Species tree estimation under joint modeling of coalescence and duplication: sample complexity of quartet methods

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Abstract

We consider species tree estimation under a standard stochastic model of gene tree evolution that incorporates incomplete lineage sorting (as modeled by a coalescent process) and gene duplication and loss (as modeled by a branching process). Through a probabilistic analysis of the model, we derive sample complexity bounds for widely used quartet-based inference methods that highlight the effect of the duplication and loss rates in both subcritical and supercritical regimes.

1 Introduction

Estimating phylogenies from the molecular sequences of existing species is a fundamental problem in computational biology that has been the subject of significant practical and theoretical work [SS03, Fel03, Gas05, Yan14, Ste16, War17, Roc19]. Rigorous statistical guarantees for inference methodologies often involve the probabilistic analysis of Markov models on trees. In particular, these analyses have uncovered deep connections with phase transitions in related statistical physics models [Mos03, MP03, Mos04b, Mos04a, MS04, BCMR06, Roc10, DMR11, MRS11, MHR13, AHR17, RS17, FR18, GZ19].

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In modern datasets, however, phylogeny estimation is confounded by heterogeneity across the genome from processes such as incomplete lineage sorting (ILS), gene duplication and loss (GDL), lateral gene transfer (LGT), and others [Mad97]. Inferred trees depicting the evolution of individual loci in the genome are referred to as gene trees, while the tree representing the speciation history is called the species tree. Current sequencing technology allows phylogenetic estimates of species relationships for many genes, and a major challenge in reconstructing species trees is that gene trees often disagree for the reasons mentioned above. There is a burgeoning literature on the many ways of extracting speciation histories from collections of gene trees [DR09, Nak13, Ste16, War17, SDG20].

In this phylogenomic context, the design and analysis of species tree estimation methods require the use of a variety of stochastic processes beyond Markov models on trees, including coalescent processes [RY03], branching processes [ALS09], random subtree prune-and-regraft operations [Gal07, LRvH07, RS13], and tree mixtures [MS07, MR12]. In fact, there is increasing realization in the phylogenetics community that ILS, GDL, LGT, etc. should not be studied in isolation [RW15, Deg18, SDP20, SS20] and, as a result, there has been a push to consider more complex models that combine many sources of uncertainty and discordance [MK09, SLA16, RK12, DNR15, RS15, MR17, RNW18, RNW19, ALR19, ABR19, LGSC20, ME20]. We study here a joint coalescent and branching process unifying ILS and GDL, as introduced in [RK12].

Much is known about estimating species trees in the presence of ILS alone [RELY20], as modeled by the multispecies coalescent (MSC) [RY03]. The latter model posits that, on a fixed species tree, gene trees evolve backwards in time on each branch according to the Kingman coalescent [Kin82] (see Section 2 for more details). Bayesian approaches are a natural choice under such complex models of evolution [DR07]. However they do not scale well to large datasets and more computationally efficient procedures have been developed that combine inferred gene trees, sometimes referred to as summary methods [War17].

One such method is to deduce the species tree by a plurality vote across gene trees. Unfortunately, that approach is not statistically consistent, that is, it may not converge to the true species tree as the number of gene trees grows to infinity. Indeed, for unrooted species trees with more than four species, the most frequently occurring gene tree topology need not coincide with that of the species tree [DR06]. However, for every 4-tuple of species—
also referred to as quartet—and every locus, the most probable unrooted
gene tree topology matches the species tree topology [ADR11]. This implies
in particular that the species tree topology is identifiable from the distribu-
tion of gene trees. That is, different species tree topologies necessarily
produce different gene tree distributions. Further, quartet-based algorithms
for combining gene trees [LKDA10, MRB+14] are known to be statistically
consistent. Tight bounds on their sample complexity, that is, how many gene
trees are needed to recover the true species tree with high probability, have
also been established [SRM18].

Less is known about estimating species trees in the presence of GDL
alone. The model in [ALS09] posits that, on a fixed species tree, the number
of copies in a gene family evolves forward in time on each branch accord-
ing to continuous-time branching process [AN72] (see Section 2 for more
details). Recently, the identifiability of the species tree in the presence of
GDL alone was established in [LMWR19] by showing that, similarly to the
ILS alone case, for every quartet the most frequent unrooted gene tree topol-
ogy matches that of the species tree. As a result, quartet-based inference
methods [RSM19] were also shown to be statistically consistent. To date, no
sample complexity results have been derived under this GDL model however.

In this paper, we investigate the gene tree evolution model of [RK12],
which unifies the multispecies coalescent and the branching process model of
gene duplication and loss discussed above. Given that quartet-based methods
have strong guarantees under these models separately, it is natural to consider
their performance under the joint model as well (see Section 2 for more
details on the methods). Numerical experiments in [DHN19, LMWR19] provide
some evidence for the accuracy of certain quartet-based methods. In [ME20],
the authors give a proof of statistical consistency for one such method. In our
main result, we give the first known upper bounds on the sample complexity
of species tree estimation methods under the joint effect of ILS and GDL.
Our proof, which highlights the somewhat counter-intuitive role played by
the duplication and loss rates in the supercritical regime (see Section 2), is
complicated by the simultaneous forward-in-time/backward-in-time nature
of the process.

The rest of the paper is organized as follows. In Section 2, after defining
the model and inference methods, we state and discuss our results. In Sec-
tion 3, we give a proof of identifiability including new quantitative estimates
that play a role in our proof of sample complexity. The rest of the proof can
be found in Section 4.
2 Background and main results

We first describe the model and then state our results formally.

2.1 Problem and model

Our input is a collection $\mathcal{T} = \{t_i\}_{i=1}^k$ of $k$ multi-labeled gene trees given without estimation error. We explain the terminology. The process of gene duplication creates multiple copies of a gene within the same individual in a species. We refer to these duplicated genomic segments as gene copies and we refer to collections of gene copies from different unrelated genes as gene families. A gene tree is a depiction of the parental lineages of a gene or multiple gene copies from individuals across several species. Roughly speaking, it shows the joint ancestral history of these related gene copies. In contrast, a species tree is a depiction of the evolutionary relationships of a group of species. Roughly speaking, it shows the sequence of speciation events that have produced the current species. By multi-labeled, we mean that each leaf of a gene tree is associated with a label from the set $S$ of $n$ species of interest and that the leaf labels need not be unique. That is, multiple leaves of a gene tree may be associated with the same species; this corresponds to observing multiple paralogous copies (i.e., which have arisen from gene duplication) of a gene within the genome. In practice, gene trees are estimated from the molecular sequences of the corresponding genomic segments using a variety of phylogenetic reconstruction methods [SS03, Fel03, Gas05, Yan14, Ste16, War17]. For the sake of our results, we assume that gene trees are provided without estimation error for a large number of gene families. Our main modeling assumption is that these gene trees have been drawn independently from a distribution for which some $n$-species tree $T$ is taken as a fixed (i.e., non-random)—but unknown—parameter. Our goal is to output this unknown $n$-species tree $T$. We define the model more precisely next.

Model We assume that the gene trees in $\mathcal{T}$ are independent and identically distributed, with each gene tree generated under the DLCoal model [RK12], a unified model of gene duplication, loss, and coalescence. The process for generating a gene tree under DLCoal involves two steps which are described below; an example realization of these steps is provided in Figure 1. In particular, we introduce yet another tree, a locus tree, which is an unobserved
intermediate step in the model generating each gene tree. The process below is repeated independently for each $i = 1, \ldots, k$, thereby producing an independent gene tree $t_i$.

1. **Locus tree: Birth-death process of gene duplication and loss with daughter edges.**

   We fix a rooted $n$-species tree $T$ with edges $E$ directed from root to leaves (i.e. from top to bottom) and edge lengths $\{\eta_e\}_{e \in E}$. In Figure 1, this is the four-species tree on the top left. Note that the species tree is unknown to us.

   Starting with a single ancestral copy of a gene at the root of $T$, a tree is generated by a top-down birth-death process [ALS09] within the species tree. That is, on every edge in $T$, each gene copy independently duplicates at exponential rate $\lambda \geq 0$ and is lost at exponential rate $\mu \geq 0$. In addition, whenever a gene copy reaches a speciation event $T$, it bifurcates into two child copies, one for each of the descendant edges on $T$. Both duplications and speciations are indicated in the resulting locus tree by a bifurcation. The locus tree is then pruned of lost copies to give an (unobserved) rooted tree, which we refer to as locus tree. In this manner, we obtain a rooted $n'$-individual locus tree $L$ with edge lengths. Species labels are associated to each leaf of $L$ from the species set $S$. These steps for generating $L$ are illustrated in the top-middle and top-right trees of Figure 1.

   Furthermore, for each vertex in $L$ which corresponds to a gene duplication event—but not vertices corresponding to speciation bifurcations—we distinguish between the two child edges by choosing one of them uniformly at random to be the daughter edge and the other to be the mother edge. This distinction plays an important role in the next step.

2. **Gene tree: Coalescent process on a locus tree.**

   Gene trees are generated by a backward-in-time (i.e., bottom-up) coalescent process [Kin82, RY03] within the locus tree $L$. The coalescent process begins with exactly one gene copy in each leaf of $L$. Copies at the bottom of a directed edge in the locus tree undergo the Kingman coalescent process for a time equal to the length of the directed edge. Edge lengths here are assumed to be in so-called coalescent time units, which means that each pair of lineages independently coalesces after
an exponential time with mean 1, unless the end of the edge is reached first. Further, we condition on the event that all gene copies at the bottom of any daughter edge of the locus tree necessarily coalesces underneath the top of the edge. Continuing upward along ancestral edges of the locus tree, this process eventually yields a gene tree.

This process, which generates a gene tree from a locus tree, is termed the multilocus coalescent (MLC) in [RK12]. A realization of this coalescent process within a locus tree is illustrated in the bottom left of Figure 1, and the tree at the bottom right depicts the resulting gene tree without the overlying locus tree.

Intuitively, in Step 1 above the species tree $T$ is a fat tree that ‘contains’ the birth-death process which generates the skinny tree $L$. In Step 2, we forget about the species tree and ‘zoom in’ on $L$ so that $L$ becomes the fat tree, with each edge of $L$ containing one or more edges (or parts of edges) of the gene tree.

Species tree estimation methods Next we describe two quartet-based species tree methods: ASTRAL-one and ASTRAL-multi [MRB+14, DHN19, RSM19]. Recall that a quartet refers to a 4-tuple of species. It is known that the unrooted topology of a species tree is entirely characterized by the collection of its quartet topologies (see, e.g., [SS03]), and hence a large number of quartet-based approaches to species tree estimation have been developed. Both ASTRAL-one and ASTRAL-multi are practical variants of an intuitive idea (which in the ILS/GDL context is motivated by the results of [ADR11, LMWR19]; see also Propositions 1 and 2 below): (1) for each quartet of species, find the most common topology across gene trees and (2) reconstruct an $n$-species tree that coincides with as many resulting quartet topologies as possible. The input is a collection of $k$ multi-labeled gene trees $T = \{t_i\}_{i=1}^k$. Let $S$ be the set of $n$ species and $\Sigma$ be the set of $m$ labels (or gene copies). The tree $t_i$ is labeled by the set $\Sigma_i \subset \Sigma$. For any species tree $T$ labeled by $S$, the extended tree $T_{\text{ext}}$ labeled by $\Sigma$ is built by adding to each leaf of $T$ all gene copies corresponding to that species as a polytomy (i.e., as a vertex with degree possibly higher than 3).

Under ASTRAL-one, we pick one gene copy of each species uniformly at random and restrict the gene tree to these copies, producing a new gene tree $\tilde{t}_i$. For any collection of gene copies $J = \{a, b, c, d\}$, let $T^J$ be the restriction
Figure 1: A species tree with balanced topology and leaf species $A, B, C, D$ is given in (a). From (a), we obtain a realization of the gene duplication and loss (GDL) process with deaths not yet pruned. The pre-pruned locus tree and its generating species tree are shown in (b). Non-extant lineages in the tree are then pruned to obtain the locus tree, shown in (c). The gene tree obtained from the multi-species coalescent (MSC) process and its generating locus tree are shown in (d). The gene tree we actually observe is shown in (e).
of any tree $T$ to those copies. Then the quartet score of every candidate species tree $\tilde{T}$ is

$$Q_k(\tilde{T}) = \sum_{i=1}^{k} \sum_{J=\{a,b,c,d\} \subset \Sigma_i} 1(\tilde{T}_{ext}^J, t_i^J),$$

where $1(T_1, T_2)$ is the indicator for the event that $T_1$ and $T_2$ have the same topology. ASTRAL-one selects the candidate tree $\tilde{T}$ that maximizes that score.

ASTRAL-multi treats copies of a gene in a species as multiple alleles within the species. So, we do not replace $t_i$ with any restricted gene tree $\tilde{t}_i$. The quartet score of $T$ with respect to $\mathcal{T}$ is

$$Q_k(T) = \sum_{i=1}^{k} \sum_{J=\{a,b,c,d\} \subset \Sigma_i} 1(\tilde{T}_{ext}^J, t_i^J).$$

The candidate tree $\tilde{T}$ that maximizes this score is chosen by ASTRAL-multi.

Both procedures can be performed in exact mode or in a default constrained mode, which restricts the number of candidate species trees to those displaying the bipartitions in the given gene trees.

### 2.2 Statement of main results

We present a theoretical bound on the number of gene trees needed for ASTRAL-one to reconstruct the model species tree with high probability under the DLCoal model. Similarly to the case of the MSC model [SRM18], the sample complexity depends on the length of the shortest species tree branch in coalescent time units. We denote this length by $f$. However we also highlight the influence of other relevant parameters in the sample complexity: the depth of the species tree, $\Delta$; the duplication and loss rates, $\lambda$ and $\mu$. For simplicity, we assume throughout that $\mu \neq \lambda$.

**Theorem 1** (Main result: Sample complexity of ASTRAL-one). Consider a model species tree whose minimum branch length $f$ is finite and assume gene trees are generated under the DLCoal model. Then, for any $\epsilon > 0$, there are universal positive constants $C, C'$ such that the exact version of ASTRAL-one returns the true species tree with probability at least $1 - \epsilon$ if the number
of input error-free gene trees satisfies:

\[ k \geq C' \frac{1}{f^2} \frac{e^{C|\mu - \lambda| \Delta}}{(1 - \frac{\lambda}{\mu} \land \frac{\mu}{\lambda})^c} \log \frac{n}{\epsilon}. \]  

(1)

Somewhat surprisingly the subcritical (\( \mu > \lambda \)) and supercritical (\( \mu < \lambda \)) regimes exhibit a similar behavior. Indeed one naturally expects a higher sample complexity in the subcritical case as \( \mu/\lambda \) becomes large because the absence of any gene copy in a species becomes more likely and leads to the need for more gene trees in order to extract signal. That prediction is borne out in (1). However the sample complexity similarly increases in the supercritical regime as \( \lambda/\mu \) becomes large. As the proof shows, the reasons for this behavior are different in that regime. They have to do with the fact that a large number of copies at the most recent common ancestor of a species quartet tends to produce large numbers of conflicting gene tree quartet topologies, thereby obscuring the signal. It is an open problem whether other inference methods (perhaps not based on quartets) are less sensitive to this last phenomenon.

Our proof of Theorem 1 involves a delicate probabilistic analysis of the DLCoal model. Along the way, we prove other results of interest. First, we show that the unrooted species tree is identifiable from the distribution of multi-labeled gene trees \( T \) under the DLCoal model over \( T \). Formally, we show that two distinct unrooted species trees produce different gene tree distributions. The result is a generalization of [LMWR19, Theorem 1], where only GDL is considered. Theorem 2 was first claimed in [ME20]. Our novel contribution here lies in the proofs of Propositions 1 and 2 below, which give a quantitative version of the identifiability result and play a role in deriving the sample complexity of ASTRAL-one.

In fact, even in the absence of ILS (i.e., in the presence of GDL alone), no sample complexity results were previously available. While identifiability is established through symmetry arguments (see, e.g., Lemma 1 below, whose simple proof closely mirrors the core argument in [LMWR19]), sample complexity bounds require a quantitative analysis that is significantly more involved. Our arguments here (see Sections 3 and 4) combine symmetry arguments, explicit computations and a detailed case analysis of well-chosen events. In addition we incorporate ILS, which further complicates the analysis, in part because of the forward-in-time/backward-in-time nature of the combined process. We expect that the types of arguments developed here
will prove useful in analyzing other reconstruction methods under related complex models of genome evolution.

Our main identifiability result is stated next.

**Theorem 2** (Identifiability of species tree). *Let T be a model species tree with at least \( n \geq 4 \) leaves. Then T, without its root, is identifiable from the distribution of gene trees \( T \) under the DLCoal model over \( T \).*

This identifiability result is established by showing that, for each quartet in the species tree, the most likely gene tree matches the species tree. As in [LMWR19, ME20], a direct consequence of this proof is the statistical consistency of the ASTRAL-one.

**Theorem 3** (Consistency of ASTRAL-one). *As the number of input gene trees tends toward infinity, the output of ASTRAL-one converges to \( T \) almost surely, when run in exact mode or in its default constrained version.*

We use a similar reasoning to prove the consistency of ASTRAL-multi. This result is new.

**Theorem 4** (Consistency of ASTRAL-multi). *As the number of input gene trees tends toward infinity, the output of ASTRAL-multi converges to \( T \) almost surely, when run in exact mode or in its default constrained version.*

## 3 First step: a proof of identifiability of the species tree under the DLCoal model

Let \( Q = \{A, B, C, D\} \) and assume, without loss of generality, that \( T^Q \) has unrooted quartet topology \( AB|CD \), that is, it has the topology depicted in the top left of Figure 2 (ignoring the root). Let \( t \) be a gene tree generated under the DLCoal model on \( T \) and let \( t^Q \) be its restriction to the gene copies from the species in \( Q \). The high-level idea behind our proof of Theorem 2 is the following:

*Conditioning on the number of copies in species \( A, B, C, D \) in the species in \( Q \), independently pick a uniformly random gene copy \( a, b, c, d \) in species \( A, B, C, D \) and let \( q \) be the corresponding quartet topology under \( t^Q \). We show that the most likely outcome is \( q = ab|cd \).*
Figure 2: The top row shows the two cases of root location on $T^Q$: the root of $T^Q$ may occur either on the internal edge (left) or the pendant edge adjacent to $D$ (right). In both cases the unrooted quartet topology of $T^Q$ is $AB|CD$. However, as shown in the bottom row, the two choices of root location leads to two different rooted topologies: the balanced case (left) and the caterpillar case (right), which we consider separately.

This is the same approach as that used in [LMWR19], but the analysis of the model is more involved.

Define $X = (A, B, C, D)$ to be the number of copies in species $A, B, C, D$, respectively. For example, Figure 1 depicts a realization in which $X = (2, 2, 2, 2)$ since the locus tree (and hence also the gene tree) has exactly two leaves in each of the species $A, B, C$ and $D$. We will let $\mathbb{P}'$ be the probability measure subject to conditioning on the random vector $X$.

Although we seek to reconstruct an unrooted species tree, under the DL-Coal model, the locus trees and gene trees are in fact generated from a rooted species tree. Therefore when restricting the species tree to four species, there are two cases of root location to consider: when the root of the species quartet $T^Q$ is located on the internal quartet edge of $T^Q$ (the “balanced case”) or on a pendant edge of $T^Q$ (the “caterpillar case”). In the caterpillar case we may take $D$ to be the pendant edge incident with the root. See Figure 2.

For gene copies $a, b, c, d$ from $A, B, C, D$, define the following events:

$$Q_1 = \{q = ab|cd\}, \quad Q_2 = \{q = ac|bd\}, \quad Q_3 = \{q = ad|bc\}.$$
3.1 Balanced case

We first consider the balanced case. Without loss of generality, assume the species tree restricted to $Q$ has rooted topology as in the bottom left of Figure 2. Let $R$ be the most recent common ancestor of $Q$ in the species quartet $T^Q$ and $I$ be the number of locus copies exiting $R$ forward in time. Let $P''$ be the probability measure indicating conditioning on $I$ as well as on $A,B,C,D$. For any selection of copies $(a,b,c,d)$ from each species in the quartet, let $i_x \in \{1,\ldots, I\}$ be the ancestral lineage on the locus tree of $x \in \{a,b,c,d\}$ in $R$. By the law of total probability, we have

$$P[Q_i] = \mathbb{E}[P''[Q_i]], \quad i = 1, 2, 3.$$  \hspace{1cm} (2)

Hence, in order to show identifiability of the species quartet, it is sufficient to show that

$$P''[Q_1] > \max\{P''[Q_2], P''[Q_3]\}$$

when the copies of $(A,B,C,D)$ are chosen uniformly at random.

We let $\mathcal{X} \geq \bar{I}$ be the event that each of $(A,B,C,D)$ has at least one copy to select from. On the complement of $\mathcal{X} \geq \bar{I}$ (that is, at least one of $(A,B,C,D)$ fails to have a copy to select), then ASTRAL-one selects $Q_1, Q_2, Q_3$ each with probability 0. So we consider the case $\mathcal{X} \geq \bar{I}$. We will use the notation $z_1 \land z_2 = \min\{z_1, z_2\}$.

**Proposition 1** (Quartet identifiability: Balanced case). Let $x = P''[i_a = i_b]$ and $y = P''[i_c = i_d]$. On the events $\mathcal{X} \geq \bar{I}$ and $I \geq 1$, we have almost surely

$$P''[Q_1] - P''[Q_2] > \frac{1}{12} \left( x - \frac{1}{I} \right) \land \left( y - \frac{1}{I} \right).$$

The proof of Proposition 1 is in Section 3.1.4. We first establish a series of lemmas.

The next lemma shows that $x,y \geq 1/I$, similarly to [LMWR19, Lemma 1]. In that work, it is proved that the probabilities of $\{i_a = i_b\}$ and $\{i_c = i_d\}$ are each at least $1/I$ under a different conditional probability measure. Our proof is otherwise identical.

**Lemma 1.** Let $x = P''[i_a = i_b]$ and $y = P''[i_c = i_d]$. On the events $\mathcal{X} \geq \bar{I}$ and $I \geq 1$, we have almost surely

$$x \land y \geq \frac{1}{I}.$$
Proof. For \( j \in \{1, \ldots, I\} \), let \( N_j \) be the number of gene copies descending from \( j \) in \( R \) that survive to the most recent common ancestor of \( A \) and \( B \). Conditioning on \((N_j)\), the choice of \( a \) and \( b \) in the locus tree is independent as in [LMWR19]. So \( i_a \) and \( i_b \) are picked proportionally to the \( N_j \)'s by symmetry. Then

\[
P''[i_a = i_b] = \mathbb{E}''[P''[i_a = i_b|(N_j)]] = \mathbb{E}'' \left[ \frac{\sum_{j=1}^{I} N_j^2}{(\sum_{j=1}^{I} N_j)^2} \right] \geq \frac{1}{I},
\]

as in [LMWR19, Lemma 1]. The same holds for \( y \), completing the proof of the lemma. \( \square \)

### 3.1.1 Ancestral locus configurations

Conditioned on \( X \) and \( I \), we will characterize the occurrence of \( Q_1, Q_2, Q_3 \) based on how \( i_x, x \in \{a, b, c, d\} \), are picked at the root \( R \), that is, based on which pairs \( i_x, i_y \) are equal. Then, in a worst-case scenario, we will analyze events of the coalescent process above \( R \). For an arbitrary quartet \((a, b, c, d)\), we relate the likelihood of \( Q_1, Q_2, Q_3 \) under each of the following events:

\[
E = a - b - c - d \\
F_{ab} = ab - c - d \\
F_{bc} = bc - a - d \\
G_{ab} = ab - cd \\
H_{abc} = abc - d \\
K = abcde,
\]

where \(-\) indicates separate lineages at \( R \) for the chosen copies from \( A, B, C, D \). For example, the event \( F_{ab} \) indicates that the \( i_c \) and \( i_d \) are different and are different from the common ancestral lineage for \( i_a \) and \( i_b \). See Figure 3. These events are disjoint and mutually exhaustive. Letting \( \mathcal{E} \) run across all the above events, the law of total probability implies

\[
P''[Q_i] = \sum_{\mathcal{E}} P''[Q_i | \mathcal{E}] P''[\mathcal{E}]. \tag{4}
\]
Figure 3: An example of a locus tree $L$ satisfying the event $F_{ab}$ for selected gene copies $a, b, c, d$. In particular, the edges of $L$ are depicted as residing within the fat edges of $T^Q$, however only the portion of $L$ coinciding with the edges of $T^Q$ is shown; the portion of $L$ above $R$ is omitted.

3.1.2 Reduction to coalescence above $R$

For the rooted locus quartet implied by the four copies $a, b, c, d$, let $\mathcal{NC}$ be the event that no coalescence event occurs beneath $R$ among the four corresponding lineages. The following lemma shows that conditioning on $\mathcal{NC}$ reduces the probability of $Q_1$ while increasing that of $Q_2$ and $Q_3$.

**Lemma 2.** For any $I$ and any $X \geq \overline{1}$ and any event $E \in \{E, F_{ab}, \ldots, G_{ab}, \ldots, H_{abc}, \ldots\}$,

we have

$$
\mathbb{P}''[Q_1|E] \geq \mathbb{P}''[Q_1|E \cap \mathcal{NC}] \quad \text{and} \quad \mathbb{P}''[Q_i|E] \leq \mathbb{P}''[Q_i|E \cap \mathcal{NC}], \quad i \in \{2, 3\},
$$

almost surely.

**Proof.** Under $\mathcal{NC}^c$, by definition, at least one of the pairs $\{a, b\}$ or $\{c, d\}$ coalesces below $R$. Both cases immediately lead to the same quartet topology, so $Q_1$ is guaranteed. See Figure 5 below for an illustration when $E = K$. Then the law of total probability implies

$$
\mathbb{P}''[Q_1|E] = \mathbb{P}''[\mathcal{NC}^c|\mathcal{E}] + \mathbb{P}''[Q_1|E \cap \mathcal{NC}] \mathbb{P}''[\mathcal{NC}|\mathcal{E}] \geq \mathbb{P}''[Q_1|E \cap \mathcal{NC}].
$$

Similarly

$$
\mathbb{P}''[Q_i|E] = \mathbb{P}''[Q_i|E \cap \mathcal{NC}] \mathbb{P}''[\mathcal{NC}|\mathcal{E}] \leq \mathbb{P}''[Q_i|E \cap \mathcal{NC}],
$$

for $i \in \{2, 3\}$. \qed
The event $K$ will play a special role in the proof and we treat it separately. For the other terms, combining (4) and Lemma 2, we have
\[
\mathbb{P}''[Q_1] - \mathbb{P}''[Q_2] = (\mathbb{P}''[Q_1 | K] - \mathbb{P}''[Q_2 | K])\mathbb{P}''[K] \\
+ \sum_{\mathcal{E} \neq K} (\mathbb{P}''[Q_1 | \mathcal{E}] - \mathbb{P}''[Q_2 | \mathcal{E}]) \mathbb{P}''[\mathcal{E}]
\geq (\mathbb{P}''[Q_1 | K] - \mathbb{P}''[Q_2 | K])\mathbb{P}''[K] \\
+ \sum_{\mathcal{E} \neq K} (\mathbb{P}''[Q_1 | \mathcal{E} \cap \mathcal{N}\mathcal{C}] - \mathbb{P}''[Q_2 | \mathcal{E} \cap \mathcal{N}\mathcal{C}]) \mathbb{P}''[\mathcal{E}].
\]

To prove Proposition 1, we derive an explicit bound on this last sum.

Under $\mathbb{P}''$, the events $E, H_{abc}, H_{abd}, H_{bcd}, H_{acd}$ are symmetric in the sense that switching the roles of $a$ and $c$ or the roles of $a$ and $d$ does not change the conditional probability of $Q_1$ and $Q_2$. Hence
\[
\mathbb{P}''[Q_1 | \mathcal{E} \cap \mathcal{N}\mathcal{C}] = \mathbb{P}''[Q_2 | \mathcal{E} \cap \mathcal{N}\mathcal{C}], \quad \forall \mathcal{E} \in \{E, H_{abc}, H_{abd}, H_{bcd}, H_{acd}\}
\]
and using this above we get
\[
\mathbb{P}''[Q_1] - \mathbb{P}''[Q_2] \geq (\mathbb{P}''[Q_1 | K] - \mathbb{P}''[Q_2 | K])\mathbb{P}''[K] \\
+ \sum_{j \in \{ab,ac,ad\}} (\mathbb{P}''[Q_1 | G_j \cap \mathcal{N}\mathcal{C}] - \mathbb{P}''[Q_2 | G_j \cap \mathcal{N}\mathcal{C}]) \mathbb{P}''[G_j] \\
+ \sum_{j \in \{ab,...,cd\}} (\mathbb{P}''[Q_1 | F_j \cap \mathcal{N}\mathcal{C}] - \mathbb{P}''[Q_2 | F_j \cap \mathcal{N}\mathcal{C}]) \mathbb{P}''[F_j].
\]

(5)

3.1.3 The $F$ and $G$ events

We now consider the events $\{F_{ab}, F_{cd}, F_{ac}, F_{bd}\}$ and $\{G_{ab}, G_{ac}\}$.

Event probabilities In the next lemma, we compute the probabilities of a given locus tree quartet satisfying the events in $\{F_{ab}, ..., F_{cd}, G_{ab}, G_{ac}\}$.

Lemma 3. Let $x = \mathbb{P}''[i_a = i_b]$ and $y = \mathbb{P}''[i_c = i_d]$. For $I \geq 2$ and any
\( X \geq 1 \), the following hold almost surely:

\[
\begin{align*}
\mathbb{P}''[F_{ab}] & = \frac{I - 2}{I} x (1 - y) \\
\mathbb{P}''[F_{cd}] & = \frac{I - 2}{I} (1 - x) y \\
\mathbb{P}''[F_{ac}] & = \mathbb{P}''[F_{bd}] = \frac{I - 2}{I} (1 - x)(1 - y) \\
\mathbb{P}''[G_{ab}] & = \frac{I - 1}{I} xy \\
\mathbb{P}''[G_{ac}] & = \frac{1}{I(I - 1)} (1 - x)(1 - y)
\end{align*}
\]

**Proof.** The calculations for \( F_{ab} \) and \( F_{cd} \) are similar, except that we condition on different events. Indeed, note that

\[
\begin{align*}
\mathbb{P}''[F_{ab}] & = \mathbb{P}''[F_{ab}|i_a = i_b, i_c \neq i_d] \mathbb{P}''[i_a = i_b] \mathbb{P}''[i_c \neq i_d] \\
\mathbb{P}''[F_{cd}] & = \mathbb{P}''[F_{cd}|i_a \neq i_b, i_c = i_d] \mathbb{P}''[i_a \neq i_b] \mathbb{P}''[i_c = i_d].
\end{align*}
\]

The conditional probability of \( F_{ab} \) is then obtained by considering that given the placement of the pair \((i_c, i_d)\) among the \( I \) ancestral lineages, the shared lineage \( i_a = i_b \) has \( I - 2 \) choices where they do not intersect \( \{i_c, i_d\} \). The result in the statement follows. Similarly, for \( F_i \) with \( i \in \{ac, bd\} \), we have

\[
\mathbb{P}''[F_i] = \mathbb{P}''[F_i|i_a \neq i_b, i_c \neq i_d] \mathbb{P}''[i_a \neq i_b] \mathbb{P}''[i_c \neq i_d].
\]

In this case, out of \( I(I - 1) \) choices for \( i_a \) and \( i_b \), the choice of \( i_c \) is determined and there are \( I - 2 \) remaining choices for \( i_d \), implying the result.

We use the same principle for \( G_{ab} \) and \( G_{ac} \). Keeping this in mind, we have

\[
\begin{align*}
\mathbb{P}''[G_{ab}] & = \mathbb{P}''[G_{ab}|i_a = i_b, i_c = i_d] \mathbb{P}''[i_a = i_b] \mathbb{P}''[i_c = i_d] \\
\mathbb{P}''[G_{ac}] & = \mathbb{P}''[G_{ac}|i_a \neq i_b, i_c \neq i_d] \mathbb{P}''[i_a \neq i_b] \mathbb{P}''[i_c \neq i_d],
\end{align*}
\]

and we proceed as before to get the result. \( \square \)

Using the previous lemma, we collect further bounds on the probabilities of events at the root of the locus tree.

**Lemma 4.** Letting again \( x = \mathbb{P}''[i_a = i_b] \) and \( y = \mathbb{P}''[i_c = i_d] \), the following statements hold.
(a) If \( I = 2 \) then
\[
\mathbb{P}''[G_{ab}] - \mathbb{P}''[G_{ac}] \geq \left( x - \frac{1}{2} \right) \wedge \left( y - \frac{1}{2} \right)
\]

(b) If \( I \geq 3 \) and \( x \wedge y \geq 1/2 \), then
\[
\mathbb{P}''[G_{ab}] - \mathbb{P}''[G_{ac}] \geq 1/8.
\]

(c) If \( I = 2 \),
\[
\mathbb{P}''[F_{ab}] - \mathbb{P}''[F_{ac}] - \mathbb{P}''[F_{bd}] + \mathbb{P}''[F_{cd}] = 0.
\]

(d) If \( I \geq 3 \) and \( x \wedge y \leq 1/2 \), then
\[
\mathbb{P}''[F_{ab}] - \mathbb{P}''[F_{ac}] - \mathbb{P}''[F_{bd}] + \mathbb{P}''[F_{cd}] \geq \frac{1}{4} \left( x - \frac{1}{I} \right) \wedge \left( y - \frac{1}{I} \right).
\]

Proof. By Lemma 3, \( \mathbb{P}''[G_{ab}] - \mathbb{P}''[G_{ac}] = \frac{1}{2}(x + y - 1) \) which implies part (a).

To prove (b), observe that by Lemma 3 again,
\[
\mathbb{P}''[G_{ab}] - \mathbb{P}''[G_{ac}] = \frac{I - 1}{I} xy - \frac{1}{I(I - 1)}(1 - x)(1 - y)
\]
\[
\geq \frac{1}{2} \left( \frac{I - 1}{I} y - \frac{1}{I(I - 1)}(1 - y) \right)
\]
\[
\geq \frac{1}{4} \left( \frac{I - 1}{I} - \frac{1}{I(I - 1)} \right)
\]
\[
= \frac{1}{4} \left( \frac{I - 2}{I - 1} \right)
\]
where the inequalities are justified by the assumption \( x \wedge y \geq 1/2 \). Since \( I \geq 3 \), it follows that \( \mathbb{P}''[G_{ab}] - \mathbb{P}''[G_{ac}] \geq 1/8 \).

To prove (c) and (d), observe that by Lemma 3,
\[
\mathbb{P}''[F_{ab}] - \mathbb{P}''[F_{ac}] - \mathbb{P}''[F_{bd}] + \mathbb{P}''[F_{cd}]
\]
\[
= \frac{I - 2}{I} (x(1 - y) + y(1 - x)) - 2\frac{I - 2}{I(I - 1)}(1 - x)(1 - y)
\]
\[
= \frac{I - 2}{I(I - 1)} ((1 - y)((I - 1)x - (1 - x)) + (1 - x)((I - 1)y - (1 - y)))
\]
\[
= \frac{I - 2}{I(I - 1)} ((1 - y)(Ix - 1) + (1 - x)(Iy - 1)).
\]
Clearly if $I = 2$, the right-hand side is zero, which proves (c). Furthermore, since $x, y \geq 1/I$ by Lemma 1, it follows that both $(1 - y)(I x - 1) \geq 0$ and $(1 - x)(I y - 1) \geq 0$, and therefore

$$P''[F_{ab}] - P''[F_{ac}] - P''[F_{bd}] + P''[F_{cd}] \geq \frac{I - 2}{I(I - 1)} (1 - u)(Iv - 1)$$

for $(u, v) \in \{(x, y), (y, x)\}$. Taking $u = \min(x, y)$ and $v = \max(x, y)$ gives

$$P''[F_{ab}] - P''[F_{ac}] - P''[F_{bd}] + P''[F_{cd}] \geq \frac{I - 2}{I(I - 1)} (1 - \min(x, y))(I \max(x, y) - 1)$$

$$\geq \frac{I - 2}{I - 1} (1 - \min(x, y)) \left( \max(x, y) - \frac{1}{I} \right)$$

$$\geq \frac{1}{2} (1 - \min(x, y)) \left( \max(x, y) - \frac{1}{I} \right)$$

$$\geq \frac{1}{4} \left( \max(x, y) - \frac{1}{I} \right)$$

which implies (d). \hfill \Box

**Conditional probabilities of quartet topologies** In the following lemma, we give expressions for $P''[Q_i | E \cap NC]$ across the events $\{F_{ab}, F_{cd}, F_{ac}, F_{bd}\}$ and $\{G_{ab}, G_{ac}\}$.

**Lemma 5.** (a) For any $I$ and any $X \geq 1/I$, we have

$$P''[Q_1 | F_{ab} \cap NC] = P''[Q_1 | F_{cd} \cap NC] = P''[Q_2 | F_{ac} \cap NC] = P''[Q_2 | F_{bd} \cap NC] = : \phi''_+$$

and

$$P''[Q_2 | F_{ab} \cap NC] = P''[Q_2 | F_{cd} \cap NC] = P''[Q_1 | F_{ac} \cap NC] = P''[Q_1 | F_{bd} \cap NC] = : \phi''_-. $$

(b) For any $I$ and any $X \geq 1/I$, we have

$$P''[Q_1 | G_{ab} \cap NC] = 1$$

and

$$P''[Q_2 | G_{ac} \cap NC] = 1.$$
Proof. (a) The quantities $\phi_+''$ and $\phi_-''$ are indeed well-defined as above by symmetry. (b) By switching the roles of $b$ and $c$, we observe that $\mathbb{P}''[Q_1|G_{ab} \cap \mathcal{NC}] = \mathbb{P}''[Q_2|G_{ac} \cap \mathcal{NC}]$. To see why $\mathbb{P}''[Q_1|G_{ab} \cap \mathcal{NC}] = 1$, we again examine the topology above the root with leaves $ab$ and $cd$. At least one of these leaves descends from a daughter edge, which implies $Q_1$ is constructed with probability 1. This completes the proof of the lemma. \hfill $\square$

The following lemma establishes that, conditioned on $F_{ab}$ and $\mathcal{NC}$, the difference in probability between $Q_1$ and $Q_2$ is at least $1/3$.

**Lemma 6.** For $I \geq 1$ and any $X \geq \bar{I}$, we have

$$\phi_+'' - \phi_-'' \geq \frac{1}{3}.$$ 

Proof. By definition of $\phi_+''$ and $\phi_-''$, it suffices to show $\mathbb{P}''[Q_1|F_{ab} \cap \mathcal{NC}] - \mathbb{P}''[Q_2|F_{ab} \cap \mathcal{NC}] \geq 1/3$. Conditioned on $F_{ab} \cap \mathcal{NC}$, no coalescence event between the chosen lineages occurs beneath $R$. So, we examine the topology of the locus tree above the root with leaf set being the three leaves implied by $F_{ab}$. Using the law of total probability, we condition further across the three possible rooted locus topologies on the three leaves $ab$, $c$, and $d$. For each $i \in \{ab, c, d\}$, let $\tau_i$ be the rooted topology on three leaves in which the outgroup is labeled by $i$ and the other two leaves are labelled from the remaining letters in $\{a, b, c, d\}$. For example, $\tau_{ab}$ has $c$ and $d$ as siblings with $ab$ as the outgroup. Then

$$\mathbb{P}''[Q_1|F_{ab} \cap \mathcal{NC}] - \mathbb{P}''[Q_2|F_{ab} \cap \mathcal{NC}] = \frac{1}{3} \sum_i \left(\mathbb{P}''[Q_1|\tau_i, F_{ab} \cap \mathcal{NC}] - \mathbb{P}''[Q_2|\tau_i, F_{ab} \cap \mathcal{NC}]\right),$$

where we used the fact that $\mathbb{P}''[\tau_i|F_{ab} \cap \mathcal{NC}] = 1/3$ for each $i$. Now we compute the summands. If $i = ab$, then either $ab$ descends from a daughter lineage or the pair $(c, d)$ descends from a daughter lineage, meaning we observe $Q_1$ with probability 1 and $Q_2$ with probability 0. In the other two cases, let $p > 0$ be the probability that $a$ and $b$ coalesce along the pendant edge for $ab$. If they do not coalesce along the pendant edge, then the lineages from $a$ and $b$ live in the same population as that of, say, $c$. Then there is probability $1/3$ that the first coalescing pair among $a, b, c$ is $a, b$. So the probability of observing
$Q_1$ is $p + \frac{1}{3}(1 - p)$. There is probability $1/3$ that the first coalescing pair among $a, b, c$ is $a, c$, so the probability of observing $Q_2$ is $\frac{1}{3}(1 - p)$. Then

$$\phi_+ - \phi_- = \frac{1}{3} \left( 1 - 0 + 2 \left( p + \frac{1}{3}(1 - p) - \frac{1}{3}(1 - p) \right) \right) = \frac{1}{3}(1 + 2p) \geq \frac{1}{3}.$$ 

\[\Box\]

### 3.1.4 Proof of Proposition 1

With that we can prove Proposition 1.

**Proof of Proposition 1.** In the $I = 1$ case, $P''[K] = 1$ so

$$P''[Q_1] - P''[Q_2] = P''[Q_1|K] - P''[Q_2|K] > 0,$$  \hspace{0.5cm} (6)

where we used that, under $K \cap NC$, the quartets $Q_1$ and $Q_2$ occur with equal probability under $P''$. Since $x - 1/I = y - 1/I = 0$, the claim follows.

For $I \geq 2$, (5) and Lemma 5 implies that

$$P''[Q_1] - P''[Q_2] \geq (P''[Q_1|K] - P''[Q_2|K])P''[K]$$

$$+ P''[G_{ab}] - P''[G_{ac}] + (\phi''_+ - \phi''_-) (P''[F_{ab}] + P''[F_{cd}])$$

$$- (\phi''_+ - \phi''_-) (P''[F_{ac}] + P''[F_{bd}])$$

$$> P''[G_{ab}] - P''[G_{ac}]$$

$$+ (\phi''_+ - \phi''_-) (P''[F_{ab}] - P''[F_{ac}] - P''[F_{bd}] + P''[F_{cd}]),$$

where again we used that, under $K \cap NC$, the quartets $Q_1$ and $Q_2$ occur with equal probability. If $I = 2$, then by Lemma 6 and Lemma 4 parts (a) and (c), this leads to

$$P''[Q_1] - P''[Q_2] > \left( x - \frac{1}{7} \right) \land \left( y - \frac{1}{7} \right).$$ \hspace{0.5cm} (7)

If $I \geq 3$, then by Lemma 4 parts (b) and (d),

$$P''[Q_1] - P''[Q_2] > \begin{cases} 1/8 & \text{if } x \land y \geq 1/2 \\ 1/12 \left( x - \frac{1}{7} \right) \land \left( y - \frac{1}{7} \right) & \text{if } x \land y \leq 1/2 \end{cases}$$ \hspace{0.5cm} (8)

It follows that $P''[Q_1] - P''[Q_2] > \frac{1}{12} \left( x - \frac{1}{7} \right) \land \left( y - \frac{1}{7} \right)$, finishing the proof of the main claim in the balanced case. \[\Box\]
3.2 Caterpillar case

We now consider the caterpillar case. Without loss of generality, assume the species tree restricted to $Q$ has rooted topology as in the bottom right of Figure 2. Let $R$ be the most recent common ancestor of $A, B, C$ and let $I$ be the number of locus copies exiting $R$ (forward in time). Let $\mathbb{P}''$ be the probability measure indicating conditioning on $I$ and $X$. Let $i_x \in \{1, ..., I\}$ be the ancestral lineage of $x \in \{a, b, c\}$ in $R$. As with the balanced case, on the complement of $X \geq \vec{1}$, ASTRAL-one selects $Q_1, Q_2, Q_3$ each with probability 0. To prove

$$\mathbb{P}[Q_1] > \max\{\mathbb{P}[Q_2], \mathbb{P}[Q_3]\},$$

it is sufficient to prove Proposition 2 below for $X \geq \vec{1}$.

**Proposition 2** (Quartet identifiability: Caterpillar case). Let $x = \mathbb{P}''[i_a = i_b]$. On the events $I \geq 1$ and $X \geq \vec{1}$, we have almost surely

$$\mathbb{P}''[Q_1] - \mathbb{P}''[Q_2] > \frac{1}{3} \left( x - \frac{1}{I} \right).$$

Similarly to the balanced case, in order to prove this proposition we consider the following events:

$$E = a - b - c$$
$$G_{ab} = ab - c \quad G_{ac} = ac - b \quad G_{bc} = bc - a$$
$$K = abc,$$

where $-$ indicates separation of lineages in $R$ of the selected copies of $A, B, C$. Letting $\mathcal{E}$ run across all events, the law of total probability implies

$$\mathbb{P}''[Q_i] = \sum_{\mathcal{E}} \mathbb{P}''[Q_i|\mathcal{E}]\mathbb{P}''[\mathcal{E}].$$  \hspace{1cm} (9)

Let $\mathcal{N}C$ be the event that no coalescent event occurs beneath $R$ between the three lineage corresponding to $a, b, c$. Analogues to Lemmas 1, 2, 3, 4, and 5 hold with similar proofs.

**Lemma 7.** Let $x = \mathbb{P}''[i_a = i_b]$. On the events $X \geq \vec{1}$ and $I \geq 1$, we have almost surely

$$x \geq \frac{1}{I}.$$
Lemma 8. Let the species tree be a rooted caterpillar on four leaves $A, B, C, D$. For all $I \geq 1$ and $X \geq \overline{1}$, and any event $E \in \{E, G_{ab}, G_{ac}, G_{bc}\}$,

$$P''[Q_1|E] \geq P''[Q_1|E \cap NC] \quad \text{and} \quad P''[Q_i|E] \leq P''[Q_i|E \cap NC], \quad i \in \{2, 3\}. $$

Lemma 9. For $I \geq 2$ and any $X \geq \overline{1}$, let $x = P''[i_a = i_b]$. Then the following hold:

$$P''[G_{ab}] = \frac{I - 1}{I} x$$

$$P''[G_{ac}] = P''[G_{bc}] = \frac{1}{I} (1 - x).$$

Lemma 10. On $I \geq 1$, almost surely

$$P''[G_{ab}] - P''[G_{ac}] = x - \frac{1}{I}.$$

Lemma 11. For any $I$ and any $X \geq \overline{1}$, we have

$$P''[Q_1|G_{ab} \cap NC] = P''[Q_2|G_{ac} \cap NC] =: \psi''$$

and

$$P''[Q_2|G_{ab} \cap NC] = P''[Q_1|G_{ac} \cap NC] =: \psi'''.$$

3.2.1 The $G$ events

The following lemma bounds the conditional probability difference for the $G$ events.

Lemma 12. On the events $I \geq 1$ and $X \geq \overline{1}$, we have almost surely

$$\psi'' - \psi''' \geq \frac{1}{3}.$$

Proof. By definition of $\psi''$ and $\psi'''$, it suffices to show that $P''[Q_1|G_{ab} \cap NC] - P''[Q_2|G_{ab} \cap NC] \geq 1/3$. The proof of this inequality involves decomposing $G_{ab} \cap NC$ into a number of subcases, depicted in Figure 4, and computing the probabilities of $Q_1$ and $Q_2$ in each subcase. Let $S$ be the most recent common ancestor of $Q$ in the species tree. Let $\Lambda$ be the event that the $ab$ and $c$ individuals in $R$ descend from a common ancestor in $S$ and let $q = P''[\Lambda|G_{ab} \cap NC]$. There are two cases:
1. (Condition on $\Lambda$) Let $\mathcal{C}$ be the event that gene copies $a$ and $b$ coalesce above $R$ and below the MRCA of loci $i_{ab} = i_a = i_b$ and $i_c$. Let $q' = \mathbb{P}''[\mathcal{C}|\Lambda, G_{ab}, \mathcal{NC}]$. We claim that $q' \geq 1/2$. To see this, observe that conditional on $G_{ab} \cap \mathcal{NC}$ the loci $i_{ab}$ and $i_c$ share the same ancestral locus at $S$ only if there occurred a duplication event between $S$ and $R$ which is ancestral to both of them. Therefore with probability at least $1/2$, the gene copies $a, b$ coalesce along their shared pendant edge in the rooted topology between $R$ and $S$, proving the claim. Furthermore, it is obvious that

$$\mathbb{P}''[Q_1|\mathcal{C}, \Lambda, G_{ab}, \mathcal{NC}] - \mathbb{P}''[Q_2|\mathcal{C}, \Lambda, G_{ab}, \mathcal{NC}] = 1.$$  \hspace{1cm} (10)

On the other hand, conditional on $\mathcal{C}^c$, the copies of $a, b$ and $c$ enter the same population and are then symmetric, and hence

$$\mathbb{P}''[Q_1|\mathcal{C}^c, \Lambda, G_{ab}, \mathcal{NC}] - \mathbb{P}''[Q_2|\mathcal{C}^c, \Lambda, G_{ab}, \mathcal{NC}] = 0.$$  \hspace{1cm} (11)

2. (Condition on $\Lambda^c$) Let $\mathcal{H}_j$ be the event that copies $d$ and $j$ share the same ancestor in the locus tree at $S$, and define $\mathcal{H} = (\mathcal{H}_{ab} \cup \mathcal{H}_{c})^c$ and $r = \mathbb{P}''[\mathcal{H}|\Lambda^c, G_{ab}, \mathcal{NC}]$. Then by symmetry, $\mathbb{P}''[\mathcal{H}_{ab}|\Lambda^c, G_{ab}, \mathcal{NC}] = \mathbb{P}''[\mathcal{H}_{c}|\Lambda^c, G_{ab}, \mathcal{NC}] = 1/2$. By a further symmetry argument similar to that made in Case 1 we have

$$\mathbb{P}''[Q_1|\mathcal{H}_{ab}, \Lambda^c, G_{ab}, \mathcal{NC}] - \mathbb{P}''[Q_2|\mathcal{H}_{ab}, \Lambda^c, G_{ab}, \mathcal{NC}] \geq 0,$$  \hspace{1cm} (12)
where the inequality accounts for the possibility that the lineages from \(a\) and \(b\) coalesce between \(R\) and \(S\). Let \(\tau\) be the topology of the locus tree restricted to the copies \(ab, c, d\) and restricted to the portion above \(S\) (and suppressing nodes of degree 2). Conditioned on \(\mathcal{H}_c\), we have \(\tau = (ab, cd)\), so it must be the case that either \(ab\) descends from a daughter lineage or \(cd\) descends from a daughter lineage, meaning we observe \(Q_1\) with probability 1 and \(Q_2\) with probability 0. Therefore

\[
P''[Q_1|\mathcal{H}_c, \Lambda^c, G_{ab}, \mathcal{NC}] - P''[Q_2|\mathcal{H}_c, \Lambda^c, G_{ab}, \mathcal{NC}] = 1. \tag{13}
\]

It remains to consider the case \(\mathcal{H} = (\mathcal{H}_{ab} \cup \mathcal{H}_c)^c\). Let \(\mathcal{K}\) be the event that \(\tau = (ab, (c, d))\). By symmetry, the three possible topologies are equally likely, so \(P''[\mathcal{K}|\mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] = 1/3\). Conditioned on \(\mathcal{K}\), either \(ab\) descends from a daughter lineage or the pair \((c, d)\) descends from a daughter lineage, and therefore

\[
P''[Q_1|\mathcal{K}, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] - P''[Q_2|\mathcal{K}, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] = 1. \tag{14}
\]

Conditioned on \(\mathcal{K}^c\), let \(p\) denote the probability that gene copies \(a\) and \(b\) coalesce along the pendant edge for \(ab\) in the rooted triple above \(S\). Then

\[
P''[Q_1|\mathcal{K}^c, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] = p + \frac{1}{3}(1 - p),
\]

and

\[
P''[Q_2|\mathcal{K}^c, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] = \frac{1}{3}(1 - p),
\]

and hence

\[
P''[Q_1|\mathcal{K}^c, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] - P''[Q_2|\mathcal{K}^c, \mathcal{H}, \Lambda^c, G_{ab}, \mathcal{NC}] \geq p, \tag{15}
\]

where again the inequality accounts for the possibility that the lineages from \(a\) and \(b\) coalesce between \(R\) and \(S\).

Finally, applying the law of total probability and using equations (10)-(15) gives

\[
P''[Q_1|G_{ab}, \mathcal{NC}] - P''[Q_2|G_{ab}, \mathcal{NC}] \geq q'q + \left(\frac{1 - r}{2} + \frac{1}{3}r + \frac{2}{3}qr\right)(1 - q)
\]

\[
\geq \frac{1}{2}q + \frac{1}{3}(1 - q)
\]

\[
\geq \frac{1}{3},
\]

where the second inequality follows from \(q' \geq 1/2\) and \(r \geq 0\). \(\Box\)
3.2.2 Proofs of Proposition 2 and Theorems 2 and 3

With that, the proof of Proposition 2 is similar to that of Proposition 1. In both the balanced and caterpillar cases, observe that $P''[Q_1] - P''[Q_3] = P''[Q_1] - P''[Q_2]$ by switching the roles of $c$ and $d$. By Propositions 1 and 2, all species quartet topologies are identifiable and hence we have verified Theorem 2.

Theorem 3 then follows, along similar lines as [LMWR19, Theorem 2], from the law of large numbers.

3.3 Proof of consistency for ASTRAL-multi

Before finishing the proof of Theorem 1, we give a proof of Theorem 4.

Proof of Theorem 4. Let $N_{AB|CD}$ (respectively $N_{AC|BD}, N_{AD|BC}$) be the number of choices consisting of one gene copy in the gene tree from each species in $Q = \{A, B, C, D\}$ whose corresponding restriction in $t_1$ agrees with $AB|CD$ (respectively $AC|BD, AD|BC$). Similarly to [LMWR19, Theorem 3], it suffices to show that

$$E[N_{AB|CD}] > \max \{E[N_{AC|BD}], E[N_{AD|BC}]\}. \tag{16}$$

Letting again $X = (A, B, C, D)$, by taking expectation with respect to $I$ in Propositions 1 and 2, we have on the event $X \geq \vec{1}$ that

$$P[q = AB|CD | X] > \max\{P[q = AC|BD | X], P[q = AD|BC | X]\}, \tag{17}$$

where $q$ is the topology of a uniformly chosen quartet among $A, B, C, D$. Let $M = ABCD$ be the number of quartet choices and let $q_i, i = 1, \ldots, M$ be the corresponding topologies ordered arbitrarily. Because $q$ is a uniform choice, we have

$$P[q = AB|CD | X] = \frac{1}{M} \sum_{i=1}^{M} P[q_i = AB|CD | X], \tag{18}$$

and similarly for the other topologies. Since

$$N_{AB|CD} = \sum_{i=1}^{M} 1\{q_i = AB|CD\},$$

and similarly for the other topologies, taking expectations and using (17) and (18) gives (16) as claimed. \qed
4 Proof of sample complexity bound

To prove Theorem 1, our sample complexity result for ASTRAL-one, we use a union bound over all quartets and build on the analysis of Section 3. In particular, the key step of the proof is a more careful analysis of the event $K$ that appeared in the proof of Theorem 2. We first discuss a number of quantities that play an important role in the analysis.

4.1 Bounds on branching process quantities

We highlight the role of a number of parameters in the sample complexity: the shortest branch length in the species tree, $f$; the depth of the species tree, $\Delta$; and the duplication and loss rates, $\lambda$ and $\mu$. These parameters enter the analysis through three quantities of significance:

- **Coalescence of a pair of lineages on an edge:** In the standard coalescent, the probability that a pair of lineages has coalesced by time $f$ is
  \[ \gamma = 1 - e^{-f}. \]

- **Survival probability of a quartet:** For a quartet $Q = \{A, B, C, D\}$, let $X_Q = (A, B, C, D)$ be the number of gene copies in the corresponding species. The smallest probability over all quartets that a gene family contains a copy in each species will be denoted by
  \[ \sigma := \min_Q \mathbb{P}[X_Q \geq \vec{1}]. \]

- **Expected number of lineages at a vertex:** For any vertex $R$ in the species tree, let $I_R$ be the number of copies at $R$ in a single gene family. The largest expectation of $I_R$ over all vertices will be denoted by
  \[ \alpha = \max_R \mathbb{E}[I_R]. \]

These last two quantities can be controlled using branching process theory. See e.g. [Ste16, Section 9.2] for the relevant results in the phylogenetic context. We use the notation $z_1 \lor z_2 = \max\{z_1, z_2\}$.

**Lemma 13.** The following hold:

\[ \text{26} \]
• When $\mu > \lambda$,

$$\sigma \geq \left[ \frac{1}{e^{(\mu-\lambda)\Delta}} \left( 1 - \frac{\lambda}{\mu} \right) \right]^4.$$ 

• When $\lambda > \mu$,

$$\sigma \geq \left[ 1 - \frac{\mu}{\lambda} \right]^4.$$ 

**Proof.** Let $Q = \{A, B, C, D\}$ be a quartet and let as before $X_Q = (A, B, C, D)$ be the number of gene copies in the corresponding species. Assume the species tree topology on $Q$ is balanced (the argument in the caterpillar case being similar). The probability that $A \geq 1$ is given by

$$\mathbb{P}[A \geq 1] = 1 - \frac{\mu}{\lambda} q(\Delta),$$

where

$$q(t) = \frac{1 - e^{-(\lambda-\mu)t}}{\lambda - \mu e^{-(\lambda-\mu)t}}.$$ 

It can be checked that, whether $\mu > \lambda$ or $\mu < \lambda$, the function $q(t)$ is increasing in $t$. Conditioned on $\{A \geq 1\}$, there is at least one copy in each vertex along the path between the root and $A$. Hence

$$\mathbb{P}[D \geq 1 \mid A \geq 1] \geq 1 - \frac{\mu}{\lambda} q(\Delta).$$

Repeating this argument for $B$ and $C$ gives

$$\mathbb{P}[X_Q \geq 1] \geq \left( 1 - \frac{\mu}{\lambda} q(\Delta) \right)^4.$$ 

It remains to bound the right-hand side. When $\lambda > \mu$, $q(t) \to 1$ as $t \to +\infty$, which implies $q(t) \leq 1$ by monotonicity. So $1 - \frac{\mu}{\lambda} q(\Delta) \geq 1 - \frac{\mu}{\lambda}$. On the other hand, when $\mu > \lambda$,

$$1 - \frac{\mu}{\lambda} q(\Delta) = \frac{\lambda}{\lambda - \mu e^{-(\lambda-\mu)\Delta}} \left( 1 - \frac{\lambda}{\mu} \right) \geq \left( 1 - \frac{\lambda}{\mu} \right).$$

$\square$
Lemma 14. We have

\[ \alpha \leq 1 \lor e^{(\lambda - \mu)\Delta}. \]

Proof. Recall that we assume there is a single lineage at the top pendant vertex of the species tree. If the time elapsed between this vertex and another vertex \( U \) is \( d \), then the expectation number of lineages at \( U \) is \( e^{(\lambda - \mu)d} \). The result follows from the fact that \( d \leq \Delta \) by considering separately the cases \( \mu > \lambda \) and \( \lambda > \mu \).

\[ \square \]

4.2 Sufficient effective number of samples

For a quartet \( Q \), let \( \mathcal{K}_Q \) be the set of gene trees such that each species in \( Q \) has at least one gene copy. For \( k^* \geq 1 \), let

\[ S_{k^*} = \{ \vert \mathcal{K}_Q \vert \geq k^* : \forall Q \}. \]

Lemma 15. For any \( k^* \geq 1 \) and \( \epsilon \in (0, 1) \), it holds that \( \mathbb{P}[S_{k^*}] \geq 1 - \epsilon \) provided

\[ k \geq \left\{ \frac{2k^*}{\sigma} \right\} \lor \left\{ \frac{8}{\sigma^2} \log \frac{n}{\epsilon} \right\}. \]

Proof. For any \( Q \), by the definition of \( \mathcal{K}_Q \), if \( k \) is the number of loci then

\[ \mathbb{E}[\mathcal{K}_Q] \geq k\sigma. \]

Assume \( k \) is large enough that \( \frac{1}{2}k\sigma \geq k^* \). Then by the union bound and Hoeffding’s inequality (see, e.g. [Ver18]), using the fact that the gene trees are independent,

\[ \mathbb{P}[S_{k^*}] \leq \sum_Q \mathbb{P}[\vert \mathcal{K}_Q \vert < k^*] \]

\[ \leq \sum_Q \mathbb{P}[\vert \mathcal{K}_Q \vert < \frac{1}{2}k\sigma] \]

\[ \leq \sum_Q \mathbb{P}[\mathbb{E}[\mathcal{K}_Q] - \vert \mathcal{K}_Q \vert > \frac{1}{2}k\sigma] \]

\[ \leq n^4 \exp \left( -2\left(\frac{k\sigma/2}{k}\right)^2 \right) \]

\[ \leq \epsilon, \]

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if
\[ k \geq \frac{2}{\sigma^2} \log \frac{n^4}{\epsilon}, \]
and since \( \epsilon < 1 \) this inequality holds whenever
\[ k \geq \frac{8}{\sigma^2} \log \frac{n}{\epsilon}. \]
That proves the claim. \( \square \)

4.3 The event \( K \)

Using the notation of Section 3, fix a quartet of species \( Q = \{A, B, C, D\} \), let \( \mathbb{P}' \) denote the conditional probability given the event \( \{\mathcal{X}_Q \geq \vec{1}\} \), and define \( \delta' = \mathbb{P}'[Q_1] - 1/3 \). Since \( \mathbb{P}'[Q_2] = \mathbb{P}'[Q_3] \), we have
\[ \delta' = \mathbb{P}'[Q_1] - \frac{\mathbb{P}'[Q_1] + \mathbb{P}'[Q_2] + \mathbb{P}'[Q_3]}{3} = \frac{2}{3} (\mathbb{P}'[Q_1] - \mathbb{P}'[Q_2]) \] (19)

We seek to bound the right-hand side. Assume first that \( T^Q \) is balanced. Let \( \mathbb{P}'_i \) indicate \( \mathbb{P}' \) conditioned on \( \{I = i\} \). By the proof of Proposition 1, specifically the argument leading up to (6), (7) and (8), we have
\[ \mathbb{P}''[Q_1] - \mathbb{P}''[Q_2] \geq (\mathbb{P}''[Q_1|K] - \mathbb{P}''[Q_2|K])\mathbb{P}'[K]. \]

By further conditioning on the event \( \{\mathcal{X}_Q \geq \vec{1}\} \) (which has positive probability when \( I \geq 1 \)), we get for all \( i \geq 1 \)
\[ \mathbb{P}'_i[Q_1] - \mathbb{P}'_i[Q_2] \geq (\mathbb{P}'_i[Q_1|K] - \mathbb{P}'_i[Q_2|K])\mathbb{P}'[K]. \]

On the event \( K \cap \mathcal{NC}, Q_1 \) and \( Q_2 \) are equally likely by symmetry. On \( K \cap \mathcal{NC}^c, \) at least one of the pairs \( \{a, b\} \) or \( \{c, d\} \) coalesces below \( R \), guaranteeing \( Q_1 \) (similarly to the proof of Lemma 2). See Figure 5 for an illustration. Hence, we have
\[
\begin{align*}
\mathbb{P}'_i[Q_1] - \mathbb{P}'_i[Q_2] &\geq \mathbb{P}'_i[Q_1 \cap K] - \mathbb{P}'_i[Q_2 \cap K] \\
&= \mathbb{P}'_i[Q_1 \cap K \cap \mathcal{NC}] - \mathbb{P}'_i[Q_2 \cap K \cap \mathcal{NC}] \\
&\quad + \mathbb{P}'_i[Q_1 \cap K \cap \mathcal{NC}^c] - \mathbb{P}'_i[Q_2 \cap K \cap \mathcal{NC}^c] \\
&\geq \mathbb{P}'_i[K \cap \mathcal{NC}^c].
\end{align*}
\]
To bound the right-hand side, we consider the event $C_{ab}$ that the lineages picked from $A$ and $B$ coalesce below $R$. Notice in particular that $C_{ab}$ implies \( \{i_a = i_b\} \). The event $K \cap NC^c$ is implied by $C_{ab}$ together with $\{i_c = i_d = i_a\}$, which are conditionally independent. By Lemma 1, the latter has probability at least \( P'_i[i_c = i_d] \geq \frac{1}{i^2} \). Hence,

\[
P'_i[Q_1] - P'_i[Q_2] \geq P'_i[C_{ab}] \cdot \frac{1}{i^2}.
\]

(20)

By a similar argument in the caterpillar case, we have

\[
P'_i[Q_1] - P'_i[Q_2] \geq P'_i[K \cap NC^c]
\]

\[
\geq P'_i[C_{ab}] \cdot \frac{1}{i}
\]

\[
\geq P'_i[C_{ab}] \cdot \frac{1}{i^2}.
\]

(21)

It remains to bound $P'_i[C_{ab}]$.

**Lemma 16.** We have

\[
P'_i[C_{ab}] \geq \left\{ \gamma \wedge \frac{1}{8} \right\} \cdot \frac{1}{i}.
\]

*Proof.* Similarly to the proof of Lemma 1, for copy $\ell$ at $R$, let $N_\ell$ be the number of its descendant copies at $R'$, the most recent common ancestor of $A$ and $B$, and let $J = \sum_{\ell=1}^i N_\ell$. We consider two cases for $N_\ell$:

1. In the case $N_\ell = 1$ and $\{i_a = i_b = \ell\}$, let $Y_\ell$ be the indicator function of the event that the lineages from $a$ and $b$ coalesce before $R$. So $Y_\ell = 1$ if the lineages from $a$ and $b$ coalesce before $R$, and 0 otherwise. Under the standard coalescent, the probability of that coalescent event is at least $\gamma$. Here, though, we are working under the bounded coalescent. In the case that a daughter edge is ancestral to the lineages from $a$ and $b$, the additional conditioning on complete coalescence only increases the probability that $a$ and $b$ coalesce before $R$. So $\gamma$ remains a lower bound.

2. In the case $N_\ell \geq 2$ and $\{i_a = i_b = \ell\}$, let $Z_\ell$ be the indicator function of the event that the lineages from $a$ and $b$ coalesce before $R$. So
Figure 5: An example realization of $K \cap \mathcal{NC}^c$. In particular, a locus tree satisfying event $K$ for copies $a, b, c, d$ is depicted in the case where $T^Q$ is balanced; additionally, a realization of the gene tree is shown (in blue) within the locus tree. This realization satisfies $K \cap \mathcal{NC}^c$ since at least one coalescence event—in this case between the ancestors of gene copies $c$ and $d$—has occurred on the gene tree below $R$. Since copies $c$ and $d$ are therefore not separated by an internal edge on the gene tree, the event $Q_1$ occurs.

$Z_\ell = 1$ if the lineages from $a$ and $b$ coalesce before $R$, and 0 otherwise. Since $N_\ell \geq 2$, there is at least one duplication below $\ell$ before $R'$. By symmetry, there is probability at least $1/2$ that the first duplication produces a daughter edge with at least half of the descendants of $\ell$ below it at $R'$. Under $\{i_a = i_b = \ell\}$, there is then a probability at least $1/4$ that $a$ and $b$ descend from copies at $R'$ below that daughter edge. So overall there is probability at least $1/8$ that $Z_\ell = 1$ in that case.
Putting these two cases together, we get

\[
\mathbb{P}'_i[C_{ab}] = \mathbb{E}'_i \left[ \mathbb{E}'_i \left[ \sum_{\ell, N_\ell = 1} \left( \frac{N_\ell}{J} \right)^2 Y_\ell + \sum_{\ell, N_\ell > 1} \left( \frac{N_\ell}{J} \right)^2 Z_\ell \right] \right]
\]

\[
= \mathbb{E}'_i \left[ \sum_{\ell, N_\ell = 1} \left( \frac{N_\ell}{J} \right)^2 \mathbb{E}'_i \left[ Y_\ell \mid N_\ell \right] + \sum_{\ell, N_\ell > 1} \left( \frac{N_\ell}{J} \right)^2 \mathbb{E}'_i \left[ Z_\ell \mid N_\ell \right] \right]
\]

\[
\geq \left\{ \gamma \wedge \frac{1}{8} \right\} \mathbb{E}'_i \left[ \sum_{\ell, N_\ell = 1} \left( \frac{N_\ell}{J} \right)^2 + \sum_{\ell, N_\ell > 1} \left( \frac{N_\ell}{J} \right)^2 \right]
\]

\[
\geq \left\{ \gamma \wedge \frac{1}{8} \right\} \frac{1}{\ell},
\]

as in [LMWR19, Lemma 1], proving the claim. \( \square \)

Lemma 17. We have

\[
\delta' \geq \frac{2}{3} \left\{ \gamma \wedge \frac{1}{8} \right\} \frac{\sigma^3}{\ell^3}.
\]

Proof. By (19), (20), (21), and Lemma 16,

\[
\delta' = \frac{2}{3} (\mathbb{P}'[Q_1] - \mathbb{P}'[Q_2])
\]

\[
\geq \frac{2}{3} \left\{ \gamma \wedge \frac{1}{8} \right\} \mathbb{E}' \left[ \frac{1}{\ell^3} \right]
\]

\[
\geq \frac{2}{3} \left\{ \gamma \wedge \frac{1}{8} \right\} \frac{1}{\mathbb{E}'[I]^3}, \tag{22}
\]

where the last line follows from Jensen’s inequality. Moreover

\[
\alpha \geq \mathbb{E}[I]
\]

\[
= \mathbb{E} \left[ I \mid \mathcal{X}_Q \geq \bar{I} \right] \mathbb{P} \left[ \mathcal{X}_Q \geq \bar{I} \right] + \mathbb{E} \left[ I \mid \{ \mathcal{X}_Q \geq \bar{I} \}^c \right] \mathbb{P} \left[ \{ \mathcal{X}_Q \geq \bar{I} \}^c \right]
\]

\[
\geq \mathbb{E}'[I] \sigma.
\]

Plugging back into (22) gives the claim. \( \square \)
4.4 Final analysis

Proof of Theorem 1. The following, adapted from [SRM18, Lemmas A.1 and A.2], gives a bound on the $k^*$ required to reconstruct the correct species tree with probability $1 - \epsilon$ in terms of $\delta'$

$$k^* > 2 \log \left( \frac{n^4}{\epsilon} \right) \frac{1}{(\delta')^2}.$$  

In particular, for $\epsilon < 1$ this inequality holds whenever

$$k^* > 8 \log \left( \frac{n}{\epsilon} \right) \frac{1}{(\delta')^2}.$$  

By Lemmas 15 and 17, it suffices to have

$$k \geq \left\{ \frac{16}{\sigma} \log \left( \frac{n}{\epsilon} \right) \frac{1}{(\frac{2}{3} \{\gamma \wedge \frac{1}{8}\} \frac{\sigma^3}{\alpha^3})^2} \right\} \lor \left\{ \frac{8}{\sigma^2} \log \frac{n}{\epsilon} \right\} \geq \frac{2304\alpha^6}{\sigma^2\gamma^2} \log \frac{n}{\epsilon}.$$  

The claim follows from Lemmas 13 and 14.  

5 Concluding remarks

Through a probabilistic analysis of the DLCoal model, we established identifiability of the model species tree and statistical consistency of quartet-based species tree estimation methods ASTRAL-one and ASTRAL-multi. In our main new result, we derived an upper bound on the required number of gene trees to reconstruct the species tree with high probability. In particular, we highlighted the roles of the branching process parameters $\lambda$ and $\mu$ as well as the tree depth $\Delta$. These parameters enter naturally through two relevant quantities: the minimum survival probability of a quartet ($\sigma$) and the maximum expected number of lineages at a vertex ($\alpha$).

Our results suggest many open problems. First, can we derive a lower bound (and matching upper bound) on the required number of gene trees for ASTRAL-one and similar methods (including ASTRAL-multi)? In particular, is our dependence on $\alpha$ (in the supercritical regime) and $\sigma$ (in the subcritical regime) optimal? An improvement on the polynomial dependence
on $\alpha$ (see Lemma 17) is likely possible with a more detailed analysis of the events in Section 3. But, perhaps more importantly, are there alternative ways of processing multi-labeled gene trees (not necessarily quartet-based) that dampen or even exclude the effect of $\alpha$?

More generally, it would be interesting to obtain statistical consistency and sample complexity results for models also including LGT, under which at low enough rates quartet-based methods have also been shown to be consistent [RS13]. One such more general model was recently introduced in [LGSC20].

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