by one rather than multiple diseases. .

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Probabilities in Naïve Bayes

$$P(C_k|x_{1,} \ldots, x_n)$$

- C_k: Class to predict i.e. diagnosis with a disease
- $X_1, \ldots, x_n$: Symptoms/risk factors of a disease

$$P(C_k|x) = \frac{P(C_k)p(x|C_k)}{p(x)} \Rightarrow \frac{P(C_k)\prod_{i=1}^{n} p(x_i|C_k)}{p(x)}$$

Baye's rule **Naïve assumption**

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PGMs as Machine Learning Methods

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Supervised Learning

Learn probabilities from a sample of observation data and encode prior knowledge

Problem: Incomplete data

Approximation methods: Monte Carlo Sampling methods, Gaussian Approximation, MAP and MI approximations and EM algorithm

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Unsupervised Learning

- Apply the learning technique to select a model with no hidden variables
- Look for sets of mutually dependent variables in the model
- Create a new model with a hidden variable
- Score new models possibly finding one better than the original

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Constructing a (temporal) BN

- using only data (data-driven BNs)
- using only knowledge (expert-driven BNs), and
- using a combination of both data and knowledge (hybrid BNs)

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Structure Learning of a (temporal) BN

- Greedy hill-climbing
- Tabu search
- Constraint-based methods i.e PC algorithm
- Genetic algorithms (Orphanou et al., GECCO 2018)

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Clinical Decision Support Systems

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Clinical Decision Support Systems (CDSS)

- Knowledge-based systems
  - Categorical
  - Rule-based systems
  - Probabilistic graphical models
- Data-based systems
  - Machine-learning systems
  - Deep-learning systems

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Evaluation of Clinical Decision Support Systems

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Evaluation of Medical Systems – What to consider?

- Safety
- Accuracy
- Feasibility
- Usability
- Social, economic, and ethical consequences
- Transparency

Evaluation by physicians/patients

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Evaluation Metrics

- To measure accuracy and efficiency of the system:
  - Recall, Precision, F-score
  - Confusion matrix
  - ROC curves and Area under the curve (AUC)
  - Correlation coefficients
- To measure usability:
  - Auditing using logs of user interaction (i.e. eye tracking)
  - Usability surveys and questionnaires (i.e. Likert scale measures)
  - Qualitative metrics
- Fairness scores to measure bias and discrimination in the system

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Sometimes we need a doctor…

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Evaluation: Expert’s Knowledge

- Medical literature
- Interview with experts to represent knowledge on systems:
  - symptoms/risk factors of the disease
  - diseases profile
- Validation of the classification model results based on medical expert’s knowledge
- Medical experts will make the final decision for diagnosis/treatment selection/medical advices to the patients
- Use expert’s knowledge to generalise the representation and generation of knowledge

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Transparency and Interpretability

- Important for **interpreting the results** and evaluating the feasibility of the systems
- **ML methods:** Decision trees, probabilistic graphical models
- Association rules (Frequent Pattern Mining or Pattern Discovery)
- Feature importance and feature selection methods to interpret the features
- Explainability techniques for deep learning and other black-box methods

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Measuring Accuracy

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Threshold Metrics - Confusion Matrix

- **True positive (TP):** a positive test result obtained for a patient in whom the disease is present (the test result correctly classifies the patient as having the disease).
- **True negative (TN):** a negative test result obtained for a patient in whom the disease is absent (the test result correctly classifies the patient as not having the disease).
- **False positive (FP):** a positive test result obtained for a patient in whom the disease is absent (the test result incorrectly classifies. the patient as having the disease).
- **False negative (FN):** a negative test result obtained for a patient in whom the disease is present (the test result incorrectly classifies the patient as not having the disease).

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Contingency Table for Test Results

Results of test	Disease present	Disease absent	Total
Positive result	TP	FP	TP + FP
Negative result	FN	TN	FN + TN
	TP + FN	FP + TN	

TP: true positive, TN: true negative, FP: false positive, FN: false negative

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Threshold Metrics in Clinical Systems

- True-positive rate (TPR) or sensitivity (recall): the likelihood that a diseased patient has a positive test.

$$\text{TPR} = \frac{number\ of\ diseased\ patients\ with\ positive\ test}{total\ number\ of\ diseased\ patients}$$

- True-negative rate (TNR) or specificity: the likelihood that a non-diseased patient has a negative test result.

$$\text{TNR} = \frac{number\ of\ non-diseased\ patients\ with\ negative\ test}{total\ number\ of\ non-diseased\ patients}$$

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Threshold Metrics in Clinical Systems

- Precision(P): likelihood that a patient has a positive test

$$\text{Precision(P)} = \frac{True\ Positive\ (TP)}{True\ Positive\ (TP) + False\ Positive\ (FP)}$$

- F1-score: harmonic mean between recall (R) and precision (P)

$$F_1 \text{ score} = 2 \text{ x } \frac{P\ x\ R}{P + R}$$

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Evaluation Metrics for Imbalanced Data

- Balanced Accuracy
- Geometric Mean
- F-beta score

- Python library: [scikit-learn]

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Ranking Metrics – ROC Curves

- Comparison of the performance of two medical tests.
- Understanding **ROC curves** is important in understanding test selection and data interpretation.
- Physicians should not necessarily, always choose the test with the most discriminating ROC curve.
- Consider the test with the **highest sensitivity and specificity**, provided that other factors, such as cost and risk to the patient are equal.
- The higher the sensitivity and specificity of a test, the more the results of that test will reduce uncertainty about probability of disease.

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Pathognomonic Tests

- Characteristic for particular disease – its presence immediately prove the disease diagnosis
- **Examples:** Pap test, PCR test
- Unfortunately, there are few pathognomonic tests in medicine and they are often of relatively low sensitivity
- Few patients with a specific condition may actually have the findings/features that the test will recognize
- Some features may be associated with multiple diseases or other factors
  - i.e. Elevation of a patient's white blood cell count
    - Either because of Leukemia
    - Or because of the use of certain medications

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SUMMARY

- All PGMs are based on Baye’s rule
- Bayesian networks have been widely applied in clinical systems due to their interpretable format and their ability to deal with uncertainty in both data and prior knowledge.
- Bayesian networks and Dynamic Bayesian networks also incorporate causality
- Dynamic Bayesian network is the most popular TBN
- Bayesian networks can be applied for differential medical diagnosis
- Naïve Bayes assumes that the occurrence of a particular feature is independent of the occurrence of other features given a particular disease.
- Evaluation of any clinical decision support system requires both evaluation metrics and evaluation by medical experts.

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Discussion

- How important is uncertainty in different medical domains/tasks?
- How do you think knowledge in those domains can support inference about the likelihood of particular events and conditions?

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