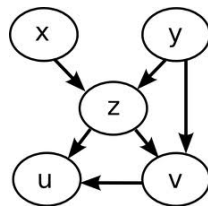


# Bayesian Networks

Guilherme J. M. Rosa

University of Wisconsin-Madison



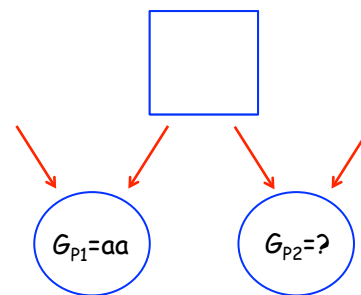
## Marginal and Conditional Independence

$$Z \rightarrow X \rightarrow Y$$

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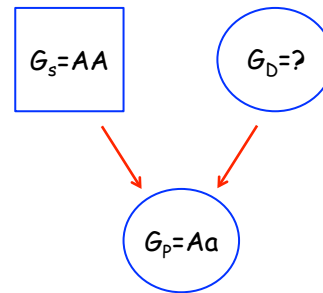
- ✓ Z & Y marginally not independent
- ✓ Conditioned on X they become independent



$$\Pr(G_{P2}) \neq \Pr(G_{P2}|G_{P1})$$

$$Z \rightarrow X \leftarrow Y$$

- ✓ Z & Y marginally independent
- ✓ Conditioned on X they are not independent
- ✓ Concept of collider, V-structure



$$\Pr(G_D=aa|G_S=AA) = \Pr(G_D=aa) = q^2$$

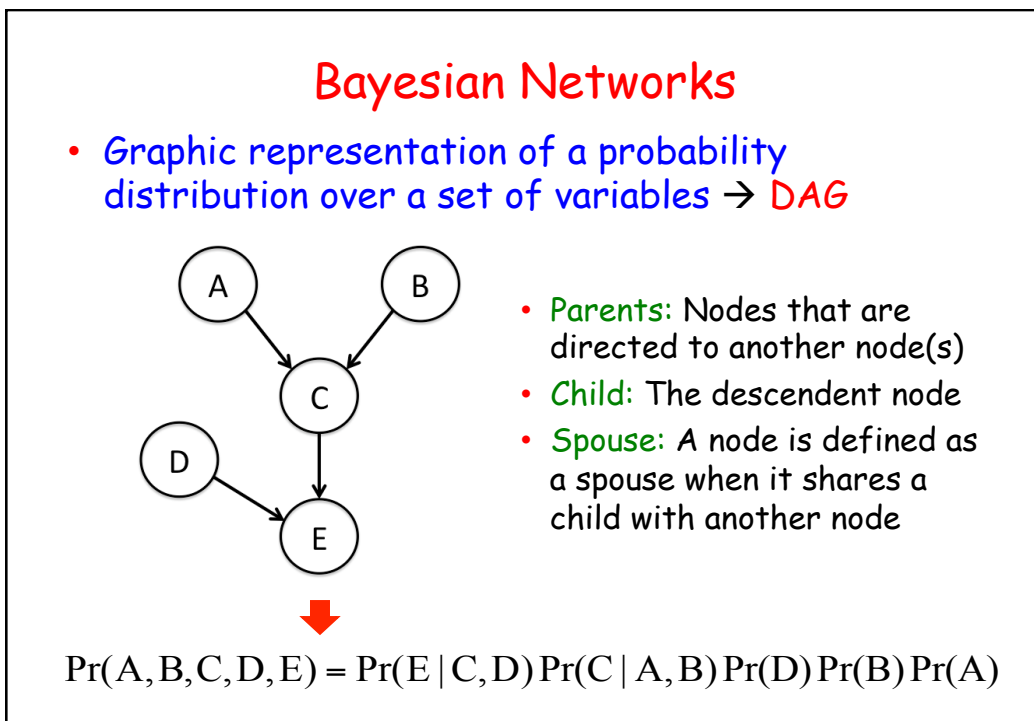
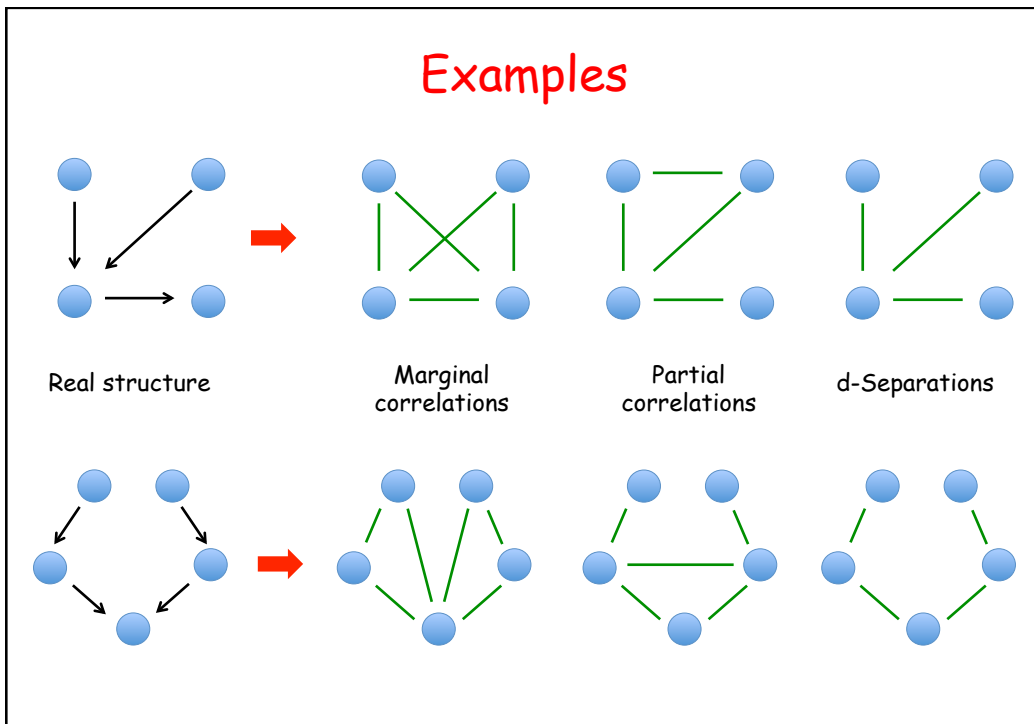
$$\Pr(G_D=aa|G_S=AA, G_P=Aa) = q \quad [\neq \Pr(G_D=aa|G_P=Aa)]$$

## 'Directed' Separation

⇒ *d-Separation* concept:

Two variables X and Y are said to be d-separated by Q if there is no active path between any X and Y conditionally on Q

(Verma and Pearl 1988, Pearl 1998, Geiger et al. 1990)



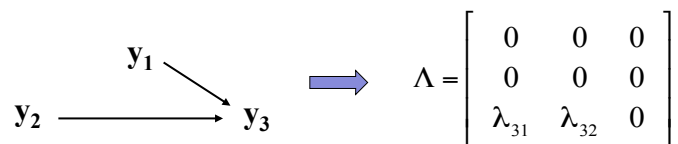
## Bayesian Networks

- Usefulness and Applications
  - Flow of information from DNA to phenotype
  - Parsimonious models for multi-trait analysis
  - Prediction, Markov Blanket
  - Causal inference
  - Visualization and model selection tool

## Inference Steps

### ① Structure Learning

- Score-based algorithms
- Constraint-based algorithms



### ① Parameter Estimation

$$y_3 = \mu + \lambda_{31}y_1 + \lambda_{32}y_2 + e$$

Maximum Likelihood or Bayesian Inference

## Structure Learning

- **Constraint-based algorithms**
  - IC, PC - Spirtes et al. (2001)
  - Grow-Shrink (GS) - Margaritis (2003)
  - Incremental Association Markov Blanket (IAMB) - Tsamardinos et al. (2003)
  - Max-Min Parents & Children (MMPC)
- **Score-based algorithms**
  - Hill Climbing (HC) - Bouckaert (1995)
  - Tabu Search (Tabu)
- **Hybrid structure learning algorithms**
  - Sparse Candidate (SC) - Friedman et al (1999)
  - Max-Min Hill Climbing (MMHC) - Tsamardinos et al. (2006)

## Constraint-based algorithms

- **Series of conditional independence tests (parametric, semiparametric and permutation)**
  - Linear correlation or Fisher's Z (continuous data; multivariate normal distribution)
  - Pearson's  $X^2$  or mutual information (categorical data; multinomial distribution)
  - Jonckheere-Terpstra (ordinal data)

## Score-based algorithms

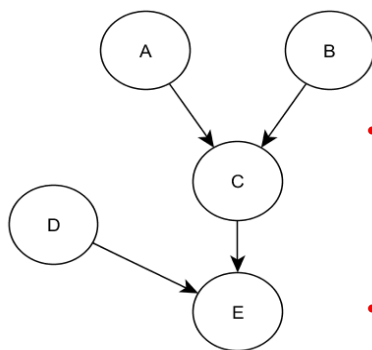
- **Different score functions**
  - Akaike Information Criterion (AIC)
  - Bayesian Information Criterion (BIC)
  - multinomial log-likelihood, Dirichlet posterior density (BDe) or K2 score (categorical data)

## The IC Algorithm (Inductive Causation; Verma and Pearl 1991)

- Step 1:** Undirected graph (search for d-separations;  
connect adjacent variables)
- Step 2:** Partially oriented graph (search for colliders)
- Step 3:** Attempt to orient remaining undirected edges such  
that no new colliders or cycles are generated

**Step 1:** skeleton; **Step 2:** V structures

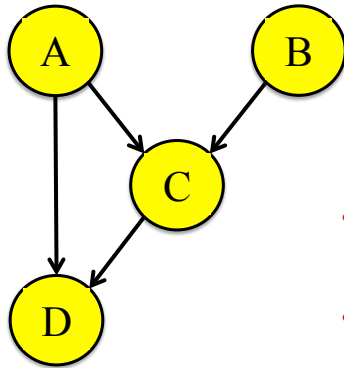
## Markov Blanket



- **Markov Blanket (MB):** a MB of a node is defined as the set containing its parent(s), child(ren) and spouse(s)
- **Conditionally on its MB, a node is independent from all other nodes**

**Examples:**  $MB(D) = \{C, E\}$ ;  $MB(E) = \{C, D\}$ ;  $MB(C) = \{A, B, D, E\}$

## Causal Inference



- **Arrows:** Causal interpretation; consequences of intervention
- **Direct, indirect and total effects**
- **Additional assumptions:** Markov condition, faithfulness and causal sufficiency assumptions

## Causal Inference

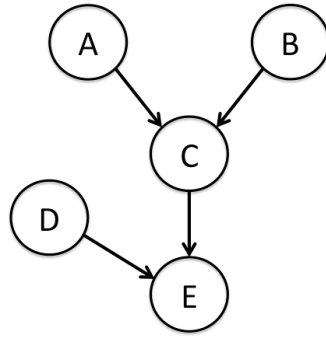
- **Prediction of the result of an intervention** (gene knockout, management decision, treatment effect)
- **Estimation of causal effects:**

If the causal DAG is known and the distribution is multivariate Gaussian, then the causal effect ( $\beta$ ) of  $X$  on  $Y$  can be estimated from the regression :

$$E[Y] = m + \beta X + pa(X)$$

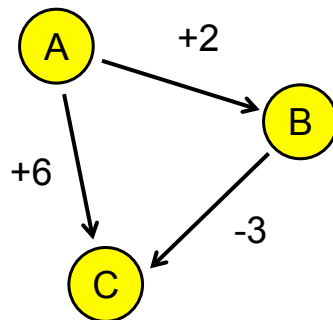
- **i.e., DAG determines adjustment variables** [backdoor adjustment; Pearl (1993)]

## Markov Condition



$$\Pr(A, B, C, D, E) = \Pr(E | C, D) \Pr(C | A, B) \Pr(D) \Pr(B) \Pr(A)$$

## Faithfulness





## Causal Sufficiency

