Learning Nonsingular Phylogenies
and Hidden Markov Models

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Abstract

In this paper, we study the problem of learning phylogenies and hidden Markov models. We call
the Markov model nonsingular if all transition matrices have determinants bounded away from 0 (and 1).
We highlight the role of the nonsingularity condition for the learning problem. Learning hidden Markov
models without the nonsingularity condition is at least as hard as learning parity with noise. On the other
hand, we give a polynomial-time algorithm for learning nonsingular phylogenies and hidden Markov
models.

Keywords: Hidden Markov models, evolutionary trees, phylogenetic reconstruction, PAC learning.
1 Introduction

This paper studies the problem of learning hidden Markov models and more generally learning phylogenies. Phylogenies are Markov models defined on trees where all the internal degrees are exactly 3. Hidden Markov models are a special case of phylogenies where only “caterpillar” trees are allowed, see Figure 1.

The reconstruction of phylogenies is one of the most important tasks of molecular biology, see e.g. [Fe04, SS03]. Many works have been devoted to the reconstruction of phylogenies in the learning setting. In particular, in [AD+97] the authors obtain almost optimal upper and lower bounds for learning a phylogenetic “star” tree on 2-state space given its topology (i.e. the underlying graph structure), where the mutation matrices are symmetric. In [CGG02], a polynomial-time algorithm is obtained for general Markov models on 2-state spaces. The authors of [CGG02] also make the claim that their technique extends to the general $k$-state model, but then restrict themselves to $k = 2$.

The framework of Markov models on trees have several special cases that are of independent interest. The case of Product distributions is discussed in [AD+97]. The case of mixture of product distributions is discussed in [FOS04]. Arguably, the most interesting case is that of learning hidden Markov models (HMMs). HMMs play a crucial role in many areas from speech recognition to biology, see e.g. [Ra89, CD+99].

In [AW92] it is shown that finding the “optimal” HMM for an arbitrary distribution is hard unless $RP = NP$. See also [LP01] where hardness of approximation results are obtained for problems such as comparing two hidden Markov models. Most relevant to our setting is the conjecture made in [KM+94] that learning parity with noise is hard. It is easy to see that the problem of learning parity with noise may be encoded as learning an HMM over 4 states, see subsection 1.3.

There is an interesting discrepancy between the two viewpoints taken in works concerning learning phylogenies and works concerning learning hidden Markov models. The results in phylogeny are mostly positive – they give polynomial-time algorithms for learning. On the other hand, the results concerning HMMs are mostly negative.

This paper tries to resolve the discrepancy between the two points of view by pointing to the source of hardness in the learning problem. Roughly speaking, we claim that the source of hardness for learning phylogenies and hidden Markov models are transition matrices $P$ such that $\det P = 0$ but $\operatorname{rank} P > 1$. Note that in the case $k = 2$ there are no such matrices and indeed the problem is not hard [CGG02]. We note furthermore that, indeed, in the problem of learning parity with noise all of the determinants are 0 and all the ranks are greater than 1.

The main technical contribution of this paper is to show that the learning problem is feasible once all the matrices have $1 - \beta > |\det P| > \beta$ for some $\beta > 0$. We thus present a proper PAC learning algorithm for this case. We believe that the the requirement that $|\det P| < 1 - \beta$ is an artifact of our proofs. Indeed, in the case of hidden Markov models we prove that the model can be learned under the weaker condition that $|\det P| > 1/poly(n)$. Assuming that learning parity with noise is indeed hard, this is an optimal result, see subsection 1.3.

The learning algorithms we present are based on combination of techniques from phylogeny, statistics, combinatorics and probability. We believe that these algorithms may be also extended to cases where $|\det P|$ is close to 1 and furthermore to cases where if $|\det P|$ is small, then the matrix $P$ is close to a rank 1 matrix, thus recovering the results of [AD+97, CGG02].

Interestingly, to prove our result we use and extend several previous results from combinatorial phylogeny and statistics. The topology of the tree is learned via variants of combinatorial results proved in phylogeny [ES+97]. Thus, the main technical challenge is to learn the mutation matrices along the edges.
For this we follow and extend the approach developed in statistics by Chang [Ch96]. Chang’s results allow the recovery of the mutation matrices from infinite number of samples. The reconstruction of the mutation matrices from a polynomial number of samples requires a delicate error analysis along with various combinatorial and algorithmic ideas.

The algorithm is sketched in Section 2 and the error analysis is performed in Section 3. Most details can be found in the appendix.

1.1 Definitions and Results

We let \( T_3(n) \) denote the set of all trees on \( n \) labeled leaves where all internal degrees are exactly 3. Note that if \( T = (\mathcal{V}, \mathcal{E}) \in T_3(n) \) then \( |\mathcal{V}| = 2n - 2 \) We will sometimes omit \( n \) from the notation. Below we will always assume that the leaf set is labeled by the set \([n]\). We also denote the leaf set by \( \mathcal{L} \). Two trees \( T_1, T_2 \) are considered identical if there is a graph isomorphism between them that is the identity map on the set of leaves \([n]\). We define a caterpillar to be a tree on \( n \) nodes that is defined in the following way: It consists of a path \( v_1, \ldots, v_r \) where each of the nodes \( v_i \) is attached to at least 1 of the nodes in the leaf set \([n]\). Furthermore, the degree of each of the \( v_i \)’s is 3. See Figure 1 for an example. We let \( TC_3(n) \) denote the set of all caterpillars on \( n \) labeled leaves.

In a Markov model \( T \) on a (undirected) tree \( T = (\mathcal{V}, \mathcal{E}) \) rooted at \( r \), each vertex iteratively chooses its state given the one of its parent by an application of a Markov transition rule. Consider the orientation of \( \mathcal{E} \) where all edges are directed away from the root. We note this set of directed edges to be a tree on \( n \) nodes that is defined in the following way: It consists of a path \( v_1, \ldots, v_r \) where each of the nodes \( v_i \) is attached to at least 1 of the nodes in the leaf set \([n]\). Furthermore, the degree of each of the \( v_i \)’s is 3. See Figure 1 for an example. We let \( TC_3(n) \) denote the set of all caterpillars on \( n \) labeled leaves.

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\[
\pi^T(s) = \pi^T_r(s(r)) \prod_{(u,v) \in \mathcal{E}_r} P^{uv}_{s(u)s(v)},
\]

where \( s \in C^V \), \( C \) is a finite state space, \( P^{uv} \) is the transition matrix for edge \((u,v) \in \mathcal{E}_r \) and \( \pi^T_r \) is the distribution at the root. We let \( k = |C| \). We write \( \pi^T_{s^V} \) for the marginals of \( \pi^T \) on the set \( \mathcal{W} \). Since the set of leaves is labeled by \([n]\), the marginal \( \pi^T_{|[n]} \) is the marginal on the set of leaves. We will often remove the superscript \( T \). Furthermore, for two vertices \( u, v \in \mathcal{V} \) we let \( P^{uv}_{ij} = \mathbb{P}[s(v) = j | s(u) = i] \). We will be mostly interested in nonsingular Markov models.

**Definition 1** We say that a Markov model on a tree \( T = (\mathcal{V}, \mathcal{E}) \) is nonsingular ((\( \beta, \sigma \))-nonsingular) if

I. For all \( e \in \mathcal{E}_r \) it holds that \( 1 > |\det P^e| > 0 \) (1 - \( \beta \) > \( |\det P^e| \) > \( \beta \)) and

II. For all \( v \in \mathcal{V} \) it holds that \( \pi_v(i) > 0 \) (\( \pi_v(i) > \sigma \)) for all \( i \) in \( C \).

We will call the model nonsingular ((\( \beta, \sigma \))-nonsingular) at 0 if instead of I, we have the weaker condition \( |\det P^e| > 0 \) (\( |\det P^e| \) > \( \beta \)).

It is well known that if the model is nonsingular, then for each \( w \in \mathcal{V} \), one can write

\[
\pi^T(s) = \pi^T_v(s(w)) \prod_{(u,v) \in \mathcal{E}_w} P^{uv}_{s(u)s(v)},
\]

(where now all edges \((u,v) \) are oriented away from \( w \)). In other words, the tree may be rooted arbitrarily. Indeed, in the learning algorithms discussed below, we will root the tree arbitrarily. We will actually refer to \( \mathcal{E} \) as the set of directed edges formed by taking the two orientations of all (undirected) edges in the tree. It is
easy to show that \((\beta, \sigma)\)-nonsingularity as stated above also implies that property I. holds for all \((u, v) \in E\) with appropriate values of \(\beta, \sigma\).

Note moreover, that if \(|\det P^u| > 0\) for all edges \((u, v)\) and for all \(v \in V\) the distribution of \(s(v)\) is supported on at most \(|C| - 1\) elements then one can redefine the model by allowing only \(|C| - 1\) values of \(s(v)\) at each node and deleting the corresponding row and columns from the transition matrices \(P^e\). Thus condition II. is very natural given condition I.

Given a collection \(M\) of mutation matrices \(P\), we let \(T_3(n) \otimes M\) denote all phylogenetic trees of the form (1) where \(T \in T_3\) and \(P^e \in M\) for all \(e\). We similarly define \(TC_3 \otimes M\).

We use the PAC learning framework introduced by [Va84], in its variant proposed by [KM+94]. Let \(\varepsilon > 0\) denote an approximation parameter, \(\delta > 0\), a confidence parameter, \(M\), a collection of matrices and \(T\), a collection of trees. Then, we say that an algorithm \(\mathcal{A}\) PAC-learns \(T \otimes M\) if for all \(T \in T \otimes M\), given access to samples from the measure \(\pi^T_{[n]}\), \(\mathcal{A}\) outputs a phylogenetic tree \(T'\) such that the total variation distance between \(\pi^T_{[n]}\) and \(\pi^{T'}_{[n]}\) is smaller than \(\varepsilon\) with probability at least \(1 - \delta\) and the running time of \(\mathcal{A}\) is \(\text{poly}(n, 1/\delta, 1/\varepsilon)\).

In our main result we prove the following.

**Theorem 1** For every constant \(\beta, \kappa_\pi > 0\) and every finite set \(C\), the collection of \((\beta, n^{-\kappa_\pi})\)-nonsingular Markov phylogenetic models is PAC-learnable. More formally, let \(C\) be a finite set, \(\beta, \kappa_\pi > 0\) and \(M\) denote the collection of all \(|C| \times |C|\) transition matrices \(P\) where \(1 - \beta > |\det P| > \beta\). Then there exists a PAC-learning algorithm for \(T_3 \otimes M\) whose running time is \(\text{poly}(n, 1/\varepsilon, 1/\delta)\), provided the node distributions satisfy \(\pi_v(i) > n^{-\kappa_\pi}\) for all \(v \in V\) and \(i \in C\).

For hidden Markov models we can prove more.

**Theorem 2** Let \(\phi_\delta, \phi'_\delta, \kappa_\pi > 0\) be constants. Let \(C\) be a finite set and \(M\) denote the collection of \(|C| \times |C|\) transition matrices \(P\) where \(1 - n^{-\phi_\delta} > |\det P| > n^{-\phi'_\delta}\). Then there exists a PAC-learning algorithm for \(TC_3 \otimes M\), provided the node distributions satisfy \(\pi_v(i) > n^{-\kappa_\pi}\) for all \(v \in V\) and \(i \in C\). The running time of the algorithm is \(\text{poly}(n, 1/\varepsilon, 1/\delta)\). If the topology is known, which is often the case in applications of HMMs, we can relax the assumption on determinants to \(1 \ge |\det P| > n^{-\phi_\delta}\).

### 1.2 Inferring the Topology Using Combinatorial Phylogeny

We let the topology of \(T\) denote the underlying tree \(T = (V, E)\). The task at hand can be divided into two natural subproblems. First, the topology of \(T\) needs to be recovered with high probability. Second, the transition matrices have to be estimated. Reconstructing the topology has been a major task in phylogeny. It follows from [ES+97, ES+98] that the topology can be recovered with high probability using a polynomial number of samples. Here is one formulation from [Mo04].

**Theorem 3** Let \(\beta > 0\) and suppose that \(M\) consists of all matrices \(P\) satisfying \(\beta < |\det P| < 1 - \beta\). For all \(\kappa_T > 0\), the topology of \(T \in T_3 \otimes M\) can be learned in polynomial time using \(\kappa_T n^{O(1/\beta)}\) samples with probability \(1 - n^{-2-\kappa_T}\).

We will also need a stronger result that applies only for hidden Markov models. This result essentially follows from [ES+97, ES+98]. We briefly sketch the additional arguments needed in the appendix.

**Theorem 4** Let \(\zeta, \tau > 0\) and suppose that \(M\) consists of all matrices \(P\) satisfying \(n^{-\zeta} < |\det P| < 1 - n^{-\tau}\). Then for all \(\theta > 0\), and all \(T \in TC_3 \otimes M\) one can recover from \(n^{O(\zeta+\tau+1/\theta)}\) samples the topology \(T\) with probability \(1 - n^{-2-\theta}\).
Figure 1: Hidden Markov model for noisy parity. The model computes $N \oplus \bigoplus_{i \in T} x_i$, where the $x_i$'s are uniform over $\{0, 1\}$, $T$ is a subset of $\{1, \ldots, n\}$, and $N$ is a small random noise. The $S_i$'s are the partial sums over variables included in $T$. The observed nodes are in light gray. In the hardness proof, the passage from learning a distribution (HMM) to learning a function (parity) requires the notion of an evaluator, i.e. the possibility of computing efficiently the probability of a particular state. Here this can be done by a dynamic programming type algorithm.

1.3 Hardness of learning singular Markov models

We now briefly explain why hardness of learning “parity with noise” implies that learning singular hidden Markov models is hard. Kearns’ work [Ke93] on the statistical query model leads to the following conjecture.

Conjecture 1 (Noisy Parity Assumption [Ke93]) There is a $0 < \alpha < 1/2$ such that there is no efficient algorithm for learning parity under uniform distribution in the PAC framework with classification noise $\alpha$.

In [KM+94], this is used to show that learning probabilistic finite automata with an evaluator is hard. It is easy to see that the same construction works with the probabilistic finite automata replaced by an equivalent hidden Markov model (HMM) with 4 states (this is a special case of our evolutionary tree model when the tree is linear). The proof, which is briefly sketched in Figure 1, is left to the reader. We remark that all matrices in the construction have determinant 0 and rank 2. Note moreover that from a standard coupling argument it follows that if for all edges $(u, v)$ we replace the matrix $P^{uv}$ by the matrix $(1 - n^{-\gamma})P^{uv} + n^{-\gamma}I$, then the model given in Figure 1 and its variant induces indistinguishable distributions on $k$ samples if $k \leq o(n^{\gamma-1})$. This shows that assuming that learning parity with noise is hard, Theorem 2 is tight up to the constant in the power of $n$.

2 Overview of the Algorithm

2.1 Chang’s Spectral Technique

One of the main ingredients of the algorithm is a very nice result due to Chang [Ch96]. Let $T$ be a 4-node (star) tree with a root $r$ and 3 leaves $a, b, c$. Let $P^{uv}$ be the transition matrix between vertices $u$ and $v$, i.e. $P^{uv}_{ij} = P[s(v) = j | s(u) = i]$ for all $i, j \in \mathcal{C}$. Fix $\gamma \in \mathcal{C}$. Then by the Markov property, for all $i, j \in \mathcal{C}$,

$$
P[s(c) = \gamma, s(b) = j | s(a) = i] = \sum_{h \in \mathcal{S}} P^{ar}_{ih} P^{rc}_{h\gamma} P^{rb}_{hj},
$$

or in matrix form

$$
P^{ab, \gamma} = P^{ar} \text{diag}(P^{rc}_{\gamma}) P^{rb},
$$
where the matrix $P_{ab}^{i,j}$ is defined by $P_{ij}^{ab} = \Pr[s(c) = \gamma, s(b) = j | s(a) = i]$ for all $i, j \in \mathcal{C}$. Then, noting that $(P_{ab})^{-1} = (P_{rb})^{-1}(P_{ar})^{-1}$, we have

$$P_{ab}^{i,j} = \Pr[s(b) = j | s(a) = i],$$

assuming the transition matrices are invertible. Equation (\star) is an eigenvalue decomposition where the l.h.s. involves only the distribution at the leaves. Therefore, given the distribution at the leaves, we can recover from (\star) the columns of $P_{ar}$ (up to scaling) provided the eigenvalues are distinct. Note that the above reasoning applies when the edges $(r, a), (r, b), (r, c)$ are replaced by paths. Therefore, loosely speaking, in order to recover an edge $(w, w')$, one can use (\star) on star subtrees with $w$ and $w'$ as roots to obtain $P_{aw}$ and $P_{aw'}$, and then compute $P_{ww'} = (P_{aw})^{-1}P_{aw'}$. In [Ch96], under further assumptions on the structure of the transition matrices, the above scheme is used to prove the identifiability of the full model, that is that the output distributions on triples of leaves uniquely determine the transition matrices. In this paper, we show that the transition matrices can actually be approximately recovered using (\star) with a polynomial number of samples.

There are many challenges in extending Chang’s identifiability result to our efficient reconstruction claim. First, as noted above, equation (\star) uncovers only the columns of $P_{ar}$. The leaves actually give no information on the labelings of the internal nodes. To resolve this issue, Chang assumes that the transition matrices come in a canonical form that allows to reconstruct them once the columns are known. For instance, if in each row, the largest entry is always the diagonal one, this can obviously be performed. This assumption is a strong and unnatural restriction on the model we wish to learn and therefore we seek to avoid it. The point is that relabeling all internal nodes does not affect the output distribution, and therefore the internal labelings can be made arbitrarily (in the PAC setting). The issue that arises is that those arbitrary labelings have to be made consistently over all edges sharing a node. Another major issue is that the leaf distributions are known only approximately through sampling. This requires a delicate error analysis and many tricks which are detailed in Section 3. The two previous problems are actually competing. Indeed, one way to solve the consistency issue is to fix a reference leaf $\omega$ and do all computations with respect to the reference leaf, that is choose $a = \omega$ in every spectral decomposition. However, this will necessitate the use of long paths on which the error builds up uncontrollably. Our solution is to partition the tree in smaller subtrees, reconstruct consistently the subtrees using one of their leaves as a reference, and patch up the subtrees by fixing the connecting edges properly afterwards. We refer to the connecting edges as separators. The algorithm FULLRECON thus consists of two phases as follows. It assumes the knowledge of distributions on triples of leaves. These are known approximately through sampling. Exactly how this is done is detailed in Section 3.

**Input:** Topology and distributions on triples of leaves;  
**Output:** Mutation matrices;  

- **Phase 1: Subtree Reconstruction**  
  - Use subroutine LEAFRECON to simultaneously: 1) partition the tree into 'small’ subtrees $T_{a_0}, \ldots, T_{a_T}$; and 2) reconstruct consistently the mutation matrices of all subtrees using their respective reference leaves $a_0, \ldots, a_T$;  
- **Phase 2: Patching Up**  
  - Reconstruct the separators with subroutine SEPRECON.  

The two subroutines are described next. A more detailed version of the algorithm appears in the appendix. The correctness of the algorithm uses the error analysis and is therefore left for Section 3.
Figure 2: Schematic representation of the execution of LEAFRECON. The only edges shown are separators.

2.2 Subtree Reconstruction and Patching Up

We need the following notations to describe the subroutines. If $e = (u, v)$, let $d_1(e)$ (resp. $d_2(e)$) be the length of the shortest path (in number of edges) from $u$ (resp. $v$) to a leaf in $L$ not using edge $e$. Then the depth of $T$ is

$$
\Delta = \max_{e \in T} \{ \max\{d_1(e), d_2(e)\} \}.
$$

It is easy to argue that $\Delta \leq \log n$ (see Section 3). Also, for a set of vertices $W$ and edges $S$, denote $\mathcal{N}(W, S)$ the set of nodes not in $W$ that share an edge in $E \setminus S$ with a node in $W$ ("outside neighbors" of $W"$ without using edges in $S"$). Let $\mathcal{B}_a^\Delta$ be the subset of nodes in $V$ at distance at most $\Delta$ from leaf $a$.

**Subroutine LEAFRECON.** The subtree reconstruction phase is performed by the algorithm LEAFRECON depicted in Figure 2. The purpose of LEAFRECON is to partition the tree into subtrees each of which has the property that all its nodes are at distance at most $\Delta$ from one of its leaves (same leaf for all nodes in the subtree), that we refer to as the reference leaf of the subtree. The correctness of the algorithm, proved in Section 3, thus establishes the existence of such a partition. This partition serves our purposes because it allows 1) to reconstruct mutation matrices in a consistent way (in each subtree) using reference leaves, and 2) to control the build up of the error by using short paths to the reference leaf. The matrix reconstruction is performed simultaneously by LEAFRECON, as the partition is built. At the call of LEAFRECON, we consider the subgraph $T'$ of $T$ where edges previously labeled as separators have been removed. We are given a reference leaf $a$ and restrict ourselves further to the (connected) subtree $T_a$ of $T'$ consisting of nodes at distance at most $\Delta$ from $a$. Moving away from $a$, we recover edge by edge the mutation matrices in $T_a$ by Chang’s spectral decomposition. At this point, it is crucial 1) that we use the transition matrices previously computed to ensure consistency in the labeling of internal nodes, and 2) in order to control error, that we choose the leaves $b$ and $c$ (in the notations of the previous subsection) to be at distance at most $\Delta + 1$ from the edge currently reconstructed (which is always possible by definition of $\Delta$). Note that the paths from the current node to $b$ and $c$ need not be in $T_a$. Once $T_a$ is reconstructed, edges on the “outside boundary” of $T_a$ (edges in $T'$ with exactly one endpoint in $T_a$) are added to the list of separators, each with a new reference leaf taken from the unexplored part of the tree (at distance at most $\Delta$). The algorithm LEAFRECON is then run on those new reference leaves, and so on until the entire tree is recovered (see Figure 2). More precisely, the algorithm goes as follows (see also the appendix for a more detailed version). Some steps are detailed in Section 3. We denote estimates with hats, e.g. the estimate of $P^\text{ar}$ is $\hat{P}^\text{ar}$.
As pointed out in the previous section, the distribution on the leaves is known only approximately through sampling. The purpose of this section is to account for the error introduced by this approximation. We obtain modifications are described where needed in the course of the analysis. For a vertex \(v\), the all-one vector (the size is usually clear from the context). For any vector \(x\), \(\|x\|_1 = \sum_i |x_i|\), and for a matrix \(X\), \(\|X\|_1 = \max_j \sum_i |x_{ij}|\). Recall that \(\|L\|\) stands for the set of leaves. We assume throughout that the model is \((\beta, n^{-\kappa_\pi})\)-nonsingular, for \(\beta, \kappa_\pi > 0\) constant, and that the topology is known. We prove the following theorem.

**Theorem 5** For all \(\varepsilon, \delta > 0\) and \(n\) large enough, our reconstruction algorithm produces a Markov model satisfying

\[
\|\hat{\pi}_L - \pi_L\|_1 \leq \varepsilon,
\]

with probability at least \(1 - \delta\). The running time of the algorithm is polynomial in \(n, 1/\varepsilon, 1/\delta\).

From here on, all \(\kappa\)'s are meant to be exponents of \(n\) (not depending themselves on \(n\)). We use the expression with high probability (w.h.p.) to mean with probability at least \(1 - 1/poly(n)\). Likewise we say negligible to mean at most \(1/poly(n)\). In both definitions it is implied that \(poly(n) = O(n^K)\) for a constant \(K\) that can be made as large as one wants if the number of samples is \(O(n^{K'})\) with \(K'\) large enough. The proofs from this section appear in the appendix. In all proofs, we assume that the bounds proved in previous lemmas hold.
apply, even though they actually occur only with high probability. We also assume in all lemmas that the various \(\kappa\)'s from previous lemmas have been chosen large enough for the results not to be vacuous. This is always possible if the number of samples is taken (asymptotically) large enough. Standard linear algebra results used throughout the analysis can be found e.g. in [HJ85].

3.1 Approximate Spectral Argument

In this subsection, we address several issues arising from the application of Chang’s spectral technique to an approximate distribution on the leaves. Our discussion is summarized in Proposition 1. The precise statements of the lemmas and their proofs can be found in the appendix. We use the notations of Section 2.1.

Determinants on Paths (ref. Lemmas 1 and 2). The estimation error depends on the determinant of the transition matrices involved. Since we use Chang’s spectral technique where \(a \rightarrow r, r \rightarrow b,\) and \(r \rightarrow c\) are paths rather than edges, we need a lower bound on transition matrices over paths. This is where the use of short paths is important. Multiplicativity of determinants gives immediately that all determinants of transition matrices on paths of length \(O(\Delta)\) are at least \(1/poly(n)\).

Error on Leaf Distributions (ref. Lemmas 3 and 4). The algorithm estimates leaf distributions through sampling. We need to bound the error introduced by sampling. Let \(a, b, c\) be leaves at distance at most \(2\Delta + 1\) from each other and consider the eigenvalue decomposition \((\ast)\). We estimate \(P^{ab}\) by taking \(poly(n)\) samples and computing

\[
\hat{P}^{ab}_{ij} = \frac{N_{i,j}^{a,b}}{N_i^a},
\]

for \(i, j \in \mathcal{C}\) where \(N_i^a\) is the number of occurrences of \(s(a) = i\) and \(N_{i,j}^{a,b}\) is the number of occurrences of \(s(a) = i, s(b) = j\). Likewise for \(P^{ab,\gamma}_{ij}\), we use \(poly(n)\) samples and compute the estimate

\[
\hat{P}^{ab,\gamma}_{ij} = \frac{N_{i,j,\gamma}^{a,b,c}}{N_i^a},
\]

where \(N_{i,j,\gamma}^{a,b,c}\) is the number of occurrences of \(s(a) = i, s(b) = j, s(c) = \gamma\). We also bound the error on the 1-leaf distributions; this will be used in the next subsection. We use \(poly(n)\) samples to estimate \(\pi_a\) using empirical frequencies. Standard concentration inequalities give that \(\|P^{ab} - \hat{P}^{ab}\|_1, \|P^{ab,\gamma} - \hat{P}^{ab,\gamma}\|_1,\) and \(\|\pi_a - \hat{\pi}_a\|_1\) are negligible w.h.p.

Separation of Exact Eigenvalues (ref. Lemma 5). In Section 2, it was noted that the eigenvalues in \((\ast)\) need to be distinct to guarantee that all eigenspaces have dimension 1. This is clearly necessary to recover the columns of the transition matrix \(P^{ar}\). When taking into account the error introduced by sampling, we actually need more. From standard results on eigenvector sensitivity it follows that we want the eigenvalues to be well separated. A polynomially small separation will be enough for our purposes. We accomplish this by multiplying the matrix \(P^{rc}\) in \((\ast)\) by a random Gaussian vector. One can think of this as adding an extra edge \((c, d)\) and using leaves \(a, b, d\) for the reconstruction, except that we do not need the transition matrix \(P^{cd}\) to be stochastic and only one line of it suffices. More precisely, let \(U\) be a vector whose \(k\) entries are independent Gaussians with mean 0 and variance 1. We solve the eigenvalue problem \((\ast)\) with \(P^{rc}\) replaced with \(Y = P^{rc}U\). This can be done by replacing \(P^{ab,\gamma}\) on the l.h.s. of \((\ast)\) with the matrix \(P^{ab,\gamma U}\) whose \((i, j)\)th entry is

\[
\sum_{\gamma \in \mathcal{C}} U_\gamma P[s(c) = \gamma, s(b) = j \mid s(a) = i].
\]
This is justified by the Markov property. A different (independent) vector $U$ is used for every triple of leaves considered by the algorithm. One can check that w.h.p. the entries of $Y$ are $1/\text{poly}(n)$-separated. Chang [Ch96] actually uses a similar idea.

**Error on Estimated L.H.S. (ref. Lemma 6).** On the l.h.s. of $(\ast)$ (modified version), we use the following estimate $\hat{P}_{ab,U} = \sum_{\gamma \in \mathcal{C}} U_\gamma \hat{P}_{ab,\gamma}$. It follows from linear algebraic inequalities and the previous bounds that the error on the l.h.s. of $(\ast)$ is negligible w.h.p.

**Separation of Estimated Eigenvalues (ref. Lemma 7).** We need to make sure that the estimated l.h.s. of $(\ast)$ is diagonalizable. By bounding the variation of the eigenvalues and relying on the gap between the exact eigenvalues, we can ensure that the eigenvalues remain distinct and therefore $\hat{P}_{ab,U}(\hat{P}_{ab})^{-1}$ is diagonalizable.

**Error on Estimated Eigenvectors (ref. Lemma 8).** From $(\ast)$, we recover $k$ eigenvectors that are defined up to scaling. Assume that for all $i \in \mathcal{C}$, $\hat{Y}_i$ is the estimated eigenvalue corresponding to $Y_i$ (see above). Denote by $\hat{X}^i$, $X^i$ their respective eigenvectors. We denote $\hat{X}$ (resp. $X$) the matrix formed with the $\hat{X}^i$’s (resp. $X^i$’s) as columns. Say we choose the estimated eigenvectors such that $\|\hat{X}^i\|_1 = 1$. This is not exactly what we are after because we need the rows to sum to 1 (not the columns). To fix this, we then compute $\eta = \hat{X}^{-1}\mathbb{1}$. This can be done because the columns of $\hat{X}$ form a basis. Then we define $\hat{X}^i = \eta_i X^i$ for all $i$ with the corresponding matrix $\hat{X}$. Our final estimate $\hat{P}_{ar} = \hat{X}$ is a rescaled version of $X$ with row sums 1. There is still the possibility that some entries of $\hat{X}$ are negative. But this is not an issue at this point. We will make sure later that (one-step) mutation matrices are stochastic. It can be proven that $\|X - \hat{X}\|_1$ is negligible w.h.p. The proof is inspired from a standard proof on the sensitivity of eigenvectors [GV96]. There is one last issue which is that $X$ is the same as $P_{ar}$ up to permutation on the states of $r$. But since relabeling internal nodes does not affect the output distribution, we assume w.l.o.g. that $P_{ar} = X$. We make sure in the next subsection that this relabeling is performed only once for each internal node. We finally get the following.

**Proposition 1** The estimate $\hat{P}_{ar}$ recovered from $(\ast)$ using $\text{poly}(n)$ samples is such that $\|\hat{P}_{ar} - P_{ar}\|_1$ is negligible w.h.p. See Lemma 8 in the appendix for a precise statement.

### 3.2 Bounding Error Propagation

The correctness of the algorithm FULLRECON proceeds from the following remarks.

**Partition (ref. Lemma 9).** We have to check that the successive application of LEAFRECON covers the entire tree, that is that all edges are reconstructed. Figure 2 helps in understanding why this is so. When uncovering a separator edge, we associate to it a new reference leaf at distance at most $\Delta$. This can always be done by definition of $\Delta$. It also guarantees that the subtree associated to this new leaf will cover the endpoint of the separator outside the subtree from which it originated. This makes the union of all subtrees explored at any point in the execution (together with their separators) connected. It follows easily that the entire tree is eventually covered.

**Consistency (ref. Lemma 10).** It is straightforward to check that all choices of labelings are done consistently. This follows from the fact that for each node, say $w$, the arbitrary labeling is performed only once. Afterwards, all computations involving $w$ use only the matrix $\hat{P}_{aw}$ where $a$ is the reference leaf for $w$.

**Subroutines (ref. Lemmas 11 and 12).** Standard inequalities show that the (unnormalized) estimates computed in LEAFRECON, and SEPRECON have negligible error w.h.p.

**Stochasticity (ref. Lemma 13)** It only remains to make the estimates of mutation matrices into stochastic matrices. Say $\tilde{P}_{ww'}$ is the (unnormalized) estimate of $P_{ww'}$. First, some entries might be negative. Define
Let $\tilde{P}_{ww'}$ to be the positive part of $\tilde{P}_{ww'}$. Then renormalize to get our final estimate

$$\hat{P}_{ww'} = \frac{(\tilde{P}_{ww'})_i}{\| (\tilde{P}_{ww'})_i \|_1},$$

for all $i \in C$. We know from Lemmas 11 and 12 that $\tilde{P}_{ww'}$ is close to $P_{ww'}$ in $L_1$. From this, it is easy to check that $\hat{P}_{ww'}$ is also close to $P_{ww'}$ and that $\| (\hat{P}_{ww'})_i \|_1$ is close to 1. Therefore $\| \hat{P}_{ww'} - P_{ww'} \|_1$ is negligible w.h.p.

**Precision and Confidence (ref. Lemma 14).** Now that all matrices have been approximately reconstructed, it is straightforward to check that the distributions on the leaves of the estimated and real models are close. The only issue is that some of the “real” transition probabilities are smaller than the tolerance of FULLRECON and will not be well approximated (relatively) by the $1/poly(n)$ additive error (see the appendix for a more precise statement). But it is easy to show that those unlikely transitions only contribute to outputs that have a very low overall probability. We show in the appendix that $\| \hat{\pi}_L - \pi_L \|_1$ is negligible w.h.p. thereby proving Theorem 5.

In the special case of the HMM (“caterpillar” tree, see Figure 1), our algorithm FULLRECON actually gives a stronger result. Here we only need to assume that determinants of mutation matrices are at least $1/poly(n)$ (instead $1$). This is proved in Appendix D.

## 4 Concluding Remarks

Many extensions of this work deserve further study.

- There remains a gap between our positive result (for general trees), where we require determinants $\Omega(1)$, and the hardness resut of [KM+94], which uses determinants exactly 0. Is learning possible in the case where determinants are $\Omega(n^{-c})$ (as it is in the case of HMMs)?

- There is another gap arising from the upper bound on the determinants. Having mutation matrices with determinant 1 does not seem like a major issue. It does not arise in the estimation of the mutation matrices. But it is tricky to analyze rigorously how the determinant 1 edges affect the reconstruction of the topology. Again, we have shown that this is not a problem in the special case of HMMs (at least if the topology is known).

- We have emphasized the difference between $k = 2$ and $k \geq 3$. As it stands, our algorithm works only for $(\beta, n^{-\kappa_n})$-nonsingular models even when $k = 2$. It would be interesting to rederive the results of [CGG02] using our technique possibly in combination with [Mo04].

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A Detailed Description of FULLRECON

Here follows a detailed description of the algorithm FULLRECON as well as the subroutines LEAFRECON and SEPRECON. See Sections 2 and 3 for notations. We assume as before that the tree is given. We also have access to an unlimited number of independent samples at the leaves.

**Algorithm FULLRECON**

*Input:* tree topology; independent samples at leaves;  
*Output:* estimated mutation matrices \( \{ \hat{P}_e \}_{e \in \mathcal{E}} \); estimated node distributions \( \{ \hat{\pi}_u \}_{u \in V} \);

- **Phase 0: Initialization**
  - Let \( \omega \) be any leaf in \( \mathcal{L} \), and initialize the set of uncovered leaves \( \mathcal{U} := \emptyset \); Set \( t := 0 \) and \( a_0 := \omega \);  
  - Initialize the separator set \( \mathcal{S} \) to \( \emptyset \);

- **Phase 1: Subtree Reconstruction**
  - While \( \mathcal{U} \neq \mathcal{L} \),
    * Perform LEAFRECON\( (a_t) \);  
    * Set \( t := t + 1 \);

- **Phase 2: Patching Up**
  - Let \( T \) be the last value taken by \( t \) above;
  - For \( t := 1 \) to \( T \), perform SEPRECON\( (a_t, (w_t, w'_t), a'_t) \).
The subroutines LEAFRECON and SEPRECON are detailed next. In both, we use estimates of various distributions at the leaves that are described in Section 3. The number of samples needed for each of these estimate is given the corresponding lemmas.

**Algorithm LEAFRECON**

**Input:** tree topology; reference leaf \( a \); uncovered leaves \( U \); separator set \( S \); independent samples at the leaves;

**Output:** estimated mutation matrices \( \hat{P}^{re} \) and estimated node distributions \( \hat{\pi}_a \) on edges and vertices in subtree associated to \( a \);

- Set \( \mathcal{W} := \{ a \}, \mathcal{U} := \mathcal{U} \cup \{ a \} \);
- While \( \mathcal{N}(\mathcal{W}, S) \cap B^a \neq \emptyset \):
  - Pick any vertex \( r \in \mathcal{N}(\mathcal{W}, S) \cap B^a \);
  - Let \( (r_0, r) \) be the edge with endpoint \( r \) in the path from \( a \) to \( r \);
  - If \( r_0 = a \), set \( \hat{P}^{aro} \) to be the identity, otherwise \( \hat{P}^{aro} \) is known from previous computations;
  - If \( r \) is not a leaf,
    - Let \( (r, r_1) \) and \( (r, r_2) \) be the other two edges out of \( r \);
    - Find the closest leaf \( b \) (resp. \( c \)) connected to \( r_1 \) (resp. \( r_2 \)) by a path not using \( (r_0, r) \);
    - Draw Gaussian vector \( U \); Perform the eigenvalue decomposition (\( \star \)) in its modified version from Section 3;
    - Build \( \tilde{P}^{aro} \) out of an arbitrary ordering of the columns recovered above;
    - Compute \( \rho = (\hat{P}^{aro})^{-1} \), and set \( \hat{P}^{aro} := \hat{P}^{aro} \, \text{diag}(\rho) \);
  - Otherwise if \( r \) is a leaf:
    - Use estimate from leaves for \( \hat{P}^{aro} \);
    - Set \( \mathcal{U} := \mathcal{U} \cup \{ r \} \);
    - Compute \( \hat{P}^{aro} := (\hat{P}^{aro})^{-1} \hat{P}^{aro} \);
    - Compute \( \hat{\pi}_r = \hat{\pi}_a \hat{P}^{aro} \);
    - Obtain \( \hat{P}^{aro} \) from \( \hat{P}^{aro}, \hat{\pi}_r \) and \( \hat{\pi}_{r_0} \) using Bayes rule;
    - Set \( \mathcal{W} := \mathcal{W} \cup \{ r \} \);
    - If \( r \) is not a leaf and the distance between \( a \) and \( r \) is exactly \( \Delta \), for \( \ell = 1, 2 \),
      - If \( \{ r, r_1 \} \) is not in \( S \), set \( t := |S| + 1 \), \( a_{\ell'} := b \) (if \( \ell = 1 \), and \( c \) o.w.), \( a'_{\ell'} := a \), \( (w_{\ell'}, w'_{\ell'}) := (r, r) \), and add \( \{ r, r_1 \} \) (as an undirected edge) to \( S \).

**Algorithm SEPRECON**

**Input:** leaves \( a, a' \) and separator edge \( \{ w, w' \} \);

**Output:** estimated mutation matrices \( \hat{P}^{ww'}, \hat{P}^{ww'} \);

- Estimate \( \hat{P}^{aa'} \);
- Compute \( \hat{P}^{ww'} := (\hat{P}^{aw})^{-1} \hat{P}^{aa'} (\hat{P}^{w'a'})^{-1} \);
- Obtain \( \hat{P}^{ww'} \) from \( \hat{P}^{ww'}, \hat{\pi}_w \) and \( \hat{\pi}_{w'} \) using Bayes rule.
B Proofs from Analysis of Spectral Argument

Lemma 1 (Bound on Depth) The depth of any full binary tree is bounded above by $\log n$.

Proof: Because the tree is full, the inequality $\Delta \geq d$ implies that there is an edge on one side of which there is a complete binary subtree of depth $d$. Since there are only $n$ vertices in the tree, we must have $\Delta \leq \log n$. ■

Lemma 2 (Determinants on Paths) Let $a, b$ be vertices at distance at most $2\Delta + 1$ from each other. Then, under the $(\beta, n^{-\kappa_{\pi}})$-nonsingularity assumption, the transition matrix between $a$ and $b$ satisfies

$$\det[P_{ab}] \geq 1/n^{\kappa_d},$$

for some constant $\kappa_d$ depending only on $\beta$.

Proof: This follows from the observation that $P_{ab}$ is the product of the mutation matrices on the path between $a$ and $b$. Those matrices have determinants bounded away from 0 (by a constant) and there are at most $O(\log n)$ of them so the multiplicativity of determinants gives the result. ■

Lemma 3 (Sampling Error) Using $n^{\kappa_{s}} = k^3(n^{\kappa_{s}^{(1)}} + n^{\kappa_{s}^{(2)}})$ samples, the estimation errors on the matrices $P^{ab}$ and $P^{ab,\gamma}$ satisfy

$$\| P_{ab} - \hat{P}_{ab} \|_1 \leq \frac{k}{n^{\kappa_{s}^{(1)}}},$$

and

$$\| P_{ab,\gamma} - \hat{P}_{ab,\gamma} \|_1 \leq \frac{k}{n^{\kappa_{s}^{(2)}}},$$

for all $\gamma \in C$ with probability at least $1 - 1/n^{\kappa_{s}^{(2)}}$ provided that for $\iota = 1, 2,$

$$n^{\kappa_{s}^{(\iota)}} \geq \max\{8n^{2\kappa_{\pi}} \log(8k^3n^{\kappa_{s}^{(2)}}), 4n^{2\kappa_{\pi}} n^{\kappa_{\pi}} \log(8k^3n^{\kappa_{s}^{(2)}})\},$$

for all $n$ large enough.

Proof of Lemma 3: We first need to argue that the number of samples is large enough to include a sufficient number of occurrences of $s(a) = i$. This is where we need the lower bound on $\pi_a$. Let $n^{\kappa_{s}^{(1)}} = (2n^{\kappa_{s}^{(1)}} \log(8k^3n^{\kappa_{s}^{(2)}}))^{1/2}$. By Azuma’s inequality

$$\mathbb{P} \left[ N_i^{\alpha} \leq \frac{n^{\kappa_{s}^{(1)}}}{n^{\kappa_{\pi}}} - n^{\kappa_{s}^{(1)}} \right] \leq \mathbb{P} \left[ N_i^{\alpha} \leq \pi_a(i) n^{\kappa_{s}^{(1)}} - n^{\kappa_{s}^{(1)}} \right]$$

$$\leq \mathbb{P} \left[ |N_i^{\alpha} - \mathbb{E}[N_i^{\alpha}]| \geq n^{\kappa_{s}^{(1)}} \right]$$

$$\leq 2 \exp \left( - \frac{n^{\kappa_{s}^{(1)}}}{2n^{\kappa_{s}^{(1)}}} \right)$$

$$\leq \frac{1}{4k^3n^{\kappa_{s}^{(2)}}}.$$
Because \[ n^{\kappa_s(1)} \geq 8(n^{\kappa_\pi})^2 \log(8k^3 n^{\kappa_p(2)}) \]
by assumption, it follows that
\[ n^{\kappa_\pi} n^{\kappa_1} \leq \frac{1}{2} n^{\kappa_s(1)}. \]
Then by the assumption
\[ n^{\kappa_s(1)} \geq 4(n^{\kappa_1(1)})^2 n^{\kappa_\pi} \log(8k^3 n^{\kappa_p(2)}) \]
we get
\[ \frac{n^{\kappa_s(1)}}{n^{\kappa_\pi}} - n^{\kappa_1} \geq \frac{n^{\kappa_s(1)}}{2n^{\kappa_\pi}} \geq 2(n^{\kappa_1(1)})^2 \log(8k^3 n^{\kappa_p(2)}). \]
Using the above inequality and a second application of Azuma, we get
\[ \mathbb{P} \left[ N_{i,j}^{a,b} - P_{i,j}^{a,b} \geq \frac{N_{i,j}^{a}}{n^{\kappa_\pi}} \right] \leq 2 \exp \left( - \frac{N_{i,j}^{a}}{2(n^{\kappa_s(1)})^2} \right) \leq 2 \exp \left( \frac{n^{\kappa_1(1)} - n^{\kappa_1}}{2(n^{\kappa_s(1)})^2} \right) \leq \frac{1}{4k^3 n^{\kappa_p(2)}}, \]
on the event
\[ \left\{ N_{i}^{a} \leq \frac{n^{\kappa_s(1)}}{n^{\kappa_\pi}} - n^{\kappa_1} \right\}. \]
The same calculations apply to the case of \( P_{i,j}^{a,b,c} \). The union bound gives the result. \qed

**Lemma 4 (Sampling Error at Leaves)** We have
\[ \| \hat{\pi}_a - \pi_a \|_1 \leq \frac{1}{n^{\kappa_\epsilon}}, \]
with probability at least \( 1 - 1/n^{\kappa_s(4)} \), provided
\[ n^{\kappa_s} \geq 2k^2 (n^{\kappa_\epsilon})^2 \log(kn^{\kappa_p(4)}), \]
for all \( n \) large enough.

**Proof of Lemma 4:** By Azuma’s inequality,
\[ \mathbb{P} \left[ |N_{i}^{a} - \mathbb{E}[N_{i}^{a}]| \geq \frac{n^{\kappa_s}}{kn^{\kappa_\epsilon}} \right] \leq \mathbb{P} \left[ |N_{i}^{a} - \mathbb{E}[N_{i}^{a}]| \geq (2n^{\kappa_s} \log(2kn^{\kappa_p(4)}))^{1/2} \right] \leq 2 \exp \left( - \frac{2n^{\kappa_s} \log(2kn^{\kappa_p(4)})}{2n^{\kappa_s}} \right) \leq \frac{1}{kn^{\kappa_p(4)}}, \]
where we have used the assumption on \( n^{\kappa_\epsilon} \). The claim follows. \qed
Lemma 5 (Eigenvalue Separation) Let $\kappa_\lambda$ be a constant such that $\kappa_d < \kappa_\lambda$. Then, with probability at least $1 - 1/n^{\kappa_d^{(1)}}$ no two entries of $Y$ are at distance less than $1/n^{\kappa_\lambda}$ provided

$$\frac{1}{n^{\kappa_\lambda^{(1)}}} \geq \frac{2k^{5/2}n^{\kappa_d}}{\sqrt{2\pi}n^{\kappa_\lambda}},$$

for all $n$ large enough.

Proof of Lemma 5: By Lemma 2, $|\det[P^{rc}]| > 1/n^{\kappa_d}$. Take any two lines $i, j$ of $P^{rc}$. The matrix, say $A$, whose entries are the same as $P^{rc}$ except that row $i$ is replaced by $P^{rc}_i - P^{rc}_j$ has the same determinant as $P^{rc}$. Moreover,

$$|\det[A]| = \left| \sum_\sigma \text{sgn}(\sigma) \prod_{h=1}^k A_{h\sigma(h)} \right| \leq \sum_\sigma \prod_{h=1}^k |A_{h\sigma(h)}| \leq \prod_{h=1}^k \|A_{h.}\|_1 \leq \|A_{h.}\|_1,$$

where the first and second sums are over all permutations of $\{1, \ldots, k\}$. Therefore

$$\|P^{rc}_i - P^{rc}_j\|_1 \geq 1/n^{\kappa_d}.$$

By the Cauchy-Schwarz inequality,

$$\|P^{rc}_i - P^{rc}_j\|_2 \geq 1/(\sqrt{kn^{\kappa_d}}).$$

Therefore, $(P^{rc}_i - P^{rc}_j)U$ is Gaussian with mean $0$ and variance at most $1/(k^{\kappa_\lambda}n^{\kappa_d})$. A simple bound on the normal distribution gives

$$\mathbb{P}\left[|(P^{rc}_i - P^{rc}_j)U| < \frac{1}{n^{\kappa_\lambda}}\right] \leq 2 \frac{1}{n^{\kappa_\lambda}} \frac{\sqrt{kn^{\kappa_d}}}{\sqrt{2\pi}}.$$

There are $O(k^2)$ pairs of lines to which we apply the previous inequality. The union bound gives the result.

Lemma 6 (Error on L.H.S.) Assume that $n^{\kappa_\lambda^{(3)}} \geq 2k^2n^{\kappa_d}$. Then we have

$$\|\tilde{P}^{ab,U} (\tilde{P}^{ab})^{-1} - P^{ab,U} (P^{ab})^{-1}\|_1 \leq \frac{1}{n^{\kappa_\lambda^{(3)}}}$$

provided

$$\frac{1}{n^{\kappa_\lambda^{(3)}}} \geq 2k^{5/2}n^{2\kappa_d} \sqrt{2\log(n^{\kappa_s})} \left( \frac{1}{n^{\kappa_\lambda^{(1)}}} + \frac{1}{n^{\kappa_\lambda^{(2)}}} + \frac{1}{n^{\kappa_\lambda^{(1)}}} \frac{1}{n^{\kappa_\lambda^{(2)}}} \right),$$

for all $n$ large enough, with probability at least $1 - 1/n^{\kappa_\lambda^{(3)}}$, where we must have

$$\frac{1}{n^{\kappa_\lambda^{(3)}}} \geq \frac{k}{(n^{\kappa_s} \sqrt{\pi \log(n^{\kappa_s})})},$$

for all $n$ large enough.
Proof of Lemma 6: Using a standard formula for the inverse, we have

\[ |(P_{ab})^{-1}_{ij}| = \frac{1}{|\det(P_{ab})||\text{adj}(P_{ab})|_{ij}} \leq \frac{1}{n^{\kappa_d}}, \]

where we have used the nonsingularity assumption, and the fact that \(\text{adj}[A]_{ij}\) is the determinant of a substochastic matrix. Then \(\|P_{ab}^{-1}\|_1 \leq k n^{\kappa_d}\). By the assumption \(n^{\kappa_3(1)} \geq 2 k^2 n^{\kappa_d}\) and a standard linear algebra result

\[ \|\hat{P}_{ab}^{-1} - P_{ab}^{-1}\|_1 \leq \frac{\|[(P_{ab})^{-1} - (\hat{P}_{ab} - P_{ab})]\|_1}{1 - \|[(P_{ab})^{-1} - (P_{ab})]\|_1}\]

where we have used Lemma 3.

We now compute the error on \(P_{ab,U}\). We have

\[ \|P_{ab,U} - \hat{P}_{ab,U}\|_1 \leq \sum_{\gamma \in \mathcal{C}} |U_{\gamma}| \|\hat{P}_{ab,\gamma} - P_{ab,\gamma}\|_1 \]

Also,

\[ \|P_{ab,U}\|_1 \leq \sum_{\gamma \in \mathcal{C}} |U_{\gamma}| \|P_{ab,\gamma}\|_1 \]

By a simple bound (see e.g. [Du96]),

\[ \mathbb{P}\left[|U_{\gamma}| \geq \sqrt{2 \log(n^{\kappa_3})}\right] \leq 2 \frac{\exp(-2 \log(n^{\kappa_3})/2)}{\sqrt{2\pi} \sqrt{2 \log(n^{\kappa_3})}} \leq \frac{1}{n^{\kappa_3} \sqrt{\pi \log(n^{\kappa_3})}}. \]

So with probability at least \(1 - k/(n^{\kappa_3} \sqrt{\pi \log(n^{\kappa_3})})\), we get \(\|U\|_1 \leq k \sqrt{2 \log(n^{\kappa_3})}\).

Putting the above bounds together, we get with a rough bound

\[ \|\hat{P}_{ab,U} - P_{ab,U}\|_1 \leq 2 k^5 n^{2\kappa_d} \sqrt{2 \log(n^{\kappa_3})}\left(\frac{1}{n^{\kappa_3}} + \frac{1}{n^{\kappa_2}} + \frac{1}{n^{\kappa_1}} + \frac{1}{n^{\kappa_2}}\right). \]

This proves the claim. \(\blacksquare\)

Lemma 7 (Sensitivity of Eigenvalues) If \(n^{\kappa_3(3)} \geq 3k^2 n^{\kappa_d} n^{\kappa_3}\) then \(\hat{P}_{ab,U}^{-1}\) is diagonalizable and all its eigenvalues are real and distinct. In particular, all eigenspaces have dimension 1.
Proof of Lemma 7: A standard theorem on eigenvalue sensitivity [HJ85] asserts that if \( \hat{Y}_j \) is an eigenvalue of \( \hat{P}^{ab,U}(\hat{P}^{ab})^{-1} \), then there is an eigenvalue \( Y_i \) of \( P^{ab,U}(P^{ab})^{-1} \) such that

\[
|\hat{Y}_j - Y_i| \leq \|P^{ar}\|_1 \|P^{ar}\|^{-1}_1 \|\hat{P}^{ab,U}(\hat{P}^{ab})^{-1} - P^{ab,U}(P^{ab})^{-1}\|_1
\]

where as in Lemma 6, we bound \( \|P^{ar}\|^{-1}_1 \leq k_\kappa^a \), and we use \( n^{(3)}_\kappa \geq 3k^2n^{(4)}n^{(5)}_\kappa \). Given that the separation between the entries of \( \hat{Y}_j \) is at least \( 1/n_{\kappa\lambda} \) we deduce that there is a unique \( Y_i \) at distance at most \( 1/(3n^{(3)}_{\kappa\lambda}) \) from \( \hat{Y}_j \). This is true for all \( j \in C \). This implies that all \( \hat{Y}_j \)'s are distinct and therefore they are real and \( \hat{P}^{ab,U}(\hat{P}^{ab})^{-1} \) is diagonalizable as claimed.

Lemma 8 (Sensitivity of Eigenvectors) We have

\[
\|\tilde{X} - X\|_1 \leq \frac{1}{n^{(5)}_{\kappa\lambda}},
\]

if

\[
\frac{1}{n^{(5)}_{\kappa\lambda}} \geq \left[ \frac{1}{n^{(5)}_{\kappa\lambda}} + k_\kappa \right] \frac{2k^3n^{(4)}_{\kappa\lambda}}{n^{(5)}_{\kappa\lambda}} + \frac{1}{n^{(5)}_{\kappa\lambda}},
\]

with

\[
\frac{1}{n^{(5)}_{\kappa\lambda}} \geq \frac{k^2}{n^{(4)}_{\kappa\lambda} - k^2},
\]

and

\[
\frac{1}{n^{(4)}_{\kappa\lambda}} \geq \frac{2k^3n^{(4)}_{\kappa\lambda}n^{(2)}_{\kappa\lambda}}{n^{(3)}_{\kappa\lambda}},
\]

for all \( n \) large enough.

Proof of Lemma 8: We want to bound the norm of \( \tilde{X} - X^i \). We first argue about the components of \( \tilde{X} - X^i \) in the directions \( X^j, j \neq i \). We follow a standard proof that can be found for instance in [GV96]. We need a more precise result than the one stated in the previous reference, and so give the complete proof here.

Because the \( X^i \)'s form a basis, we can write

\[
\tilde{X}^i - X^i = \sum_{j \in C} \rho_j X^j,
\]

for some values of \( \rho_j \)'s. Denote \( A = P^{ab,U}(P^{ab})^{-1}, E = \hat{P}^{ab,U}(\hat{P}^{ab})^{-1} - P^{ab,U}(P^{ab})^{-1} \) and \( \Delta_i = \hat{Y}_i - Y_i \). Then

\[
(A + E)\tilde{X}^i = \hat{Y}_i\tilde{X}^i,
\]

which, using \( AX^i = Y_iX^i \), implies

\[
\sum_{j \in C} Y_j \rho_j X^j + E\tilde{X}^i = Y_i \sum_{j \in C} \rho_j X^j + \Delta_i\tilde{X}^i.
\]
For all \( j \in C \), let \( Z^j \) be the left eigenvector corresponding \( X^j \). It is proved in [GV96] that \((X^j)^T Z^{j'} = 0\) for all \( j \neq j' \). Fix \( h \in C \). Multiplying both sides of the previous display by \( Z^h \) and rearranging gives

\[
\rho_h = \frac{(Z^h)^T (E \hat{X}^i) + \Delta_i (Z^h)^T \hat{X}^i}{(Y_h - Y_i)(Z^h)^T X^h}.
\]

Eigenvectors are determined up to multiple constants. Here we make the \( X^i \) be equal to the columns of \( P_{ar} \) and the \( Z^i \) equal to the columns of \((P_{ar})^T \). Recall that the \( \hat{X}^i \)s were chosen such that \( \| \hat{X}^i \|_1 = 1 \). Then using standard matrix norm inequalities, Cauchy-Schwarz inequality, and Lemmas 5, 6, and 7, we get

\[
|\rho_h| \leq \left| \frac{(Z^h)^T (E \hat{X}^i) + \Delta_i (Z^h)^T \hat{X}^i}{(Y_h - Y_i)(Z^h)^T X^h} \right| \leq \frac{n^{\kappa_\lambda} \| Z^h \|_1 \| \hat{X}^i \|_1 [\sqrt{k} \| E \|_1 + k^2 n^{\kappa_d}/n^{\kappa_e(3)}]}{2k^3 n^{\kappa_\lambda} n^{2\kappa_d}} \leq \frac{1}{n^{\kappa_e(3)}},
\]

where we have used the bound \( \| Z^h \|_1 \leq kn^{\kappa_d} \) as in Lemma 6. The bound on \( |\Delta_i| \) comes from the proof of Lemma 7. Plugging in the expansion of \( \hat{X}^i \) gives

\[
1 = \| \hat{X}^i \|_1 \leq |1 + \rho_i| \| X^i \|_1 + \sum_{j \neq i} |\rho_j| \| X^j \|_1 \leq k|1 + \rho_i| + k^2/n^{\kappa_e(4)}.
\]

Assuming that \( n^{\kappa_e(4)} \) is large enough, we get

\[
|1 + \rho_i| \geq \frac{1 - k^2/n^{\kappa_e(4)}}{k} > 0.
\]

Defining \( \bar{X}^i = \hat{X}^i/(1 + \rho_i) \) and plugging again into the expansion of \( \hat{X}_i \), we get

\[
\| \bar{X}^i - X^i \|_1 = \left\| \sum_{j \neq i} \frac{\rho_j}{1 + \rho_i} X^j \right\|_1 \leq \frac{k^2}{n^{\kappa_e(4)} - k^2} \leq \frac{1}{n^{\kappa_e(5)}},
\]

where we have used \( \| X^j \|_1 \leq k, j \neq i \).

Denote \( \bar{e} = \bar{X} \mathbf{1} \) the row sums of \( \bar{X} \), the matrix formed with the \( \bar{X}^i \)s as columns. The scaling between \( \bar{X} \) and \( \bar{X} \) is given by \( \bar{\eta} = \bar{X}^{-1} \mathbf{1} \). Indeed, because the columns of \( \bar{X} \) and \( \bar{X} \) are the same up to scaling, there is a vector \( \bar{\eta} \) such that

\[
\bar{X} = \bar{X} \text{diag}(\bar{\eta}).
\]
By the normalization of both matrices, we get

\[ X \tilde{\eta} = X \mathbb{1} = X \text{diag}(\tilde{\eta}) \mathbb{1} = X \tilde{\eta}. \]

Because \( \tilde{X} \) is invertible, \( \tilde{\eta} = \tilde{\eta} \). We want to argue that \( \tilde{\eta} \) is close to \( \mathbb{1} \), that is that \( \tilde{X} \) and \( \tilde{X} \) are close. Note that

\[
\| \tilde{\eta} - \mathbb{1} \|_1 = \| X^{-1} (1 - \tilde{e}) \|_1 \\
\leq \| X^{-1} \|_1 \| 1 - \tilde{e} \|_1.
\]

By the condition, \( \| X^i - X^j \|_1 \leq 1/n^{\kappa_e(5)} \) for all \( i \) and the fact that the row sums of \( X \) are 1, we get

\[
\| 1 - \tilde{e} \|_1 \leq \frac{k^2}{n^{\kappa_e(5)}}.
\]

To bound \( \| \tilde{X}^{-1} \|_1 \), let \( \tilde{E} = \tilde{X} - X \) and note that, using a standard theorem on the sensitivity of the inverse \([HJ85]\),

\[
\| \tilde{X}^{-1} \|_1 \leq \| (X + \tilde{E})^{-1} - X^{-1} + X^{-1} \|_1 \\
\leq \| (X + \tilde{E})^{-1} - X^{-1} \|_1 + \| (X + \tilde{E})^{-1} - X^{-1} \|_1 \\
\leq \| X^{-1} \|_1 \left[ \frac{\| X^{-1} \|_1 \| \tilde{E} \|_1}{1 - \| X^{-1} \|_1 \| \tilde{E} \|_1} \right] + \| X^{-1} \|_1 \\
\leq \frac{\| X^{-1} \|_1}{1 - \| X^{-1} \|_1 \| \tilde{E} \|_1}.
\]

As we have seen before, \( \| X^{-1} \|_1 \leq kn^{\kappa_d} \) and by the bound above, \( \| \tilde{E} \|_1 \leq 1/n^{\kappa_e(5)} \). Assuming that \( kn^{\kappa_d}/n^{\kappa_e(5)} \leq 1/2 \), we get

\[
\| \tilde{X}^{-1} \|_1 \leq 2 \| X^{-1} \|_1 \leq 2kn^{\kappa_d}.
\]

Therefore,

\[
\| \tilde{\eta} - \mathbb{1} \|_1 \leq \frac{2k^3 n^{\kappa_d}}{n^{\kappa_e(5)}}.
\]

This finally gives the bound

\[
\| \tilde{X} - X \|_1 \leq \| \tilde{X} - X \|_1 + \| \tilde{X} - X \|_1 \\
\leq \| \tilde{X} \text{diag}(\tilde{\eta}) - \tilde{X} \|_1 + \frac{1}{n^{\kappa_e(5)}} \\
\leq \| \tilde{X} \|_1 \| \tilde{\eta} - \mathbb{1} \|_1 + \frac{1}{n^{\kappa_e(5)}} \\
\leq \left[ \frac{1}{n^{\kappa_e(5)}} + k \right] \frac{2k^3 n^{\kappa_d}}{n^{\kappa_e(5)}} + \frac{1}{n^{\kappa_e(5)}} \\
\leq \frac{1}{n^{\kappa_e}}.
\]

\[ \square \]
C Proofs from Error Propagation Analysis

Lemma 9 (Partition) The successive application of LEAFRECON covers the entire tree.

Proof of Lemma 9: We need to check that the algorithm outputs a transition matrix for each edge in \( T \). Denote \( T_{a_t} \) the subtree explored by LEAFRECON applied to \( a_t \). The key point is that for all \( t \), the tree \( T_{\leq t} \) made of all \( T_{a_t} \) for \( t' \leq t \) as well as their separators is connected. We argue by induction. This is clear for \( t = 0 \). Assume this is true for \( t \). Because \( T \) is a tree, \( T_{\leq t} \) is a (connected) subtree of \( T \) and \( (w_{t+1}, w_0) \) is an edge on the “boundary” of \( T_{\leq t} \), the leaf \( a_{t+1} \) lies outside \( T_{\leq t} \). Moreover, being chosen as the closest leaf from \( w_{t+1} \), it is at distance at most \( \Delta \). Therefore, applying LEAFRECON to \( a_{t+1} \) will cover a (connected) subtree including \( w_{t+1} \). This proves the claim.

Lemma 10 (Consistency) The internal labelings are made consistently by subroutines LEAFRECON and SEPRECON.

Proof of Lemma 10: It is straightforward to check that for each node, say \( w \), the arbitrary labeling is performed only once. Afterwards, all computations involving \( w \) use only the matrix \( \hat{P}_{aw} \) where \( a \) is the reference leaf for \( w \).

Lemma 11 (Error Analysis: LEAFRECON) Assume the bounds in the previous lemmas apply. Assume also \( n^{\kappa_e} \leq n^{\kappa_e(1)} / k \). Let \( a \) be a leaf and fix \( \Delta \) as in Section 2. Then all edges \( (w, w') \) reconstructed by LEAFRECON applied to \( a \) satisfy

\[
||\hat{P}_{ww'} - P_{ww'}||_1 \leq \frac{1}{n^{\kappa_e}},
\]

where

\[
\frac{1}{n^{\kappa_e}} \geq \frac{n^{\kappa_e} n^{\kappa_v}}{n^{\kappa_e(1)} (n^{\kappa_v} - n^{\kappa_e})} + \frac{2n^{2\kappa_e}}{n^{\kappa_v} - n^{\kappa_e}},
\]

provided

\[
\frac{1}{n^{\kappa_e(1)}} \geq \frac{2k n^{\kappa_v} (2k^2 n^{2\kappa_e} + 1)}{n^{\kappa_v}},
\]

for all \( n \) large enough, for a proper relabeling of the states at \( r_0, r \), where we require \( n^{\kappa_v} = \Omega(n^{\kappa_{v+1}}) \). As for vertex distributions, they satisfy

\[
||\hat{\pi}_r - \pi_r||_1 \leq \frac{1}{n^{\kappa_v}},
\]

if

\[
\frac{1}{n^{\kappa_v}} \geq \frac{1}{n^{\kappa_e}} + \frac{k}{n^{\kappa_e}},
\]

for all \( n \) large enough.
Proof of Lemma 11: Suppose first that \((w, w') = (r_0, r)\) as in the description of the algorithm and assume that \(r\) is not a leaf. By an argument similar to that in Lemma 6, provided \(kn_e^d/n_e^c \leq 1/2\), we get

\[
\|\tilde{P}^{r_0} - P^{r_0}\|_1 = \|(\tilde{P}^{r_0})^{-1} \tilde{P}^{ar} - (P^{r_0})^{-1} P^{ar}\|_1
\]

\[
\leq 2kn_e^d \|\|\tilde{P}^{r_0})^{-1} - (P^{r_0})^{-1}\|_1 + \|\tilde{P}^{ar} - P^{ar}\|_1
\]

\[
\leq 2kn_e^d \frac{2k^2 n_e^{2d} \frac{1}{n_e^c} + \frac{1}{n_e^c}}{n_e^c}
\]

\[
\leq \frac{1}{n_e^{c(0)}},
\]

Next the algorithm computes \(\hat{\pi}_r\). In that case, we get

\[
\|\hat{\pi}_r - \pi_r\|_1 = \|\hat{\pi}_a \tilde{P}^{ar} - \pi_a P^{ar}\|_1
\]

\[
= \|\hat{\pi}_a (\tilde{P}^{ar} - P^{ar}) - (\pi_a - \hat{\pi}_a) P^{ar}\|_1
\]

\[
\leq \|\hat{\pi}_a\|_1 \|\tilde{P}^{ar} - P^{ar}\|_1 + \|\pi_a - \hat{\pi}_a\|_1 \|P^{ar}\|_1
\]

\[
\leq \frac{1}{n_e^c} + \frac{k}{n_e^c}
\]

\[
\leq \frac{1}{n_e^c},
\]

by Lemmas 3 and 4. Then applying Bayes rule, \(\tilde{P}^{r_0}\) is computed. Because the previous bound applies also to \(\hat{\pi}_r\), we get

\[
|\tilde{P}^{r_0} - P^{r_0}| = \left| \frac{\hat{\pi}_r(j) \tilde{P}^{r_0 j}}{\hat{\pi}_r(i)} - \frac{\pi_r(j) P^{r_0 j}}{\pi_r(i)} \right|
\]

\[
\leq \frac{\hat{\pi}_r(j) \tilde{P}^{r_0 j}}{\hat{\pi}_r(i)} \left( \frac{\hat{\pi}_r(j) P^{r_0 j}}{\pi_r(i)} \right) + \left| \frac{\hat{\pi}_r(j) - \pi_r(j)}{\pi_r(i)} \right|
\]

\[
\leq \frac{1}{n_e^{c(0)} \left( 1/n_e^c - 1/n_e^c \right)} + \frac{\pi_r(j) + 1/n_e^c - \pi_r(j)}{\pi_r(i) - 1/n_e^c - \pi_r(i)}
\]

\[
\leq \frac{n_e^c n_e^c}{n_e^{c(0)} - n_e^c} + \frac{2n^2 e^c}{n_e^c - n_e^c}
\]

\[
\leq \frac{1}{n_e^{c(0)}},
\]

where we have assumed \(n_e^c > n_e^c\).

The case where \(r\) is a leaf gives the bound

\[
\|\tilde{P}^{r_0} - P^{r_0}\|_1 \leq \frac{1}{n_e^{c(0)}},
\]

if \(n_e^c \leq n_e^{c(1)}/k\). Then the above bound on \(\hat{M}^{r_0}\) also follows by \(n_e^c > n_e^c\). This proves the claim. ■
Lemma 12 (Error Analysis: SEPReCON) All edges \((w, w')\) reconstructed by SEPReCON satisfy
\[
\| \tilde{P}^{ww'} - P^{ww'} \|_1 \leq \frac{1}{n^{e_M}},
\]
provided
\[
\frac{1}{n^{e_M}} \geq \frac{12k^4 n^{3e_d}}{n^{e_c}},
\]
for all \(n\) large enough.

Proof of Lemma 12: Let \(a, a'\) be the leaves used by SEPReCON to estimate \(P^{ww'}\) by
\[
\tilde{P}^{ww'} = (\hat{P}^{aw})^{-1} \hat{P}^{aa'} (\hat{P}^{w'a'}^{-1}).
\]
By a calculation we have performed many times (here applied twice), we get for \(n\) large enough
\[
\| \tilde{P}^{ww'} - P^{ww'} \|_1 \leq 2 \left\{ 2 \left( \frac{2k^3 n^{2e_d}}{n^{e_c}} + \frac{k^2 n^{e_d}}{n^{e_c}} \right) k n^{e_d} + \frac{2k^3 n^{3e_d}}{n^{e_c}} \right\}
\leq \frac{12k^4 n^{3e_d}}{n^{e_c}},
\]
assuming as before that \(n^{e_c} \leq \frac{n^{e_c(1)}}{k} \). ■

Lemma 13 (Stochasticity) Note \(n^{e_M''} = \min\{n^{e'_M}, n^{e_M''}\} \). Then the estimate \(\tilde{P}^{ww'}\) is well-defined and satisfies
\[
\| \tilde{P}^{ww'} - P^{ww'} \|_1 \leq \frac{1}{n^{e_M}},
\]
if
\[
\frac{1}{n^{e_M}} \geq \frac{1 + 2k^2}{n^{e_M'']],
\]
for all \(n\) large enough.

Proof of Lemma 13: Because \(P^{ww'}\) is nonnegative, taking the positive part of \(\tilde{P}^{ww'}\) can only make it closer to \(P^{ww'}\), that is
\[
\| \tilde{P}^{ww'} - P^{ww'} \|_1 \leq \| \tilde{P}^{ww'} - P^{ww'} \|_1.
\]
Note also that by the bound on \(\| \tilde{P}^{ww'} - P^{ww'} \|_1\), the row sums of \(\tilde{P}^{ww'}\) are at least \(1 - k/n^{e_M''}\) and taking the positive part of \(\tilde{P}^{ww'}\) can only decrease its row sums by \(k/n^{e_M'}\). Therefore, we get
\[
\| (\tilde{P}^{ww'})_i \|_1 \geq 1 - \frac{2k}{n^{e_M'}}.
\]
Thus
\[ \| \hat{P}^{ww'} - P^{ww'} \|_1 \leq \| \hat{P}^{ww'} - P^{ww'} \|_1 + \| \hat{P}^{ww'} - \hat{P}^{ww'} \|_1 \]
\[ \leq \| \hat{P}^{ww'} - P^{ww'} \|_1 + \frac{2k}{n^{\kappa_p M}} \| \hat{P}^{ww'} \|_1 \]
\[ \leq \frac{1 + 2k^2}{n^{\kappa_p M}} \leq \frac{1}{n^{\kappa_p M}}. \]

\[ \] 

**Lemma 14 (Precision and Confidence)** Denote by $1 - 1/n^{\kappa_p (T)}$ the probability that the topology of the tree is reconstructed properly. With probability at least $1 - 1/n^{\kappa_p}$ where
\[ \frac{1}{n^{\kappa_p}} \geq n^3 \left( \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} \right), \]
all previous lemmas hold for all edges and all vertices, and the reconstructed model satisfies
\[ \| \hat{\pi}_L - \pi_L \|_1 \leq \varepsilon, \]
for all $n$ large enough. This can be done, using a sample of total size at most
\[ m = n^3(n^{\kappa_L} + n^{\kappa'_L}). \]

**Proof of Lemma 14:** The bounds in Section 3.1 and 3.2 must hold for all triples of leaves used by subroutine LEAFRECON, of which there are at most $n^3$ (recall that $n$ is the number of leaves). Therefore, we need a total of $m = n^3(n^{\kappa_L} + n^{\kappa'_L})$ samples. The probability that one of the inequalities fails or that the topology is wrong is
\[ n^3 \left( \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} + \frac{1}{n^{\kappa_p}} \right) \leq \frac{1}{n^{\kappa_p}}, \]
by the union bound.

The $L_1$ distance on the leaves is bounded above by the $L_1$ distance on the entire tree. Indeed, let $r$ be the root of $T$. Denote $\hat{L} = \mathcal{V} \setminus \mathcal{L}$. For partial assignments of the states $s_L \in C^L$ and $s_\hat{L} \in C^{\hat{L}}$ on leaf and non-leaf vertices resp. we denote $s = (s_L, s_\hat{L})$ the state on the entire tree. Then
\[ \| \hat{\pi}_L - \pi_L \|_1 = \sum_{s_L \in C^L} |\hat{\pi}_L(s) - \pi_L(s)| \]
\[ = \sum_{s_L \in C^L} \left| \sum_{s_\hat{L} \in C^{\hat{L}}} \left( \hat{\pi}_{\hat{L}}(s) - \pi_{\hat{L}}(s) \right) \right| \]
\[ \leq \sum_{s_L \in C^L} \sum_{s_\hat{L} \in C^{\hat{L}}} |\hat{\pi}_\hat{L}(s) - \pi_\hat{L}(s)| \]
\[ = \| \hat{\pi}_\hat{L} - \pi_\hat{L} \|_1. \]

One issue we have to deal with before bounding $\| \hat{\pi}_\hat{L} - \pi_\hat{L} \|_1$ is that the transition probabilities smaller than $1/n^{\kappa_M}$ are not well approximated (relatively). We bound the probability associated to states arising from
such small transition probabilities. Denote by \( n_V \) the number of vertices. Fix a threshold, say \( 1/n^{\kappa_{th}} \geq 2/n^{\kappa_M} \). In a realization of the Markov model, the probability that at least one the transitions with probability at most \( 1/n^{\kappa_{th}} \) occurs is bounded above by \((n_V - 1)(k - 1)/n^{\kappa_{th}}\) by the union bound, because at each transition there is at most \( k - 1 \) possible transitions with probability less than \( 1/n^{\kappa_{th}} \). By the same argument, under \( \hat{\pi}_V \) this probability is less than \((n_V - 1)(k - 1)/n^{\kappa_{th}} + 1/n^{\kappa_M}\). So as long as \( n^{\kappa_{th}} = \omega(n) \), these probabilities are small. Denote by \( S_{th} \) the states on \( V \) that use no transition with probability at most \( 1/n^{\kappa_{th}} \).

Then, for any \( s \in S_{th} \) such that \( \hat{\pi}_V(s) > \pi_V(s) \),

\[
|\hat{\pi}_V(s) - \pi_V(s)| = \hat{\pi}_V(s(r)) \prod_{(u,v) \in E_r} \hat{P}_{uv}^{s(r)s(u)} - \pi_V(s(r)) \prod_{(u,v) \in E_r} P_{uv}^{s(u)s(v)}
\]

\[
\leq \pi_V(s(r)) \prod_{(u,v) \in E_r} P_{uv}^{s(u)s(v)} \left[ \left(1 + \frac{n^{\kappa_{th}}}{n^{\kappa_M}} \right) \left(1 + \frac{n^{\kappa_{th}}}{n^{\kappa_M}} \right)^{n_V - 1} - 1 \right],
\]

and likewise for \( \hat{\pi}_V(s) < \pi_V(s) \),

\[
|\hat{\pi}_V(s) - \pi_V(s)| \leq \pi_V(s(r)) \prod_{(u,v) \in E_r} P_{uv}^{s(u)s(v)} \left[ 1 - \left(1 - \frac{n^{\kappa_{th}}}{n^{\kappa_M}} \right) \left(1 - \frac{n^{\kappa_{th}}}{n^{\kappa_M}} \right)^{n_V - 1} \right].
\]

Provided \( \kappa_M = \Omega(n^2/n^{\kappa_{th}}) \) and \( n^{\kappa_{th}} = \Omega(n^2/n^{\kappa_{th}}) \), the two expressions in square brackets are less than \( \varepsilon/4 \) for \( n \) large enough. Then

\[
\|\hat{\pi}_V - \pi_V\|_1 \leq \sum_{s \in S_{th}} |\hat{\pi}_V(s) - \pi_V(s)| + \sum_{s \notin S_{th}} |\hat{\pi}_V(s) - \pi_V(s)|
\]

\[
\leq \frac{\varepsilon}{4} + \frac{\varepsilon}{4} + (n_V - 1)(k - 1) \left[ \frac{2}{n^{\kappa_{th}}} + \frac{1}{n^{\kappa_M}} \right]
\]

\[
\leq \varepsilon,
\]

for \( n \) large enough.

All matrices involved have constant size so the only contributions to the running time are the \( O(n) \) reconstructions and \( O(n) \) breath-first searches. The overall running time is \( O(n^2) \). ■
D  Special Case: HMM

Proof Sketch for Theorem 2: Here we only need to assume that determinants of mutation matrices are at least $1/poly(n)$ (instead $\Omega(1)$). The reason is that the HMM tree can be partitioned into subtrees of constant size in which all nodes are at distance at most a constant from a reference leaf. Take all edges leading to observed nodes as subtrees, with two extra 1-node subtrees for the extremal vertices. Proofs from the previous section apply without change. The only difference is that now all paths are of constant length and therefore we can allow mutation matrices to have $1/poly(n)$ determinants. Therefore, if the topology is known, which is often naturally true in applications of HMMs, we can reconstruct the (approximate) mutation matrices in polynomial time. If the topology is not known, then Theorem 4 allows to build $T$ provided determinants are at most $1 - 1/poly(n)$. ■
E Proof of Theorem 4

For some background on this proof, see [ES+97, ES+98].

**Proof Sketch for Theorem 4** Recall the definition of a caterpillar from Section 1.1. It is easy to see that the caterpillar topology is determined by the topologies on all quartets of leaves \( y_1, \ldots, y_4 \) all of which are among the descendants of \( v_i, v_{i+1}, v_{i+2}, v_{i+3} \) for some \( i \). Thus the main task is to find the topologies on such subtrees. Note that for all such \( y_1, \ldots, y_4 \) it holds that \( |\det P_{y_i y_j}| > n^{-5C} \). Thus we are interested in reconstructing the quartet trees corresponding to quartets \( y_1, \ldots, y_4 \) such that the empirical determinant between \( y_i \) and \( y_j \) is at least \( n^{-6C} \) (say). If we are given that all determinants satisfy \( |\det P| < 1 - n^{-4\tau} \), say, then it is easy to do so using the standard 4-point method. ■