From gene trees to species trees II: 
Species tree inference in the deep coalescence model

Louxin Zhang
Departments of Mathematics and Biochemistry
National University of Singapore
Singapore 119076
Email: matzlx@nus.edu.sg
Abstract

When gene copies are sampled from various species, the resulting gene tree might disagree with the containing species tree. The primary causes of gene tree and species tree discord include lineage sorting, horizontal gene transfer, and gene duplication and loss. Each of these events yields a different parsimony criterion for inferring the (containing) species tree from gene trees. With lineage sorting, species tree inference is to find the tree minimizing extra gene lineages that had to coexist along species lineages; with gene duplication, it becomes to find the tree minimizing gene duplications and/or losses. In this paper, we show the following results:

(i) The deep coalescence cost is equal to the number of gene losses minus two times the gene duplication cost in the reconciliation of a uniquely leaf labeled gene tree and a species tree. The deep coalescence cost can be computed in linear time for any arbitrary gene tree and species tree.

(ii) The deep coalescence cost is always no less than the gene duplication cost in the reconciliation of an arbitrary gene tree and a species tree.

(iii) Species tree inference by minimizing deep coalescences is NP-hard.

Index terms: Reconciliation of gene tree and species, deep coalescence, gene duplication and loss, parsimony criterion, NP-hardness.

1 Introduction

Gene trees are fundamental to molecular systematics. Traditionally, a gene tree is reconstructed from DNA sequence variation at individual genetic loci in a group of species and is taken as the phylogenetic tree of the species due to sequencing technology limitations. However, when gene copies are sampled from various species, the resulting gene tree might disagree with the species tree. As such, the relationship between gene trees and species trees has been the focus of many studies (see for example [5, 12, 20, 25, 27, 31, 33]). It has long been recognized that gene trees can be used to estimate species divergence time, ancestral population sizes and even the containing species tree although they may not accurately reflect the species tree [7, 15, 21].

The discord of gene trees and the containing species tree can arise from horizontal gene transfer, lineage sorting, and gene duplication and loss. The importance of these causes depends on the considered genes and species. Hence, inferring the species tree from gene trees has been investigated under various parsimony criteria. With lineage sorting (also called deep coalescence), the problem is to find the tree minimizing extra gene lineages that had to coexist along species lineages [20]; with gene duplication, it becomes to find the tree minimizing gene duplications and/or losses [12, 25, 13, 28].

Inferring the species tree from a set of gene trees has often been studied under the gene duplication cost [1, 2, 3, 6, 8, 14, 16, 18, 30, 33] until very recently. In a seminal work [20], Maddison addressed lineage sorting in the framework of coalescence theory. Coalescence theory is an active branch of population genetics concerned with tracing the genealogical
history of a present-day gene copy. For a gene sampled from two individuals, one may ask: How deep in time do these two lineages coalesce? Hence, the depth of this coalescence is a measure of the relationship between two sampled gene copies. The more deep in time coalescence occurs, the more distantly related they are. Maddison proposed to use the total number of “extra” gene lineages that fails to coalesce on a species tree to measure the inconsistence of a gene tree and species tree, called deep coalescence cost. For the gene tree and species tree shown in Figure 1, there are three gene lineages on a branch and two gene lineages on another branch that fail to coalesce, giving the deep coalescence cost of 3. Since coalescence theory provides the probability that a gene tree would exist in a species tree, it allows the inference problem to be studied in explicit statistical framework [4, 29]. This seems to give the deep coalescence model an advantage over the other models.

The paper is a sequel of [19], which studies the complexity and algorithmic issues of inferring the species tree from a set of gene trees with the gene duplication/loss cost. In this work, we present a relationship of the deep coalescence cost, the duplication cost, and the number of gene losses. Although deep coalescence and gene duplication are two different mechanisms responsible for the discord of gene trees and species trees, this relationship suggests that the deep coalescence cost and the duplication cost are closely related to each other as a similarity measure of trees. We further show that inferring species tree from gene trees is also NP-hard by minimizing the deep coalescence cost.

Figure 1: (i) A gene tree. (ii) A species tree. (iii) The reconciliation of the gene tree in (i) into the species tree in (ii) has deep coalescence cost 3.
2 Basic definitions and notations

In this section, we shall introduce basic definitions and notations on gene duplication, gene loss and deep coalescence that are used in the following sections.

2.1 Species trees and gene trees

For a set of \( n \) taxa, their evolutionary history is modeled as a rooted, full binary tree with \( n \) leaves in which leaves are labeled with taxa, representing the labeling taxa, and internal nodes are unlabeled. Here, the 'fullness' means that each internal node has exactly two children. Such a tree is called species tree. In a species tree, each unlabeled internal node is considered as a taxon family which include as its members the subordinate species represented by the leaves below it. Thus, the evolutionary relation "\( m \) is a descendant of \( n \)" is expressed using the set-theoretic notation as "\( m \subset n \)". We also call an internal node an ancestor of the species below it.

The model for gene relationship is also a rooted, full binary tree with leaves representing genes, called a gene tree. Usually, a gene tree is reconstructed from a collection of gene family members sampled from the considered species. We label the gene copies by the species from which they are sampled. Thus, leaf labels may not be unique in a gene tree as two or more gene copies might be found in a species. An internal node is corresponds to a multiset of leaf labels.

Finally, for a species or gene tree \( T \), we use \( L(T) \) to denote the set of leaf labels of it. For an internal node \( t \) in \( T \), \( a(t) \) and \( b(t) \) are used to denote its two children.

2.2 Gene duplication

Let \( G \) be a gene tree and \( S \) a species tree such that \( L(G) \subseteq L(S) \). For any nodes \( s', s'' \) in \( S \), the least common ancestor of \( s' \) and \( s'' \) is defined to be the smallest node \( s \) in \( S \) such that \( s', s'' \subseteq s \), which is denoted by \( \text{lca}(s', s'') \). To reconcile the gene tree \( G \) and the species tree \( S \), each node \( g \) of \( G \) is mapped to a unique node \( M(g) \) in \( S \) as

\[
M(g) = \begin{cases} 
g, & g \in L(G) \\
\text{lca}(M(a(g)), M(b(g))), & g \not\in L(G). 
\end{cases}
\]

This mapping \( M \) was first considered in [12] and then formulated in [25]. We call \( M \) the lca mapping or reconciliation of \( G \) in \( S \). Obviously, if \( g' \subseteq g \), \( M(g') \subseteq M(g) \).

Definition 2.1 Let \( g \) be an internal node of \( G \). If \( M(c(g)) = M(g) \) for some child \( c(g) \) of \( g \), then we say that a duplication occurs at \( M(g) \) (or more exactly in the lineage entering \( M(g) \)) in \( S \).

The total number of duplications arising in the lca reconciliation \( G \) in \( S \) is proposed to measure the discord of the gene tree and species tree and is called the duplication cost. We use \( c_{\text{dup}}(G, S) \) to denote the duplication cost for \( G \) and \( S \). Note that the duplication cost is not symmetric.
2.3 Gene loss

A subset $A$ of (internal and/or leaf) nodes of a species tree $S$ is incompatible if $x \cap y = \phi$ for any $x, y \in A$. For an incompatible subset $A$ in $S$, the restriction of $S$ on $A$ is the smallest subtree of $S$ containing $A$ as its leaf set, denoted by $R_S(A)$. It is easy to see that the root of $R_S(A)$ is the least common ancestor of the nodes from $A$. The homomorphic subtree $S|_A$ of $S$ induced by $A$ is a tree obtained from $R_S(A)$ by contracting all degree-2 nodes except for the root of $R_S(A)$.

Let $G$ be a gene tree such that $L(G) \subseteq L(S)$. $S|_{L(G)}$ is well defined. To reconcile $G$ and $S$ in this general case, we consider the the lca mapping $M$ from $G$ to $S|_{L(G)}$. For any two nodes $s$ and $s'$ of $S|_{L(G)}$ such that $s \subset s'$, we define

$$d(s, s') = |\{ h \in S|_{L(G)} | s \subset h \subset s'\}|.$$

That is, $d(s, s')$ is the number of nodes on the path from $s'$ to $s$.

Recall that $a(g)$ and $b(g)$ denote the children of $g$. The number of losses $l_g$ associated to $g$ is defined as

$$l_g = \begin{cases} 
0, & \text{if } M(g) = M(a(g)) = M(b(g)), \\
\sum_{h=a(g), b(g)} d(M(h), M(g)), & \text{if } M(a(g)) \subset M(g) = M(b(g)), \\
d(M(a(g)), M(g)) + 1, & \text{if } M(a(g)), M(b(g)) \subset M(g). 
\end{cases}$$

This definition of $l_g$ is a generalization of the loss cost given in [13]. When $L(G) = L(S)$, our definition is then identical to the one given in [13].

The gene loss cost in the reconciliation of $G$ in $S$ is defined as the total number of losses $\sum_{g \in G} l_g$. We denoted this gene loss cost for $G$ and $S$ by $c_{loss}(G, S)$.

2.4 Deep coalescence

Let $G$ be a gene tree and $S$ a species tree such that $L(G) = L(S)$. Under the lca mapping $M : G \rightarrow S$, if a branch $e$ of $S$ is on the $k$ paths from $M(g_i)$ to $M(c(g_i))$, $g_i \in G$ ($1 \leq i \leq k$), then we say that there are $k - 1$ ‘extra’ lineages on $e$ failing to coalesce on $e$. The deep coalescence (DC) cost is defined as the total number of the ‘extra’ lineages on all branches of $S$ in the reconciliation $M$ of $G$ in $S$ (see [20]), which is denoted by $c_{dc}(G, S)$. Note that the concept of deep coalescence is meaningful only if $S$ has 2 or not leaves. We assume this throughout the paper.

In general, if $L(G) \subseteq L(S)$, the deep coalescence cost $c_{dc}(G, S)$ is defined as $t_{dc}(G, S|_{L(G)})$, where $S|_{L(G)}$ is the homomorphic subtree of $S$ induced by $L(G)$. Such a generalization will be used in the study of inferring the species tree from a set of gene trees.

3 A equation of the duplication and DC costs

We have seen that deep coalescences, gene losses and duplications are inferred through the gene tree/species tree reconciliation. Actually, they are indeed closely related through a simple
Definition 3.1 Let $G$ be a gene tree and $S$ a species tree such that $L(G) \subseteq L(S)$. Under the lca mapping $M : G \rightarrow S$, an internal node $g \in G$ is of

- type-1 if $M(g') \subset M(g)$ for each child $g'$ of $g$;
- type-2 if there exists a unique child $g'$ such that $M(g') = M(g)$;
- type-3 if $M(g') = M(g)$ for each child $g'$ of $g$.

Note that type-2 or type-3 internal nodes correspond one-to-one with duplication events.

Theorem 3.1 Let $G$ be a uniquely leaf-labeled gene tree and $S$ a species tree such that $L(G) = L(S)$. Then,

$$c_{dc}(G, S) = c_{loss}(G, S) - 2c_{dup}(G, S).$$

Proof. Let $G$ and $S$ have $n$ leaves. Assume that there are $k_1$ type-1 internal nodes $g_{11}, g_{12}, \ldots, g_{1k_1}$, $k_2$ type-2 internal nodes $g_{21}, g_{22}, \ldots, g_{2k_2}$, and $k_3$ type-3 internal nodes $g_{31}, g_{32}, \ldots, g_{3k_3}$ in $G$ under the lca mapping $M : G \rightarrow S$, respectively. Since $G$ is a full binary tree with $n$ leaves, $G$ has $n - 1$ internal nodes and hence

$$k_1 + k_2 + k_3 = n - 1. \tag{1}$$

Additionally, type-2 and type-3 nodes correspond one-to-one with duplication events,

$$c_{dup}(G, S) = k_2 + k_3. \tag{2}$$

For simplicity, we assume that $g'$ and $g''$ are the children of $g$ for each type-1 internal node $g$; we also assume that $a(g)$ is the unique child such that $M(a(g)) \subset M(g)$ for each type-2 node $g$. Since we use $d(M(h), M(g))$ to denote the number of nodes on the path from $M(g)$ to $M(h)$ for a node $g$ and its child $h$, the number of lineages contained in the path is $d(M(h), M(g)) + 1$. Therefore, by Eqn. (1) and (2) and the fact that $|E(S)| = 2n - 2$,

$$c_{dc}(G, S) = \sum_{j=1}^{k_1} \left\{ d(M(g'_{1j}), M(g_{1j})) + 1 \right\} + \sum_{j=1}^{k_2} \left\{ d(M(a_{1j}), M(g_{1j})) + 1 \right\} - |E(S)|$$

$$= c_{loss}(G, S) + 2k_1 - (2n - 2)$$

$$= c_{loss}(G, S) - 2(k_2 + k_3)$$

$$= c_{loss}(G, S) - 2c_{dup}(G, S).$$
This concludes the proof. □

Remarks. (1) Following the proof of the equation in the above theorem, one can easily see that for an arbitrary gene tree $G$ in which there may be two or more gene copies are from the same species and a species tree $S$ such that $L(G) = L(S)$,

$$c_{dc}(G, S) = c_{loss}(G, S) - 2c_{dup}(G, S) + (\text{no.of genes}) - (\text{no.of species}).$$

(2) Since the number of gene duplications and losses can be calculated in linear time \[34, 19\], the first remark implies that the deep coalescence cost can also be computed in linear time.

By Thm 3.1 $c_{dc}(G, S) \leq c_{loss}(G, S)$ for a species tree $S$ and a uniquely leaf labeled gene tree $G$. Now we show that it is bounded below by the duplication cost for any arbitrary gene tree.

**Theorem 3.2** Let $G$ be a uniquely leaf-labeled gene tree and $S$ a species tree such that $L(G) = L(S)$. Then, $c_{dc}(G, S) \geq c_{dup}(G, S)$.

**Proof.** Denote the image node set of the lca mapping $M$ by $M(G)$, which is a subset of nodes in the species tree $S$. For any internal node $s \in M(G)$, we use $M^{-1}(s)$ to denote all internal nodes $g$ of the gene tree that are mapped to $s$ under $M$. For any nodes $x$ and a descendant $y$ of $x$ in the gene tree $G$, if $M(x) = M(y) = s$, then $M(g) = s$ for each node in the path from $x$ to $y$. Since $G$ is uniquely leaf labeled, all internal nodes in $M^{-1}(s)$ form a rooted subtree of $G$, denoted by $T^{-1}(s)$, as illustrated in Figure 2.

$T^{-1}(s)$ is not a full binary tree in general. In particular, its root might has degree 1. Let $n'_s, n''_s, n'''_s$ denote the number of non-root degree-1, degree-2 and degree-3 nodes in the subtree $T^{-1}(s)$, respectively. Assume that $T^{-1}(s)$ has two or more nodes. Then, by definition, the root of $T^{-1}(s)$ corresponds with a gene duplication in the reconciliation of $G$ and $S$; each degree-2 or degree-3 node of $T^{-1}(s)$ also corresponds with a gene duplication. Therefore, there are $n''_s + n'''_s + 1$ duplication events at $s$. We now consider two cases.

Case 1. The root of $T^{-1}(s)$ has degree 1. Then $T^{-1}(s)$ has $n''_s + 1$ leaves, that is $n'_s = n''_s + 1$. For each leaf of $T^{-1}(s)$, it has two children that are mapped to a node below $s$ in the species tree $S$; each non-root degree-2 node has exactly one child that is mapped to a node below $s$ and so is the root since it has degree 1. Thus, there are $2(n''_s + 1) + n''_s + 1$ image paths that contain one of the two lineages from $s$ to one of its children.

Case 2. The root has degree 2. In this case, $T^{-1}(s)$ has $n'''_s + 2$ leaves and there are $2(n'''_s + 2) + n''_s$ image paths that contain one of the two lineages from $s$ to one of its children.
Figure 2: (i) A gene tree. (ii) A species tree. In the lca reconciliation $M$ of the gene tree in the species tree, $a$ is mapped to the green node, $b, c, d, e, f$ and $r$ to the red node, and $g$ to the purple node. The nodes $b, c, d, e, f, r$ form a subtree of the gene tree.

By distributing the DC and duplication costs to each image node $s$ in $M(G)$, we obtain that

$$c_{dc}(G, S) \geq \sum_{s \in M(G) : \mid T^{-1}(s) \mid > 1} (\text{the no. of extra gene lineages on the branches leaving } s)$$

$$\geq \sum_{s \in M(G) : \mid T^{-1}(s) \mid > 1} (2n'''_s + n''_s + 1)$$

$$\geq \sum_{s \in M(G) : \mid T^{-1}(s) \mid > 1} (n'''_s + n''_s + 1)$$

$$= c_{dup}(G, S).$$

This finishes the proof. □

**Remark** The fact $c_{dc}(G, S) \geq c_{dup}(G, S)$ holds even for arbitrary gene trees in which 2 or more leaves with the same label, which represent genes sampled from the same species. In the general case, $T^{-s}$ might be a forest – a union of rooted trees. However, the estimation (4) in the proof is still valid if the sum is over all the subtrees that are mapped to a node in the species tree, i.e. $T^{-s}$ is replaced by a subtree of each resulting forest.

4 The NP-harness of the species tree problem in the DC cost

Parsimony criterion is often used for inference in biology. Hence, inferring species tree from a set of gene trees is formulated as the following algorithmic problem

**Species Tree Problem**

**INPUT:** A set of gene trees $G_i, 1 \leq i \leq n$. 
SOLUTION: A species tree $S$ that minimizes the total cost $\sum_i c(G_i, S)$, where $c(\cdot)$ is a cost function.

It is proved that the species tree problem is NP-hard for the duplication and/or loss cost in [19]. In this section, we prove the following theorem.

**Theorem 4.1** The species tree problem is NP-hard under the DC cost.

**Proof.** Given a gene tree $G$ and a species tree $S$, the DC cost $c_{dc}(G, S)$ can be computed in polynomial time since gene duplications and losses can be counted in linear time [34]. Therefore, the species tree problem is in NP.

To prove its NP-hardness, we reduce the Maximum Cut problem to the decision version of the species tree problem. Given an instance graph $G = (V, E)$ and a positive integer $I$, the Maximum Cut problem is to partition the node set $V$ into two disjoint subsets $V_1$ and $V_2$ such that there are at least $I$ edges from $E$ that have one endpoint in $V_1$ and one endpoint in $V_2$. Assume that $V = \{v_1, v_2, \ldots, v_n\}$ and $|E|$ denotes the number of edges from $E$, where $n > 3$. We construct a corresponding instance of the species tree problem as follows.

![Gene trees](image)

**Figure 3:** Gene trees defined for each edge $e = (v_i, v_j)$.

Choose $N > n^2$ and $M \ge n^2 N(N + 1) + |E|$. For each node $v_i$ ($1 \le i \le n$), we introduce a label with the same name $v_i$. We also introduce $2N + M$ extra labels $x_i, y_i, 1 \le i \le N$ and $z_j, 1 \le j \le M$. For each edge $e = (v_i, v_j) \in E$, we define two gene trees $T_{e1}$ and $T_{e2}$ as shown in Figure 3. These two trees are same except that the leaf labels $v_i$ and $v_j$ are swapped.
Figure 4: ‘Structural’ gene trees.

Let the trees shown in Figure 4 (i)-(iii) be written as $L[x_i, x_j, y_k, z_l]$, $L[y_i, y_j, x_k, z_l]$ and $F[\{x_i\}, \{y_i\}, z_l]$, respectively. Besides the ‘edge’ gene trees $T_{e1}$ and $T_{e2}$ ($e \in E$), the set $\mathcal{A}$ of gene trees in the instance of the problem to be defined also contains

\[
G_{(i,j,k,m)} = L[x_i, x_j, y_k, z_m], \quad 1 \leq i < j \leq N, \quad 1 \leq k \leq N, \quad 1 \leq m \leq M,
\]

\[
G'_{(i,j,k,m)} = L[y_i, y_j, x_k, z_m], \quad 1 \leq i < j \leq N, \quad 1 \leq k \leq N, \quad 1 \leq m \leq M,
\]

\[
G''_m = F[\{x_i\}, \{y_i\}, \{z_m\}], \quad 1 \leq m \leq M.
\]

These three classes of gene trees are introduced to restrict the topology of the optimal species tree for the defined instance of the problem. Hence, we call them ‘structural’ gene trees. The NP-completeness of the decision version of the species tree problem follows from the following two lemmas.

**Lemma 4.1** If the graph $\mathcal{G}$ has a cut of $d$ edges, there is a species tree $S_\mathcal{G}$ having the DC cost

\[
c_{dc}(\mathcal{A}, S_\mathcal{G}) = N(N + 1)|E| + |E| - d.
\]

**Proof.** Assume that the node set $V$ of the graph $\mathcal{G}$ divides into $V_1 = \{v_1, v_2, \ldots, v_p\}$ and $V_2 = \{v_{p+1}, v_{p+2}, \ldots, v_n\}$ such that there are exactly $d$ edges having one endpoint in $V_1$ and one endpoint in $V_2$. We define a species tree $S_\mathcal{G}$ as shown in Figure 5.

First, we observe that

\[
c_{dc}(G_{(i,j,k,m)}, S_\mathcal{G}) = 0, \quad c_{dc}(G'_{(i,j,k,m)}, S_\mathcal{G}) = 0, \quad c_{dc}(G''_m, S_\mathcal{G}) = 0,
\]

for each possible $i, j, k, m$. 

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Consider a non-cut edge $e = (v_i, v_j)$ ($i < j$). Since $L(T_{e1}) = L(T_{e2}) \subset L(S_G)$, $c_{dc}(T_{e1}, S_G) = c_{dc}(T_{e1}, S_G|L(T_{e1}))$ and $c_{dc}(T_{e2}, S_G) = c_{dc}(T_{e2}, S_G|L(T_{e2}))$. If $v_i, v_j \in V_1$, we have that

$$c_{dc}(T_{e1}, S_G) = \frac{1}{2}N(N-1) + N + 1, \quad c_{dc}(T_{e2}, S_G) = \frac{1}{2}N(N-1) + N.$$ 

Symmetrically, if $v_i, v_j \in V_2$, we have that

$$c_{dc}(T_{e1}, S_G) = \frac{1}{2}N(N-1) + N, \quad c_{dc}(T_{e2}, S_G) = \frac{1}{2}N(N-1) + N + 1.$$ 

For each cut edge $e = (v_i, v_j)$ ($i < j$) with one endpoint in $V_1$, say $v_i \in V_1$, and another in $V_2$, we have that

$$c_{dc}(T_{e1}, S_G) = 0, \quad c_{dc}(T_{e2}, S_G) = N(N + 1).$$ 

Therefore, we have

$$c_{dc}(\mathcal{A}, S_G) = N(N + 1)|E| + |E| - d.$$ 

This finishes the proof of the lemma. \Box

**Lemma 4.2** If there is a species tree $S$ having the DC cost $c_{dc}((\mathcal{A}, S) = N(N + 1)|E| + t$, then the graph $G$ has a cut of at least $|E| - t$ edges.

**Proof.** If $t > |E|$, the fact is trivial. Hence, without loss of generality, we may assume that $t \leq |E|$. Here, we use $\text{LTree}[a, \ldots, b, c]$ to denote the line tree with leaves labeled by $a$, $b$, $\ldots$, $c$, respectively, as shown in Figure 6 (i). Note that the leaf $a$ is a child of the root in $\text{LTree}[a, \ldots, b, c]$. For a set of trees $T', T'', \ldots, T'''$, we use $\text{LTree}[T', \ldots, T'', T''']$.
Figure 6: (i) Line tree LTree[a, . . . , b, c] to denote the tree obtained by replacing each leaf by a corresponding subtree in LTree replacing each leaf with a tree in a line tree.

(ii) The resulting tree LTree[T′, . . . , T′′, T′′′] after replacing each leaf with a tree in a line tree.

to denote the tree obtained by replacing each leaf by a corresponding subtree in LTree[a, . . . , b, c] as shown in Figure 6 (ii).

Let B be a subset of leaves in the species tree S and the least common ancestor of the leaves from B be r_B in S. Recall that the homomorphic subtree S|_B of S induced by B is the tree obtained from S by removing all the nodes and edges that are not on a path from r_B to a leaf from B and then contracting all the degree-2 node except for the root r_B. For example, for S_G defined in Lemma 4.1, S_G{(x_1, x_2, y_1)} = LTree[y_1, x_1, x_2].

Set

\[ U = \{x_1, x_2, \ldots, x_N\} \cup \{y_1, y_2, \ldots, y_N\} \cup \{v_1, v_2, \ldots, v_n\}; \]
\[ Z = \{z_1, z_2, \ldots, z_M\}. \] (6)

By replacing the children of a two-leaf root tree with S|_U and S|_Z, we obtain a species tree S′ = LTree[S|_U, S|_Z] from S. First, S′ has the following property.

**Fact 1** \( c_{dc}(A, S') \leq c_{dc}(A, S) = N(N + 1)|E| + t. \)

**Proof.** For each gene tree T = T_e1 or T_e2, we use \( f \) and \( f' \) to denote the lca mappings from T to S and S′, respectively. For each edge \( e = (u_1, u_2) \) in the spanning subtree over z_i's in T, by the definition of S|_Z, \( f(u_1) = f(u_2) \) if and only if \( f'(u_1) = f'(u_2) \) and \( d(f'(u_1), f'(u_2)) \leq d(f(u_1), f(u_2)) \). For each edge in the spanning subtree over x_i's, v_i, v_j and y_i's, the same property holds. But, the edges incident to the root of T may not satisfy the property discussed above. Let r be the root of T. Assume that \( a(r) \) is the left child of r, which is the least common anceder of x_i's and y_i's, and \( b(r) \) the right child of r. It is possible that \( f(r) = f(a(r)) \) and/or \( f(r) = f(b(r)) \). However, \( f'(r) = r', f'(a(r)) = a(r') \) and \( f'(b(r)) = b(r') \), where \( r' \) is the root of S′, a(r') and b(r') the root of S|_U and S|_Z respectively. Since no other lineages fail to coalesce with \( (r, a(r)) \) on \( (r', a(r')) \) and with \( (r, b(r)) \) on \( (r', b(r')) \) respectively, these two edges does not affect the deep coalescence cost. Thus, \( c_{dc}(T, S') \leq c_{dc}(T, S) \).
Similarly, we also have the following three inequalities

\[ c_{dc}(G(i, j, k, m), S') \leq c_{dc}(G(i, j, k, m), S) \]
\[ c_{dc}(G'(i, j, k, m), S') \leq c_{dc}(G'(i, j, k, m), S) \]
\[ c_{dc}(G''(m), S') \leq c_{dc}(G''(m), S) \]

for any \( i, j, k, m \). Thus, the fact holds. \( \square \)

**Fact 2.** In \( S|_U \), all the leaves \( x_i \) must be below one child of the root and all the leaves \( y_i \) must be below the other child of the root. In other words, \( S|_U = LTree[T_1, T_2] \), where \( T_1 \) is a tree over \( x_i \) and some \( v_i \)'s and \( T_2 \) is a tree over \( y_i \)'s and some \( v_j \)'s.

**Proof.** Assume that the fact is false. There are \( x_i, x_j \) and \( y_k \) such that \( S|_{\{x_i, x_j, y_k\}} = (S|_U)|_{\{x_i, x_j, y_k\}} = LTree[x_i, x_j, y_k] \), or there are \( y_i, y_j \) and \( x_k \) such that \( S|_{\{y_i, y_j, x_k\}} = (S|_U)|_{\{y_i, y_j, x_k\}} = LTree[y_i, y_j, x_k] \). If the former is true, then,

\[ c_{dc}(G(i,j,k,m), S') \geq 1, \quad 1 \leq m \leq M. \]

This implies that

\[ N(N+1)|E| + t \geq c_{dc}(A, S') \geq \sum_{m=1}^{M} c_{dc}(G(i,j,k,m), S') = M, \]

contradicting to the fact that \( M \geq N(N+1)n^2 \). If the latter is true, for any \( 1 \leq m \leq M \), \( c_{dc}(G'(i,j,k,m), S') \geq 1 \). Again, we have that \( c_{dc}(A, S') \geq M \), leading to a contradiction. \( \square \)

Let \( X = \{x_1, x_2, \ldots, x_N\} \) and \( Y = \{y_1, y_2, \ldots, y_N\} \). Then \( S'|_X = (S|_U)|_X \) and \( S'|_Y = (S|_U)|_Y \).

**Fact 3.** \( S'|_X = LTree[x_1, x_2, \ldots, x_N] \) and \( S'|_Y = LTree[y_1, y_2, \ldots, y_N] \).

**Proof.** Note that \( G'_m|_X = LTree[x_1, x_2, \ldots, x_N] \) and \( G'_m|_Y = LTree[y_1, y_2, \ldots, y_N] \) for any \( 1 \leq m \leq M \). If the claim is false, then, \( c_{dc}(G''(m), S') \geq 1 \) for any \( m \) and hence

\[ N(N+1)|E| + t \geq c_{dc}(A, S') \geq \sum_{m=1}^{M} c_{dc}(G''(m), S') = M, \]

a contradiction as in the proof of Fact 2. \( \square \)

Let the least common ancestor of \( x_i \)'s and \( y_i \)'s be \( r \) in \( S' \). We have shown that \( x_i \)'s are below one child of \( r \), say \( r_1 \), and \( y_i \)'s are below the other child of \( r \), say \( r_2 \). In addition, \( S'|_X \) and \( S'|_Y \) are two line trees.

**Fact 4.** For each edge \( e = (v_i, v_j) \) (\( i < j \)) such that \( v_i \) and \( v_j \) are in the same subtree as \( x_i \)'s or as \( y_i \)'s, then

\[ c_{dc}(T_{e1}, S') + c_{dc}(T_{e2}, S') \geq N(N+1) + 1. \]
Proof. Without loss of generality, we may assume that $v_i$ and $v_j$ are below $r_1$ in the same subtree as $x_i$s. We consider the following cases.

Case 1. $S_{X \cup \{v_i, v_j\}} = \text{LTree}[x_1, x_2, \ldots, x_k, v_i, x_{k+1}, \ldots, x_m, v_j, v_m+1, \ldots, x_N]$ for some $0 \leq k \leq m \leq N$. In this case, we have that

$$c_{dc}(T_{e1}, S') = \frac{1}{2}N(N-1) + N + 1 + \frac{1}{2}(N-k)(N-k-1)$$

and

$$c_{dc}(T_{e2}, S') = \frac{1}{2}N(N-1) + k + 1 + \frac{1}{2}(N-m)(N-m-1).$$

Hence,

$$c_{dc}(T_{e1}, S') + c_{dc}(T_{e2}, S')$$

$$\geq N(N-1) + 1 + \frac{1}{2}[(N-k)(N-k-1) + 2k + 2]$$

$$\geq N(N+1) + 1$$

as the minimum value of $(N-k)(N-k-1) + 2k + 2$ is $N$ (reaching at $k = N-2, N-1$).

Case 2. $S_{X \cup \{v_i, v_j\}} = \text{LTree}[x_1, x_2, \ldots, x_k, \text{LTree}[v_i, v_j], x_{k+1}, \ldots, x_{N-1}, x_N]$ for some $0 \leq k \leq N$. We have that

$$c_{dc}(T_{e1}, S') + c_{dc}(T_{e2}, S') = \frac{1}{2}N(N-1) + k + 2 + \frac{1}{2}(N-k)(N-k-1)$$

and so

$$c_{dc}(T_{e1}, S') + c_{dc}(T_{e2}, S')$$

$$\geq N(N-1) + 2k + 4 + (N-k)(N-k-1)$$

$$\geq N(N+1) + 2$$

as the minimum value of $2k + (N-k)(N-k-1)$ is $2N-2$ (reaching at $k = N-1, N-2$). The fact is proved. $\square$

Fact 5. For each edge $e = (v_i, v_j)$ such that $v_i$ is below $r_1$ in the same subtree as $x_i$ and $v_j$ is below $r_2$ in the subtree as $y_i$s. Then,

$$c_{dc}(T_{e1}, S') + c_{dc}(T_{e2}, S') \geq N(N+1).$$

Proof. Let

$$S_{X \cup \{v_i\}} = \text{LTree}[x_1, x_2, \ldots, x_k, v_i, x_{k+1}, \ldots, x_{N-1}, x_N]$$

and

$$S_{Y \cup \{v_j\}} = \text{LTree}[y_1, y_2, \ldots, y_m, v_j, y_{m+1}, \ldots, y_{N-1}, y_N].$$
We have that all the internal nodes in $T_{e2}$ are mapped onto the least common ancestor $r$ of $x_is$ and $y_js$ and thus

$$c_{dc}(T_{e2}, S') = N(N + 1).$$

Since $c_{dc}(T_{e1}, S') \geq 0$, the fact is proved. □

Let $V_1$ denote the subset of leaves $v_i$ below $r_1$ in the same subtree as $x_is$ and $V_2$ the subset of leaves $v_j$ below $r_2$ in the same subtree as $y_js$. Then $(V_1, V_2)$ is a cut of the graph $G$. Assume there are $p$ cut edges. Since there are $|E| - p$ non-cut edges,

$$N(N + 1)|E| + t = c_{dc}(A, S') \geq (|E| - p)N(N + 1) + pN(N + 1) + (|E| - p)$$

$$= N(N + 1)|E| + |E| - p,$$

which implies that $p \geq |E| - t$. This finishes the proof of Lemma 4.2. □

5 Conclusion

We conclude this paper by posing two related research problems. In this paper, we have proved that species tree inference by minimizing deep coalescences is NP-hard. This justifies the effort from different groups in seeking efficient heuristic methods for the inference problem [21, 32]. We have also discussed the relationship of the deep coalescence cost and the gene duplication cost. Is there any polynomial-time algorithm with constant approximation ratio for the species tree problem in the deep coalescence model? Note that the heuristic method developed by Than and Nakhleh in [32] seems to be effective.

In [9], Stege studied the parametric complexity of the species tree inference by minimizing gene duplications. Is it possible to develop efficient algorithm for parametric species tree inference under the deep coalescence model?

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