Rank conditions on phylogenetic networks

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Abstract

Less rigid than phylogenetic trees, phylogenetic networks allow the description of a wider range of evolutionary events. In this note, we explain how to extend the rank invariants from phylogenetic trees to phylogenetic networks evolving under the general Markov model and the equivariant models.

1 Introduction and preliminaries

In order to model the evolution of a set of DNA sequences (each representing a species), one usually considers a phylogenetic tree (whose leaves are in correspondence with the living species and interior nodes correspond to ancestral species) and a Markov process governing the substitution of nucleotides on it. In phylogenetics, invariants is the name given to the polynomials that vanish on every distribution that arises as a Markov process on the phylogenetic tree. The main idea behind finding invariants is that they might help to distinguish phylogenetic trees and phylogenetic networks and they have been successfully used in phylogenetic reconstruction [see CK14, FSC16], in solving the identifiability of certain models [AR09] and in model selection [KDGC12].

Nevertheless, trees might be too restrictive to represent the evolutionary history as they cannot take into account processes such as hybridization or horizontal gene transfer. In order to incorporate them, one can use phylogenetic networks. Invariants for phylogenetic networks have been found for the JC69 substitution model [GL18] (for networks with a single reticulation vertex) and for the 2-state symmetric model on networks with four leaves [Mit16, MSH18].

We restrict to tree-child binary networks [Ste16 §10]. That is, throughout the paper a phylogenetic network $N$ is a rooted acyclic directed graph (with no edges in parallel) satisfying:

1. the root $r$ has out-degree two,
2. every vertex with out-degree zero has in-degree one and is called a leaf,
3. all other vertices have either in-degree one and out-degree two (which are called tree vertices) or in-degree two and out-degree one (which are called reticulation vertices),
4. the child of a reticulation vertex is a tree vertex.

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Following [GL18] and [Nak11], we introduce Markov processes on phylogenetic networks. We denote by $\mathcal{V}$ the set of vertices of the network and will assume that there is a discrete random variable assigned to each vertex taking values in $\Sigma := \{A, C, G, T\}$. We assign a distribution $\pi = (\pi_A, \pi_C, \pi_G, \pi_T)$ to the root $r$ and to each edge $e$, a $4 \times 4$-transition matrix $M^e$. We write $\theta$ for the whole set of these parameters.

Let $\mathcal{N}$ be an $n$-leaf phylogenetic network and associate a $4 \times 4$ transition matrix from a nucleotide substitution model to each directed edge of $\mathcal{N}$. Suppose $\mathcal{N}$ has $m$ reticulation vertices $R = \{w_1, \ldots, w_m\}$. Each $w_i$ has indegree two, and we denote by $e^0_i$ and $e^1_i$ the two edges directed into $w_i$. Figure 1 shows an example of a phylogenetic network with 4 leaves and only one reticulation vertex $w_1$ (painted white).

Each binary vector $\sigma \in \{0, 1\}^m$ encodes the possible choices for the reticulation edges, where a 0 or a 1 in the $i$-th coordinate indicates that the edge $e^0_i$ or $e^1_i$ was deleted, respectively. Any $\sigma$ results in a $n$-leaf tree $T_\sigma$ rooted at $r$ with a collection of transition matrices corresponding to the particular edges in that tree. We call $\theta_\sigma$ the restriction of the parameters $\theta$ of the network to $T_\sigma$.

![Figure 1: On the left, a 4-leaf phylogenetic network $\mathcal{N}$ with one reticulation vertex $w_1$ painted white. The clade corresponding to leaves $A = \{1, 2\}$ has been coloured with gray. On the right, the two trees obtained when removing the edges $e^0_1$ and $e^1_1$ incident on $w_1$.](image)

For $1 \leq i \leq m$, denote by $\delta_i$ the parameter corresponding to the probability that a particular site was inherited along edge $e^1_i$. We can then define a distribution on the set $\Sigma^n$ (corresponding to characters at the leaves of the network) as follows

$$P_{\mathcal{N}, \theta} = \sum_{\sigma \in \{0, 1\}^m} \left( \prod_{i=1}^m \delta_i^{1-\sigma_i}(1-\delta_i)^{\sigma_i} \right) P_{T_\sigma, \theta_\sigma}.$$  

**def 1.1.** Let $A | B$ be a bipartition of the set of leaves of $\mathcal{N}$. Given a distribution vector $p$ on $4^n$ states, the flattening of $p$ relative to the bipartition $A | B$ is the $4^{|A|} \times 4^{|B|}$ matrix $\text{flatt}_{A|B}(p)$ whose $(i,j)$-entry is given by $p(k)$ where $k = (i,j)$ has entries matching those of $i$ and $j$ in the convenient order.

Let $T$ be a tree and let $A | B$ be a bipartition of the leaves of $T$ induced by removing an edge $e$ of $T$. Let $w$ be the vertex of $e$ adjacent to $A$. If $p$ is a distribution on $T$ given by a distribution $\pi$ at $w$ and transition matrices at the edges of $T$ oriented out from $w$, then $\text{flatt}_{A|B}(p)$ can be written as ([AR07], [Eri05])

$$\text{flatt}_{A|B}(p) = (M_A)^T D_\pi M_B,$$  

(1)
where \( D_\sigma \) is the \( 4 \times 4 \) diagonal matrix with the entries of \( \pi \) at the diagonal, \( M_A \) is the \( 4 \times 4^{|A|} \) matrix whose entry \( (x,i) \) is the probability in the subtree \( T_A \) of observing \( i \) at the leaves \( A \) given that the node \( w \) is at state \( x \) (and similarly for \( M_B \)). In the next sections we extend the well known edge invariants to phylogenetic networks. On a separate work we will study the consequences that this may have in distinguishing phylogenetic networks and phylogenetic trees.

2 Invariants for the general Markov model

Assume that there is a clade \( T_A \) in \( \mathcal{N} \) that does not contain any reticulation vertex (this is illustrated in the network of Figure 1 where the clade \( T_A \) corresponds to leaves 1 and 2). Thus \( T_A \) is a subtree of \( \mathcal{N} \) shared by all \( T_\sigma \) and the transition matrices at the edges of \( T_A \) are also shared by all \( T_\sigma \). We call \( B \) the leaves in \( \mathcal{N} \) that are not in \( A \).

**thm 2.1.** If \( p = P_{\mathcal{N},\theta} \) is a distribution on a phylogenetic network \( \mathcal{N} \) evolving under the GMM and \( T_A \) is a tree-clade in \( \mathcal{N} \), then \( \text{flatt}_{A|B}(p) \) has rank \( \leq 4 \).

**Proof.** Let \( v \) be the root of \( T_A \). To keep the proof simple we assume that \( v \) is different from \( r \). By rerooting each \( T_\sigma \) at \( v \), the edges of \( \mathcal{N} \) that are not in \( T_A \) might change their orientation, but the corresponding transition matrices can also be changed so that the joint distribution does not change. If \( \mu_\sigma \) is the new set of parameters for \( T_\sigma \), which is composed of the distribution \( \pi_\sigma \) at the vertex \( v \) and the new transition matrices, then \( P_{r_v,\theta_\sigma} = P_{r_v,\mu_\sigma} \). Note that after the rerooting process, the new transition matrices associated to the clade \( T_A \) are still the same for all \( T_\sigma \) (even if the distribution \( \pi_\sigma \) at \( v \) might be different for each \( T_\sigma \)). For each \( T_\sigma \), we write \( M_A \) for the transition matrix from \( v \) to the leaves in \( A \) and write \( M_B^\sigma \) for the transition matrix from \( v \) to the leaves in \( B \) (as in equation (I)). Then, we have

\[
\text{flatt}_{A|B}(p) = \sum_\sigma \left( \prod_{i=1}^m \delta_i^{1-\sigma_i} (1 - \delta_i)^{\sigma_i} \right) \text{flatt}_{A|B}(P_{r_\sigma,\mu_\sigma})
\]

\[
= \sum_\sigma \left( \prod_{i=1}^m \delta_i^{1-\sigma_i} (1 - \delta_i)^{\sigma_i} \right) M_A^{\sigma} D_\sigma M_B^\sigma =
\]

\[
= M_A^{\sigma} \sum_\sigma \left( \prod_{i=1}^m \delta_i^{1-\sigma_i} (1 - \delta_i)^{\sigma_i} \right) D_\sigma M_B^\sigma,
\]

where the second equality is obtained by using (I) for each \( T_\sigma \). Therefore, \( \text{flatt}_{A|B}(p) \) factorizes as a product of a \( 4^{|A|} \times 4 \) and a \( 4 \times 4^{|B|} \) matrix, and hence has rank \( \leq 4 \).

**Corollary 2.2.** If \( \mathcal{N} \) is a phylogenetic network with a tree-clade \( T_A \) as above and \( p \) is a distribution coming from a Markov process on \( \mathcal{N} \), then the \( 5 \times 5 \) minors of \( \text{flatt}_{A|B}(p) \) are invariants for \( \mathcal{N} \).

Note that these invariants are shared by all the phylogenetic networks that have the same clade \( T_A \). It is necessary to prove that the \( 5 \times 5 \) minors above do not vanish for other networks before using them with the idea of distinguishing networks.
3 Invariants for equivariant models

The construction of the first section stands for the general Markov model (GMM), where no particular structure is assumed for the transition matrices or the root distribution. This construction can be adapted by taking the substitution model more restrictive and considering evolutionary submodels of the general Markov model. A large class of these submodels are the \( G \)-equivariant models, where the transition matrices satisfy some symmetries according to a permutation group \( G < S_4 \). With precision, equivariant models only consider transition matrices that remain invariant after permuting rows and columns according to the permutations of some given permutation group (see [DK08] and [CFS10] for details). Among the \( G \)-equivariant models one finds the well known Jukes-Cantor model, Kimura 2 and 3 parameters and the strand symmetric model.

The result obtained in the previous section can be extended to \( G \)-equivariant models by using the tools introduced in [CFS10]. We explain briefly the idea. Let \( \mathcal{N} \) be a network with a tree-clade \( T_A \). If \( p \) is a distribution on \( \mathcal{N} \) arising from a \( G \)-equivariant model, then \( p \) actually lies in \( (\mathbb{C}^4)^G \), the set of points that remain invariant under the action of \( G \). If we write \( N_i \) for the irreducible representations of \( G \), the regular representation of \( G \) induces a decomposition of \( W = \mathbb{C}^4 \) into isotypic components: \( W \cong \bigoplus_{i=1}^{k} N_i \otimes \mathbb{C}^{m_i} \), for some well-defined multiplicities \( m_i \geq 0 \), and similar decompositions for every tensor power \( W \otimes l \), \( l \geq 1 \) (Maschke’s theorem). If \( \cdot | \cdot \) stands for cardinality, we can rewrite \( \text{flatt}_{A|B}(p) \) in a convenient basis of \( (\mathbb{C}^4)^G \cong \text{Hom}_G(W \otimes |A|, W \otimes |B|) \) consistent with these decompositions, so that the resulting matrix becomes block diagonal:

\[
\text{flatt}_{A|B}(p) = (B_1, \ldots, B_k).
\]

In this setting, we are able to prove the following result:

**Theorem 3.1.** If \( p \) arises from the \( G \)-equivariant model on \( \mathcal{N} \), then \( \text{rank}(B_i) \leq m_i \) for each \( i = 1, \ldots, k \).

**Corollary 3.2.** If \( \mathcal{N} \) is a phylogenetic network with a tree-clade \( T_A \) as above and \( p \) is a distribution coming from a Markov process on \( \mathcal{N} \), then the \( (m_i+1) \times (m_i+1) \)-minors of the block \( B_i \) of \( \text{flatt}_{A|B}(p) \) are invariants for \( \mathcal{N} \).

The precise technical statement and the proof will be provided in a forthcoming paper. It will be interesting to check whether these invariants arising from rank conditions coincide with some of the invariants found in [GL18] for the Jukes-Cantor model.

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