Phylogenetic invariants for stationary base composition

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Abstract

Changing base composition during the evolution of biological sequences can mislead some of the phylogenetic inference techniques in current use. However, detecting whether such a process has occurred may be difficult, since convergent evolution may lead to similar base frequencies emerging from different lineages.

To study this situation, algebraic models of biological sequence evolution are introduced in which the base composition is fixed throughout evolution. Basic properties of the associated algebraic varieties are investigated, including the construction of some phylogenetic invariants.

\textit{Key words:} phylogenetic invariants, evolution, algebraic statistics

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1 Introduction

Models of molecular evolution, such as for DNA sequences, typically assume evolution occurs along a bifurcating tree, proceeding from a root representing the common ancestral sequence, toward the leaves representing the descendant sequences. At each site in the sequence, bases mutate according to a

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probabilistic process that depends upon the edge of the tree. Usually only the
sequences at the leaves of the tree can be observed, while sequences at internal
nodes correspond to hidden variables in this graphical model. A fundamental
problem of sequence-based phylogenetics is to infer the tree topology from
observed sequences, assuming some reasonable model.

In the works of Cavendar and Felsenstein [CF87] and Lake [Lak87], the con-
nections between this problem and algebraic geometry first emerged in the
phylogenetics literature. Under many standard models of molecular evolution,
for a fixed tree topology the joint distribution of bases in the leaf sequences
are described by polynomial equations in the parameters of the model, thus
parameterizing a variety associated to the tree. The defining polynomials of
this variety, called phylogenetic invariants, are polynomials that vanish on any
joint distribution arising from the tree and model, regardless of parameter val-
ues. Finding phylogenetic invariants for various models has been of interest
both for providing theoretical understanding, and in hopes that methods of
phylogenetic inference that do not require parameter estimation may be de-
veloped. See [Fel03].

For certain models, much progress has been made on determining invariants.
Key advances for group-based models such as the Kimura three-parameter,
were made in [ES93] and [SSEW93], which built on the Hadamard conjuga-
tion introduced in [HP93]. Recently, Sturmfels and Sullivant [SS04] further
exploited the Hadamard conjugation to recognize these varieties were toric,
completing the determination of all invariants in this case. For the general
Markov model, Allman and Rhodes [AR03], found new constructions of in-
variants, though the complete determination of the ideal is still open.

In this paper, we consider models that lie between group-based models and the
general Markov model. Specifically, we assume that a fixed vector describes
the relative frequencies of the bases in sequences at every node of the tree, so
that the base composition of sequences remains stable throughout evolution.

Our motivation for this assumption is a biological one. Many of the models
currently assumed in performing inference with real data make an assump-
tion of a stable base composition (e.g., all group-based models, the general
time-reversible model). However, there are data sets in which base composition
seems to have changed during evolution, as reflected in comparisons of
the sequences at the leaves. Although the extent to which this issue is prob-
lematic in real data sets is controversial, a number of authors have pointed
out that changing base composition may mislead some methods of inference,
especially if it results in convergent mutations in different parts of the tree.
See [LAHP94,CL01,RK03] and their references.

In [KG01] a ‘disparity index’ was introduced as a simple statistical test that
might indicate inhomogeneity of the mutation process along the different edges of the tree. This index is based on a pair-wise comparison of base compositions of sequences at the leaves. It is, however, possible that all leaf sequences have the same base composition, while an internal node sequence has a different one. Indeed, this is exactly the issue with convergent mutations; base composition may appear to be the same in observed sequences, yet it differed in the common ancestral sequence. If a model is chosen only through comparing the base compositions of sequences at the leaves, it may be an inappropriate one.

Better understanding the constraints placed on the joint distribution of bases in sequences from various taxa by an assumption of a stable base distribution is therefore desirable. To begin investigating this issue, in section 2 we introduce three models of molecular evolution that include an assumption of stable base distribution. When the number of bases in the model is $\kappa = 2$, the three models are the same, and its structure allows us to give a more in-depth analysis than for general $\kappa$. This is the subject of section 3, where we give a rational map inverting the parameterization, and find the full ideal of phylogenetic invariants for the 3-taxon tree. In section 4, the case of general $\kappa$ is considered. Basic facts about the associated phylogenetic varieties, such as their dimension and irreducibility/non-irreducibility, are investigated. Although our knowledge of phylogenetic invariants is incomplete, we give constructions of some for these models.

2 The Models

Let $T$ denote an undirected bifurcating tree, with $n$ leaves labeled by the taxa $a_1, a_2, \ldots, a_n$. If $r$ is some vertex in $T$, either internal or terminal, we use $T_r$ to denote the tree rooted at $r$. We view $T_r$ as a directed graph, with all edges directed away from $r$ forming a set $\text{Edge}(T_r)$. Thus $T_r$ represents a hypothetical evolutionary history of the taxa in their descent from a common ancestor at $r$. For simplicity, we refer to $T_r$ as a rooted $n$-taxon tree.

We model the evolution along $T_r$ of sequences composed from an alphabet $[\kappa] = \{1, 2, \ldots, \kappa\}$ of bases or states (e.g., $\kappa = 4$ for DNA). A root distribution vector $p_r = (p_1, p_2, \ldots, p_\kappa)$, with $p_i \in [0, 1]$, $\sum_i p_i = 1$, describes the frequency of bases in an ancestral sequence. To each $e \in \text{Edge}(T_r)$ we associate a $\kappa \times \kappa$ Markov matrix $M_e$ (with entries in $[0, 1]$, each row summing to 1) whose $(i, j)$-entry specifies the conditional probability of base $i$ at the initial vertex mutating to base $j$ at the final vertex of the edge. Together $p_r$ and $\{M_e\}_{e \in \text{Edge}(T_r)}$ comprise the parameters of the model. If no additional requirements are placed on $p_r$ or the $M_e$, then we have described the general Markov model (GM) of sequence evolution, studied in [AR03].
Letting $X_{GM,K,T_r}$ denote the parameter space for the $\kappa$-state GM model on $T_r$, we can view $X_{GM,K,T_r}$ as a subset of $[0,1]^M$ for $M = K + \kappa^2 E$ with $E = 2n - 3$, the number of edges of $T$. We have a map

$$\phi = \phi_{GM,K,T_r} : X_{GM,K,T_r} \to [0,1]^\kappa \subset \mathbb{C}^\kappa,$$

so that $\phi(x)$ gives the joint distribution of bases in aligned sequences at the leaves arising from the parameter choice $x$. Specifically, $\phi(x) = P = (p_{j_1j_2...j_n})$, a $\kappa \times \cdots \times \kappa$ tensor with entries

$$p_{j_1j_2...j_n} = \sum_{i \in I(j_1,j_2,...,j_n)} p_{i_r} \prod_{e \in \text{Edge}(T_r), e=(v \to w)} M_e(i_v,i_w),$$

where $I(j_1,j_2,...,j_n) = \{(i_v) \mid v \in Vert(T_r), i_v \in [\kappa], i_{a_k} = j_k \} \subset [\kappa]^{2n-2}$. Note $\phi$ is a polynomial map, viewed as a function of the entries of $p_r$ and $M_e$, and extends to a polynomial map $\mathbb{C}^M \to \mathbb{C}^\kappa$ which we also denote by $\phi$.

In this paper we are interested in submodels of the GM model, in which we have stable base frequencies at all vertices in the tree. We introduce three such models, defined by imposing additional restrictions on parameters of the GM model. After formally defining the models, we will motivate their assumptions and names.

- **Stable Base Distribution Model (SBD):** 1) $p_r$ has no zero entries, and 2) $p_r$ is fixed by all $M_e$; that is, $p_r M_e = p_r$ for all edges $e$.
- **Simultaneous Diagonalization Model (SD):** In addition to the assumptions of SBD, 3) with $D_r = \text{diag}(p_r)$, all matrices in

$$\{M_e \mid e \in \text{Edge}(T_r)\} \cup \{D_r^{-1}M_e^T D_r \mid e \in \text{Edge}(T_r)\}$$

commute with one another.
- **Algebraic Time Reversible Model (ATR):** In addition to the assumptions of SD, 4) for all edges $e$, $M_e = D_r^{-1}M_e^T D_r$.

We will also need:

**Definition 1** For any model $M$ formed from the $\kappa$-state GM model by imposing additional assumptions on the parameters, and for any rooted $n$-taxon tree $T_r$, we let $X_{M,K,T_r}$ denote the parameter space of $M$ on $T_r$. Then the algebraic variety $V(M,K,T_r)$ is the Zariski closure in $\mathbb{C}^\kappa$ of $\phi(X_{M,K,T_r})$.

We now expand upon the model definitions. First, assuming $p_r$ has no zero entries, the matrices $D_r^{-1}M_e^T D_r$ are also Markov matrices fixing $p_r$. They arise naturally as follows: Consider a 2-taxon tree consisting of a single edge $e$ from vertex $r$ to vertex $s$, with model parameters $p_r$ and $M_e$. Then the joint distribution of bases in aligned sequences at $r$ and $s$ arising from these
parameter choices is given by the entries in $D_r M_e$. Assuming $M_e$ fixes $p_r$, so that $p_r$ is also the base distribution of a sequence at $s$, then the identity

$$D_r M_e = (D_r (D_r^{-1} M_e^T D_r))^T$$

shows that the model parameters $p_r, M_e$ on the 1-edge tree rooted at $r$ lead to the same joint distribution as the model parameters $p_r, D_r^{-1} M_e^T D_r$ on the 1-edge tree rooted at $s$. More generally, as shown in [SSH94, AR03] for the GM model, parameters for the SBD, SD, or ATR model on $T_r$ produce the same joint distribution as the set of parameters on $T_s$ for any other vertex $s$, simply by defining $p_s = p_r$, and for those edges whose directions have reversed in changing the root location, replacing $M_e$ by $D_r^{-1} M_e^T D_r$. In particular, we see

**Proposition 2** $\phi(X_{M,\kappa,T_r})$ and $V(M,\kappa,T_r)$ are independent of the choice of $r$ for $\mathcal{M} = \text{SBD}, \text{SD}, \text{and ATR}$. Thus, for these models, $V(M,\kappa,T)$ is well-defined without reference to $r$.

Second, the requirement for the SD and ATR model that the specified collection of matrices commute is, in fact, equivalent to an assumption that those matrices are simultaneously diagonalizable. To see this, first note that the commutation assumption is equivalent to the commutation of the collection

$$\{D_r^{1/2} M_e D_r^{-1/2} \mid e \in Edge(T_r)\} \cup \{D_r^{-1/2} M_e^T D_r^{1/2} \mid e \in Edge(T_r)\}.$$ 

But this implies in particular that each matrix $D_r^{1/2} M_e D_r^{-1/2}$ is normal, and hence diagonalizable. Commutativity then implies the existence of simultaneous eigenvectors for this collection, and hence for the original collection. Conversely, if the matrices are simultaneously diagonalizable, they certainly commute.

Third, the ATR model is related to the general time-reversible model (GTR) often used in phylogenetic studies. The GTR assumes that for each edge $e$, $M_e = \exp(R t_e)$, where $t_e$ is a scalar parameter and $R$ is a rate matrix (with rows summing to 0) common to all edges with the properties that $D_r R$ is symmetric and $p_r R = 0$ ([Fel03]). A collection of Markov matrices arising from GTR parameters thus satisfies the hypotheses of the ATR model. However, the common rate matrix assumption of the GTR imposes a relationship among the logarithms of the eigenvalues of the Markov matrices $M_e$ which the ATR does not, and thus the ATR is more amenable to algebraic analysis.

Finally, we note that the group-based models, such as the Kimura 3-parameter (KST), can be viewed as the ATR together with additional assumptions on the eigenvectors of the $M_e$. For instance, KST requires the eigenvectors be the columns of a $4 \times 4$ Hadamard matrix, with $(1, 1, 1, 1)$ the stable base distribution.

We summarize the relationships of the various algebraic models with the in-
Group-based $\subseteq$ ATR $\subseteq$ SD $\subseteq$ SBD $\subseteq$ GM.

For $\kappa \geq 3$, these inclusions are all strict, though for $\kappa = 2$ the three central models are identical, as shown below. Of course, the associated varieties are related by a reversed chain of inclusions.

### 3 The 2-base model

For $\kappa = 2$, the SBD, SD, and ATR models are all the same. To see this, and fix notation for future use, consider the SBD model, with root distribution vector $\mathbf{p}_r = (p, 1-p) = (p, q)$. Since each matrix $M_e$ has left eigenvector $\mathbf{p}_r$ and right eigenvector $(1, 1)$, both with eigenvalue 1, we readily find we can express

$$M_e = M(m_e) = \begin{pmatrix} 1 - m_e q & m_e q \\ m_e p & 1 - m_e p \end{pmatrix},$$

thus associating a single scalar parameter $m_e$ to each edge. We also see that $M_e$ satisfies the hypotheses of the ATR model as well. (In fact, for $\kappa = 2$, the ATR model and the GTR model also coincide.) The form of $M_e$ allows us to identify parameters for an $n$-taxon tree with a point $(p; \{m_e\}) \in \mathbb{R}^{2n-2}$.

We first consider a 3-taxon tree $T_r$, rooted at its central node, with three edges $e_1$, $e_2$, and $e_3$ leading from $r$ to leaves $a_1$, $a_2$, and $a_3$. Labeling the states 0 and 1, and using these as indices to refer to matrix entries corresponding to the states, the joint distribution of bases at a site in sequences at the leaves is now described by a $2 \times 2 \times 2$ tensor $P = (p_{ijk})$, where

$$p_{ijk} = pM_1(0, i)M_2(0, j)M_3(0, k) + qM_1(1, i)M_2(1, j)M_3(1, k).$$

Viewing $P$ as a polynomial function of $p, m_1, m_2, m_3$, we thus have a map $\varphi : \mathbb{C}^4 \rightarrow \mathbb{C}^8$, and readily see that the Zariski closure of the image of $\varphi$ is $V(SBD, \kappa, T)$.

For notational ease, we follow the convention that replacing an index by the symbol ‘+’ indicates marginalization over that index. For instance, $p_{ij+} = \sum_k p_{ijk}$, while $p_{i++} = \sum_{j,k} p_{ijk}$.

**Proposition 3** The following rational map provides an explicit inverse to the
parameterization of the map \( \varphi \):

\[
p = p_{0++}, \\
m_i = 1 - \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk} p(1-i)++p(1-j)++p(1-k)++}{(p_{1++} - p_{0++})d_i}, \quad i = 1, 2, 3,
\]

where \( d_1 = \det(p_{+ij}), \ d_2 = \det(p_{i+j}), \) and \( d_3 = \det(p_{ij+}) \).

**PROOF.** Define a \( 2 \times 2 \times 2 \) diagonal tensor \( D \) with \( D(0,0,0) = p, \ D(1,1,1) = q, \) and all other entries zero. We then have

\[
p_{ijk} = \sum_{l,m,n=0}^1 \sum_{i,j,k} D(l,m,n)M_1(l,i)M_2(m,j)M_3(n,k), \tag{1}
\]

expressing \( P \) as the result of an action of an element of \( GL_2 \times GL_2 \times GL_2 \) on \( D \). Also observe that each matrix \( M_i \) has as right eigenvectors \((1,1)\) and \((-q,p)\), with eigenvalues 1 and \( 1 - m_i \), respectively. Thus multiplying the tensor \( P \), whose entries are polynomials in \( p, m_1, m_2, \) and \( m_3 \), by the vector \( v = (v_0, v_1) = (-q, p) \) along each of its indices, yields

\[
g_0 = \sum_{i,j,k=0}^1 p_{ijk} v_i v_j v_k \tag{2}
\]

\[
= \sum_{i,j,k=0}^1 \sum_{l,m,n=0}^1 D(l,m,n)M_1(l,i)v_i M_2(m,j)v_j M_3(n,k)v_k.
\]

Interchanging summations, and using that \( v \) is an eigenvector of each of the \( M_i \) yields

\[
g_0 = \sum_{l,m,n=0}^1 (1 - m_1)(1 - m_2)(1 - m_3)D(l,m,n)v_l v_m v_n
\]

\[
= (1 - m_1)(1 - m_2)(1 - m_3)pq(p - q). \tag{3}
\]

Multiplying similarly, with two copies of \( v \) and one of \((1,1)\) yields

\[
g_1 = \sum_{i,j,k=0}^1 p_{ijk} v_j v_k = (1 - m_2)(1 - m_3)pq,
\]

\[
g_2 = \sum_{i,j,k=0}^1 p_{ijk} v_i v_k = (1 - m_1)(1 - m_3)pq, \tag{4}
\]

\[
g_3 = \sum_{i,j,k=0}^1 p_{ijk} v_j v_i = (1 - m_1)(1 - m_2)pq.
\]
Now since \( p = p_{0++} \), if we express the entries of \( \mathbf{v} \) as linear polynomials in the \( p_{ijk} \), we may view the \( g_i \) as polynomials in the \( p_{ijk} \) as well. Then from equations (2) and (4) we see that \( g_0 \) is of degree 4, while \( g_1, g_2 \), and \( g_3 \) are each of degree 3.

A calculation shows all four of these polynomials have a factor of \( s = \sum_{i,j,k=0}^1 p_{ijk} \), which of course evaluates to 1 on \( V \), so we may replace each \( g_i \) with \( \tilde{g}_i = g_i / s \), if desired. We also note that explicit expressions for the quadratic \( \tilde{g}_i \) as ordinary matrix determinants can be given:

\[
\tilde{g}_1 = \det(p_{+ij}), \quad \tilde{g}_2 = \det(p_{i+j}), \quad \tilde{g}_3 = \det(p_{ij+}).
\]

Equations (3) and (4) now lead directly to formulas for the \( m_i \),

\[
m_i = 1 - \frac{g_0}{(v_0 + v_1)\tilde{g}_i}, \text{ for } i = 1, 2, 3,
\]

which yield the stated map. \( \square \)

The explicit invertibility of the parameterization map for the 3-taxon tree readily extends to \( n \)-taxon trees.

**Theorem 4** Suppose \( T_r \) is a rooted \( n \)-taxon tree with \( (p; \{m_e\}_{e \in \text{Edge}(T_r)}) \in \mathbb{C}^{2n-2} \) defining \( \mathbf{p}_r = (p, 1-p) \), \( M_e = M(m_e) \) and

\[
P = (p_{i1i2...in}) = \phi(p_r; \{M_e\}_{e \in \text{Edge}(T_r)}).
\]

Then the polynomial map \( \phi : (p; \{m_e\}_{e \in \text{Edge}(T_r)}) \mapsto P \) is inverted by a rational map explicitly given by the formulas:

1) \( p = p_{0++...+} \).

2) For each terminal edge \( e_0 \), assume without loss of generality that \( e_0 = (v \rightarrow a_1) \). Choose two other taxa \( a_2, a_3 \) such that the path from \( a_2 \) to \( a_3 \) in \( T \) passes through \( v \). Then

\[
m_{e_0} = 1 - \frac{\sum_{ijk}(-1)^{i+j+k} p_ijk + ... + p(1-i)...+p(1-j)...+p(1-k)...}{(p_{1+...+} - p_{0+...+}) \det(p_{+ij...+})}.
\]

3) For each internal edge \( e_0 = (v \rightarrow w) \), chose four taxa which, without loss of generality, we assume are \( a_1, a_2, a_3, a_4 \), such that the path joining \( a_1 \) to \( a_2 \) in \( T \) passes through \( v \), but not through \( w \); and the path joining \( a_3 \) to \( a_4 \) passes through \( w \), but not \( v \). Then

\[
m_{e_0} = 1 - \frac{(\sum_{ijk}(-1)^{i+j+k} p_{ijk} + ... + p(1-i)...+p(1-j)...+p(1-k)... \det(p_{+i+j...+})}{(\sum_{ijk}(-1)^{i+j+k} p_{ijk} + ... + p(1-i)...+p(1-j)...+p(1-k)... \det(p_{ij...+})}.
\]
Likewise, considering the 3-dimensional tensor \((p_{ijk+\ldots+}) = \phi(p; M_1, M_2, M_3)\) for a 3-taxon tree \(T'_v\), where \(M_i = \prod_{e \in \text{Path}(v,a_i)} M_e\) with \(\text{Path}(v,a_i)\) the set of edges in the path joining \(v\) to \(a_i\). In particular, \(M_1 = M_{e_0}\), so applying the formula of Proposition 3 for \(m_1\) yields the desired formula.

Similarly, for an internal edge \(e_0\) as described above, start with the 3-dimensional tensor \((p_{ijk+\ldots+}) = \phi_T(p; M_1, M_2, M_3)\) for the 3-taxon tree \(T'_v\). Then, since \(M(m)M(m') = M(m'')\) is equivalent to \((1 - m)(1 - m') = 1 - m''\), by applying the formula of Proposition 3, we find

\[
(1 - m_e) = \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk+\ldots+} \prod_{i} p_{(1-i)+\ldots+} p_{(1-j)+\ldots+} p_{(1-k)+\ldots+}}{(p_{1+\ldots+} - p_{0+\ldots+}) \det(p_{ij+k+\ldots+})}.
\]

Likewise, considering the 3-dimensional tensor \((p_{ijk+\ldots+})\), we find

\[
(1 - m_e) = \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk+\ldots+} \prod_{i} p_{(1-i)+\ldots+} p_{(1-j)+\ldots+} p_{(1-k)+\ldots+}}{(p_{1+\ldots+} - p_{0+\ldots+}) \det(p_{ij+k+\ldots+})}.
\]

Since \(\prod_{e \in \text{Path}(v,a_3)} (1 - m_e) = (1 - m_{e_0}) \prod_{e \in \text{Path}(w,a_3)} (1 - m_e)\), this yields the given formula. □

Now, to determine phylogenetic invariants for the SBD model with \(\kappa = 2\), we first consider the 3-taxon tree \(T\). We seek all polynomials in the \(p_{ijk}\) that vanish on \(\varphi(\mathbb{C}^4)\), and thus define \(V(SBD, 2, T)\).

As we are considering a submodel of GM, we obtain the stochastic invariant, which defines \(V(GM, 2, T)\):

\[
f_0 = 1 - p_{+++}.
\]

Several other invariants for the stable base composition model are easily found. The distribution of bases in a sequence at \(a_i\) is given by the vector \(p_i\) where

\[
p_1 = (p_{i+}), \quad p_2 = (p_{i+i}), \quad p_3 = (p_{+++}).
\]

Since each leaf sequence must have the same base composition for SBD, we set \(p_1 - p_2 = p_1 - p_3 = 0\), obtaining two linear invariants

\[
f_1 = p_{010} + p_{011} - p_{100} - p_{101}, \quad f_2 = p_{001} + p_{011} - p_{100} - p_{110}.
\]
whose span includes that arising from $p_2 - p_3 = 0$. These are the invariants underlying the disparity index of [KG01].

From equations (3) and (4) we can also see that

$$h = \tilde{g}_0^2v_0v_1 + \tilde{g}_1\tilde{g}_2\tilde{g}_3(v_0 + v_1)^2$$

(5)

is an invariant of degree 8. However, while $h \not\in (f_0, f_1, f_2)$, the ideal $(f_0, f_1, f_2, h)$ is not the full ideal of invariants. Using Macaulay 2 [GS] to find the kernel of the ring map associated to $\varphi$ quickly yields a single invariant of degree 6 with 258 terms, which together with $f_0$, $f_1$, and $f_2$ generates the full ideal defining $V(SBD, 2, T)$.

In fact, this invariant can be explained through the hyperdeterminants of [GKZ94]. For a $2 \times 2 \times 2$ tensor such as $P$, the hyperdeterminant is given explicitly as

$$\text{Det}(P) = (p_{000}p_{111} + p_{001}p_{110} + p_{010}p_{101} + p_{011}p_{100})$$

$$- 2(p_{000}p_{011}p_{110} + p_{000}p_{010}p_{101} + p_{000}p_{011}p_{100} + p_{000}p_{011}p_{111} + p_{001}p_{010}p_{111} + p_{001}p_{011}p_{100} + p_{010}p_{011}p_{110} + p_{010}p_{011}p_{100})$$

$$+ 4(p_{000}p_{011}p_{111} + p_{001}p_{010}p_{111} + p_{010}p_{011}p_{110} + p_{010}p_{011}p_{100}).$$

Now reasoning from equation (1) and using the invariance properties of $\text{Det}(P)$ under the $SL_2 \times SL_2 \times SL_2$ action, one finds that in terms of model parameters,

$$\text{Det}(P) = p^2q^2(1 - m_1)^2(1 - m_2)^2(1 - m_3)^2.$$

Thus $\tilde{g}_1\tilde{g}_2\tilde{g}_3 - pq \text{Det}(P) = 0$, and so $\tilde{g}_1\tilde{g}_2\tilde{g}_3 + v_1v_0 \text{Det}(P)$, viewed as a degree 6 polynomial in the $p_{ijk}$, is an invariant. Expressing this explicitly in terms of the $p_{ijk}$, we have the invariant

$$f_3 = \text{det}(p_{+ij}) \text{det}(p_{+i}) \text{det}(p_{+j}) - p_{0+++}p_{1++} \text{Det}(p_{ijk}).$$

A computation with Macaulay 2 now yields the following:

**Theorem 5** The ideal of phylogenetic invariants vanishing on $V(SBD, 2, T)$ for the 3-taxon tree is

$$(f_0, f_1, f_2, f_3).$$

We thank a reviewer for pointing out that the $2 \times 2 \times 2$ hyperdeterminant was introduced into a phylogenetic context in [SJ04], where it is called the tangle. That paper considers the 2-base GM model on a 3-taxon tree rooted along an edge, and proposes the hyperdeterminant as a generalized ‘distance.’
For an \(n\)-taxon tree, determining the full ideal of invariants for the 2-base SBD model remains open. Of course, this model inherits the invariants of the GM model, which have been conjectured in [PS04] to be generated by ‘edge invariants’ arising from rank conditions on 2-dimensional flattenings of the tensor. This issue for GM will be dealt with in [AR04]. Additional invariants for SBD arise from applying the invariants of Theorem 5 to all 3-dimensional marginalizations of the \(n\)-dimensional tensor \(P\). One might suspect these generate the full ideal, but even for the 4-taxon tree we have been unable to confirm this computationally.

4 The \(\kappa\)-base models, arbitrary \(\kappa\)

**Proposition 6** Let \(T\) be an \(n\)-taxon tree, with \(E = E(n) = 2n - 3\) the number of its edges. Denoting the dimension of the variety \(V(\mathcal{M}, \kappa, T)\) by \(d(\mathcal{M}, \kappa, T)\),

\[
\begin{align*}
\quad d(SBD, \kappa, T) &= (\kappa - 1) + (\kappa - 1)^2 E, \\
\quad d(SD, \kappa, T) &= d(ATR, \kappa, T) = \frac{\kappa(\kappa - 1)}{2} + (\kappa - 1)E.
\end{align*}
\]

**PROOF.** For fixed \(\kappa\) and \(T\), choose a root \(r\) for \(T\). For each model \(\mathcal{M}\) we consider here, the parameter space \(X_{\mathcal{M}, \kappa, T_r} \subset X_{GM, \kappa, T_r}\) is a semialgebraic subset of \(\mathbb{R}^M\) with \(M = \kappa + \kappa^2 E\), as all our model assumptions are polynomial equalities or inequalities placing restrictions on the entries of \(p_r\) and the \(M_e\).

For each \((\mathcal{M}, \kappa, T_r)\), we will find a complex quasi-projective variety \(X\) of dimension \(d = d(\mathcal{M}, \kappa, T)\) and a generically finite map \(\psi : X \to \mathbb{C}^M\), such that \(X_{\mathcal{M}, \kappa, T_r} \subset \psi(X)\) and \(\psi^{-1}(X_{\mathcal{M}, \kappa, T_r}) = X\). These conditions imply

\[
\psi(X) = X_{\mathcal{M}, \kappa, T_r},
\]

so applying the map \(\phi = \phi_{GM, \kappa, T_r} : \mathbb{C}^M \to \mathbb{C}^\kappa\), we find

\[
\phi \circ \psi(X) = \phi(X_{\mathcal{M}, \kappa, T_r}) = V(\mathcal{M}, \kappa, T).
\]

Since the results of [AR03] show \(\phi\) is generically finite, the general fiber \((\phi \circ \psi)^{-1}(P)\) is of dimension zero. Using a standard result on the dimension of fibers of regular maps (see [Har92], for instance), we conclude that the dimension of \(X\) and \(V(\mathcal{M}, \kappa, T)\) are the same.

We begin with the SBD model, so \(d = (\kappa - 1) + (\kappa - 1)^2 E\). Let

\[
X = \{x \in \mathbb{C}^d \mid \sum_{i=1}^{\kappa-1} x_i \neq 1\},
\]
and define the map $\psi$ as follows: $\psi(x) = (p, \{M_e\})$ where $p_i = x_i$ for $i = 1, \ldots, \kappa - 1$, and the upper left $(\kappa - 1) \times (\kappa - 1)$ blocks of each $M_e$ are given by successive entries in $x$. Use the conditions that $\sum_i p_i = 1$, $\sum_j M_e(i,j) = 1$, and $p M_e = p$ to give rational formulas for the remaining entries of $p$ and $M_e$ in terms of $x$. Clearly $\psi$ is 1-1 and $X_{SD, \kappa, T_r} \subset \psi(X)$. Moreover, $\psi^{-1}(X_{SD, \kappa, T_r})$ is dense in $X$ since it contains a Euclidean-open subset of the real points of $X$, which is Zariski dense in $X$.

For the ATR model, with $d = \frac{\kappa(\kappa - 1)}{2} + (\kappa - 1)E$, let

$$X = \{(Q,u) \mid Q \in O_\kappa(\mathbb{C}), \ Q = (q_{ij}), \ q_{i1} \neq 0, \ u \in \mathbb{C}^{(\kappa - 1)E}\}.$$

Here $O_\kappa(\mathbb{C})$ is the variety of complex orthogonal $\kappa \times \kappa$ matrices, which has dimension $\frac{\kappa(\kappa - 1)}{2}$. Define $\psi$ by: $\psi(Q, u) = (p, \{M_e\})$ where $p = (q_{11}^2, q_{21}^2, \ldots, q_{\kappa 1}^2)$, and, with $D = \text{diag}(q_{i1}, q_{21}, \ldots, q_{\kappa 1})$,

$$M_{e_i} = D^{-1}Q \text{diag}(1, x_{j+1}, x_{j+2}, \ldots, x_{j+\kappa-1})Q^T D,$$

where $j = (\kappa - 1)(i - 1)$. That $\psi$ is generically finite is clear, and that $X_{ATR, \kappa, T_r} \subset \psi(X)$ follows from the discussion in section 2. Also, the set $\psi^{-1}(X_{ATR, \kappa, T_r})$ is dense in $X$ since it contains a Euclidean-open subset of the real points of $X$, which is Zariski dense in $X$.

Finally, for the SD model, recall [Jac75] that a family of real commuting normal matrices $A_{e_i}$ can be simultaneously expressed as $A_i = QB_iQ^T$, with $Q \in O_\kappa(\mathbb{R})$, and the $B_i$ real block diagonal matrices with the same block structure, where each diagonal block is either $1 \times 1$ or $2 \times 2$ of the form $\begin{pmatrix} a & b \\ -b & a \end{pmatrix}$. The block structures we need to consider will have $n$ $1 \times 1$ blocks, the first of which is 1, followed by $m$ $2 \times 2$ blocks, where $n \geq 1$, $m \geq 0$, and $n + 2m = \kappa$. Proceeding similarly to the case of the ATR model, for each of these $\binom{\kappa+1}{2}$ possible block structures $B$, we let $X_B$ denote a copy of $X$ as defined for ATR, and define a map $\psi_B : X_B \rightarrow \mathbb{C}^M$ similar to the ATR map, where the entries in $u$ give the independent block entries in $B_i$ and $M_{e_i} = D^{-1}QB_iQ^TD$. Letting $X$ be the disjoint union of the $X_B$, we obtain a map $\psi : X \rightarrow \mathbb{C}^M$. The rest of the argument is similar to that for the ATR model.

The construction of the varieties $X$ and maps $\psi$ in this proof also yield

**Proposition 7** The varieties $V(SBD, \kappa, T)$ and $V(ATR, \kappa, T)$ are irreducible, but $V(SD, \kappa, T)$ is the union of $\binom{\kappa+1}{2}$ distinct irreducible components, one of which is $V(ATR, \kappa, T)$.

For each of the SBD, SD, and ATR $\kappa$-base models on a tree $T$, we can construct a few phylogenetic invariants, though we are far from a full understanding of the ideals and varieties. Since any submodel inherits all invariants of a
supermodel, and \( SBD \supseteq SD \supseteq ATR \), we consider the models in that order. In addition, since these are all submodels of GM, all GM invariants on \( T \), such as those of \([AR03,AR04]\), are also invariants of these models.

**SBD model:** We first consider the 3-taxon tree \( T_r \), rooted at the central node and reason similarly to section 3. If \( (p_{ijk}) = \phi(p_r; M_1, M_2, M_3) \), then we have

\[
p_{i++} = p_{i+i} = p_{++i},
\]

giving \( 2(\kappa - 1) \) independent linear invariants expressing equality of base distributions at the leaves. We can also construct an invariant from the hyperdeterminant \( \text{Det}(p_{ijk}) \) on \( \kappa \times \kappa \times \kappa \) tensors. Letting \( m = m(\kappa) \) denote the degree of this polynomial (so \( m(3) = 36 \) and \( m(4) = 272 \)), then as before we find

\[
\text{Det}(p_{ijk}) = (\det(M_1) \det(M_2) \det(M_3) \det(D_r))^{m/\kappa}.
\]

Similarly,

\[
\begin{align*}
\det(p_{ij+}) &= \det(M_1) \det(M_2) \det(D_r), \\
\det(p_{i+j}) &= \det(M_1) \det(M_3) \det(D_r), \\
\det(p_{+ij}) &= \det(M_2) \det(M_3) \det(D_r),
\end{align*}
\]

so

\[
(\det(p_{ij+}) \det(p_{i+j}) \det(p_{+ij}))^{m/(2\kappa)} - (\prod_i p_{i++})^{m/(2\kappa)} \text{Det}(p_{ijk})
\]

is an invariant for the SBD model, since \( 2\kappa \) divides \( m \), as can be shown from formulas in \([GKZ94]\).

To see this is not an invariant for the GM model, we check that it does not vanish for some GM parameters. Indeed, if the parameters are chosen so the entries of \( p_r \) and \( p_{i++} \) are positive, the \( M_i \) are non-singular, and \( \det(D_r) \neq \prod_i p_{i++} \), then the invariant will be non-zero.

A similar construction replacing \( \text{Det} \) with any relative invariant \( h \) of \( GL_\kappa \times GL_\kappa \times GL_\kappa \) acting on \( \mathbb{C}^\kappa \otimes \mathbb{C}^\kappa \otimes \mathbb{C}^\kappa \) produces a phylogenetic invariant, provided \( h \) does not vanish on all diagonal tensors. If \( h \) does vanish on diagonal tensors, then \( h \) is already an invariant of the GM model.

To obtain \( n \)-taxon invariants we can of course compose 3-taxon invariants with any marginalization map of \( n \)-dimensional tensors to 3-dimensional ones.

**SD model:** Note that for any choice of 2 taxa \( a_j, a_k \) on \( T \), if \( P = \phi(p_r, \{M_e\}) \), where \( (p_r, \{M_e\}) \in X_{SD,\kappa,T} \), then the 2-dimensional marginalization \( \tilde{P}^{jk} \) of \( P \) obtained by summing over indices corresponding to all other taxa will be of the form \( D_r M \), where \( M \) is a product of matrices in the collection

\[
\{M_e \mid e \in \text{Edge}(T_r)\} \cup \{D_r^{-1}M_e^T D_r \mid e \in \text{Edge}(T_r)\},
\]
and $D_r = \text{diag}(p_r) = \text{diag}(p_1, p_2, \ldots, p_{\kappa})$. Thus all matrices in the collection

$$\{D_r^{-1}\tilde{P}^{jk} \mid 1 \leq j < k \leq n\} \cup \{D_r^{-1}(\tilde{P}^{jk})^T \mid 1 \leq j < k \leq n\}$$

will commute. For each pair chosen from this set, we get a collection of polynomials of degree $\kappa + 1$ from the statement of commutativity: For instance,

$$(D_r)^{-1}\tilde{P}^{jk}(D_r)^{-1}\tilde{P}^{lm} = (D_r)^{-1}\tilde{P}^{lm}(D_r)^{-1}\tilde{P}^{jk},$$

gives invariants from the entries of

$$\tilde{P}^{jk}(\det(D_r)(D_r)^{-1})\tilde{P}^{lm} - \tilde{P}^{lm}(\det(D_r)(D_r)^{-1})\tilde{P}^{jk}.$$ 

That some of these are not invariants of the SBD model when $\kappa > 2$ can be verified, most easily for a 2-taxon tree by a generic choice of SBD parameters.

**ATR model:** We consider first a 2-taxon tree, with $P \in \phi(X_{\text{ATR},\kappa,T_r})$. Then $P = D_r M_e$, so the condition $M_e = D_r^{-1}M_e^TD_r$ implies $P = P^T$. The entries of this matrix equation then give linear invariants, which are not invariants of the SD model for $\kappa > 2$, since there exist parameters for the SD model with $M_e \neq D_r^{-1}M_e^TD_r$. Composing these invariants with 2-dimensional marginalization maps gives linear invariants for an $n$-taxon tree.

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