Some context-specific graphical models for discrete longitudinal data

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Outline

Introduction

Automata

Maximum likelihood estimation

State merging

Hypothesis tests

Model selection

APFA equivalent to conventional Markov models

Summary and conclusion

Introduction

- Acyclic probabilistic finite automata[1] (APFA) are a rich family of models for discrete longitudinal data.
- An APFA
  - embodies a set of context-specific conditional independence relations
  - may be represented as a directed multigraph.
  - and is a context-specific graphical model.
- The methodology is highly scalable and is routinely used for high-dimensional genomic data in the Beagle software[2].
- Here we describe the models and methods from a statistical perspective.

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[1] Ron, Singer and Tishby (1998). On the learnability and usage of acyclic finite automata. J. Comp. Syst. Sci, 56, 133-52.

[2] Browning and Browning (2007). Rapid and accurate haplotype phasing and missing-data inference for whole-genome association studies by use of localized haplotype clustering. Am. J. Hum. Gen., 81, 1084-1097

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- **Automata**
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- APFA equivalent to conventional Markov models
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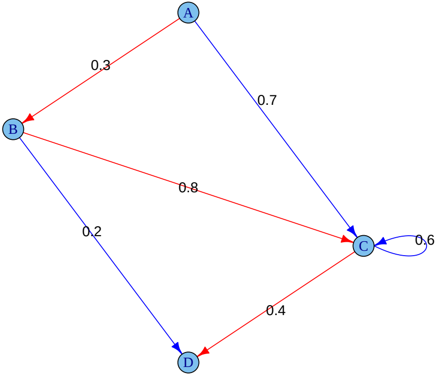
Automata

- Automata are devices that either input strings (as in parsing) or output strings.
- They are used in computer science and machine learning:
  - to represent formal languages and regular expressions;
  - for speech recognition,
  - natural language processing,
  - machine translation.
- We first consider the more general probabilistic finite automata (PFA) before focussing on the subclass of APFA.

Probabilistic Finite Automata

- A PFA is a device to generate random strings of symbols.
- It may be displayed as a directed multigraph, in which
  - nodes are called states,
  - there is one initial or root state with only outgoing edges, and one final or sink state with only incoming edges,
  - self-loops (edges from a state to itself) are allowed,
  - each edge e has a symbol $\sigma(e)$ and a probability $\pi(e)$, and
  - outgoing edges from each state have distinct symbols and the sum of their probabilities is unity.

A PFA



(a)

red='1'; blue='2'

How a PFA generates strings

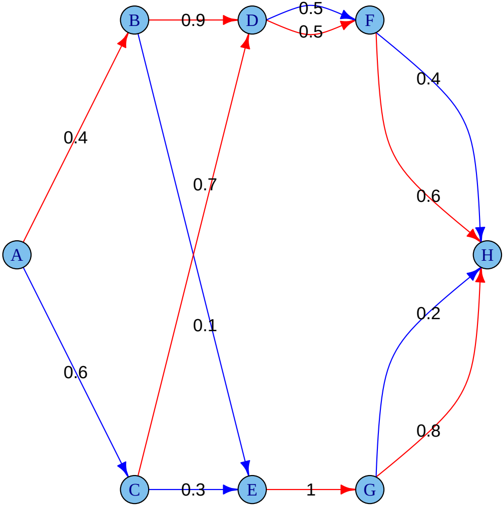
It starts at the root, then repeatedly

- chooses an outgoing edge at random according to the edge probabilities,
- emits the edge symbol,
- traverses the edge to the next state,

until it reaches the sink.

This generates symbol strings of possibly variable length.

An APFA



Acyclic Probabilistic Finite Automata

- An APFA $\mathcal{A}$ is a PFA that generates strings of constant length.
- So all root-to-sink paths have the same length p.
- So all paths from the root to any specific state have the same length, called the level of the state.
- Regard the strings as realizations of a random p-vector $\mathbf{X} = (X_1, X_2, \dots X_p)$.
- Distinct root-to-sink paths $\mathbf{e} = (e_1, e_2, \dots, e_p)$ generate distinct realizations of $\mathbf{X} = \sigma(\mathbf{e}) = (\sigma(e_1), \sigma(e_2), \dots, \sigma(e_p))$.
- The sample space of $\mathbf{X}$ is $\mathbb{X}(\mathcal{A}) = \{\sigma(\mathbf{e}) : \mathbf{e} \in \mathcal{E}(\mathcal{A})\}$, where $\mathcal{E}(\mathcal{A})$ is the set of root-to-sink paths in $\mathcal{A}$.
- For any $\mathbf{x} \in \mathbb{X}(\mathcal{A})$ there exists a unique root-to-sink path $\mathbf{e}$ such that $\mathbf{x} = \sigma(\mathbf{e})$: we write this as $\mathbf{e} = \sigma^{-1}(\mathbf{x})$.

A little theory

- The sample space of X_i, $\mathbb{X}_i$, is the set of symbols on edges incoming to a level i state.
- The parameters are the edge probabilities $\pi = \{\pi(e) : e \in E(\mathcal{A})\}$.
- The $\pi(\mathbf{e})$ specify the right-hand side of

$$\Pr(\mathbf{X} = \mathbf{x}) = \Pr(X_1 = x_1) \prod_{i=2\ldots p} \Pr(X_i = x_i | X_{<i} = x_{<i}) \quad (1)$$

where $\mathbf{X}_{<i} = (X_1, \ldots, X_{i-1})$, $\mathbf{x}_{\geq i} = (x_i, \ldots, x_p)$, $\mathbf{Y}_{\geq i; \leq j} = (Y_i, \ldots, Y_j)$ etc.

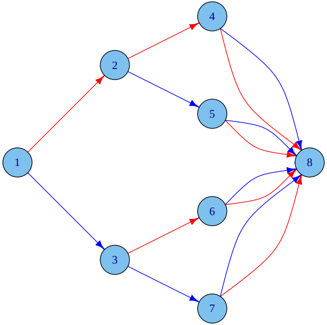
Context-specific conditional independences

- When the data generating process arrives at a level i state w, the distribution of $\mathbf{X}_{>i}$ does not depend on the path the process took to arrive at w. So

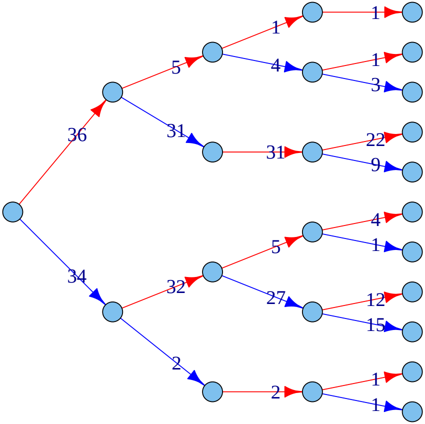
$$\mathbf{X}_{>i} \perp\!\!\!\perp \mathbf{X}_{\leq i} | \mathbf{X}_{\leq i} \in \mathcal{C}(w) \tag{2}$$

  where $\mathcal{C}(w) = \{\sigma(\mathbf{e}) : \mathbf{e} \in \mathcal{P}(w)\}$, and $\mathcal{P}(w)$ is the set of paths from the root to w.
- Thus an APFA expresses a set of context-specific conditional independence constraints on the distribution of $\mathbf{X}$.

Maximal and minimal APFA for three binary variables



A sample tree for $N = 70$ observations of 4 binary variables



To derive the maximal (unrestricted) APFA, contract the states at the last level.

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Likelihood

We draw independent samples $\mathbf{x}^{(v)}$ for $v = 1 \dots N$ from $\mathcal{A}$, and want to estimate the $\pi(e)$. We have

$$\Pr(\mathbf{x}) = \prod_{i=1 \dots p} \pi(e_i)$$

where $\mathbf{e} = \sigma^{-1}(\mathbf{x})$ so that the likelihood of the sample is

$$\prod_{v=1 \dots N} \prod_{i=1 \dots p} \pi(e_i^{(v)})$$

where $\mathbf{e}^{(v)} = \sigma^{-1}(\mathbf{x}^{(v)})$. This can be re-written as

$$\prod_{e \in E(\mathcal{A})} \pi(e)^{n(e)}$$

where $n(e)$ is the edge count, i.e. the number of observations in the sample whose root-to-sink path traverses the edge e.

Maximum likelihood

- So the log-likelihood is:

$$\ell(\mathcal{A}) = \sum_{e \in E(\mathcal{A})} n(e) \log \pi(e).$$

- which is easy to maximize:

$$\hat{\pi}(e) = \frac{n(e)}{n(v)}, \tag{3}$$

where $n(v)$ is the node count of v, the source node of e.

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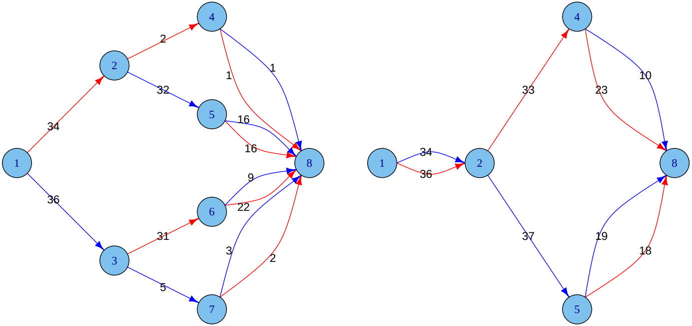
State merging

- Simplifying APFA involves state merging.
- Only states at the same level may be merged.
- Suppose we wish to merge state w into state v.
- That is, redirect all incoming edges to w to v instead, and all outgoing edges from w to outgo from v instead.
- This can lead to the existence of outgoing edges from v with duplicate symbols.
- Any such edges must therefore also be merged, and if their target nodes are distinct, these must also be merged.
- So the operation is recursive.
- Write $\mathcal{L}(s)$ for the merge-list induced by merging s. E.g. $\mathcal{L}(\{2,3\}) = \{2,3\}, \{5,7\}, \{4,6\}$.

An example of state merging



(a)

(b)

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Likelihood ratio tests

- We can construct likelihood ratio tests of nested hypotheses, that is of $\mathcal{A}_0$ versus $\mathcal{A}$, where $\mathcal{A}_0$ is a submodel of $\mathcal{A}$.
- For example, for the APFA shown 3 slides back, the deviance is

$$\begin{aligned} G^2 &= -2[\hat{\ell}(\mathcal{A}) - \hat{\ell}(\mathcal{A}_0)] \qquad (4) \\ &= 53.1228 \qquad (5) \end{aligned}$$

- Under $\mathcal{A}_0$, $G^2 \sim \chi^2(k)$ where k is the difference in model dimension (number of free parameters) between the models.
- By inspection we see that $\mathcal{A}$ has 7 free parameters and $\mathcal{A}_0$ has 4, so $k = 3$, and clearly $\mathcal{A}_0$ fits very poorly.

Likelihood ratio tests continued

- The same test can be computed by applying a standard contingency table test of independence to the table

source	(1,1)	(1,2)	(2,1)	(2,2)
2	2	3	22	9
3	16	16	1	1

- Recall that for an $r \times c$ table of counts $\{n_{ij}\}_{i=1\ldots r; j=1\ldots c}$ the deviance is

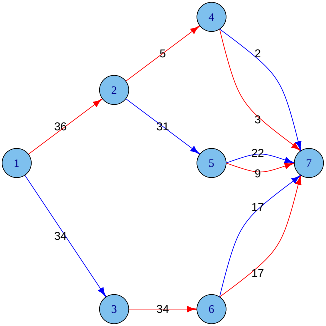
$$G^2 = 2 \sum_{i,j} n_{ij} \log \frac{n_{ij} n_{++}}{n_{i+} n_{+j}} \tag{6}$$

with degrees of freedom given as

$$k = (\#\{i : n_{i+} > 0\} - 1)(\#\{j : n_{+j} > 0\} - 1) \tag{7}$$

where n_{i+} and n_{+j} are the row and column totals, respectively.

Another example



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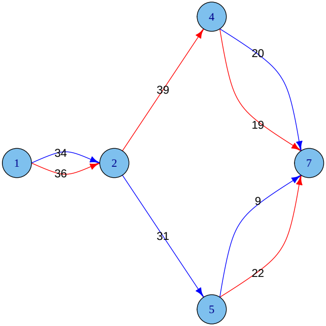

(b)

Adjusted degrees of freedom

- As before we can construct the contingency table

source	(1,1)	(1,2)	(2,1)	(2,2)
2	2	3	22	9
3	17	17	0	0

and find $G^2 = 67.288$ on 3 d.f.

- or we can decompose the test

element of $\mathcal{L}(2,3)$	2×2 table		G^2	df
(2,3)	5 34	31 0	67.112	1
(4,6)	2 17	3 17	0.176	1
sum			67.288	2

and find $G^2 = 67.288$ on 2 d.f.

- This is a sharper result that takes account of inestimability.
- We call these the adjusted degrees of freedom.
- For large APFA the unadjusted and adjusted degrees of freedom can differ considerably.

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The model selection algorithm of Ron et al (1998)

- The sample tree is constructed and then simplified in a series of state merging operations.
- Two nodes v and w at level i are merged

  $$\Pr(\text{future}|\mathbf{X}_{\leq i} \text{ goes through } v) = \Pr(\text{future}|\mathbf{X}_{\leq i} \text{ goes through } w).$$

  or in other words if $\forall\ \mathbf{x}_{>i}$,

  $$\Pr(\mathbf{X}_{>i} = \mathbf{x}_{>i}|\mathbf{X}_{\leq i} \in \mathcal{C}(v)) = \Pr(\mathbf{X}_{>i} = \mathbf{x}_{>i}|\mathbf{X}_{\leq i} \in \mathcal{C}(w)).$$

- The decision is based on a measure of similarity $\delta(v, w)$ between nodes v and w, and a fixed threshold, μ.
- v and w are called similar if $\delta(v, w) < \mu$: otherwise they are called dissimilar. Dissimilar nodes are not merged.

The algorithm

1. Start with the sample tree.
2. From level 1 to $p-1$:
   Repeatedly merge similar nodes until all the resulting nodes are pairwise dissimilar.
3. Merge all nodes at level p.

Similarity scores

- Ron et al proposed the similarity score

$$\delta_R(v,w) = \max_{k=i+1\dots p} \max_{\mathbf{x}_{i+1,\dots,k}}$$
$$|\hat{\Pr}(\mathbf{X}_{i+1,\dots,k} = \mathbf{x}_{i+1,\dots,k} | \mathbf{X}_{\leq i} \in \mathcal{C}(v)) - \hat{\Pr}(\mathbf{X}_{i+1,\dots,k} = \mathbf{x}_{i+1,\dots,k} | \mathbf{X}_{\leq i} \in \mathcal{C}(w))|$$

- We propose instead a score based on the penalized likelihood criterion

$$IC(\mathcal{A}) = -2\hat{\ell}(\mathcal{A}) + \alpha \dim(\mathcal{A}) \tag{8}$$

namely

$$\begin{aligned} \delta_{IC}(v,w) &= IC(\mathcal{A}_0) - IC(\mathcal{A}) \\ &= G^2 - \alpha k \end{aligned} \tag{9}$$

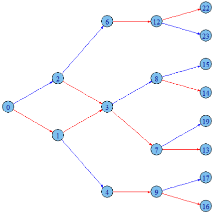
We set $\mu = 0$, so that two nodes are similar whenever merging them decreases the IC.

- Thus the selection algorithm seeks to minimize the IC.
- We are currently comparing the performance of this algorithm with the one in Beagle.

An example

Level	Node pair	G^2	k	δ_{IC}	Action
1	1,2	53.98	5	32.74	go to next level
2	3,4	20.78	3	8.03	
2	3,5	1.03	3	-11.71	
2	3,6	5.60	3	-7.14	
2	4,5	58.49	3	45.74	
2	4,6	0.36	1	-3.89	
2	5,6	7.43	3	-5.31	merge 5 into 3
2	3,4	61.36	3	48.62	
2	3,6	7.60	3	-5.15	
2	4,6	0.36	1	-3.89	merge 6 into 3
2	3,4	56.60	3	43.85	go to next level
3	7,8	2.88	1	-1.37	
3	7,9	0.05	1	-4.19	
3	8,9	5.40	1	1.15	merge 9 into 7
3	7,8	6.41	1	2.16	stop

An example



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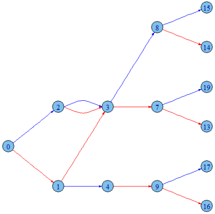

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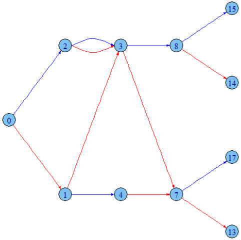

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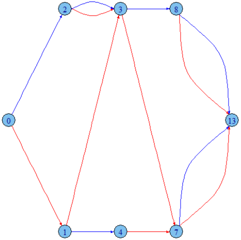

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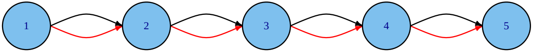
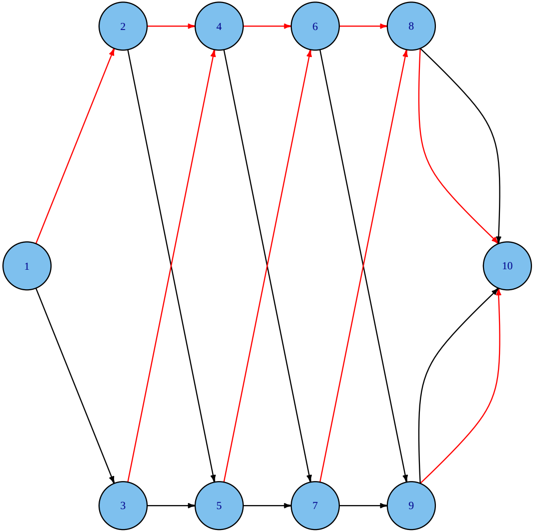
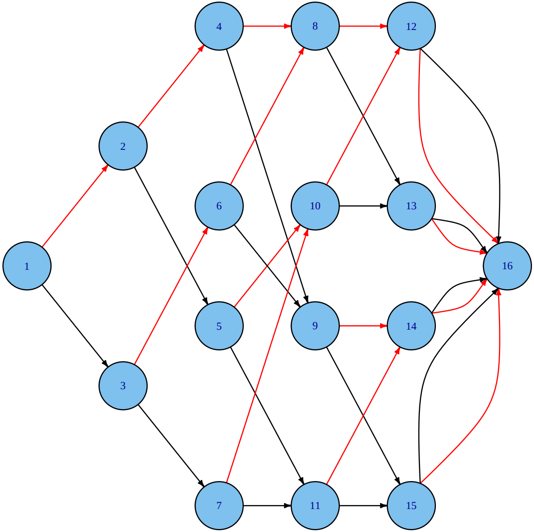
Independence



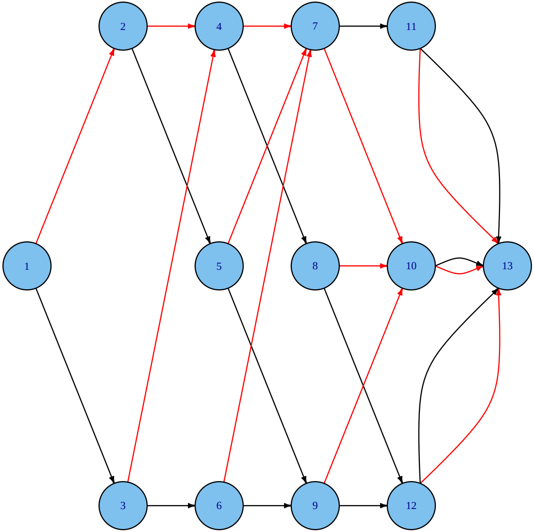
First order Markov



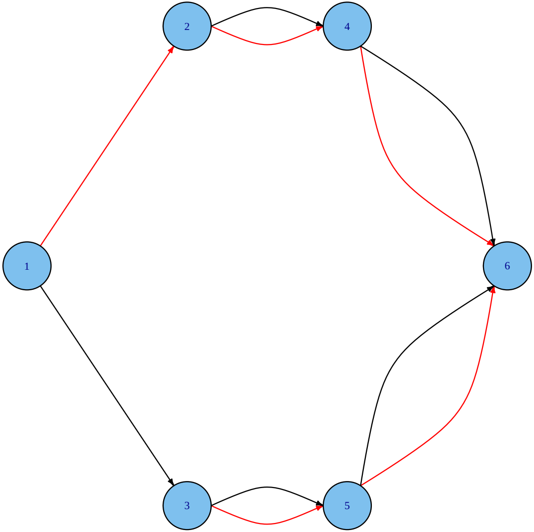
Second order Markov



Variable order Markov



Memory gap Markov



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- This talk has tried to describe APFA as statistical models.
- An APFA embodies a set of context-specific conditional independence relations, and may be represented as a directed multigraph.
- So it may be called a context-specific graphical model.
- APFA form a very rich class of models for discrete longitudinal data.
- We have shown how likelihood ratio tests may be constructed, and used this to modify the selection algorithm of Ron et al. (1998).
- We are preparing an R package to work with the models.