



Species notions that combine phylogenetic trees and phenotypic partitions

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Abstract

A recent paper (Manceau and Lambert in bioRxiv, 2017. <https://doi.org/10.1101/075580>) developed a novel approach for describing two well-defined notions of ‘species’ based on a phylogenetic tree and a phenotypic partition. In this paper, we explore some further combinatorial properties of this approach and describe an extension that allows an arbitrary number of phenotypic partitions to be combined with a phylogenetic tree for these two species notions.

Keywords Phylogenetic tree · Partition lattice · Species

Mathematics Subject Classification 05C05 · 06A07 · 92D15

1 Introduction

In biological classification, there have been numerous attempts to define notions of ‘species’, from Aristotle through to the present (De Queiroz and Donoghue 1988; Wheeler and Meier 2000). Today, phylogenetic trees and networks provide a framework for addressing this question (Baum 1992; Baum and Donoghue 1995; Rosenberg 2007); however, phylogeny alone captures only part of the concept of species. For example, two taxa may appear as different leaves in a tree (e.g. because of genetic differences in a neutral gene) but still be virtually identical (and perhaps even able to interbreed to produce viable offspring in the case where the taxa are diploid) and so would not be considered as belonging to different species. Thus, phenotypical characters also play a role in the concept of species (Sneath 1976).

However, phenotypical characters alone are also not sufficient to delineate species, because events such as convergent evolution can result in different species appearing to

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be quite similar. For example, two species separated by a long period of evolution may share many morphological traits because they have evolved under similar selection pressures (Holland et al. 2010).

If we regard species classification as the construction of a partition of a set X of taxa (e.g. organisms, populations, etc.) into disjoint sets ('species'), then a natural question arises: How can one construct such a partition in a canonical way so as to satisfy various desirable criteria by using a phylogenetic tree, and one or more phenotypic characters? A novel approach to this question was explored recently by Manceau and Lambert (2017), and provides the motivation and focus for this paper.

Suppose we are given a rooted binary phylogenetic tree T on a sample X of individuals (or, equivalently, as a 'hierarchy' on X , defined shortly), together with a partition \mathcal{P} of X , called a phenotypic partition (such a partition is induced by a phenotypic character by grouping taxa in the same state together). The desired output is a 'species partition' \mathcal{S} of X that satisfies three desirable properties (defined shortly) which should be satisfied by a classification into species, namely, *heterotypy* between species, *homotypy* within species and *monophyly* of each species. Manceau and Lambert (2017) showed that all three properties cannot generally be simultaneously satisfied; however, any pair of them can be, in which case there are two canonical constructions of a species partition \mathcal{S} of X . Related combinatorial notions were also considered in a somewhat different setting in Aldous et al. (2008); see also Alexander (2013), Dress et al. (2010), Kornet et al. (1995) and Kwok (2011) for combinatorial approaches to the species problem.

In biology, each species will typically have many associated phenotypic characters, and while it is possible to consider a single associated phenotype (e.g. by taking the product of the associated state spaces) such a transformation will generally lose information (i.e. different combinations of phenotypes could lead to the same phenotype partition). Thus, it is desirable to extend the concepts above to allow more than one phenotypic partition, in concert with a hierarchy.

In this paper, we generalize some of the main results from Manceau and Lambert (2017), by allowing an arbitrary number of phenotypic partitions, and by also lifting the requirement that the input tree T be binary. We also provide a short proof of a key result from that paper by using lattice theory, and, in the final section, establish conditions under which phenotypic partitions can be realized by state changes (indicated by 'marks') on edges of the tree T .

1.1 Preliminaries

We first review some basic notions from phylogenetic combinatorics [further details can be found in Dress et al. (2011) or Steel (2016)].

A *rooted phylogenetic tree* T on leaf set X is a rooted tree in which each non-leaf vertex (including the root) is unlabelled and has at least two outgoing edges. Given any rooted phylogenetic tree T , there is a partial order \leq_T on the vertices of T , defined by $u \leq_T v$ if $u = v$ or if u lies on the path from the root to v . Given a rooted tree $T = (V, E)$ and a non-empty subset C of V , there is a (unique) vertex v for which $v \leq_T c$ for all $c \in C$ and which is maximal (under \leq_T) with respect to this property; this vertex v is called the *lowest common ancestor* (*lca*) of C in T and is denoted $\text{lca}_T(C)$.

A *hierarchy* \mathcal{H} is a collection of non-empty subsets of X that contains X and that satisfies the following *nesting* property:

$$\text{For all } A, B \in \mathcal{H}, A \cap B \in \{\emptyset, A, B\}.$$

If a hierarchy also has the property that the singleton $\{x\}$ is an element of \mathcal{H} for every $x \in X$ then we say that \mathcal{H} is a *hierarchy on* X .

An element of a hierarchy \mathcal{H} is also called a *cluster* (it is sometimes also referred to in the literature as a ‘clade’). Given a hierarchy on X , there is a unique rooted phylogenetic tree T on X with the property that for each cluster there is a unique vertex in T such that the cluster consists of the labels of exactly the leaves descending from this vertex. In this way, hierarchies on X and rooted phylogenetic trees on leaf set X are essentially equivalent and will be used interchangeably.

A *partition* of a set X is a division of X into a set of non-empty subsets, called *blocks*, such that every element of X is in exactly one block. Let $\mathbb{P}(X)$ denote the set of all partitions of X . A *phenotypic partition* \mathcal{P} is a partition of X , such that the blocks of the partition correspond to the different ‘phenotypes’ of the individuals in X according to some biological characteristics. For example, birds can have the phenotype ‘able to fly’ or ‘not able to fly’, resulting in a phenotypic partition with two blocks.

A *species partition* \mathcal{S} is a partition of X in which the blocks are called ‘species’. We are particularly interested in species partitions that satisfy one or more of the following three desirable properties from Manceau and Lambert (2017):

- (A) *Heterotypy between species*. Individuals in different species are phenotypically different. Formally, for each phenotype $P \in \mathcal{P}$ and for each species $S \in \mathcal{S}$, either $P \subseteq S$ or $P \cap S = \emptyset$.
- (B) *Homotypy within species*. Individuals in the same species are phenotypically identical. Formally, for each phenotype $P \in \mathcal{P}$ and for each species $S \in \mathcal{S}$, either $S \subseteq P$ or $P \cap S = \emptyset$.
- (M) *Monophyly* All species are monophyletic. Formally, each species is a cluster of T (i.e. $S \subseteq \mathcal{H}$).

For any species partition \mathcal{S} of X satisfying property (M) for a given hierarchy \mathcal{H} on X , there is a corresponding hierarchy $\mathcal{H}_{\mathcal{S}}$ on \mathcal{S} which is defined in Manceau and Lambert (2017) by

$$\mathcal{H}_{\mathcal{S}} := \{H \in \mathcal{H} : \text{there exists some } S \in \mathcal{S} \text{ with } S \subseteq H\}. \quad (1)$$

Thus, the rooted tree associated with $\mathcal{H}_{\mathcal{S}}$ is obtained from the rooted tree associated with \mathcal{H} by removing all clusters that lie strictly within a block of \mathcal{S} (i.e. collapsing the tree structure below the ‘species level’ determined by \mathcal{S}). We will discuss an example of this later in relation to Fig. 1.

1.2 The lattice of partitions of X

Since we want to apply combinatorial arguments, we first recall some terms from lattice theory. Let (Y, \preceq) be a *partially ordered set* (poset) consisting of a set Y and a

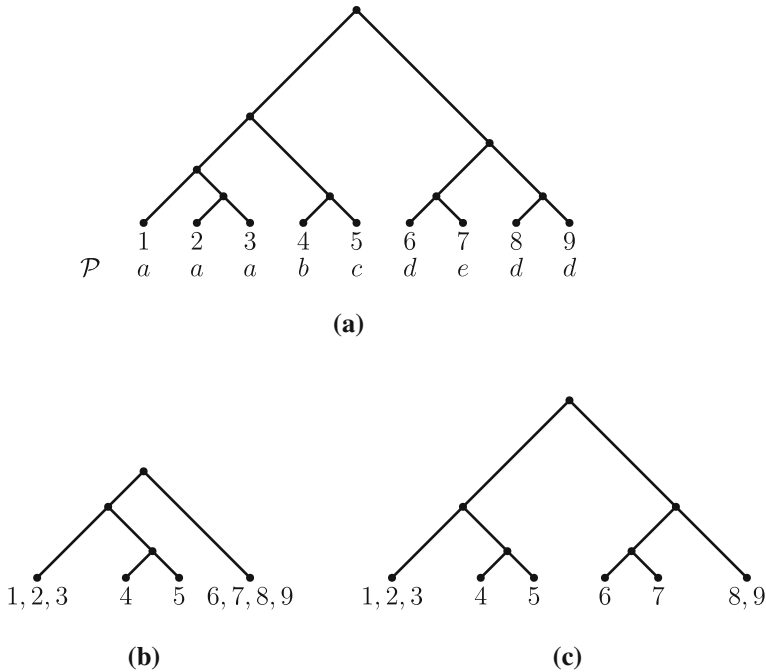


Fig. 1 **a** A hierarchy \mathcal{H} and a phenotypic partition \mathcal{P} indicated by the states at the leaves. **b** The corresponding loose species partition $\mathcal{S}_{\text{loose}}$ and its associated hierarchy $\mathcal{H}_{\text{loose}}$ **c** The corresponding lacy species partition $\mathcal{S}_{\text{lacy}}$ and its associated hierarchy $\mathcal{H}_{\text{lacy}}$

partial order \preceq . Given a set $A \subseteq Y$ with $A \neq \emptyset$, we say that $p \in Y$ is an *upper bound* for A if $a \preceq p$ for all $a \in A$. The *least upper bound* for A ($\text{lub}(A)$) is an upper bound such that for any other upper bound p one has $\text{lub}(A) \preceq p$. The *lower bound* and *greatest lower bound* ($\text{glb}(A)$) are defined analogously. A *lattice* is a partially ordered set (Y, \preceq) with the property that for all non-empty subsets A of Y , $\text{lub}(A)$ and $\text{glb}(A)$ exist and both are unique [for further background and details, see Bóna (2011)].

Next, we recall some important properties of the poset $\mathbb{P}(X)$ (of partitions of X), beginning with notion of *refinement* of partitions. Let \mathcal{S} and \mathcal{S}' be two partitions of the set X . We say that \mathcal{S} is *finer* than \mathcal{S}' and \mathcal{S}' is *coarser* than \mathcal{S} , denoted $\mathcal{S} \preceq \mathcal{S}'$, if for each $S \in \mathcal{S}$ and each $S' \in \mathcal{S}'$, either $S \subseteq S'$ or $S \cap S' = \emptyset$. The relation \preceq is a partial order but not a linear order (i.e. two partitions \mathcal{S} and \mathcal{S}' of the set X cannot always be compared; for example, consider $\mathcal{S} = \{\{a, b\}, \{c\}, \{d\}\}$ and $\mathcal{S}' = \{\{a\}, \{b\}, \{c, d\}\}$).

It can be shown that the set of all partitions of the set X ordered by refinement [i.e. $(\mathbb{P}(X), \preceq)$] is a lattice. In other words, the lub and the glb of any subset of $\mathbb{P}(X)$ exist (Bóna 2011). We describe the glb and lub explicitly now. Given k partitions $\Sigma_1, \dots, \Sigma_k \in \mathbb{P}(X)$, their glb is the set of non-empty intersections of blocks each chosen from a different partition. That is, $\text{glb}(\Sigma_1, \dots, \Sigma_k) = \{\bigcap_{i=1}^k B^{(i)} : B^{(i)} \in \Sigma_i\} - \{\emptyset\}$. To define the lub , first define the relation \sim by $x \sim y$ if there exists $i \in \{1, \dots, k\}$ such that $\{x, y\} \subseteq B \in \Sigma_i$. In other words, $x \sim y$ if x and y are in the same block in at least one of the k partitions of X . Let \approx be the transitive closure of

the relation \sim . The lub of $\Sigma_1, \dots, \Sigma_k$ is then defined as the set of equivalence classes of X under \approx . Note, that

$$\mathcal{S}_0 = \{\{x\} : x \in X\} \text{ and } \mathcal{S}_1 = \{X\}$$

are the minimal and maximal elements of $\mathbb{P}(X)$, respectively, under the partial order \preceq .

2 Discussion of earlier results

In this section, we describe some of the main results of Manceau and Lambert (2017), which we have organised into two theorems. The first theorem has a short proof, as presented in that paper, the second theorem involved a more complicated argument, and we present a shorter lattice-theoretic proof. From now on, \mathcal{H} always denotes a hierarchy of X , \mathcal{P} always denotes a phenotypic partition, and \mathcal{S} always denotes a species partition.

Theorem 1 (Manceau and Lambert 2017) *Given a phenotypic partition \mathcal{P} of X and a hierarchy \mathcal{H} on X :*

- (i) \mathcal{S} satisfies property (A) if and only if $\mathcal{P} \preceq \mathcal{S}$, and \mathcal{S} satisfies property (B) if and only if $\mathcal{S} \preceq \mathcal{P}$. In particular, if \mathcal{S} satisfies properties (AB), then $\mathcal{S} = \mathcal{P}$.
- (ii) Unless \mathcal{P} and \mathcal{H} satisfy $\mathcal{P} \subseteq \mathcal{H}$ (i.e. each phenotype is a cluster), there is no species partition \mathcal{S} that satisfies all three properties (ABM).
- (iii) Any two properties from (A), (B) and (M) can be satisfied by at least one species partition \mathcal{S} . Specifically, $\mathcal{S} = \mathcal{P}$ satisfies (AB), \mathcal{S}_1 satisfies (AM), and \mathcal{S}_0 satisfies (BM).

Theorem 1 raises an interesting question. One can satisfy (AM) by the very coarsest partition \mathcal{S}_1 , and (BM) by the very finest partition \mathcal{S}_0 . However, neither of these is particularly relevant for biology. Rather, one would like to find a finest partition satisfying (AM) and a coarsest partition satisfying (BM). Ideally, such partitions should also be uniquely determined by those properties. Fortunately, this turns out to be the case. A second main theorem from Manceau and Lambert (2017) is the following.

Theorem 2 (Manceau and Lambert 2017) *Given \mathcal{P} and \mathcal{H} :*

- (i) There is a unique finest partition of X satisfying (AM) (heterotypy between species and monophyly).
- (ii) There is a unique coarsest partition of X satisfying (BM) (homotypy within species and monophyly).

Following Manceau and Lambert (2017), we will refer to the unique species partitions referred to in parts (i) and (ii) of Theorem 2 as the *loose* and *lacy* species partitions, respectively, and denote them as $\mathcal{S}_{\text{loose}}$ ($= \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P})$) and $\mathcal{S}_{\text{lacy}}$ ($= \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P})$), respectively.

Proposition 1 (below) provides a more explicit description of the loose and lacy species partitions. Roughly speaking, the reader can regard $\mathcal{S}_{\text{loose}}$ as the species partition that allows as many species as possible such that different species have different phenotypes, while $\mathcal{S}_{\text{lacy}}$ is the species partition that allows as few species as possible such that each species has a single phenotype.

We will describe example of these two partitions in the next section, but first we provide a short lattice-theoretic proof of Theorem 2. This proof relies on the following lemma.

Lemma 1 *Given a non-empty subset \mathbb{P} of partitions of X (i.e. $\mathbb{P} \subseteq \mathbb{P}(X)$) and a hierarchy \mathcal{H} on X , if $\Sigma \subseteq \mathcal{H}$ for each $\Sigma \in \mathbb{P}$ then $\text{glb}(\mathbb{P}) \subseteq \mathcal{H}$ and $\text{lub}(\mathbb{P}) \subseteq \mathcal{H}$.*

Proof Consider $\text{glb}(\mathbb{P})$. Because $\Sigma \subseteq \mathcal{H}$ for each $\Sigma \in \mathbb{P}$, and \mathcal{H} satisfies the nesting property of a hierarchy, if we select a block from each partition in \mathbb{P} , then the resulting sets $B_1, B_2, \dots, B_{|\mathbb{P}|}$ satisfy $\bigcap_{i=1}^{|\mathbb{P}|} B_i \in \{B_1, \dots, B_{|\mathbb{P}|}, \emptyset\}$. Hence, all elements of $\text{glb}(\mathbb{P})$ are also elements of \mathcal{H} and thus, $\text{glb}(\mathbb{P}) \subseteq \mathcal{H}$. Next consider $\text{lub}(\mathbb{P})$. Because $\Sigma \subseteq \mathcal{H}$ for all $\Sigma \in \mathbb{P}$, the relation \sim is already transitive. To see this, observe that if two blocks B_1 and B_2 of two different partitions from \mathbb{P} share at least one element, then the nesting property of hierarchies implies that one of those blocks (B_1 or B_2) contains the other block. Consequently, if $x \sim y$ and $y \sim z$ with $x, y \in B_1$ and $y, z \in B_2$, then either $x \in B_2$ or $z \in B_1$, and thus, $x \sim z$. Thus, \sim is transitive and so is equal to the equivalence relation \approx whose equivalence classes are the blocks of $\text{lub}(\mathbb{P})$. Consequently, each block of the partition $\text{lub}(\mathbb{P})$ is also an element of \mathcal{H} and thus, $\text{lub}(\mathbb{P}) \subseteq \mathcal{H}$. \square

We can now provide a short proof of Theorem 2.

Proof Let

$$\mathbb{S}_{AM} := \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \mathcal{P} \preceq \Sigma\}, \text{ and } \mathbb{S}_{BM} := \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \Sigma \preceq \mathcal{P}\}.$$

Since $\mathbb{S}_{AM}, \mathbb{S}_{BM} \subseteq \mathbb{P}(X)$, and $(\mathbb{P}(X), \preceq)$ is a lattice, $\text{glb}(\mathbb{S}_{AM})$ and $\text{lub}(\mathbb{S}_{BM})$ exist. Therefore, to establish Theorem 2, it suffices to show that:

$$\text{glb}(\mathbb{S}_{AM}) \in \mathbb{S}_{AM} \text{ and } \text{lub}(\mathbb{S}_{BM}) \in \mathbb{S}_{BM}, \tag{2}$$

from which it follows that $\mathcal{S}_{\text{loose}} = \text{glb}(\mathbb{S}_{AM})$ and $\mathcal{S}_{\text{lacy}} = \text{lub}(\mathbb{S}_{BM})$. We establish Eq. (2) as follows. The two sets $\text{glb}(\mathbb{S}_{AM})$ and $\text{lub}(\mathbb{S}_{BM})$ are not empty because $\mathcal{S}_1 \in \mathbb{S}_{AM}$ and $\mathcal{S}_0 \in \mathbb{S}_{BM}$. Since all elements of \mathbb{S}_{AM} and of \mathbb{S}_{BM} satisfy property (M) (i.e. $\mathcal{S} \subseteq \mathcal{H}$ for all $\mathcal{S} \in \mathbb{S}_{AM}, \mathbb{S}_{BM}$), Lemma 1 implies that $\text{glb}(\mathbb{S}_{AM})$ and $\text{lub}(\mathbb{S}_{BM})$ satisfy property (M). Also, because elements of \mathbb{S}_{AM} satisfy property (A) (i.e. $\mathcal{P} \preceq \mathcal{S}$ for all $\mathcal{S} \in \mathbb{S}_{AM}$), it follows that \mathcal{P} is a lower bound for \mathbb{S}_{AM} . Since $\text{glb}(\mathbb{S}_{AM})$ is the greatest lower bound we know that it is coarser than all other lower bounds and, in particular, $\mathcal{P} \preceq \text{glb}(\mathbb{S}_{AM})$. Thus, $\text{glb}(\mathbb{S}_{AM})$ satisfies property (A) as well. The proof that $\text{lub}(\mathbb{S}_{BM})$ satisfies property (B) is analogous. In summary, $\text{glb}(\mathbb{S}_{AM}) \in \mathbb{S}_{AM}$ and $\text{lub}(\mathbb{S}_{BM}) \in \mathbb{S}_{BM}$. \square

3 Constructing the loose and lacy species partitions

We now describe an explicit and readily computable way to construct the loose and lacy species partitions [our treatment differs in some details from the construction proposed by Manceau and Lambert (2017)]. The process involves first constructing subsets \mathcal{H}_1 and \mathcal{H}_2 of \mathcal{H} . The loose and lacy species partitions are then the maximal elements (under set inclusion) of \mathcal{H}_1 and \mathcal{H}_2 respectively. We first introduce some additional notation. For $P \in \mathcal{P}$, let

$$h_P = \bigcap_{h \in \mathcal{H}: P \subseteq h} h. \quad (3)$$

Notice that $P \subseteq h_P \in \mathcal{H}$, and that h_P is the unique minimal cluster in \mathcal{H} that contains P (it is the cluster in \mathcal{H} that corresponds to $\text{lca}_T(P)$ where T is the phylogenetic X -tree associated with \mathcal{H}). It is also possible that $h_P = h_Q$ for different blocks P and Q of \mathcal{P} .

Next, given \mathcal{P} and \mathcal{H} , define the following two sets:

$$\mathcal{H}_1 := \{h_P : P \in \mathcal{P}\}, \text{ and } \mathcal{H}_2 := \{h \in \mathcal{H} : \exists P \in \mathcal{P} : h \subseteq P\}.$$

Observe that \mathcal{H}_1 and \mathcal{H}_2 are both subsets of \mathcal{H} ; in particular, \mathcal{H}_2 is the set of clusters in \mathcal{H} that are contained in a block of the phenotypic partition \mathcal{P} . The following proposition provides an explicit description of the loose and lacy partitions; its proof is provided in the ‘‘Appendix’’.

Proposition 1 *Given a phenotypic partition \mathcal{P} of X and a hierarchy \mathcal{H} on X ,*

$\mathcal{S}_{\text{loose}}$ is the set of maximal elements of \mathcal{H}_1 ;

$\mathcal{S}_{\text{lacy}}$ is the set of maximal elements of \mathcal{H}_2 .

In the phylogenetic tree T corresponding to the hierarchy \mathcal{H} the elements in \mathcal{H}_1 are the lowest common ancestors in T of each element in \mathcal{P} . The loose species partition contains, for each leaf of T , the first last common ancestor which is in \mathcal{H}_1 that lies on the path from the root to the leaf.

The elements of \mathcal{H}_2 correspond to the vertices of the phylogenetic tree T associated with \mathcal{H} for which all descended leaves have the same phenotype. The lacy species partition contains, for each leaf of T , the first element which is in \mathcal{H}_2 that lies on the path from the root to the leaf.

An example [based on Fig. 4 from Manceau and Lambert (2017)] is shown in Fig. 1, for the set $X = \{1, 2, 3, 4, 5, 6, 7, 8, 9\}$ together with the hierarchy \mathcal{H} on X defined by

$$\begin{aligned} \mathcal{H} = & \{\{x\} : x \in X\} \cup \{\{2, 3\}, \{1, 2, 3\}, \\ & \{4, 5\}, \{6, 7\}, \{8, 9\}, \{1, 2, 3, 4, 5\}, \{6, 7, 8, 9\}, X\} \end{aligned}$$

along with the phenotypic partition $\mathcal{P} = \{\{1, 2, 3\}, \{4\}, \{5\}, \{6, 8, 9\}, \{7\}\}$. In this example,

$$\mathcal{H}_1 = \{\{1, 2, 3\}, \{4\}, \{5\}, \{7\}, \{6, 7, 8, 9\}\}, \text{ and so}$$

$$\mathcal{S}_{\text{loose}} = \{\{1, 2, 3\}, \{4\}, \{5\}, \{6, 7, 8, 9\}\}.$$

In addition,

$$\mathcal{H}_2 = \{\{x\} : x \in X\} \cup \{\{2, 3\}, \{1, 2, 3\}, \{8, 9\}\}, \text{ and so}$$

$$\mathcal{S}_{\text{lacy}} = \{\{4\}, \{5\}, \{6\}, \{7\}, \{1, 2, 3\}, \{8, 9\}\}.$$

The corresponding induced hierarchies [given by Eq. (1)] are given by:

$$\mathcal{H}_{\text{loose}} = \mathcal{S}_{\text{loose}} \cup \{\{1, 2, 3, 4, 5\}, X\}, \text{ and}$$

$$\mathcal{H}_{\text{lacy}} = \mathcal{S}_{\text{lacy}} \cup \{\{4, 5\}, \{6, 7\}, \{1, 2, 3, 4, 5\}, \{6, 7, 8, 9\}, X\}.$$

4 Extending the theory to $k \geq 2$ phenotypic partitions

Suppose now that one has $k \geq 2$ phenotypic partitions $\mathcal{P}_1, \dots, \mathcal{P}_k$ with $\mathcal{P}_i \in \mathbb{P}(X)$ for $i = 1, \dots, k$ and, as before, a hierarchy \mathcal{H} on X .

Define the sets of species partitions satisfying properties (AM), (BM) as the following:

$$\mathbb{S}_{AM}^{(k)} := \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \mathcal{P}_i \leq \Sigma, \text{ for each } i = 1, \dots, k\}, \text{ and}$$

$$\mathbb{S}_{BM}^{(k)} := \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \Sigma \leq \mathcal{P}_i, \text{ for each } i = 1, \dots, k\}.$$

Some natural questions arise now: Is $\text{glb}(\mathbb{S}_{AM}^{(k)}) \in \mathbb{S}_{AM}^{(k)}$? Is $\text{lub}(\mathbb{S}_{BM}^{(k)}) \in \mathbb{S}_{BM}^{(k)}$? And if so, how are these two partitions related to $\mathcal{S}_{\text{loose}}$ and $\mathcal{S}_{\text{lacy}}$? We will show that the case $k \geq 2$ can be reduced to the earlier case where $k = 1$.

Theorem 3 *Given a hierarchy \mathcal{H} on X and phenotypic partitions $\mathcal{P}_1, \dots, \mathcal{P}_k$ of X , let*

$$\mathcal{P}^+ := \text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_k) \text{ and } \mathcal{P}^- := \text{glb}(\mathcal{P}_1, \dots, \mathcal{P}_k).$$

Then $\text{glb}(\mathbb{S}_{AM}^{(k)}) = \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}^+) \in \mathbb{S}_{AM}^{(k)}$,

and $\text{lub}(\mathbb{S}_{BM}^{(k)}) = \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}^-) \in \mathbb{S}_{BM}^{(k)}$.

Proof First, we show that:

$$\mathbb{S}_{AM}^{(k)} = \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \mathcal{P}^+ \leq \Sigma\}. \tag{4}$$

By the definition of $\mathbb{S}_{AM}^{(k)}$, it follows that $\mathcal{P}_i \leq \Sigma$ for all $\Sigma \in \mathbb{S}_{AM}^{(k)}$ and for all $i = 1, \dots, k$. Furthermore, for all $i = 1, \dots, k$ we have $\mathcal{P}_i \leq \mathcal{P}^+$. By the definition of the lub, there is no $\mathcal{P} \in \mathbb{P}(X)$ with $\mathcal{P} \subseteq \mathcal{H}$ and with \mathcal{P} lying strictly between \mathcal{P}_i

and \mathcal{P}^+ under the refinement partial order \preceq for all $i = 1, \dots, k$. Therefore, we obtain Eq. (4). For $\mathbb{S}_{BM}^{(k)}$ an analogous argument applies, using glb instead of lub. Specifically:

$$\mathbb{S}_{BM}^{(k)} = \{\Sigma \in \mathbb{P}(X) : \Sigma \subseteq \mathcal{H}, \Sigma \preceq \mathcal{P}^-\}.$$

Now, \mathcal{P}^+ and \mathcal{P}^- are partitions of X , and for each of these two phenotypic partitions Eq. (2) ensures that $\text{glb}(\mathbb{S}_{AM}^{(k)}) = \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}^+) \in \mathbb{S}_{AM}^{(k)}$ and $\text{lub}(\mathbb{S}_{BM}^{(k)}) = \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}^-) \in \mathbb{S}_{BM}^{(k)}$, as claimed. \square

4.1 Combining properties for $k \geq 2$ phenotypic partitions

Until now, we were looking for species partitions that are subsets of the given hierarchy and either finer or coarser than the given phenotypic partitions, in other words, satisfying either (AM) or (BM). We now consider the setting of $k = 2$ phenotypic partitions \mathcal{P}_1 and \mathcal{P}_2 with $\mathcal{P}_1 \preceq \mathcal{P}_2$. Such a situation arises when, for example, \mathcal{P}_1 is the partition of X induced by the phenotypes of individuals in X , while \mathcal{P}_2 is a coarser partition of X into disjoint ‘types’, in which individuals having the same phenotype have the same type (but a type might also allow some variation of phenotypes within it). In this setting we can ask when is there a subset Σ of \mathcal{H} for which $\mathcal{P}_1 \preceq \Sigma \preceq \mathcal{P}_2$? In other words, is there a species partition that satisfies (AM) for one phenotypic partition and (BM) for a second one? The answer to this question is provided in the following result.

Theorem 4 *Given phenotypic partitions $\mathcal{P}_1, \mathcal{P}_2$ of X and a hierarchy \mathcal{H} on X , there is a subset Σ of \mathcal{H} with $\mathcal{P}_1 \preceq \Sigma \preceq \mathcal{P}_2$ if and only if $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$.*

Proof (\Rightarrow) First suppose that $\mathcal{P}_1 \preceq \Sigma \preceq \mathcal{P}_2$ holds for some $\Sigma \in \mathbb{P}(X)$, $\Sigma \subseteq \mathcal{H}$. This implies the following:

- $\mathcal{P}_1 \preceq \Sigma \subseteq \mathcal{H}$, so Σ is an element of the set \mathbb{S}_{AM} for \mathcal{P}_1 and $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \Sigma$ (since $\mathcal{S}_{\text{loose}}$ is a lower bound to \mathbb{S}_{AM} for \mathcal{P}_1);
- $\mathcal{H} \supseteq \Sigma \preceq \mathcal{P}_2$, so Σ is an element of the set \mathbb{S}_{BM} for \mathcal{P}_2 and $\Sigma \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$ (since $\mathcal{S}_{\text{lacy}}$ is an upper bound to \mathbb{S}_{BM} for \mathcal{P}_2).

Consequently, $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$, which establishes the forward implication.

(\Leftarrow) Conversely, suppose that $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$ holds. Since

$$\mathcal{P}_1 \preceq \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \text{ and } \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2) \preceq \mathcal{P}_2,$$

by the definitions of $\mathcal{S}_{\text{loose}}$ and $\mathcal{S}_{\text{lacy}}$, we have $\mathcal{P}_1 \preceq \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2) \preceq \mathcal{P}_2$. Moreover, $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1), \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2) \subseteq \mathcal{H}$, by definition. Therefore, by choosing either $\Sigma := \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1)$ or $\Sigma := \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$, it holds that:

$$\mathcal{P}_1 \preceq \mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1) \preceq \Sigma \preceq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2) \preceq \mathcal{P}_2.$$

This establishes the reverse implication. \square

If we focus on the set of all species partitions that lie between the two given phenotypic partitions (i.e. $\mathbb{S} := \{\Sigma \subseteq \mathcal{H} : \mathcal{P}_1 \leq \Sigma \leq \mathcal{P}_2\}$), then we automatically see that $\mathcal{S}_{\text{loose}}(\mathcal{H}, \mathcal{P}_1)$ is the finest element of \mathbb{S} and $\mathcal{S}_{\text{lacy}}(\mathcal{H}, \mathcal{P}_2)$ is the coarsest.

We now extend this problem to $k \geq 2$ phenotypic partitions. The question is: Given phenotypic partitions $\mathcal{P}_1, \dots, \mathcal{P}_k$ and an integer l with $0 \leq l \leq k$ is there a subset Σ of \mathcal{H} for which $\mathcal{P}_1, \dots, \mathcal{P}_l \leq \Sigma \leq \mathcal{P}_{l+1}, \dots, \mathcal{P}_k$? The answer to this question is provided as follows.

Corollary 1 *There exists a $\Sigma \subseteq \mathcal{H}$ with $\mathcal{P}_1, \dots, \mathcal{P}_l \leq \Sigma \leq \mathcal{P}_{l+1}, \dots, \mathcal{P}_k$ for a given integer l with $0 \leq l \leq k$ if and only if $\mathcal{S}_{\text{loose}}(\mathcal{H}, \text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l)) \leq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k))$.*

Proof (\Rightarrow) If Σ satisfies $\mathcal{P}_1, \dots, \mathcal{P}_l \leq \Sigma \leq \mathcal{P}_{l+1}, \dots, \mathcal{P}_k$ then

$$\text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l) \leq \Sigma \leq \text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k).$$

If, in addition, $\Sigma \subseteq \mathcal{H}$ then $\mathcal{S}_{\text{loose}}(\mathcal{H}, \text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l)) \leq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k))$ by Theorem 4.

(\Leftarrow) Conversely, suppose that

$$\mathcal{S}_{\text{loose}}(\mathcal{H}, \text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l)) \leq \mathcal{S}_{\text{lacy}}(\mathcal{H}, \text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k)).$$

By Theorem 4 there exists a set $\Sigma \subseteq \mathcal{H}$ with $\text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l) \leq \Sigma \leq \text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k)$. Since $\mathcal{P}_1, \dots, \mathcal{P}_k \leq \text{lub}(\mathcal{P}_1, \dots, \mathcal{P}_l)$ and $\text{glb}(\mathcal{P}_{l+1}, \dots, \mathcal{P}_k) \leq \mathcal{P}_{l+1}, \dots, \mathcal{P}_k$ it follows that

$$\mathcal{P}_1, \dots, \mathcal{P}_l \leq \Sigma \leq \mathcal{P}_{l+1}, \dots, \mathcal{P}_k.$$

□

Note that if $l = 0$, the properties (AM) hold for k phenotypic partitions, while if $l = k$, the properties (BM) hold for k phenotypic partitions.

5 Generating phenotypic partitions from edge-marked trees

In this final section, we return to the setting of single partitions and ask when a phenotypic partition of X can be realized by marking some edges of a given phylogenetic X -tree $T = (V, E)$, and assigning two individuals to the same phenotype if there is no marked edge on the path between them.

This process has a clear biological interpretation: a marked edge denotes that a new phenotype evolves to replace the existing phenotype, and it is assumed here that a phenotype that has already appeared will not appear a second time; in other words, the evolution of the phenotype is homoplasy-free. Therefore, we define a *marking map* $m : E \rightarrow \{0, 1\}$ on T as follows:

$$m(e) = \begin{cases} 1, & \text{if } e \in E \text{ is marked;} \\ 0, & \text{otherwise.} \end{cases}$$

Fig. 2 A rooted phylogenetic tree on which the phenotypic partition $\mathcal{P} = \{\{1, 3\}, \{2, 4\}\}$ cannot be realized by marking any subset of edges

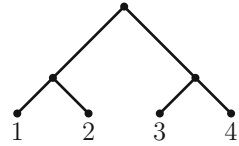
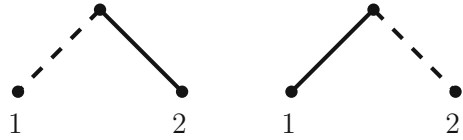


Fig. 3 A rooted phylogenetic tree with different labeled edges which induce the phenotypic partition $\mathcal{P} = \{\{1\}, \{2\}\}$. The dashed line is marked and the solid line is not



For $x, y \in X$ we define the relation \sim_m by $x \sim_m y$ if $m(e) = 0$ for every edge e on the path from x to y . This induces a corresponding phenotypic partition \mathcal{P}_m as the set of equivalence classes of X under \sim_m . Not all possible partitions of X can be realized in this way; an example for such a case is given in Fig. 2.

In general, a partition \mathcal{P} of X can be realized by a marking map on a given rooted phylogenetic tree T on X if and only if that partition is ‘convex’ on T (i.e. the minimal subtrees of T that connect the leaves in each block of \mathcal{P} are vertex-disjoint), for details, see Steel (2016). In general, there can be more than one marking map that leads to the same phenotypic partition, as Fig. 3 shows. For this reason, we define the relation \simeq by $m \simeq m'$ if $\mathcal{P}_m = \mathcal{P}_{m'}$.

There is a close connection between the property (M) for the phenotypic partition and its realization by marking maps, as we now describe.

Proposition 2 *Suppose that m is a marking map on a rooted phylogenetic tree T on X with associated hierarchy \mathcal{H} . $\mathcal{P}_m \subseteq \mathcal{H}$ if and only if for any two distinct leaves x, y with $x \sim_m y$, we have $m(e) = 0$ for every edge e in the subtree of T with root $\text{lca}_T(x, y)$.*

Proof For this proof, we introduce some further notation. Given a rooted phylogenetic tree T and a vertex v of T , let $\text{subtree}(T, v)$ denote the rooted phylogenetic tree which has root v and contains all vertices and edges which are descendants from v in T .

(\Rightarrow) Suppose that $\mathcal{P}_m \subseteq \mathcal{H}$ holds, and that x, y are distinct leaves of X with $x \sim_m y$. Let P be the block of \mathcal{P}_m containing x and y . Then $P \in \mathcal{H}$ (since $\mathcal{P}_m \subseteq \mathcal{H}$) and so $m(e) = 0$ for each edge in the subtree $(T, \text{lca}_T(P))$. Since $\text{lca}_T(P) \preceq_T \text{lca}_T(\{x, y\})$ it follows that $m(e) = 0$ for every edge e in the subtree $(T, \text{lca}_T(\{x, y\}))$, as claimed.

(\Leftarrow) Suppose that $\mathcal{P}_m \not\subseteq \mathcal{H}$ holds. Then there exists $P \in \mathcal{P}_m$ with $P \notin \mathcal{H}$. This means that there exists a leaf $x \in X$ in the subtree $(T, \text{lca}_T(P))$ with $x \notin P$. Select $y \in P$. Hence, there exists an edge e on the path from x to y , with $m(e) = 1$ and with e present in the subtree $(T, \text{lca}_T(\{x, y\}))$. \square

We now state a necessary condition, followed by a sufficient condition for a phenotypic partition to be realizable by a marking map on a given tree.

Proposition 3 *Let \mathcal{P} be a phenotypic partition of X and T be a rooted phylogenetic tree on X , with corresponding hierarchy \mathcal{H} .*

- (i) If \mathcal{P} can be realized by a marking map on T then at least one block P of \mathcal{P} is in \mathcal{H} .
- (ii) If $\mathcal{P} \subseteq \mathcal{H}$, then the phenotypic partition \mathcal{P} can be realized by a marking map.

Proof Part (i): Let \mathcal{P} be a phenotypic partition that can be realized by a marking map on T . If no edge is marked at all, then $\mathcal{P} = \{X\}$ and $X \in \mathcal{H}$ by definition. If there exists at least one marked edge, then at least one marked edge e has no marked edge descendant from it (i.e. there exists a block $P \in \mathcal{P}$ that consists of all descending leaves) and so $P \in \mathcal{H}$.

Part (ii): Suppose that $\mathcal{P} \subseteq \mathcal{H}$ holds. If $\mathcal{P} = \{X\}$ then set $m(e) = 0$ for each edge e of T . Otherwise, if $\mathcal{P} \neq \{X\}$, for each block $P \in \mathcal{P}$, let e_P be the edge of T that is directed into the vertex $\text{lca}_T(P)$. Then set $m(e) = 1$, if $e = e_P$ for some $P \in \mathcal{P}$ and set $m(e) = 0$ otherwise. This gives a marking map that realizes \mathcal{P} . \square

5.1 Maximum marking maps and the glb and the lub of multiple markings

In order to extend the study to multiple marking maps in partitions, we first require some further notation.

Given a partition \mathcal{P} and a rooted phylogenetic tree T we define a marking map $m_{\mathcal{P}}$ on T by:

$$m_{\mathcal{P}}(e) = \begin{cases} 0, & \text{if } e \text{ is on a path from } x \text{ to } y, x \sim y \text{ for } \mathcal{P}, x \neq y; \\ 1, & \text{otherwise.} \end{cases}$$

Recall here that $x \sim y$ for \mathcal{P} means that x, y are in the same block of the partition \mathcal{P} . It can be checked that the partition associated with $m_{\mathcal{P}}$ (i.e. $\mathcal{P}_{m'}$ for $m' = m_{\mathcal{P}}$) is equal to or refines \mathcal{P} in the lattice of partitions of X (i.e. $\mathcal{P}_{m'} \leq \mathcal{P}$) though $\mathcal{P}_{m'}$ need not equal \mathcal{P} (for example, in Fig. 2 with $\mathcal{P} = \{\{1, 3\}, \{2, 4\}\}$, the partition associated with $m_{\mathcal{P}}$ is $X = \{1, 2, 3, 4\}$).

Notice that any marking map m for a rooted tree $T = (V, E)$ is determined by the set

$$\mathcal{E}(m) := \{e \in E : m(e) = 1\},$$

consisting of the edges of T that are marked.

For a marking map m on T , we let \bar{m} be the marking map $m_{\mathcal{P}}$ in which \mathcal{P} is taken to be \mathcal{P}_m . Thus, $\bar{m}(e)$ takes the value 1, unless there is some pair $x, y \in X$ with $x \sim_m y$ for which e lies on the path connecting x and y . In particular, \bar{m} is the marking map that has the maximal number of edges assigned to state 1, while still inducing the same partition as \mathcal{P}_m (this is formalized in part (ii) of Lemma 2).

An example to illustrate this concept is provided in Fig. 4.

Lemma 2 *Let m and m' be marking maps on a rooted phylogenetic T on X with associated hierarchy \mathcal{H} .*

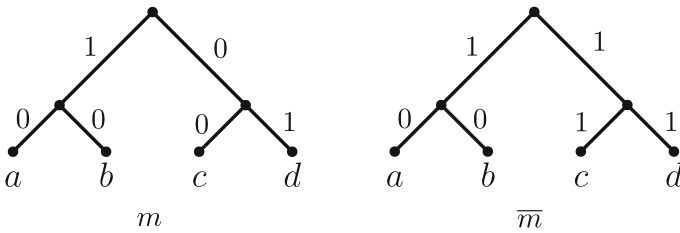


Fig. 4 For the marking map m described on the left, we have $\mathcal{P}_m = \{\{a, b\}, \{c\}, \{d\}\}$. The associated marking map \bar{m} is shown on the right

- (i) If $\mathcal{E}(m) \subseteq \mathcal{E}(m')$, then $\mathcal{P}_{m'} \preceq \mathcal{P}_m$.
- (ii) $m \simeq \bar{m}$. Moreover, if $m' \simeq m$, then $\mathcal{E}(m') \subseteq \mathcal{E}(\bar{m})$.
- (iii) Suppose that $\mathcal{P}_m \subseteq \mathcal{H}$ and that e, e' are edges of T with e' on the path from the root of T to e . If $e \in \mathcal{E}(\bar{m})$, then $e' \in \mathcal{E}(\bar{m})$.

Proof The proof of parts (i) and (ii) is straightforward. For part (iii), suppose that e' is not in $\mathcal{E}(\bar{m})$. Then, by definition of \bar{m} , there exist leaves $x, x' \in P \in \mathcal{P}_m$ for which the path from x to x' includes e' . By part (ii) of this lemma, $\mathcal{P}_m = \mathcal{P}_{\bar{m}}$ and so, by Proposition 2, $\bar{m}(e'') = 0$ for every edge e'' in the subtree of T with root $\text{lca}_T(x, x')$. However, edge e lies in this subtree, yet $\bar{m}(e) = 1$ so $e \notin \mathcal{E}(\bar{m})$. \square

Given marking maps m_1, \dots, m_k on a tree T , we let $m_1 \vee m_2 \vee \dots \vee m_k$ and $m_1 \wedge m_2 \wedge \dots \wedge m_k$ be the marking maps on T defined by:

$$\mathcal{E}(m_1 \vee m_2 \vee \dots \vee m_k) = \bigcup_{i=1}^k \mathcal{E}(m_i),$$

and

$$\mathcal{E}(m_1 \wedge m_2 \wedge \dots \wedge m_k) = \bigcap_{i=1}^k \mathcal{E}(m_i).$$

In words, $m_1 \vee m_2 \vee \dots \vee m_k$ is the marking map that assigns 1 to all edges of T that are marked 1 by at least one m_i ($1 \leq i \leq k$), while $m_1 \wedge m_2 \wedge \dots \wedge m_k$ assigns 1 only to the edges of T that are marked 1 by every m_i ($1 \leq i \leq k$).

The following result describes the glb and lub of a collection of partitions (relevant to Theorem 3) in the setting where each partition is derived from a marking map on a given tree, in terms of a single marking map on that tree.

Theorem 5 Consider a rooted phylogenetic tree $T = (V, E)$ with associated hierarchy \mathcal{H} , and marking maps $m_i : E \rightarrow \{0, 1\}$, for $i = 1, \dots, k$. Then

- (i) $\text{glb}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k}) = \mathcal{P}_{m_1 \vee m_2 \vee \dots \vee m_k}$.
- (ii) Provided that $\mathcal{P}_{m_i} \subseteq \mathcal{H}$ for each $1, \dots, k$, we also have:

$$\text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k}) = \mathcal{P}_{\bar{m}_1 \wedge \bar{m}_2 \wedge \dots \wedge \bar{m}_k}.$$

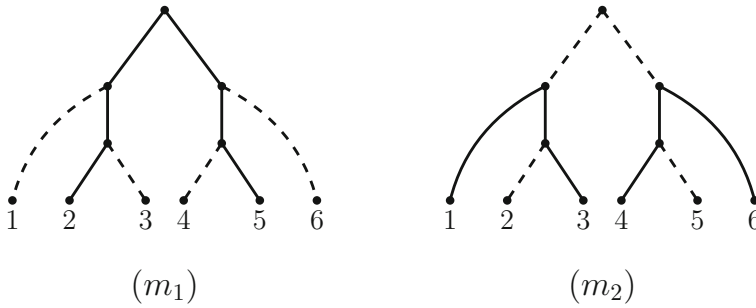


Fig. 5 Left: The marking map m_1 (where a dashed edge e corresponds to $m_1(e) = 1$) induces the phenotypic partition \mathcal{P}_{m_1} in which the only block of size > 1 is $\{2, 5\}$. Right: The marking map m_2 (where a dashed edge e corresponds to $m_2(e) = 1$) induces the phenotypic partition \mathcal{P}_{m_2} in which the only blocks of size > 1 are $\{1, 3\}$ and $\{4, 6\}$. Thus, the least upper bound (lub) of these two partitions contains the three blocks $\{1, 3\}, \{2, 5\}, \{4, 6\}$. However, the marking map $m_1 \wedge m_2$ assigns 0 to all the edges of the tree shown, and so $\mathcal{P}_{m_1 \wedge m_2} = X$. Note that in this example, $m_1 = \overline{m_1}$ and $m_2 = \overline{m_2}$

Before proceeding to the proof, we note that the additional assumption ($\mathcal{P}_{m_i} \subseteq \mathcal{H}$) in (ii) is required, even in the special case where $k = 2$ and $m_i = \overline{m_i}$ for $i = 1, 2$. An example to demonstrate this is shown in Fig. 5. In this example, $\text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k})$ is not realized by any marking map on T . Notice that Lemma 1 and Proposition 3(ii) ensure that when $\mathcal{P}_{m_i} \subseteq \mathcal{H}$ for each $1, \dots, k$, then $\text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k})$ is realized by at least some marking map on T , and Theorem 5(ii) gives an explicit description of such a map. Note also, in part (ii) that we have replaced m_i by $\overline{m_i}$ on the right-hand side. Without such a replacement, the identity in (ii) does not hold, as Fig. 3 shows.

Proof of Theorem 5 Part (i): First, we consider the case $k = 2$ of just two marking maps m_1 and m_2 on T . Since $\mathcal{E}(m_i) \subseteq \mathcal{E}(m_1 \vee m_2)$, for $i = 1, 2$, Lemma 2(i) gives $\mathcal{P}_{m_1 \vee m_2} \leq \mathcal{P}_{m_i}$ for $i = 1, 2$, and therefore:

$$\mathcal{P}_{m_1 \vee m_2} \leq \text{glb}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}).$$

To replace \leq with equality it remains to show that $\text{glb}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}) \leq \mathcal{P}_{m_1 \vee m_2}$, which means that if $S \in \text{glb}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2})$ and $S' \in \mathcal{P}_{m_1 \vee m_2}$ with $S \cap S' \neq \emptyset$ then $S \subseteq S'$. Recall (Sect. 1.2) that S is a block of the glb of two given partitions in the poset $\mathbb{P}(X)$ precisely if S is the (non-empty) intersection of a block from the first partition with a block from the second. Thus, we may suppose that $S = \beta_1 \cap \beta_2$ where $\beta_1 \in \mathcal{P}_{m_1}$ and $\beta_2 \in \mathcal{P}_{m_2}$. Since $S \cap S' \neq \emptyset$ select $x \in S \cap S'$. Then S' is the set of leaves of T whose path to x does not cross an edge marked 1 by either m_1 or m_2 (or both). On the other hand, β_i is the set of leaves of T whose path to x does not cross an edge marked 1 by m_i . Thus, if $y \in \beta_1 \cap \beta_2 = S$, then $y \in S'$, and so $S \subseteq S'$ as claimed.

The case where $k > 2$ now follows by the associativity of \vee on the lattice $(\mathbb{P}(X), \leq)$. We have

$$\text{glb}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k}) = \text{glb}(\mathcal{P}_{m_1}, \text{glb}(\mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k})),$$

and so part (i) follows by induction, from the case where $k = 2$. This establishes part (i) of Theorem 5.

Part (ii): Again, we first consider the case of $k = 2$ marking maps m_1 and m_2 on T . Note that $\mathcal{E}(\overline{m_1 \wedge m_2}) \subseteq \mathcal{E}(\overline{m_i})$ for $i = 1, 2$. So Lemma 2 gives $\mathcal{P}_{m_i} = \mathcal{P}_{\overline{m_i}} \preceq \mathcal{P}_{\overline{m_1 \wedge m_2}}$ for $i = 1, 2$ and therefore:

$$\text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}) \preceq \mathcal{P}_{\overline{m_1 \wedge m_2}}.$$

To replace \preceq with equality it remains to show that $\mathcal{P}_{\overline{m_1 \wedge m_2}} \preceq \text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2})$, which means that if $S \in \mathcal{P}_{\overline{m_1 \wedge m_2}}$ and $S' \in \text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2})$ with $S \cap S' \neq \emptyset$ then $S \subseteq S'$. To this end, suppose that $x \in S \cap S'$ and that $x \sim_{\overline{m_1 \wedge m_2}} y$. We will establish the following:

Claim 1 Given $\mathcal{P}_{m_1}, \mathcal{P}_{m_2} \subseteq \mathcal{H}$, if $x \sim_{\overline{m_1 \wedge m_2}} y$, then either $x \sim_{\overline{m_1}} y$ or $x \sim_{\overline{m_2}} y$ (or both) holds.

It follows from Claim 1 that $x \approx y$ (where \approx is the equivalence relation in the definition of lub from Sect. 1.2) and so $y \in S' \in \text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2})$, as required to show that $\mathcal{P}_{\overline{m_1 \wedge m_2}} \preceq \text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2})$, and thereby to establish part (ii) in the case $k = 2$.

Thus, for the $k = 2$ case, it remains to prove Claim 1. We do this by assuming that Claim 1 is false, and derive a contradiction. Now, if Claim 1 is false, then the path from x to y crosses at least one edge e_1 with $\overline{m_1}(e_1) = 1$ and at least one edge e_2 with $\overline{m_2}(e_2) = 1$, and there is no edge e on this path with $\overline{m_1}(e) = \overline{m_2}(e) = 1$ (since $x \sim_{\overline{m_1 \wedge m_2}} y$).

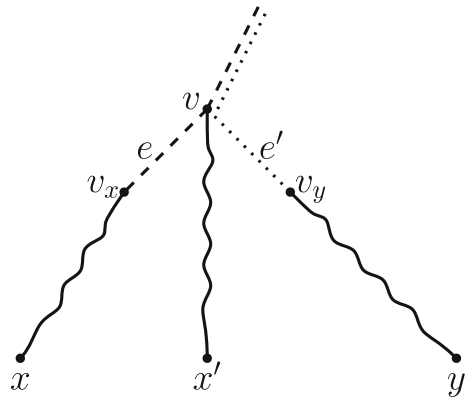
Let $v = \text{lca}_T(\{x, y\})$ and let v_x (respectively, v_y) be vertex adjacent to v that is on the path from v to x (respectively, to y). By Lemma 2(iii), it follows that the edge $e = (v, v_x)$ has $\overline{m_i}(e) = 1$ and $\overline{m_j}(e) = 0$, while the edge $e' = (v, v_y)$ has $\overline{m_i}(e) = 0$ and $\overline{m_j}(e) = 1$, where $\{i, j\} = \{1, 2\}$. Without loss of generality, we may assume that $i = 1$ and $j = 2$. The condition that $\overline{m_2}(e) = 0$ then implies (by the definition of $\overline{m_2}$) that e lies on a path connecting two leaves of T (which lie in the same block of $\mathcal{P}_{m_2} = \mathcal{P}_{\overline{m_2}}$) and every edge in that path has $\overline{m_2}$ value equal to zero. One of these two leaves is in the subtree of T below v_x , the other leaf x' must also lie below v since the edge e'' of T that ends in v has $\overline{m_1}(e'') = \overline{m_2}(e'') = 1$ by Lemma 2(iii). Moreover, x' cannot lie in the subtree with root v_y (since the path would then cross e' which has $\overline{m_2}(e') = 1$), nor can it be in the subtree with root v_x (since the path from x' to x has to cross e). Thus, x' must lie in a third pendant subtree that attaches below v , as indicated in Fig. 6.

But then Proposition 2 implies that $\mathcal{P}_{m_2} = \mathcal{P}_{\overline{m_2}}$ is not a subset of \mathcal{H} , since $x \sim_{\overline{m_2}} x'$ but $\overline{m_2}$ is not zero on every edge in the subtree $(T, \text{lca}_T(\{x, x'\}))$ since this subtree included the edge e' for which $\overline{m_2}(e') = 1$. This contradiction establishes Claim 1, and thereby the proof in the case where $k = 2$.

The case where $\overline{k} > 2$ now follows by the associativity of \wedge on the lattice $(\mathbb{P}(X), \preceq)$ and the fact that $\overline{\overline{m_i}} = \overline{m_i}$. We have

$$\text{lub}(\mathcal{P}_{m_1}, \mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k}) = \text{lub}(\mathcal{P}_{m_1}, \text{lub}(\mathcal{P}_{m_2}, \dots, \mathcal{P}_{m_k})),$$

Fig. 6 For the two leaves x, y for which $x \sim_{\overline{m_1} \wedge \overline{m_2}} y$ the edge e incident with $v = \text{lca}_T(\{x, y\})$ has $\overline{m_1}(e) = 1$ and $\overline{m_2}(e) = 0$ while the edge e' has $\overline{m_1}(e') = 0$ and $\overline{m_2}(e') = 1$ (each edge above v has an $\overline{m_1}$ and $\overline{m_2}$ value equal to 1). Every edge on the path from v to x , and on the path from v to x' has $\overline{m_2}$ value equal to 0, and so $x \sim_{\overline{m_2}} x'$



and so part (ii) follows by induction, from the case where $k = 2$. This establishes part (ii) of Theorem 5. \square

Concluding comments

Our purpose in this paper was twofold. First, we have represented some of the main results of Manceau and Lambert (2017) within the framework of lattice theory, which has allowed for more concise proofs, and elucidated the key combinatorial principles. Second, we have extended these results to the more general setting of $k \geq 2$ phenotypic partitions, and we have also explored the combinatorial aspects of representing phenotypic partitions by marking maps.

An interesting project for future development could be to investigate the loose and lacy species partitions arising when the phenotypic character(s) evolve on a tree T with leaf set X , and \mathcal{H} is the hierarchy associated with T . The evolution of phenotypic characters on T is typically modelled by discrete-state, continuous-time Markov processes that are already widely used in molecular phylogenetics (Felsenstein 2004). Finite state Markov models can lead to homoplasy (convergent or reverse changes), while infinite state models always produce homoplasy-free character states at the leaves. Under either model, the character states at the leaves then induce a phenotypic partition of X , and the stochastic properties of the loose and lacy species partition could then be studied.

A first step would be to simply calculate the probability that the loose and lacy partitions species coincide (i.e. a species partition satisfying all three properties (ABM), or, equivalently, each phenotype corresponds to a cluster of \mathcal{H}). It would also be of interest to predict the distribution of block sizes in species partitions, a topic that has long been of interest to biologists, dating back to Yule (1925) [see, for example, Scotland and Sanderson (2004)].

Finally, the definition and computation of loose and lacy species partitions based on phylogenetic networks (rather than phylogenetic trees) could be a further interesting direction for future work.

Acknowledgements We thank Amaury Lambert for helpful discussions, Mareike Fischer for comments on an earlier version of this paper, and the (former) Allan Wilson Centre for funding this work. We also thank the two anonymous reviewers for several helpful comments.

Appendix: Proof of Proposition 1

Let $\tilde{\mathcal{S}}_{\text{loose}}$ be the set of maximal elements of \mathcal{H}_1 and let $\tilde{\mathcal{S}}_{\text{lacy}}$ be set of maximal elements of \mathcal{H}_2 . We show that $\tilde{\mathcal{S}}_{\text{loose}} = \mathcal{S}_{\text{loose}}$ and $\tilde{\mathcal{S}}_{\text{lacy}} = \mathcal{S}_{\text{lacy}}$, by establishing the following properties:

- (i) $\tilde{\mathcal{S}}_{\text{loose}}$ and $\tilde{\mathcal{S}}_{\text{lacy}}$ are partitions of X ,
- (ii) $\tilde{\mathcal{S}}_{\text{loose}}$ satisfies (AM) and $\tilde{\mathcal{S}}_{\text{lacy}}$ satisfies (BM),
- (iii) $\tilde{\mathcal{S}}_{\text{loose}}$ is the finest partition of X that satisfies (AM) and $\tilde{\mathcal{S}}_{\text{lacy}}$ is the coarsest partition of X that satisfies (BM).

Proof Part (i):

1. Proof that $\tilde{\mathcal{S}}_{\text{loose}}$ is a partition of X .
Suppose that $x \in X$. Since \mathcal{P} is a partition of X , there is a set $P \in \mathcal{P}$ with $x \in P$, so $x \in h_P$ [where h_P is as in Eq. (3)]. Thus, x is contained in at least one maximal element of \mathcal{H}_1 (and so in some set of $\tilde{\mathcal{S}}_{\text{loose}}$). Moreover, two different maximal elements of \mathcal{H}_1 , say h and h' , have empty intersection. For otherwise, since $h, h' \in \mathcal{H}$, the nesting property of hierarchies implies that either $h \subsetneq h'$ or $h' \subsetneq h$ which is impossible if both sets are maximal.
2. Proof that $\tilde{\mathcal{S}}_{\text{lacy}}$ is a partition of X .
Suppose that $x \in X$. Then $x \in P$ for some set $P \in \mathcal{P}$, and since $h = \{x\} \in \mathcal{H}$, and $h \in \mathcal{P}$, \mathcal{H}_2 has a maximal set h' containing x , so $x \in h' \in \mathcal{H}_2$. Moreover, two different maximal elements of \mathcal{H}_2 have empty intersection, for the same reason as holds for \mathcal{H}_1 .

Part (ii):

1. Proof that $\tilde{\mathcal{S}}_{\text{loose}}$ satisfies (AM):
For any $h \in \tilde{\mathcal{S}}_{\text{loose}}$ we have $h = h_P$ for some $P \in \mathcal{P}$. Suppose that $Q \in \mathcal{P}$ has $Q \cap h \neq \emptyset$. Then $h_Q \cap h_P \neq \emptyset$ and so $h_Q \subseteq h_P$ or $h_P \subseteq h_Q$ (since h_Q and h_P are both elements of a hierarchy). The maximality condition in the definition of $\tilde{\mathcal{S}}_{\text{loose}}$ applied to $h = h_P$ now ensures that $h_Q \subseteq h_P$, so $Q \subseteq h_Q \subseteq h_P = h$ and so property (A) holds for $\tilde{\mathcal{S}}_{\text{loose}}$; moreover, since $h \in \mathcal{H}$, property (M) holds also.
2. Proof that $\tilde{\mathcal{S}}_{\text{lacy}}$ satisfies (BM):
For any $h \in \tilde{\mathcal{S}}_{\text{lacy}}$ there is a set $P \in \mathcal{P}$ with $h \subseteq P$. For any $Q \in \mathcal{P}$ with $Q \neq P$ we must have $Q \cap h = \emptyset$, since otherwise $P \cap Q \neq \emptyset$. Thus, property (B) holds for $\tilde{\mathcal{S}}_{\text{loose}}$; moreover, since $h \in \mathcal{H}$, property (M) holds also.

Part (iii):

1. Proof that $\tilde{\mathcal{S}}_{\text{loose}}$ is the finest species partition satisfying (AM):
This means for $\mathcal{P} \preceq \mathcal{W} \preceq \tilde{\mathcal{S}}_{\text{loose}}$ with $\mathcal{W} \subseteq \mathcal{H}$ it holds that $\mathcal{W} = \tilde{\mathcal{S}}_{\text{loose}}$.
Suppose this is not the case (we will derive a contradiction). Then there must exist a set $h_P \in \tilde{\mathcal{S}}_{\text{loose}}$ for which h_P is the disjoint union of $k \geq 2$ non-empty

sets h_1, h_2, \dots, h_k in \mathcal{W} . Since $\mathcal{P} \preceq \mathcal{W}$, if $P \cap h_i \neq \emptyset$ we have $P \subseteq h_i$ and so $h_P \subseteq h_i$, which implies that $h_P = h_i$. Thus, P can intersect at most one of the sets h_i , and so it must intersect exactly one set, say h_1 , since the union of h_1, \dots, h_k is h_P which contains P . But this implies that each of the remaining $k - 1$ sets is empty (since the union is disjoint), contradicting the requirement that the sets h_i are non-empty.

2. Proof that $\tilde{\mathcal{S}}_{\text{lacy}}$ is the coarsest species partition satisfying (BM):

This means for $\tilde{\mathcal{S}}_{\text{lacy}} \preceq \mathcal{V} \preceq \mathcal{P}$ with $\mathcal{V} \subseteq \mathcal{H}$ it holds that $\mathcal{V} = \tilde{\mathcal{S}}_{\text{lacy}}$.

Suppose this is not the case (we will derive a contradiction). Then there must exist some element $h \in \mathcal{V}$ and some element $h' \in \tilde{\mathcal{S}}_{\text{lacy}}$ with $h' \subsetneq h$. Let P be an element of \mathcal{P} with $h \subseteq P$ (such an element must exist since $h \in \mathcal{V} \preceq \mathcal{P}$). Then h and h' are both elements of \mathcal{H}_2 (since they are both elements of \mathcal{H} and are contained within P) and since $h' \in \tilde{\mathcal{S}}_{\text{lacy}}$ is a maximal element of \mathcal{H}_2 we cannot have $h' \subsetneq h$, which provides the required contradiction. \square

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