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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.

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1. Introduction

Models of molecular evolution, such as for DNA sequences, typically assume that evolution occurs along a bifurcating tree, proceeding from a root representing the common ancestral sequence, toward the leaves representing the descendent sequences. At each site in the sequence, bases mutate according to a 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 how to infer the tree topology from observed sequences, assuming some reasonable model.

In the works of Cavender and Felsenstein (1987) and Lake (1987), the connections 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 distributions 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 values. 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 developed. See Felsenstein (2003).

For certain models, much progress has been made in determining invariants. Key advances for group-based models such as the Kimura 3-parameter one, were made in Evans and Speed (1993) and Steel et al. (1993), which built on the Hadamard conjugation introduced in Hendy and Penny (1993). Recently, Sturmfels and Sullivan (2005) further exploited the Hadamard conjugation to recognize that these varieties are toric, completing the determination of all invariants in this case. For the general Markov model, Allman and Rhodes (2003), found new constructions of invariants, 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 assumption 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 problematic 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 Lockhart et al. (1994), Conant and Lewis (2001), Rosenberg and Kumar (2003) and their references.

In Kumar and Gadagkar (2001) 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 pairwise 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 distribution. When the number of states 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 κ . 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 κ 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, \dots, 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 $Edge(T_r)$. 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 $[\kappa] = \{1, 2, \dots, \kappa\}$ of states (e.g., $\kappa = 4$ for the bases of DNA). A *root distribution vector* $\mathbf{p}_r = (p_1, p_2, \dots, p_\kappa)$, with $p_i \in [0, 1]$, $\sum_i p_i = 1$, describes the frequency of states in an ancestral sequence. With each $e \in Edge(T_r)$ 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 state i at the initial vertex mutating to state j at the final vertex of the edge. Together \mathbf{p}_r and $\{M_e\}_{e \in Edge(T_r)}$ comprise the parameters of the model. If no additional requirements are placed on \mathbf{p}_r or the M_e , then we have described the general Markov model (GM) of sequence evolution, studied in Allman and Rhodes (2003).

Letting X_{GM, κ, T_r} denote the parameter space for the κ -state GM model on T_r , we can view X_{GM, κ, T_r} as a subset of $[0, 1]^M$ for $M = \kappa + \kappa^2 E$ with $E = 2n - 3$, the number of edges of T . We have a map

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

so that $\phi(x)$ gives the joint distribution of states in aligned sequences at the leaves arising from the parameter choice x . Specifically, $\phi(x) = P = (p_{j_1 j_2 \dots j_n})$, a $\kappa \times \dots \times \kappa$ tensor with entries

$$p_{j_1 j_2 \dots j_n} = \sum_{i \in \mathcal{I}(j_1, j_2, \dots, j_n)} p_{i_r} \prod_{\substack{e \in Edge(T_r), \\ e = (v \rightarrow w)}} M_e(i_v, i_w),$$

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

In this paper we are interested in submodels of the GM model, in which we have stable state 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) \mathbf{p}_r has no zero entries, and 2) \mathbf{p}_r is fixed by all M_e ; that is, $\mathbf{p}_r M_e = \mathbf{p}_r$ for all edges e .
- Simultaneous Diagonalization Model (SD): In addition to the assumptions of SBD, 3) with $D_r = \text{diag}(\mathbf{p}_r)$, 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 \mathcal{M} formed from the κ -state GM model by imposing additional assumptions on the parameters, and for any rooted n -taxon tree T_r , we let $X_{\mathcal{M}, \kappa, T_r}$ denote the parameter space of \mathcal{M} on T_r . Then the algebraic variety $V(\mathcal{M}, \kappa, T_r)$ is the Zariski closure in \mathbb{C}^{κ^n} of $\phi(X_{\mathcal{M}, \kappa, T_r})$.

We now expand upon the model definitions. First, assuming that \mathbf{p}_r has no zero entries, the matrices $D_r^{-1} M_e^T D_r$ are also Markov matrices fixing \mathbf{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 \mathbf{p}_r and M_e . Then the joint distribution of states in aligned sequences at r and s arising from these parameter choices is given by the entries in $D_r M_e$. Assuming that M_e fixes \mathbf{p}_r , so that \mathbf{p}_r is also the state distribution of a sequence at s , the identity

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

shows that the model parameters \mathbf{p}_r, M_e on the 1-edge tree rooted at r lead to the same joint distribution as the model parameters $\mathbf{p}_r, D_r^{-1} M_e^T D_r$ on the 1-edge tree rooted at s . More generally, 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 $\mathbf{p}_s = \mathbf{p}_r$, 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. $\phi(X_{\mathcal{M}, \kappa, T_r})$ and $V(\mathcal{M}, \kappa, T_r)$ are independent of the choice of r for $\mathcal{M} = \text{SBD}, \text{SD}, \text{ and ATR}$. Thus, for these models, $V(\mathcal{M}, \kappa, T)$ is well defined without reference to r .

We note that independence of root location is a property also shared by the varieties associated with group-based models and the GM model (Steel et al., 1994; Allman and Rhodes, 2003).

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

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

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 , $M_e = \exp(Rt_e)$, 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 $\mathbf{p}_r R = \mathbf{0}$ (Felsenstein, 2003). 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 one (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 4×4 Hadamard matrix, with $(1, 1, 1, 1)$ the stable state distribution.

We summarize the relationships of the various models by

$$\begin{array}{c} \text{Group-based} \subset \text{ATR} \subseteq \text{SD} \subseteq \text{SBD} \subset \text{GM} \\ \subseteq \\ \text{GTR} \end{array}$$

The models on the main row all have algebraic definitions. Of course, the ideals associated with these models and their varieties are related by a reversed chain of inclusions. For $\kappa \geq 3$, the inclusions are all strict, though for $\kappa = 2$ equalities hold.

We have placed the GTR off the main row in this diagram since if $\kappa \geq 3$ it is not within the class of models with strictly algebraic definitions. Note also that for certain rate matrices a GTR model may be a submodel of a group-based one.

3. The 2-state 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 write

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

thus associating a single scalar parameter m_e with 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 states 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 φ 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 φ :*

$$p = p_{0++},$$

$$m_l = 1 - \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk} p_{(1-i)++} p_{(1-j)++} p_{(1-k)++}}{(p_{1++} - p_{0++})d_l}, \quad l = 1, 2, 3,$$

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

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

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 $\mathbf{v} = (v_0, v_1) = (-q, p)$ 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 \mathbf{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 \mathbf{v} and one of $(1, 1)$, yields

$$\begin{aligned}
 g_1 &= \sum_{i,j,k=0}^1 p_{ijk} v_j v_k = (1 - m_2)(1 - m_3) p q, \\
 g_2 &= \sum_{i,j,k=0}^1 p_{ijk} v_i v_k = (1 - m_1)(1 - m_3) p q, \\
 g_3 &= \sum_{i,j,k=0}^1 p_{ijk} v_i v_j = (1 - m_1)(1 - m_2) p q.
 \end{aligned}
 \tag{4}$$

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 Eqs. (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:

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

Eqs. (3) and (4) now lead directly to formulas for the m_i ,

$$m_i = 1 - \frac{g_0}{(v_0 + v_1)g_i}, \quad \text{for } i = 1, 2, 3,$$

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*

$$P = (p_{i_1 i_2 \dots i_n}) = \phi(\mathbf{p}_r; \{M_e\}_{e \in \text{Edge}(T_r)}).$$

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

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

$$m_{e_0} = 1 - \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk\dots+} + P_{(1-i)\dots+} + P_{(1-j)\dots+} + P_{(1-k)\dots+}}{(p_{1\dots+} - p_{0\dots+}) \det(p_{+ij\dots+})}.$$

- (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

$$m_{e_0} = 1 - \frac{\left(\sum_{ijk} (-1)^{i+j+k} p_{ijk+\dots+P(1-i)+\dots+P(1-j)+\dots+P(1-k)+\dots}\right) \det(p_{+i+k+\dots+})}{\left(\sum_{ijk} (-1)^{i+j+k} p_{+ijk+\dots+P(1-i)+\dots+P(1-j)+\dots+P(1-k)+\dots}\right) \det(p_{ij+\dots+})}.$$

Proof. The formula for p is clear. For the remaining formulas, by Proposition 2 the root location may be changed without changing the variety. Moreover, while moving the root may change a direction of an edge e , the matrix M_e is unchanged.

For a terminal edge e_0 as described above, the 3-dimensional tensor $(p_{ijk+\dots+}) = \phi(\mathbf{p}; M_1, M_2, M_3)$ 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 $(p_{ijk+\dots+}) = \phi_{T'}(\mathbf{p}; M_1, M_2, M_3)$ 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

$$\prod_{e \in \text{Path}(v, a_3)} (1 - m_e) = \frac{\sum_{ijk} (-1)^{i+j+k} p_{ijk+\dots+P(1-i)+\dots+P(1-j)+\dots+P(1-k)+\dots+}}{(p_{1+\dots+} - p_{0+\dots+}) \det(p_{ij+\dots+})}.$$

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

$$\prod_{e \in \text{Path}(w, a_3)} (1 - m_e) = \frac{\sum_{ijk} (-1)^{i+j+k} p_{+ijk+\dots+P(1-i)+\dots+P(1-j)+\dots+P(1-k)+\dots+}}{(p_{1+\dots+} - p_{0+\dots+}) \det(p_{+i+k+\dots+})}.$$

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. \square

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 $V(\text{SBD}, 2, T)$.

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

$$f_0 = 1 - p_{+++}.$$

Several other invariants for the SBD model are easily found. The distribution of states in a sequence at a_i is given by the vector \mathbf{p}_i where

$$\mathbf{p}_1 = (p_{i++}), \quad \mathbf{p}_2 = (p_{+i+}), \quad \mathbf{p}_3 = (p_{++i}).$$

Since each leaf sequence must have the same state composition, we set $\mathbf{p}_1 - \mathbf{p}_2 = \mathbf{p}_1 - \mathbf{p}_3 = \mathbf{0}$, 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 $\mathbf{p}_2 - \mathbf{p}_3 = 0$. These are the invariants underlying the disparity index of Kumar and Gadagkar (2001).

From Eqs. (3) and (4) we can also see that

$$h = \tilde{g}_0^2 v_0 v_1 + \tilde{g}_1 \tilde{g}_2 \tilde{g}_3 (v_0 + v_1)^2 \quad (5)$$

is an invariant of degree 8. However, while $h \notin (f_0, f_1, f_2)$, the ideal (f_0, f_1, f_2, h) is not the full ideal of invariants. Using Macaulay 2 (Grayson and Stillman, 2002) to find the kernel of the ring map associated with φ 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 Gel'fand et al. (1994). For a $2 \times 2 \times 2$ tensor such as P , the hyperdeterminant is given explicitly as

$$\begin{aligned} \text{Det}(P) = & (p_{000}^2 p_{111}^2 + p_{001}^2 p_{110}^2 + p_{010}^2 p_{101}^2 + p_{011}^2 p_{100}^2) \\ & - 2(p_{000} p_{001} p_{110} p_{111} + p_{000} p_{010} p_{101} p_{111} + p_{000} p_{011} p_{100} p_{111} \\ & + p_{001} p_{010} p_{101} p_{110} + p_{001} p_{011} p_{110} p_{100} + p_{010} p_{011} p_{101} p_{100}) \\ & + 4(p_{000} p_{011} p_{101} p_{110} + p_{001} p_{010} p_{100} p_{111}). \end{aligned}$$

Now reasoning from Eq. (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,

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

Thus $\tilde{g}_1 \tilde{g}_2 \tilde{g}_3 - pq \text{Det}(P) = 0$, and so $\tilde{g}_1 \tilde{g}_2 \tilde{g}_3 + v_1 v_0 \text{Det}(P)$, 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

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

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 Sumner and Jarvis (2004), where it is called the *tangle*. That paper considers the 2-state 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-state SBD model remains open. Of course, this model inherits the invariants of the GM model, which have been conjectured in Pachter and Sturmfels (2004) 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 Allman and Rhodes (2004). 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 that these generate the full ideal, but even for the 4-taxon tree we have been unable to confirm this computationally.

4. The κ -state models, arbitrary κ

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 $d(\mathcal{M}, \kappa, T)$,*

$$d(\text{SBD}, \kappa, T) = (\kappa - 1) + (\kappa - 1)^2 E,$$

$$d(\text{SD}, \kappa, T) = d(\text{ATR}, \kappa, T) = \frac{\kappa(\kappa - 1)}{2} + (\kappa - 1)E.$$

The varieties $V(\text{SBD}, \kappa, T)$ and $V(\text{ATR}, \kappa, T)$ are irreducible, but $V(\text{SD}, \kappa, T)$ is the union of $\lfloor \frac{\kappa+1}{2} \rfloor$ distinct irreducible components, one of which is $V(\text{ATR}, \kappa, T)$.

Proof. For fixed κ and T , choose a root r for T . For each model \mathcal{M} that 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 $\psi : X \rightarrow \mathbb{C}^M$, such that $X_{\mathcal{M}, \kappa, T_r} \subset \psi(X)$ and $\overline{\psi^{-1}(X_{\mathcal{M}, \kappa, T_r})} = X$. These conditions imply $\overline{\psi(X)} = \overline{X_{\mathcal{M}, \kappa, T_r}}$, so applying the map $\phi = \phi_{GM, \kappa, T_r} : \mathbb{C}^M \rightarrow \mathbb{C}^{\kappa^n}$, we find

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

Since the results of Allman and Rhodes (2003) show that ϕ 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 Harris, 1992, for instance), we conclude that the dimensions 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

$$X = \left\{ \mathbf{x} \in \mathbb{C}^d \mid \sum_{i=1}^{\kappa-1} x_i \neq 1 \right\},$$

and define the map ψ as follows: $\psi(x) = (\mathbf{p}, \{M_e\})$ where $p_i = x_i$ for $i = 1, \dots, \kappa - 1$, and the upper left $(\kappa - 1) \times (\kappa - 1)$ blocks of each M_e are given by successive entries in \mathbf{x} . Use the conditions that $\sum_i p_i = 1$, $\sum_j M_e(i, j) = 1$, and $\mathbf{p}M_e = \mathbf{p}$ to give rational formulas for the remaining entries of \mathbf{p} and M_e in terms of \mathbf{x} . Clearly ψ is 1–1 and $X_{\text{SBD}, \kappa, T_r} \subset \psi(X)$. Moreover, $\psi^{-1}(X_{\text{SBD}, \kappa, T_r})$ 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

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

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

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

where $j = (\kappa - 1)(i - 1)$. That ψ is generically finite is clear, and that $X_{ATR,\kappa,T_r} \subseteq \psi(X)$ 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 (Jacobson, 1975) that a family of real commuting normal matrices A_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 1×1 or 2×2 of the form $\begin{pmatrix} a & b \\ -b & a \end{pmatrix}$. The block structures we need to consider will have n 1×1 blocks, the first of which is 1, followed by m 2×2 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 $\lfloor \frac{\kappa+1}{2} \rfloor$ possible block structures \mathcal{B} , we let $X_{\mathcal{B}}$ denote a copy of X as defined for ATR, and define a map $\psi_{\mathcal{B}} : X_{\mathcal{B}} \rightarrow \mathbb{C}^M$ similar to the ATR map, where the entries in \mathbf{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 $X_{\mathcal{B}}$, we obtain a map $\psi : X \rightarrow \mathbb{C}^M$. The rest of the argument is similar to that for the ATR model. \square

For each of the SBD, SD, and ATR κ -state 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 Allman and Rhodes (2003, 2004), 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 in Section 3. If $(p_{ijk}) = \phi(\mathbf{p}; M_1, M_2, M_3)$, then we have

$$p_{i++} = p_{+i+} = p_{++i},$$

giving $2(\kappa - 1)$ independent linear invariants expressing equality of state 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,

$$\begin{aligned} \det(p_{ij+}) &= \det(M_1) \det(M_2) \det(D_r), \\ \det(p_{i+j}) &= \det(M_1) \det(M_3) \det(D_r), \\ \det(p_{+ij}) &= \det(M_2) \det(M_3) \det(D_r), \end{aligned}$$

so

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

is an invariant for the SBD model, since 2κ divides m , as can be shown from formulas in Gel'fand et al. (1994).

To see that 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 \mathbf{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 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 two taxa a_j, a_k on T_r , if $P = \phi(\mathbf{p}_r, \{M_e\})$, where $(\mathbf{p}_r, \{M_e\}) \in X_{SD,\kappa,T_r}$, 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 $D_r = \text{diag}(\mathbf{p}_r) = \text{diag}(p_{1+\dots+}, p_{2+\dots+}, \dots, p_{\kappa+\dots+})$. 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 $P \in \phi(X_{ATR,\kappa,T_r})$. Then $P = D_r M_e$, so the condition $M_e = D_r^{-1} M_e^T D_r$ 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 $M_e \neq D_r^{-1} M_e^T D_r$. Composing these invariants with 2-dimensional marginalization maps gives linear invariants for an n -taxon tree.

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