

# Logic and Bayesian Networks

## Part 5: Applications

Jinbo Huang

NICTA and ANU

- ▶ Medical diagnosis
- ▶ Genetic linkage analysis
- ▶ Probabilistic planning

# Constructing Bayesian Networks

- ▶ Define network variables & their domains
  - ▶ Query & evidence variables, usually from problem statement
  - ▶ Intermediary variables, harder to determine
- ▶ Define network structure (edges)
  - ▶ What variables are *direct causes* of  $X$ ?
- ▶ Define network CPTs
  - ▶ Determined objectively from problem statement
  - ▶ Reflection of subjective beliefs
  - ▶ Estimated from data

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- ▶ Cold associated with chilling, can cause sore throat
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- ▶ Flu, cold, tonsillitis; chilling, body ache, sore throat, fever



# Medical Diagnosis: Network Structure

Cold

Flu

Tonsillitis

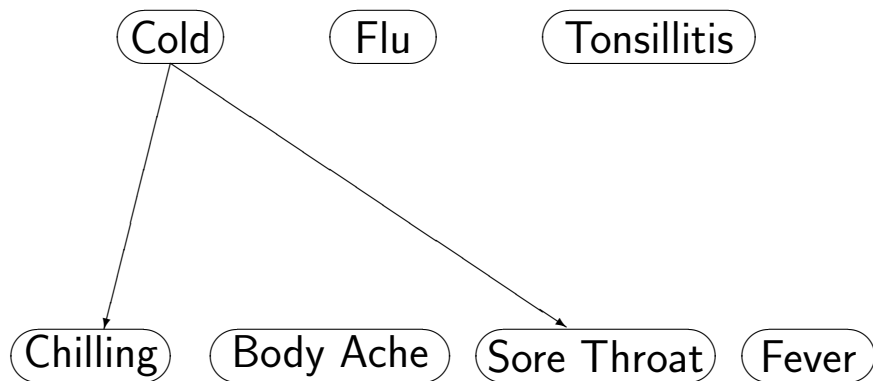
Chilling

Body Ache

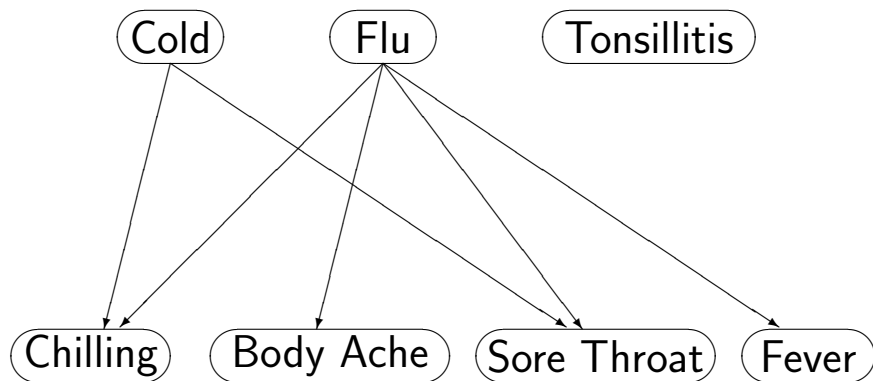
Sore Throat

Fever

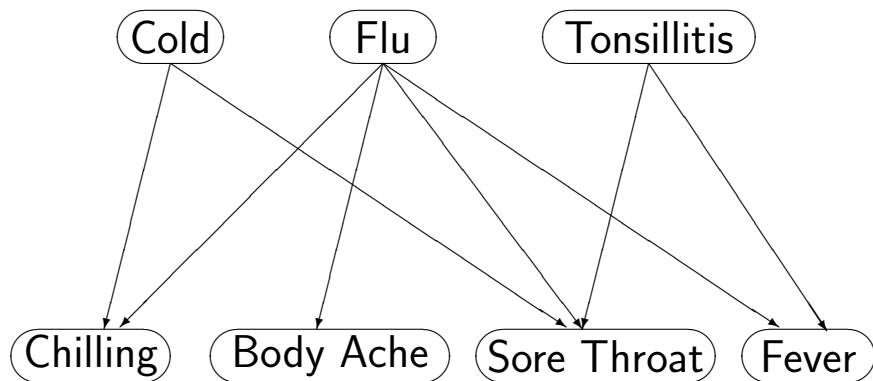
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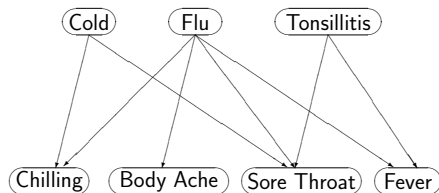
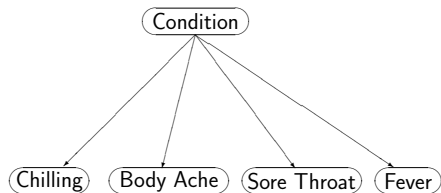


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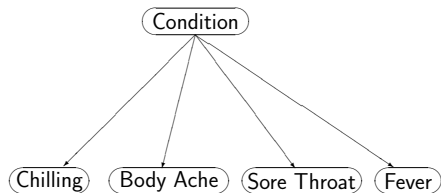


# Medical Diagnosis: Naive Bayes

Condition: normal, cold, flu, tonsillitis

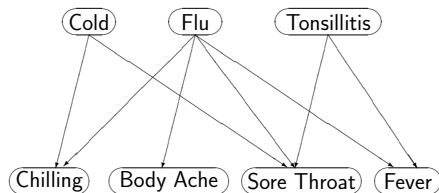


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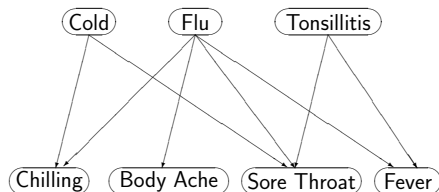
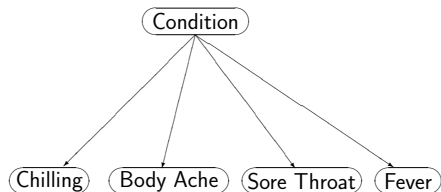


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Model inaccuracy: *single fault* assumption



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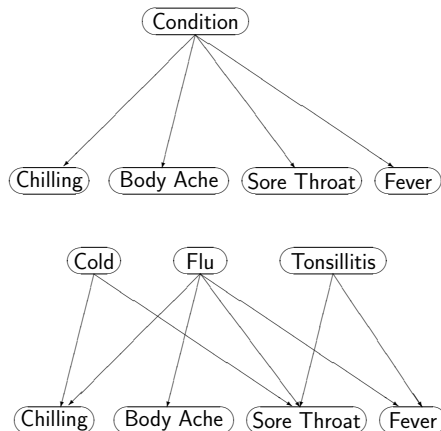


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- ▶ Cold  $\Rightarrow$  Fever & Sore Throat independent

# Medical Diagnosis: Naive Bayes



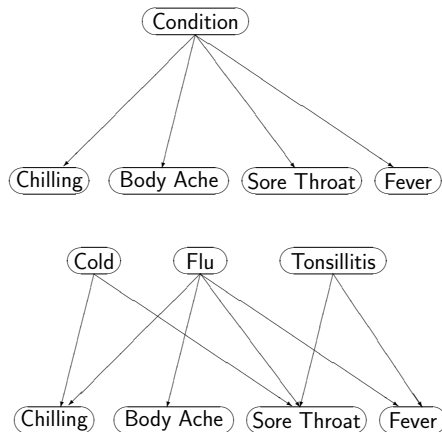
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- ▶ Cold  $\Rightarrow$  Fever & Sore Throat independent
- ▶ Body Ache  $\Rightarrow$  Flu  $\uparrow$   
 $\Rightarrow$  Cold & Tonsillitis  $\downarrow$



# Medical Diagnosis: Naive Bayes



Condition: normal, cold, flu, tonsillitis

Model inaccuracy: *single fault* assumption

- ▶ Cold  $\Rightarrow$  Fever & Sore Throat independent
- ▶ Body Ache  $\Rightarrow$  Flu  $\uparrow$   $\Rightarrow$  Cold & Tonsillitis  $\downarrow$
- ▶ No Fever  $\Rightarrow$  Cold  $\uparrow$

# Medical Diagnosis: CPTs

From medical experts, based on known stats or subjective beliefs

- ▶ CPTs for conditions (roots of graph):  $\Pr(\textit{condition})$  without knowledge of symptoms
- ▶ CPTs for symptoms:  $\Pr(\textit{Chilling} | \textit{Cold}, \textit{Flu})$  etc

# Medical Diagnosis: CPTs

From medical experts, based on known stats or subjective beliefs

- ▶ CPTs for conditions (roots of graph):  $\Pr(\textit{condition})$  without knowledge of symptoms
- ▶ CPTs for symptoms:  $\Pr(\textit{Chilling}|\textit{Cold}, \textit{Flu})$  etc

Estimated directly from medical records

Case	Cold	Flu	Tonsillitis	Chilling	Body Ache	Sore Throat	Fever
1	T	F	?	T	F	F	F
2	F	T	F	T	T	F	T
3	?	?	T	F	?	T	F
...	...						

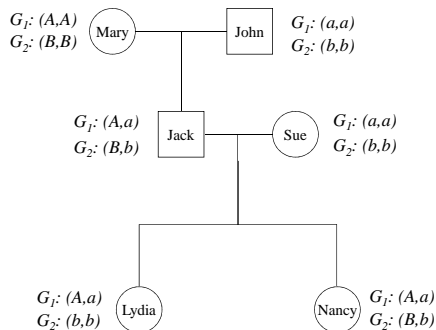
Maximize 
$$\prod_{i=1}^N \Pr(\mathbf{d}_i)$$

Given symptoms (chilling, body ache, sore throat, fever), pose MAP query on Cold, Flu, & Tonsillitis

Reduces to MPE if evidence covers all four symptoms

- ▶ Goal: map genes onto chromosome (i.e., determine location of genes on chromosome)
- ▶ Useful for detecting and predicting diseases
- ▶ Input: pedigree, observed genotype/phenotype

# Pedigree



- ▶ Genes:  $G_1$ ,  $G_2$
- ▶ Gene has *alleles* (states):  
 $G_1(A, a)$ ,  $G_2(B, b)$
- ▶ Individual carries two alleles per gene, from father & mother
- ▶ *Genotype*: collection of alleles of individual, composed of two *haplotypes*
- ▶ *Phenotype*: observable traits of individual

# Genotype and Phenotype

Genotype	Phenotype
A/A	blood type A
A/B	blood type AB
A/O	blood type A
B/B	blood type B
B/O	blood type B
O/O	blood type O

- ▶ Deterministic
- ▶ However, not in reverse direction: Blood type A  $\Rightarrow$  A/A or A/O

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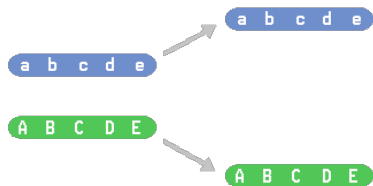
- ▶ Deterministic
- ▶ However, not in reverse direction: Blood type A  $\Rightarrow$  A/A or A/O

Genotype	Phenotype
H/H	healthy
H/D	healthy
D/D	ill with probability .9

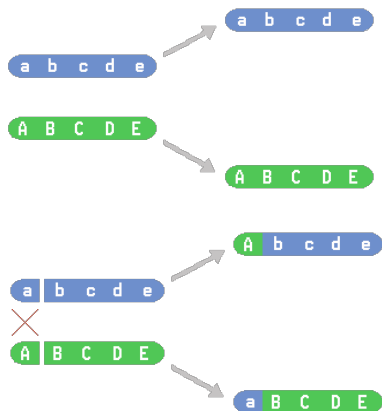
- ▶ Not deterministic
- ▶ *Penetrance*:  $\Pr(\text{phenotype} \mid \text{genotype})$



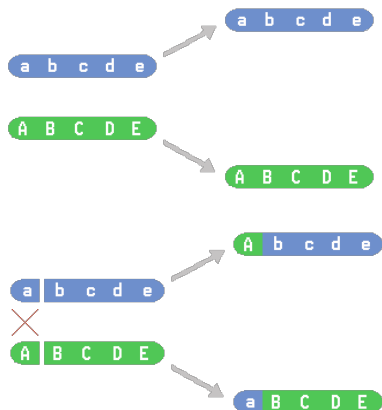
# Recombination



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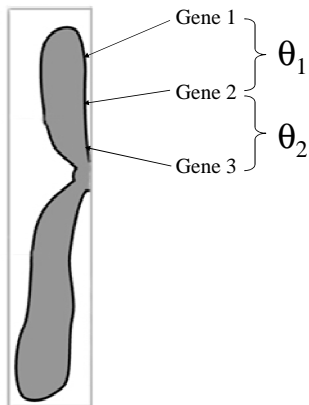


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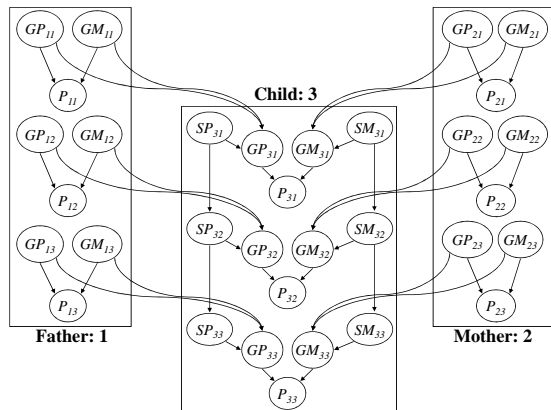


- ▶ Independent genes:  
*recombination frequency*  
 $\theta = 1/2$
- ▶ *Linked* genes:  $\theta < 1/2$
- ▶ Closer on chromosome  $\approx$   
more linked
- ▶ Genetic linkage analysis:  
Estimate distances between,  
and hence locations of,  
genes on chromosome based  
on data containing  
recombination information

# Recombination Frequencies

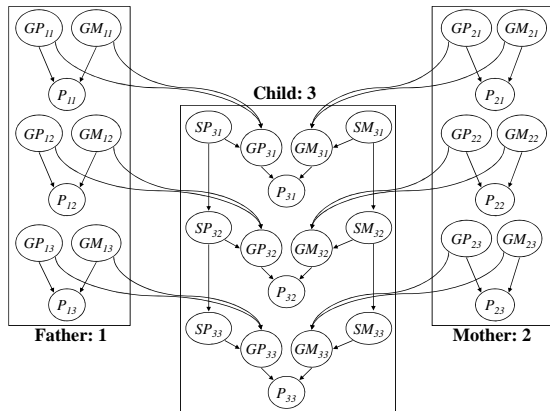


# Pedigree as Bayesian Network



- ▶ 3 genes:  $GP_{i1}/GM_{i1}$ ,  $GP_{i2}/GM_{i2}$ ,  $GP_{i3}/GM_{i3}$
- ▶ Phenotype:  $P_{i1}, P_{i2}, P_{i3}$
- ▶ Selectors:  $SP_{31}/SM_{31}$ ,  $SP_{32}/SM_{32}$ ,  $SP_{33}/SM_{33}$

# Pedigree as Bayesian Network

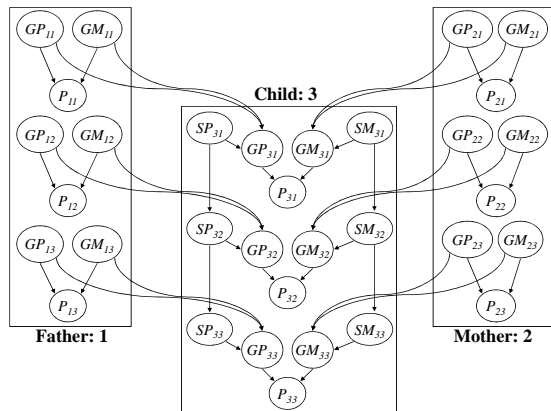


Encoding recombination frequencies

$SP_{31}$	$SP_{32}$	$\theta_{sp_{32} sp_{31}}$
$p$	$p$	$1 - \theta_{12}$
$p$	$m$	$\theta_{12}$
$m$	$p$	$\theta_{12}$
$m$	$m$	$1 - \theta_{12}$

Priors on  $GP/GM$  of founders obtained from population stats

# Pedigree as Bayesian Network



Given  $\mathbf{e}$  on subset of genotype ( $GP, GM$ ) and phenotype ( $P$ ),  $\Pr(\mathbf{e})$  indicates likelihood of  $\hat{\theta}$  (recombination frequencies)

Can compute preferred  $\hat{\theta}$ , or search for most likely  $\hat{\theta}$

Given  $\hat{\theta}$  and relative order of genes, determine their locations on chromosome

- ▶ Planning: Find plan (sequence of actions) to go from initial to goal state
- ▶ Probabilistic planning: In uncertain domains, find plan to reach goal with high probability



# Slippery Griper



Goal: Block painted and held,  
gripper clean

Probabilistic initial state

- ▶ Block not painted, not held
- ▶ Gripper clean, but dry with probability 0.7

Probabilistic action effects

- ▶ Paint: Paints block w/p 1; makes gripper dirty w/p 1 if it holds block, w/p 0.1 if not
- ▶ Pick-up: Succeeds w/p 0.5 if gripper wet, 0.95 if gripper dry
- ▶ Dry: Dries wet gripper w/p 0.8; doesn't affect dry gripper

- ▶ Probabilistic initial state and action effects
- ▶ Conformant: Action effects not observable (can't decide next action by observing environment)
- ▶ Need straight-line plan with max probability of success, for given horizon (number of steps)
- ▶ Example 2-step plan: [paint, pickup] (succeeds with probability 0.7335)

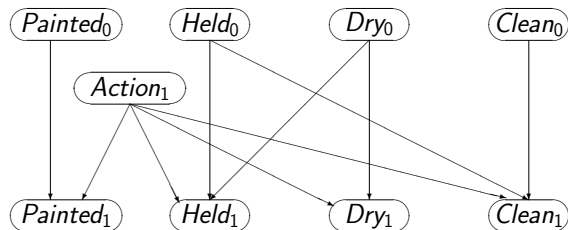
# Brute-force Approach

- ▶ Compute success probability for all plans of length one
- ▶ Given success probabilities of plans of length  $i$ , compute probability of success for plans of length  $i + 1$
- ▶ Iterate to planning horizon  $n$
- ▶ Pick  $n$ -step plan with max success probability
- ▶ Exponential in planning horizon

- ▶ Suppose *action variable*  $A_i$  represents action taken at step  $i$
- ▶ Planning  $\Leftrightarrow \max \Pr(\text{goal} | A_1, \dots, A_n)$   
 $\Leftrightarrow \max \Pr(\text{goal}, A_1, \dots, A_n)$

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- ▶ How to define Bayesian network?

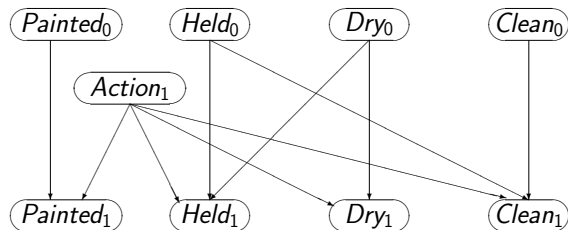
# Planning Domain as Bayesian Network



Paint: Paints block with probability 1

$Painted_0$	$Action_1$	$Painted_1$	Pr(.)
F	paint	T	1
T	*	T	1
...	...	...	...

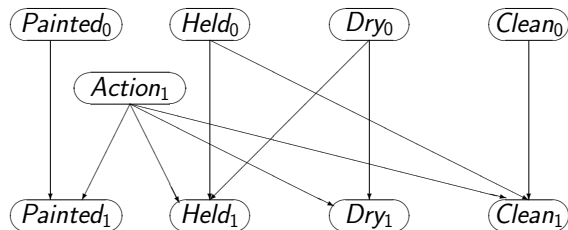
# Planning Domain as Bayesian Network



Paint: Makes gripper dirty w/p 1 if it holds block, w/p 0.1 if not

$Clean_0$	$Held_0$	$Action_1$	$Clean_1$	$Pr(.)$
T	T	paint	F	1
T	F	paint	F	.1
F	*	*	F	1
...	...	...	...	...

# Planning Domain as Bayesian Network



Add one layer per time step

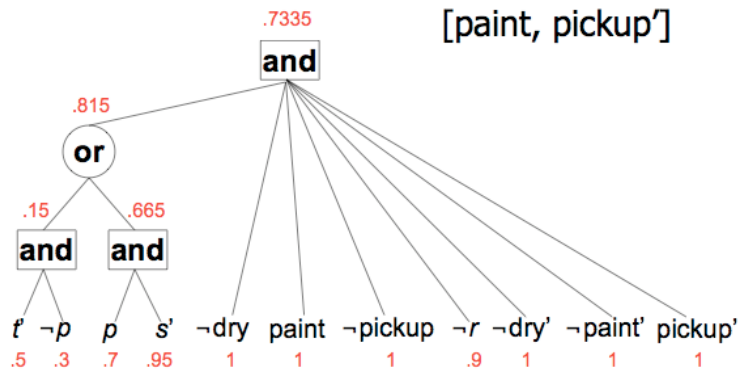
Initial state  $\rightarrow$  priors on  $Painted_0, Held_0, Dry_0, Clean_0$

Goal state  $\rightarrow$  evidence  $\mathbf{e}$  on  $Painted_n, Held_n, Dry_n, Clean_n$

Optimal plan  $\rightarrow \operatorname{argmax} \Pr(\mathbf{e}, A_1, \dots, A_n)$

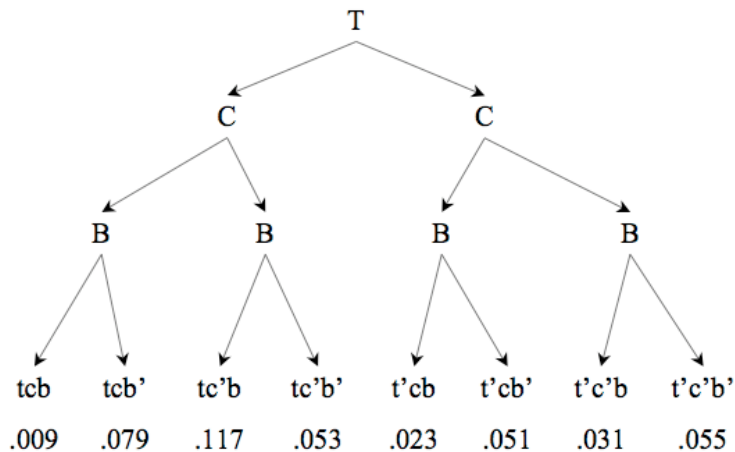
Priors on  $Action_i$ ; irrelevant



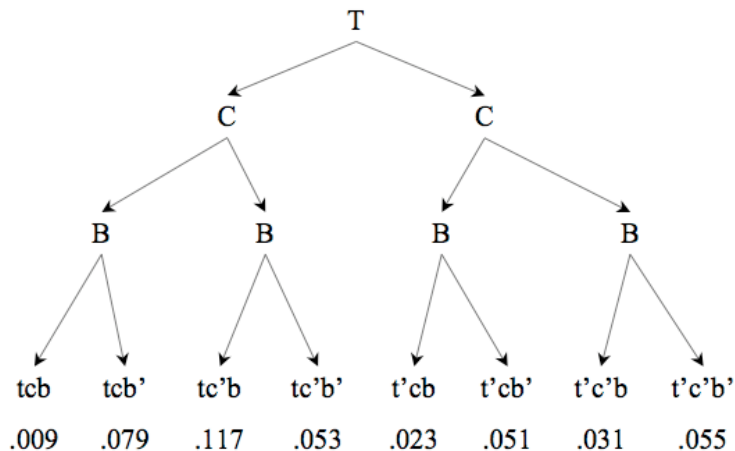


$$\Pr(\mathbf{e}, A_1, \dots, A_n)$$

# Plan Search

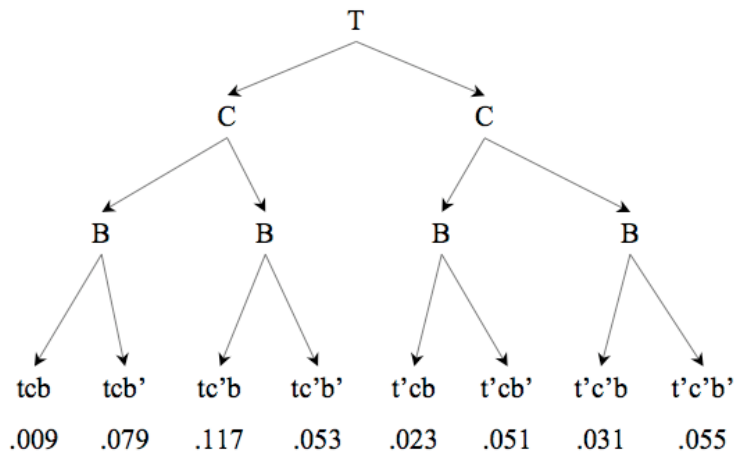


# Plan Search



$$\max_{C, B} \Pr(\mathbf{e}, t', C, B) \leq$$

# Plan Search



$$\max_{C, B} \Pr(\mathbf{e}, t', C, B) \leq .101$$

$$\max_{A_{k+1}, \dots, A_n} \Pr(\mathbf{e}, a_1, \dots, a_k, A_{k+1}, \dots, A_n)$$

$$\max_{A_{k+1}, \dots, A_n} \Pr(\mathbf{e}, a_1, \dots, a_k, A_{k+1}, \dots, A_n)$$

Some OR-nodes are decision nodes on  $A_{k+1}, \dots, A_n$

Turn these into max (instead of +)

Root of arithmetic circuit gives upper bound

- ▶ Bayesian networks as compact representations of probability distributions
- ▶ Inference by logical encoding and compilation to arithmetic circuits
- ▶ Inference by variable elimination
- ▶ Applications