GEOMETRY OF THE KIMURA 3-PARAMETER MODEL

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ABSTRACT. The Kimura 3-parameter model on a tree of $n$ leaves is one of the most used in phylogenetics. The affine algebraic variety $W$ associated to it is a toric variety. We study its geometry and we prove that it is isomorphic to a geometric quotient of the affine space by a finite group acting on it. As a consequence, we are able to study the singularities of $W$ and prove that the biologically meaningful points are smooth points. Then we give an algorithm for constructing a set of minimal generators of the localized ideal at these points, for an arbitrary number of leaves $n$. This leads to a major improvement of phylogenetic reconstruction methods based on algebraic geometry.

1. Introduction

The goal of phylogenetic algebraic geometry is to translate the knowledge of algebraic geometry into new tools for phylogenetic inference problems. The dictionary used in this translation is based on algebraic statistics, which allows viewing statistical evolutionary models as algebraic varieties. The first approaches in this direction are due to Allman and Rhodes [AR03] and Pachter and Sturmfels [PS04]. Since then, many other authors have contributed to the development of phylogenetic algebraic geometry, either from the more geometric point of view (see for instance [ERSS05], [SS05], [AR07], [WB06], [CS05]) or from the applied standing point ([ES93], [Eri05], [CFS07]). The base of algebraic statistics for computational biology were finally set up in the book [PS05].

The applications of algebraic geometry to phylogenetics rely on the computation of the generators of the ideal of the algebraic variety associated to a statistical evolutionary model on a phylogenetic tree $T$. In phylogenetics, these generators are called phylogenetic invariants as they are useful to infer the topology of the tree $T$ (note that in phylogenetics, topology refers to the topology of the graph $T$ with labels at the leaves). Phylogenetic invariants have been given for some algebraic evolutionary models, namely the general Markov model ([AR07]) and group-based models (Kimura models [Kim80], [Kim81], and Jukes-Cantor model [JC69]).
In this paper, we deal with the Kimura 3-parameter model. As it was shown in [SS05], the Kimura 3-parameter model on a tree of $n$ species is a toric variety in a suitable coordinate system (Fourier coordinates). Sturmfels and Sullivant gave an algorithm to construct a set of generators of the ideal of this variety for any number of species $n$. For example, for four species a set of minimal generators contains 8002 binomials of degrees 2, 3 and 4. In a previous paper, we proved that this set of binomials can be successfully used for phylogenetic inference (see [CFS07]). However, this is a large number of generators if one considers that the codimension of the variety is 48. Moreover, as the number of species increases, the codimension increases exponentially but the number of generators given in [SS05] increases more than exponentially. This makes phylogenetic reconstruction methods based on this set of generators unfeasible for larger trees.

The main goal here is to prove that the points of biological interest are smooth points of the algebraic variety and to provide the generators of a local complete intersection at these points. To this end, we prove that this Kimura variety $W$ is isomorphic to the quotient of a certain affine space under the action of a finite group (Corollary 3.7). This result allows the study of the singular locus of $W$ and shows that there are no singularities in the points with biological meaning (Corollary 3.13). We use this result in section 4, where we provide a recursive procedure to give a minimal sequence of generators for the variety $W$ near these points (Theorem 4.5). As an example, the whole list of these generators in the case of trees with 4 leaves is given (Example 4.8).

In the paper [SSEW93] the authors also provided a local complete intersection for the Kimura 3-parameter model (they called it a complete collection of invariants). In their case the degree of the generators increases exponentially on the number of leaves $n$ and this makes it unfeasible to be used in a phylogenetic reconstruction method for large trees. Our set of generators for the local complete intersection consists of binomials of degree 2 and 4 for any number of leaves $n$ and leads to some hope for the generalization of the method given in [CGS05] to arbitrary trees. It is worth mentioning that Hagedorn [Hag00] also realized that there exists an open set of the variety in which it is sufficient to consider a local complete intersection (this is clear if one knows that the set of singular points on a variety form a Zarisky closed subset). However he did not specify the open subset nor the set of generators.

This paper is organized as follows. In section 2 we review the relation between algebraic geometry and statistical evolutionary models for phylogenetic inference. In this section as well, we recall the discrete Fourier transform (or Hadamard conjugation) introduced by Evans and Speed (see [ES93]) as a linear change of coordinates which diagonalizes group-based models. Then we introduce the algebraic varieties we are interested in and we set up notation used in the sequel. Section 3 is devoted to the global study of the geometry of the Kimura variety and to determine its
singular points. In section 4 we perform a local study of the variety at the biological meaningful points and we give an algorithm to obtain the generators of a local complete intersection at these points.

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2. Preliminaries

Let $T$ be a tree (i.e. a connected undirected acyclic graph) of $n$ leaves labelled as $1, 2, \ldots, n$. The degree of a node in $T$ is defined as the number of edges incident to it. Nodes of degree one are leaves $L(T)$, while the others are internal nodes $N(T)$. We assume that our trees are trivalent, i.e. internal nodes have degree 3, and we call $E(T)$ the set of edges in $T$. An edge in $T$ is said to be terminal if it contains one leaf. We write $e_l$ for the terminal edge ending at leaf $l$, $l \in \{1, \ldots, n\}$. It is easy to see that the number of internal nodes is $|N(T)| = n - 2$ and the number of edges is $|E(T)| = 2n - 3$.

2.1. Algebraic evolutionary models. In phylogenetics, a tree $T$ represents the ancestral relationships (edges) among a set of species (nodes). The leaves $L(T)$ represent the current species whose phylogenetic history we wish to infer. The input data is an alignment of $n$ sequences in the alphabet $\Sigma := \{A, C, G, T\}$ (representing nucleotides) of length $N$, and one needs to infer the correct phylogenetic tree that produced the observed alignment.

In order to explain the relationship between phylogenetic inference and algebraic geometry it is useful to assume for the moment that the tree is rooted. That is, the graph $T$ is directed and it has a unique not trivalent node called the root of the tree with two edges emerging from it. This assumption will be removed in subsection 2.3.

From the biological standing point, Kimura 3-parameter model is a stationary Markov model of evolution. Kimura [Kim81] proposed a statistical model of evolution under the following assumptions: all sites in the $n$ sequences evolve equally and independently, the distribution of nucleotides at the root is uniform and the tree is stationary (and hence all nodes of the tree have uniform distribution of nucleotides), the evolution of a species depends only of the node immediately preceding it, mutations occur randomly and with strictly positive probabilities, and transitions (mutations between purines $A, T$ or between pyrimydines $C, G$) occur more often than transversions (mutations between purines and pyrimydines). As all sites evolve independently and in the same way, one restricts the model to one site. We describe here an algebraic version of this model (see the books [PS05] and [AR04] for an introduction to the algebraic versions of evolutionary models).
In statistical evolutionary models, to each node \( v \) of the tree \( T \) we associate a discrete random variable \( X_v \) that takes values on \( \Sigma = \{A, C, G, T\} \). The parameters of algebraic evolutionary models are the substitution probabilities of nucleotides along each edge. These parameters are written in a matrix indexed by the alphabet elements \( \Sigma \) so that the matrix \( S^e \) associated to the edge \( e \) is

\[
S^e = \begin{pmatrix}
A & C & G & T \\
A & P(A|A, e) & P(C|A, e) & P(G|A, e) & P(T|A, e) \\
C & P(A|C, e) & P(C|C, e) & P(G|C, e) & P(T|C, e) \\
G & P(A|G, e) & P(C|G, e) & P(G|G, e) & P(T|G, e) \\
T & P(A|T, e) & P(C|T, e) & P(G|T, e) & P(T|T, e)
\end{pmatrix}
\]

where \( S^e_{x,y} = P(x \mid y, e) \) is the probability that nucleotide \( y \) at the parent of edge \( e \), \( s(e) \), mutates to nucleotide \( x \) at the descendant node \( t(e) \). Then the probability of observing nucleotides \( x_1 \ldots x_n \) at the leaves of the tree is given by a Markov process:

\[
p_{x_1 \ldots x_n} = \sum_{\{x_v \in \Sigma \mid v \in N(T)\}} \prod_{e \in E(T)} S^e_{x_{t(e)}, x_{t(e)}}
\]

where we assume that if \( e = e_1 \) is a terminal edge, then \( x_{t(e)} = x_1 \). In the Kimura 3-parameter model the substitution matrices have the following form

\[
S^e = \begin{pmatrix}
a^e & b^e & c^e & d^e \\
b^e & a^e & d^e & c^e \\
c^e & d^e & a^e & b^e \\
d^e & c^e & b^e & a^e
\end{pmatrix}
\]

where \( a^e + b^e + c^e + d^e = 1 \). This model includes the more restrictive models of Jukes-Cantor (where \( b^e = c^e = d^e \), [JC69]) and Kimura 2-parameter model (\( b^e = d^e \), [Kim80]).

In the algebraic geometry setting, the Kimura 3-parameter model is given by the polynomial map

\[
\prod_{e \in E(T)} \Delta^3 \rightarrow \Delta^{4^n - 1}
\]

\[
((a^e, b^e, c^e, d^e))_{e \in E(T)} \mapsto (p_{x_1 \ldots x_n})_{x_1, \ldots, x_n \in \Sigma^n}
\]

where \( p_{x_1 \ldots x_n} \) is given by (2.1) and \( \Delta^d \) denotes the standard \( d \)-dimensional simplex in \( \mathbb{R}^{d+1} \). As we are interested in algebraic varieties, instead of restricting to the simplex, we also consider this polynomial map as

\[
\prod_{e \in E(T)} \mathbb{C}^4 \rightarrow \mathbb{C}^{4^n}
\]

\[
((a^e, b^e, c^e, d^e))_{e \in E(T)} \mapsto (p_{x_1 \ldots x_n})_{x_1, \ldots, x_n \in \Sigma^n}.
\]

One of the goals in phylogenetic algebraic geometry is determining the ideal of the closure of the image of this polynomial map. Knowing the generators of this ideal provides tools for phylogenetic inference. See for example [CFS07] and [Eri05].
where some of these methods for phylogenetic inference have been proposed. In order to find the generators of this ideal, it is extremely useful to perform change of coordinates as we explain below.

2.2. Fourier transform. The models described above are known as group-based models because if the nucleotides are thought of as the elements of the group

\[ H = \mathbb{Z}/(2) \times \mathbb{Z}/(2) \]

(namely \( A = (0, 0), C = (1, 0), G = (0, 1), T = (1, 1) \)) then the entries \( \{ S_{g,h}^e \}_{g,h \in H} \) in the substitution matrices \( S^e \) can be expressed as functions of the group \( f^e(h - g) \) (see \[SS05\] for details). For the Kimura 3-parameter model, the function \( f^e \) is

\[
\begin{cases}
  a^e & \text{if } h = (0, 0) \\
  b^e & \text{if } h = (1, 0) \\
  c^e & \text{if } h = (0, 1) \\
  d^e & \text{if } h = (1, 1)
\end{cases}
\]

As a consequence, probabilities \( p_{x_1 \ldots x_n} \) can also be thought of as functions on \( H \times \cdots \times H \). In what follows, when we add nucleotides, we mean addition in the group \( H \).

One of the main properties of group-based models is that a discrete Fourier transform simplifies the expression in the probabilities (2.1). We briefly recall how this Fourier transform works and we refer to \[SS05\] and \[CGS05\] for more details. Given a function \( f : H \rightarrow \mathbb{C} \), its discrete Fourier transform is the function \( \hat{f} : H^\lor = \text{Hom}(H, \mathbb{C}^*) \rightarrow \mathbb{C} \) defined by

\[
\hat{f}(\chi) = \sum_{g \in H} \chi(g) f(g).
\]

The Fourier transform turns convolution into multiplication and this allows to simplify the expression of joint probabilities:

**Theorem 2.1** (Evans-Speed \[ES93\]). Let \( p(g_1, \ldots, g_n) \) be the joint distribution of a group-based model for a phylogenetic tree \( T \), then its Fourier transform has the form

\[
q(\chi_1, \ldots, \chi_n) = \prod_{e \in E(T)} \hat{f}^e \left( \prod_{l \in \{ \text{leaves below } e \}} \chi_l \right)
\]

As \( H \) and its dual \( H^\lor \) are isomorphic groups we can identify \((\chi_1, \ldots, \chi_n)\) with the corresponding tuple \((g_1, \ldots, g_n)\). From now on, \( q(\chi_1, \ldots, \chi_n) \) will be denoted as \( q_{g_1 \ldots g_n} \). In the additive notation of the group \( H \), one can rewrite expression (2.2) as

\[
q_{g_1 \ldots g_n} = \prod_{e \in E(T)} \hat{f}^e(m(e))
\]
where \( m(e) = \sum_{l \in \text{leaves below } e} g_l \). The Fourier transform is a linear coordinate change given by

\[
q_{g_1 \ldots g_n} = \sum_{j_1, \ldots, j_n} \chi^{g_1}(j_1) \cdots \chi^{g_n}(j_n)p_{j_1 \ldots j_n}
\]

where \( \chi^j \) is the character of the group associated to the \( i \)th group element:

|   | \( A \) | \( C \) | \( G \) | \( T \) |
|---|---|---|---|---|
| \( \chi^A \) | 1 | 1 | 1 | 1 |
| \( \chi^C \) | 1 | -1 | 1 | -1 |
| \( \chi^G \) | 1 | 1 | -1 | -1 |
| \( \chi^T \) | 1 | -1 | -1 | 1 |

In this new coordinate system, the Fourier transforms \( \hat{f}^e \) of the substitution functions are the new parameters of the model. For the Kimura 3-parameter model, these Fourier transforms become

\[
\hat{f}^e(h) = \begin{cases} 
a^e + b^e + c^e + d^e & \text{if } h = (0, 0) 
a^e - b^e + c^e - d^e & \text{if } h = (1, 0) 
a^e + b^e - c^e - d^e & \text{if } h = (0, 1) 
a^e - b^e - c^e + d^e & \text{if } h = (1, 1) 
\end{cases}
\]

As before, we think of these substitution functions as matrices. Therefore, the parameters in Fourier coordinates are diagonal matrices

\[
P^e = \begin{pmatrix} P_A^e & 0 & 0 & 0 
0 & P_C^e & 0 & 0 
0 & 0 & P_G^e & 0 
0 & 0 & 0 & P_T^e \end{pmatrix}
\]

where \( P_A^e = a^e + b^e + c^e + d^e \), \( P_C^e = a^e - b^e + c^e - d^e \), \( P_G^e = a^e + b^e - c^e - d^e \), \( P_T^e = a^e - b^e - c^e + d^e \).

From now on, \( P^e \) will indistinctly denote this diagonal matrix or the vector \((P_A^e, P_C^e, P_G^e, P_T^e)\) and we will restrict to Fourier coordinates. It is not difficult to see that if \( g_1 + \cdots + g_n \neq 0 \), then \( q_{g_1 \ldots g_n} = 0 \) (cf. Proposition 29 of [SS05]). Therefore, the polynomial map we are interested in is

\[
\varphi : \prod_{e \in E(T)} \mathbb{C}^4 \rightarrow \mathbb{C}^{4^n-1}
\]

\[
(P_A^e, P_C^e, P_G^e, P_T^e) \mapsto (q_{x_1, \ldots, x_n})_{x_1, \ldots, x_n}
\]

where \( x_1 + \cdots + x_n = 0 \), \( q_{x_1, \ldots, x_n} = \prod_{e \in E(T)} P_{m(e)}^e \), and \( m(e_l) = x_l \) if \( e_l \) ends at leaf \( l \).

**Notation 2.2.** The image of the standard simplex \( \Delta^d \) under the Fourier change of coordinates will be denoted as \( \Delta^d \). For a picture of \( \Delta^3 \), see figure \[\text{figure 3}\]. Notice that the Fourier change of parameters transforms the hyperplane \( a^e + b^e + c^e + d^e = 1 \) into \( P_A^e = 1 \), for all \( e \in E(T) \). As we will be interested in coordinates \( \{q_{x_1, \ldots, x_n}\}_{x_1 + \cdots + x_n = 0} \), we
will focus on the simplex $\Delta^{4^{n-1}}$, which coincides with the projection of the Fourier transform of the simplex $\triangle^{4^{n-1}}$ onto this set of coordinates. As before, note that the Fourier change of coordinates transforms the hyperplane $\sum_{i} p_{i} = 1$ into $q_{A...A} = 1$.

2.3. Kimura variety. In subsections 2.1 and 2.2 we have assumed that the tree is rooted. However, in the Kimura 3-parameter model the matrices $S^e$ are symmetric and therefore parameterization (2.4) does not depend on the orientation of the tree or the position of the root. One can even think of the root as being one of the leaves and in this case the root is one of the observed variables. In what follows we will consider unrooted trees. This is due to the issue of *identifiability* that induces the use of unrooted trees for phylogenetic inference. Roughly speaking, the question addressed by identifiability is whether observation data of character states at the leaves of the tree contain enough information in order to uncover the topology and the parameters of the model (see [Cha96]). This means that there precisely exists one topology and one set of parameters of the model that explain the data. The identifiability of the Kimura 3-parameter model has been established by Steel, Hendy and Penny in [SHP98].

The reformulation of the parameterization in Fourier coordinates for unrooted trees is given by the lemma below. Before stating it, we introduce some notation:

**Notation 2.3.** Given an interior node $v \in N(T)$, denote by $e_{v}^{1}, e_{v}^{2}, e_{v}^{3}$ the three edges coincident at $v$ (see (a) in figure 1). Given three elements of the group $x_{e_{v}^{1}}, x_{e_{v}^{2}}, x_{e_{v}^{3}} \in H$ associated to these edges, we define

$$x(v) := x_{e_{v}^{1}} + x_{e_{v}^{2}} + x_{e_{v}^{3}}$$

as a sum in $H$. 

![Figure 1](image-url)
Lemma 2.4. Let $T$ be an (unrooted) tree with $n$ leaves. Then the parameterization of the Kimura 3-parameter model in Fourier coordinates is given by

$$q_{x_1 \ldots x_n} = \prod_{e \in E(T), x(v) = 0 \forall v \in N(T)} P_{x_e}^e,$$

where $x_1 + \cdots + x_n = 0$ and $x_{e_i} = x_i$ if $e_i$ is the terminal edge corresponding to the leaf $l$.

Proof. We already know that the parameterization for rooted trees is independent of the root placement, so we root the tree $T$ at leaf 1. Then we need to prove that

$$\prod_{e \in E(T), x(v) = 0 \forall v \in N(T)} P_{x_e}^e = \prod_{e \in E(T)} P_{m(e)}^e.$$

In other words, we want to prove that the condition $x(v) = 0$ for all interior nodes $v \in N(T)$ is equivalent to $x_e = m(e)$ for all edges $e \in E(T)$.

We first assume that for all interior nodes $v$, $x(v) = 0$. We proceed by induction on the number of leaves of the tree. Let $w$ be the only node next to the leaf 1 and let $e^1_w$ be the terminal edge connecting 1 and $w$. Write $T_2$ and $T_3$ for the two connected components obtained when removing $e^1_w$ from $T$ and adding $W$ as a root (see (b) of figure 1). By induction hypotheses on $T_2$ and $T_3$, we have $x_{e^2_w} = m(e^2_w)$, $x_{e^3_w} = m(e^3_w)$, and $x_e = m(e)$ for all edges but $e_1$. It remains to check that $x_{e_1} = m(e_1)$. This follows using the hypothesis that $x(w) = 0$. Indeed, $x(w) = 0$ implies $x_1 = x_{e^2_w} + x_{e^3_w}$, which in turn is equal to $m(e^2_w) + m(e^3_w)$ and hence equal to $x_2 + \cdots + x_n$.

In order to prove the converse we assume that $x_e = m(e)$ for all edges $e \in E(T)$. By induction hypothesis on the trees $T_2$ and $T_3$, the condition $x(v) = 0$ holds for all interior nodes in $T$ but $w$. We just need to show that $x_{e_1} + x_{e^2_w} + x_{e^3_w} = 0$. Our hypothesis implies that $x_{e_1} + x_{e^2_w} + x_{e^3_w} = m(e_1) + m(e^2_w) + m(e^3_w)$, and this vanishes because $m(e_1) = m(e^2_w) + m(e^3_w)$ by definition of $m$. \[\blacksquare\]

Remark 2.5. In expression (2.5), the indices $x_e$ associated to edge $e$ are completely determined by condition $x(v) = 0$, $\forall v \in N(T)$. Indeed, as at the terminal edges $e_l$ we have imposed $x_e = x_l$, condition $x(v) = 0$ for nodes that join a cherry to the tree determines the value $x_e$ at those edges that join a cherry to the tree. Performing the same process from the exterior of the tree to the interior, one assigns a unique value to every edge. Condition $x_1 + \cdots + x_n = 0$ guarantees that this assignment is consistent at all interior nodes.

Example 2.6. For the unrooted 3-leaf claw tree (see (a) of figure 2), the parameterization $\varphi$ of (2.4) in Fourier coordinates is given by

$$q_{x_1 x_2 x_3} = P_{x_1}^{e_1} P_{x_2}^{e_2} P_{x_3}^{e_3}$$

if $x_1 + x_2 + x_3 = 0$. 

Example 2.7. For the unrooted tree with 4 leaves (see (b) of figure 2) the parameterization in Fourier coordinates is given by

\begin{equation}
q_{x_1x_2x_3x_4} = P_{x_1}^{e_1} P_{x_2}^{e_2} P_{x_3}^{e_3} P_{x_4}^{e_4}
\end{equation}

if \(x_1 + x_2 + x_3 + x_4 = 0\).

Notation 2.8. From now on, we write \(V\) for the closure of the image of

\begin{equation}
\varphi : \prod_{e \in E(T)} \mathbb{C}^4 \longrightarrow \mathbb{C}^{4^n-1}
\end{equation}

\( (P_{A}^{e}, P_{C}^{e}, P_{G}^{e}, P_{T}^{e})_{e} \mapsto (q_{x_1...x_n})_{x_1+...+x_n=0} \)

where

\begin{equation}
q_{x_1...x_n} = \prod_{e \in E(T), x(v)=0 \forall v \in N(T)} P_{x_e}^{e},
\end{equation}

and \(x_{e_l} = x_l\) if \(e_l\) is the terminal edge corresponding to leaf \(l\). We will denote \(Q_n\) the following set of indeterminates

\begin{equation}
Q_n = \{q_{x_1...x_n} | x_1 + \cdots + x_n = 0 \text{ in } H\}.
\end{equation}

The affine coordinate ring \(A(V)\) of \(V\) is isomorphic to the \(\mathbb{C}\)-algebra

\begin{equation}
\mathbb{C}[\prod_{e \in E(T)} P_{x_e}^{e} | x_{e_l} = x_l \forall l \in N(T)]_{x_1...x_n \in \Sigma},
\end{equation}

because \(A(V)\) is defined as the image of the morphism of \(\mathbb{C}\)-algebras

\begin{equation}
\theta : \mathbb{C}[Q_n] \longrightarrow \mathbb{C}[\{P_{x_e}^{e} | e \in E(T), x \in \Sigma\}]
\end{equation}

\[ q_{x_1...x_n} \mapsto \prod_{e \in E(T), x(v)=0 \forall v \in N(T)} P_{x_e}^{e} \]
where \( x_{e_l} = x_l \) for all \( l \in N(T) \). The toric ideal defining \( V \) is the kernel of this morphism and we denote it as \( I_V \). Sturmfels and Sullivant gave an algorithm to construct a set of generators of \( I_V \) in \([SS05]\). We note that as the map \( \theta \) is homogeneous, \( V \) is actually a cone over a projective variety.

The variety we are interested in is \( V \cap \{q_{A\ldots A} = 1\} \) because the simplex in Fourier coordinates is contained in the hyperplane \( q_{A\ldots A} = 1 \).

**Definition 2.9.** The Kimura variety of the phylogenetic tree \( T \) is

\[ W := V \cap \{q_{A\ldots A} = 1\} \]

**Lemma 2.10.** The Kimura variety \( W \) is the closure of the parameterization \( \varphi \) restricted to \( \prod_{e \in E(T)} (\mathbb{C}^4 \cap \{P_A^e = 1\}) \).

**Proof.** Write \( \varphi_1 \) for this restriction, and let \( \rho \) be the morphism of \( \mathbb{C} \)-algebras defined as:

\[
\begin{align*}
\rho : \mathbb{C}[Q_n] &\longrightarrow \mathbb{C}[Q_n \setminus \{q_{A\ldots A}\}] \\
f(q_{A\ldots A}, q_{A\ldots C}, \ldots) &\mapsto f(1, q_{A\ldots C}, \ldots)
\end{align*}
\]

Then the affine coordinate ring of \( W \) is

\[
A(W) = \mathbb{C}[Q_n]/(I_V + (q_{A\ldots A} - 1)) \cong \mathbb{C}[Q_n \setminus \{q_{A\ldots A}\}]/(\rho(I_V)).
\]

On the other hand, note that \( \varphi_1 \) induces a morphism of \( \mathbb{C} \)-algebras

\[
\theta_1 : \mathbb{C}[Q_n \setminus \{q_{A\ldots A}\}] \longrightarrow \mathbb{C}[\{P_x^e \mid e \in E(T) \text{ and } x \in \Sigma \setminus \{A\}\}]
\]

so that the following diagram commutes

\[
\begin{array}{ccc}
0 & \longrightarrow & I_V \\
\downarrow \rho & & \downarrow \rho \\
0 & \longrightarrow & \mathbb{C}[Q_n \setminus \{q_{A\ldots A}\}] \\
& & \downarrow \rho' \\
& & \mathbb{C}[\{P_x^e \mid e \in E(T) \text{ and } x \in \Sigma \setminus \{A\}\}]
\end{array}
\]

Here \( \rho' \) sends \( P_A^e \) to 1 for each \( e \in E(T) \) and is the identity on the other indeterminates. Write \( X \subset \mathbb{C}^4 \) for the closure of the image of \( \varphi_1 \). Then, the affine coordinate ring of \( X \) is \( A(X) = \mathbb{C}[Q_n]/\text{Ker}(\rho' \circ \theta) \). Since the above diagram commutes, we have that

\[
\rho' \circ \theta = \theta_1 \circ \rho = \rho^{-1}(\rho(I_V)) = I_V + (q_{A\ldots A} - 1)
\]

and \( X = W \). \( \square \)

### 2.4. Biologically meaningful points

Here, we introduce some notation and we give a biological interpretation on the points of some polytopes in the simplices defined in subsection 2.2. As above, let \( T \) be a phylogenetic tree with \( n \) leaves and write \( E(T) \) for the set of its edges. For any \( e \in E(T) \), let \( \Delta^3 \) be the parameter simplex associated to it (see Notation 2.2). Write

\[
\Delta^3_0 = \{ P = (1, P_C, P_G, P_T) \in \Delta^3 \mid P_C, P_G, P_T \geq 0 \}
\]
Figure 3. The polytope $\Delta^3_0$ in the simplex $\Delta^3$. It represents the points of $\Delta^3$ having non-negative Fourier coordinates.

for the set of non-negative points in $\Delta^3$ i.e. the polytope delimited by the vertex $(1, 1, 1, 1)$, the points $(1, 1, 0, 0), (1, 0, 1, 0), (1, 0, 0, 1)$ and the centroid $(1, 0, 0, 0)$ of $\Delta^3$ (see figure 3), all of them written in Fourier coordinates. We write also $\Delta^3_+ = \{ P \in \Delta^3 \mid P^c, P^g, P^t > 0 \}$. Any point of $\Delta^3_0$ represents a substitution matrix $S^e$ satisfying that the probability of no mutation ($a^e$) plus the probability of any mutation in the site ($b^e, c^e$ or $d^e$) is bigger or equal than $1/2$. In particular, the probability of no mutation is bigger or equal than the probability of any particular mutation. Since this is a reasonable hypothesis if we work with realistic data, we will call the points in $\prod_{e \in E(T)} \Delta^3_+$ the *biological meaningful parameters of the model*. We also write

\[\varphi_+ : \prod_{e \in E(T)} \Delta^3_+ \rightarrow W\]

\[\left( P^a, P^c, P^g, P^t \right)_e \mapsto (q_{x_1\ldots x_n})_{x_1\ldots x_n}\]

for the restriction of $\varphi$ in (2.4) to these parameters. Its image is contained in $W_+ := W \cap \Delta_+$, where $\Delta_+ = \{ q \in \Delta^{4^n-1} \mid q_{x_1\ldots x_n} > 0, \sum_i x_i = 0 \}$ is the set of points with positive coordinates of the polytope delimited by the point $1_n = (1, \ldots, 1)$ (which is one of the vertices on the simplex $\Delta^{4^n-1}$), the points $q_i = (1, e_i)_{i=1,\ldots,4^n-1}$ where $e_i = (0, \ldots, 1, \ldots, 0) \in \mathbb{C}^{4^n-1}$ and the centroid $(1,0,\ldots,0)$ of $\Delta^{4^n-1}$. We call the points of $W_+$ the *biologically meaningful points of the model*. This name will be justified in forthcoming Remark 3.12 where we show that $W_+$ equals the image of $\varphi_+$.

3. The geometry of Kimura 3-parameter model

Let $V \subset \mathbb{C}^{4^n}$ be the affine variety associated to a tree $T$ of $n$ leaves, as defined above. In this section we are going to determine the singular points of the Kimura
variety \( W = V \cap \{ q_A \ldots A = 1 \} \). To this aim, we will first prove that \( V \) is isomorphic to the quotient of \( (\mathbb{C}^4)^{2n-3} \) by the action of a certain abelian group.

In order to simplify notation, we briefly recall the notion of **multigrading** and refer to Chapter 8 of [MS05] for a nice introduction to multigraded polynomial rings (we also refer to [MS05] for the correspondence between toric varieties and quotients, although we need little preliminary knowledge in this subject for the results of this section.)

**Notation 3.1.** Let \( M \) be a monomial in \( S = \mathbb{C}[\{ P^e_x \mid e \in E(T) \text{ and } x \in \Sigma \}] \). Then, \( M \) has the form \( M = \prod_{e \in E(T), x \in \Sigma} (P^e_x)^{i(e)_x} \) where each \( i(e) = (i(e)_A, i(e)_C, i(e)_G, i(e)_T) \) is composed of natural numbers. This notation means that

\[
M = \prod_{e \in E(T), x \in \Sigma} (P^e_x)^{i(e)_x}.
\]

We call \( \text{deg}(i(e)) = i(e)_A + i(e)_C + i(e)_G + i(e)_T \). Each indeterminate in \( S \) has a natural multidegree in \( \mathbb{Z}^{\left| E(T) \right|} \) defined as

\[
\text{deg}(P^e_x) = (0, \ldots, 0, 1, 0, \ldots, 0) \quad \text{for any } x \in \Sigma.
\]

Given a monomial \( M \in S \) as above, we call \( \text{deg}(M) := (\text{deg}(i(e)))_{e \in E(T)} \). Note that the image of \( \theta \) of (2.6) is generated by monomials of degree \( d \cdot (1, \ldots, 1) \), so that they are multi-homogeneous with respect to the given grading.

**Notation 3.2.** From now on, \( \mathbb{Z}/(2) \) means additive group whereas \( \mathbb{Z}_2 \) means multiplicative group.

### 3.1. The 3-leaves case

We start by studying the case \( n = 3 \) (see example 2.6). We call \( V_3 \) the corresponding affine variety in \( \mathbb{C}^{16} \). The parameterization \( \varphi \) in this case is:

\[
\varphi : \mathbb{C}^{12} \to \mathbb{C}^{16} \hspace{1cm} (\text{or} (P^e_A, P^e_C, P^e_G, P^e_T)) \to (P^{e_1}_A P^{e_2}_C P^{e_3}_G P^{e_3}_T)_{\{x+y+z=0\mid x,y,z \in \mathbb{Z}_2\}}
\]

In the next result we prove that \( V_3 \) is an affine GIT quotient [MS05] chapter 10.

**Proposition 3.3.** \( V_3 \) is isomorphic to the affine GIT quotient \( \mathbb{C}^{12}/G \) where the group

\[
G = \{ (\lambda_1, \lambda_2, \lambda_3, \varepsilon, \delta) \mid | \lambda_1 \in \mathbb{C}^*, (\varepsilon, \delta) \in \mathbb{Z}_2 \times \mathbb{Z}_2, \lambda_1 \lambda_2 \lambda_3 = 1 \} \simeq (\mathbb{C}^*)^2 \times \mathbb{Z}_2 \times \mathbb{Z}_2
\]

acts on \( \mathbb{C}^{12} \) sending \( (P^e_A, P^e_C, P^e_G) \) to

\[
(\lambda_1(P^e_A, \varepsilon P^e_A, \delta P^e_A), \lambda_2(P^e_C, \varepsilon P^e_C, \delta P^e_C), \lambda_3(P^e_G, \varepsilon P^e_G, \delta P^e_G)).
\]

In order to prove this proposition, we need a technical lemma that we state separately for future reference.
Lemma 3.4. Let \( i = (i_A, i_C, i_G, i_T) \), \( j = (j_A, j_C, j_G, j_T) \), \( k = (k_A, k_C, k_G, k_T) \) be 4-tuples in \( \mathbb{N}^4 \). Then the set of indices \( (i, j, k) \) that satisfy

\[
\begin{cases}
i_A + i_C + i_G + i_T = 1 \\
j_A + j_C + j_G + j_T = 1 \\
k_A + k_C + k_G + k_T = 1 \\
i_C + i_T + j_C + j_T + k_G + k_T = 0 \text{ in } \mathbb{Z}/(2) \\
i_G + i_T + j_G + j_T + k_G + k_T = 0 \text{ in } \mathbb{Z}/(2)
\end{cases}
\]

is equal to

\[
\{(i, j, k) \mid i_x = 1, j_y = 1, k_z = 1, \deg(i) = \deg(j) = \deg(k) = 1, x + y + z = 0 \text{ in } H\}.
\]

Proof. Let \( (i, j, k) \) satisfy (3.1). The first three equations imply that for each index, there is just one letter in \( \Sigma \) such that the corresponding entry is non-zero, and in fact, equal to one. We write \( i_x = 1 \), \( j_y = 1 \), \( k_z = 1 \). As in the first section we think of the letters in \( \Sigma \) as elements in \( \mathbb{Z}/(2) \times \mathbb{Z}/(2) \): \( A = (0, 0) \), \( C = (1, 0) \), \( G = (0, 1) \), \( T = (1, 1) \). We call

\[
\begin{align*}
I_{AC} &= i_A + i_C, & I_{CT} &= i_C + i_T, & I_{GT} &= i_G + i_T, \\
J_{AC} &= j_A + j_C, & J_{CT} &= j_C + j_T, & J_{GT} &= j_G + j_T, \\
K_{AC} &= k_A + k_C, & K_{CT} &= k_C + k_T, & K_{GT} &= k_G + k_T.
\end{align*}
\]

In this setting, \( i_x = 1 \) if and only if \( x = (I_{CT}, I_{GT}) \) in \( \mathbb{Z}/(2) \times \mathbb{Z}/(2) \). Similarly, \( j_y = 1 \) (resp. \( k_z = 1 \)) if and only if \( y = (J_{CT}, J_{GT}) \) (resp. \( z = (K_{CT}, K_{GT}) \)) in \( \mathbb{Z}/(2) \times \mathbb{Z}/(2) \). The last two equations in (3.1) can be written as

\[
\begin{cases}
I_{CT} + J_{CT} + K_{CT} = 0 \text{ in } \mathbb{Z}/(2) \\
I_{GT} + J_{GT} + K_{GT} = 0 \text{ in } \mathbb{Z}/(2)
\end{cases}
\]

They imply that \( x + y + z = 0 \) in \( \mathbb{Z}/(2) \times \mathbb{Z}/(2) \).

As for the other inclusion, if \( (i, j, k) \) are three 4-tuples in \( \mathbb{N}^4 \) of degree 1 (\( \deg(i) = \deg(j) = \deg(k) = 1 \)) whose non-vanishing indices \( i_x = 1, j_y = 1, j_z = 1 \) satisfy \( x + y + z = 0 \). Then the first three equations in (3.1) are clearly satisfied. The last two equations also hold because \( (i_C + i_T, i_G + i_T) = x, (j_C + j_T, j_G + j_T) = y, (k_C + k_T, k_G + k_T) = z \) and \( x + y + z = 0 \) in \( \mathbb{Z}/(2) \times \mathbb{Z}/(2) \). \( \square \)

We prove the proposition above.

Proof of Proposition 3.3. Recall that \( \mathbb{C}^{12}/G \) is defined as \( \text{Spec}(S^G) \), the spectrum of the ring of invariants \( S^G \) where \( S = \mathbb{C}[P_A^{e_1}, \ldots, P_T^{e_1}, P_A^{e_2}, \ldots, P_T^{e_2}, P_A^{e_3}, \ldots, P_T^{e_3}] \). This ring is generated as a \( \mathbb{C} \)-algebra by those monomials invariant by the action of \( G \).

Monomials in \( S \) are of the form \( (P^{e_1})^i(P^{e_2})^j(P^{e_3})^k \) where \( i, j, k \) are sets of natural numbers \( i = (i_A, i_C, i_G, i_T), j = (j_A, j_C, j_G, j_T), k = (k_A, k_C, k_G, k_T) \) and \( (P^{e_1})^i \) means \( (P_A^{e_1})^i(P_C^{e_1})^i(P_G^{e_1})^i(P_T^{e_1})^i \). A monomial is invariant under the action of \( G \).
if and only if for any \((\lambda_1, \lambda_2, \lambda_3, \sigma = (\varepsilon, \delta))\) in \(G\) we have
\[
\lambda_1^{i_A + \cdots + i_T} \lambda_2^{j_A + \cdots + j_T} \lambda_3^{k_A + \cdots + k_T} \varepsilon^{i_C + i_T + j_C + j_T + k_C + k_T} \delta^{i_G + i_T + j_G + j_T + k_G + k_T} = 1
\]
This happens if and only if
\[
\begin{cases}
  i_A + \cdots + i_T = j_A + \cdots + j_T = k_A + \cdots + k_T \\
  i_C + i_T + j_C + j_T + k_C + k_T = 0 \text{ in } \mathbb{Z}_2 \\
  i_G + i_T + j_G + j_T + k_G + k_T = 0 \text{ in } \mathbb{Z}_2
\end{cases}
\]
Therefore \(S^G\) is minimally generated as a \(\mathbb{C}\)-algebra by those monomials \((P^e_1)^i (P^e_2)^j (P^e_3)^k\) that satisfy
\[
\begin{cases}
  i_A + i_C + i_G + i_T = 1 \\
  j_A + j_C + j_G + j_T = 1 \\
  k_A + k_C + k_G + k_T = 1 \\
  i_C + i_T + j_C + j_T + k_C + k_T = 0 \text{ in } \mathbb{Z}_2 \\
  i_G + i_T + j_G + j_T + k_G + k_T = 0 \text{ in } \mathbb{Z}_2
\end{cases}
\]
By the lemma following this proof, this set of monomials is precisely
\[
\{P^e_1 P^e_2 P^e_3 | x + y + z = 0 \text{ in } \mathbb{Z}/(2) \times \mathbb{Z}/(2)\}.
\]
Therefore \(S^G\) is the finitely generated \(\mathbb{C}\)-algebra
\[
\mathbb{C}[\{P^e_1 P^e_2 P^e_3 | x + y + z = 0 \text{ in } \mathbb{Z}/(2) \times \mathbb{Z}/(2)\}]
\]
which is isomorphic to the affine coordinate ring of \(V\). \hfill \Box

### 3.2. The general case.

Now we generalize Proposition 3.3 to trees with an arbitrary number \(n\) of leaves. Recall that the number of edges in such a tree is \(2n - 1\).

By a path \(\sigma\) in \(T\) we mean a minimal subgraph of \(T\) connecting two nodes (interior nodes or leaves). We write \(\sigma = \{s_1, \ldots, s_r\}\) for the sequence of edges in \(\sigma\). Let
\[
G \subset (\mathbb{C}^* \times \mathbb{Z}_2 \times \mathbb{Z}_2)^{2n-3}
\]
be defined as the subset composed of the elements \((\lambda_e, \varepsilon_e, \delta_e)_{e \in E(T)}\) such that \(\prod_{e \in E(T)} \lambda_e = 1\) and satisfying the following condition
\[
(*) \text{ for any path } \sigma = \{s_1, \ldots, s_r\} \text{ between two leaves in } T, \prod_{i=1}^{r} \varepsilon_{s_i} = 1 \text{ and } \prod_{i=1}^{r} \delta_{s_i} = 1.
\]
It is immediate to see that the natural product induces a group structure in \(G\).

Moreover, we have

**Lemma 3.5.** The group \(G\) is isomorphic to \((\mathbb{C}^*)^{2n-4} \times (\mathbb{Z}_2 \times \mathbb{Z}_2)^{n-2}\).

**Proof.** For any interior node \(v\) of \(T\), let \(e^1_v, e^2_v, e^3_v\) be the edges incident at \(v\), and write \(\varepsilon(v) = \{\varepsilon_v, \delta_v\}_{v \in E(T)}\) by taking \(\varepsilon_{e^1_v} = \varepsilon_{e^2_v} = \varepsilon_{e^3_v} = -1\), \(\varepsilon_v = 1\) for the remaining edges. Similarly, we define \(\delta(v)\). Let \(e_0 \in E(T)\) and take the ring homomorphism
\[
\psi: (\mathbb{C}^*)^{|E(T)|-1} \times (\mathbb{Z}_2 \times \mathbb{Z}_2)^{|N(T)|} \rightarrow (\mathbb{C}^* \times \mathbb{Z}_2 \times \mathbb{Z}_2)^{2n-3}
\]

**Proof.** For any interior node \(v\) of \(T\), let \(e^1_v, e^2_v, e^3_v\) be the edges incident at \(v\), and write \(\varepsilon(v) = \{\varepsilon_v, \delta_v\}_{v \in E(T)}\) by taking \(\varepsilon_{e^1_v} = \varepsilon_{e^2_v} = \varepsilon_{e^3_v} = -1\), \(\varepsilon_v = 1\) for the remaining edges. Similarly, we define \(\delta(v)\). Let \(e_0 \in E(T)\) and take the ring homomorphism
\[
\psi: (\mathbb{C}^*)^{|E(T)|-1} \times (\mathbb{Z}_2 \times \mathbb{Z}_2)^{|N(T)|} \rightarrow (\mathbb{C}^* \times \mathbb{Z}_2 \times \mathbb{Z}_2)^{2n-3}
\]
defined by mapping \(((\mu_e)_{e \in E(T)}, (\varepsilon(v), \delta_e)_{v \in V(T)})\) to \((\lambda_e, \varepsilon_e, \delta_e)_{e \in E(T)}\), where \(\lambda_e = \mu_e\) if \(e \neq e_0\), \(\lambda_{e_0} = (\prod_{e \neq e_0} \lambda_e)^{-1}\) and \(\varepsilon_e = \prod_{v \in e} \varepsilon(v)\), \(\delta_e = \prod_{v \in e} \delta(v)\). The image of \(\psi\) is \(G\) and it is easy to check that \(\psi\) is a monomorphism. The claim follows. □

The main result of this section is the following theorem.

**Theorem 3.6.** Let \(T\) be a tree with \(n\) leaves and let \(G\) be the group defined above. Let \(G\) act on \(\prod_{e \in E(T)} \mathbb{C}^4\) by sending \((P_A^e, P_C^e, P_G^e, P_T^e)_{e \in E(T)}\) to

\[
(\lambda_e(P_A^e, \varepsilon_eP_C^e, \delta_eP_G^e, \varepsilon_e\delta_eP_T^e))_{e \in E(T)}.
\]

Then \(V\) is isomorphic to \((\mathbb{C}^4)^{2n-3}/\!\!/G\).

**Proof.** We need to check that the affine coordinate rings of \(V\) and \((\mathbb{C}^4)^{2n-3}/\!\!/G\) are isomorphic. If \(S\) is the algebra \(S = \mathbb{C}[[\{P_x^e \mid e \in E(T)\} \text{ and } x \in \Sigma]]\), we need to check that the ring of invariants \(S^G\) is isomorphic to

\[
\mathbb{C}[\prod_{e \in E(T), x(v) = 0 \forall v \in N(T)} P_x^e \mid x_l \in \Sigma, x_{e_l} = x_l \forall l \in \{1, \ldots, n\}].
\]

Let \(M \in S\) be a monomial. Then, \(M\) has the form

\[
M = \prod_{e \in E(T)} (P_e)^{i(e)},
\]

with the notation introduced in 3.1. This monomial is invariant by the action of \(G\) if and only if for any \((\lambda_e, \varepsilon_e, \delta_e)_{e \in E(T)} \in G\) we have

\[
1 = \prod_{e \in E(T)} \lambda_e^{\deg(i(e))} \prod_{e \in E(T)} \varepsilon_e^{i(e)C + i(e)T} \prod_{e \in E(T)} \delta_e^{i(e)G + i(e)T}.
\]

As \(\prod_{e \in E(T)} \lambda_e = 1\), equation (3.2) implies \(\deg(i(e)) = \deg(i(e'))\) for all \(e, e' \in E(T)\) (in the language of the previous section, this means that \(M\) is multi-homogeneous). Therefore the algebra \(S^G\) is generated by monomials that satisfy \(\deg(i(e)) = 1\) for all edges \(e \in E(T)\). We assume from now on that \(M\) satisfies this condition.

Let \(v\) be an interior node of \(T\), and let \(e_1^v, e_2^v, e_3^v\) be the edges incident at \(v\). If we take \(\varepsilon_{e_1} = \varepsilon_{e_2} = \varepsilon_{e_3} = -1\) (resp. \(\delta_{e_1} = \delta_{e_2} = \delta_{e_3} = -1\)) and \(\varepsilon = 1\) (resp. \(\delta = 1\)) for the remaining edges, condition (*) is satisfied. For this particular choice, equation (3.2) implies

\[
\begin{align*}
\begin{cases}
i(e_1^v)_C + i(e_1^v)_T + i(e_2^v)_C + i(e_2^v)_T + i(e_3^v)_C + i(e_3^v)_T = 0 \text{ in } \mathbb{Z}/(2) \\
i(e_1^v)_G + i(e_1^v)_T + i(e_2^v)_G + i(e_2^v)_T + i(e_3^v)_G + i(e_3^v)_T = 0 \text{ in } \mathbb{Z}/(2)
\end{cases}
\end{align*}
\]

Lemma 3.3 tells us that \(M = \prod_{e \in E(T), x(v) = 0 \forall v \in N(T)} P_x^e\).

We need to prove the converse: if \(M = \prod_{e \in E(T), x(v) = 0 \forall v \in N(T)} P_x^e\) for some given \(x_1, \ldots, x_n\), we shall check that it is invariant by the action of \(G\). In other words, if
\[\{i_e\}_{e \in E(T)}\] denotes the set of exponents in \(M\), we are going to check that equation (3.2) holds. By lemma 3.4, condition \(x_{e_1} + x_{e_2} + x_{e_3} = 0\) is equivalent to

\[(3.3)\]

\[i(e_1)C + i(e_1)T + i(e_2)G + i(e_3)G + i(e_3)T = 0 \text{ in } \mathbb{Z}/(2)\]

\[i(e_1)G + i(e_1)T + i(e_2)G + i(e_2)T + i(e_3)T = 0 \text{ in } \mathbb{Z}/(2)\]

**Claim:** If condition (3.3) holds for any \(v \in N(T)\), the sets \(\gamma_{CT} = \{e \in E(T) \mid i(e)C + i(e)T = 0\}\) and \(\gamma_{GT} = \{e \in E(T) \mid i(e)G + i(e)T = 0\}\) are unions of disjoint paths between leaves of \(T\).

**Proof:** If \(x_i = A\) for all leaves, then the set of conditions \(x_{e_1} + x_{e_2} + x_{e_3} = 0\) lead to \(x_e = A\) for all \(e \in E(T)\), so there is nothing to prove in this case. We assume that there is a leaf \(v\) such that \(x_i \neq A\) and we assume that \(x_i \in \{C, T\}\) (if \(x_i \in \{G, T\}\) we proceed analogously). Then \(e_i\) belongs to \(\gamma_{CT}\).

Let \(v\) be the interior node connecting the edge \(e_i\) to the rest of the tree. As \(e_i\) is one of the edges intersecting at \(v\), condition \(i(e_i)C + i(e_i)T + i(e_i)G + i(e_i)T = 0 \text{ in } \mathbb{Z}/(2)\) implies that one of the other two edges emerging from \(v\) also belongs to \(\gamma_{CT}\). We call this edge \(e_vw\) and \(w\) is the other extreme of the edge. Then for \(w\) condition \(i(e_v)C + i(e_v)T + i(e_v)G + i(e_v)T = 0 \text{ in } \mathbb{Z}/(2)\) again implies that one of the other two edges emerging from \(w\) belongs to \(\gamma_{CT}\). We repeat this process until we end at another leaf of \(T\). We note that any two paths obtained this way are disjoint because an interior node cannot have three edges in \(\gamma_{CT}\). Therefore the claim is proved.

The claim immediately implies that the monomial \(M\) satisfies equation (3.2), since elements in the group \(G\) are defined by the condition (*). \(\square\)

**Corollary 3.7.** The coordinate ring of the Kimura variety \(W = V \cap \{q_{A...A} = 1\}\) is isomorphic to \(S^G\) where

\[S' = \mathbb{C}[\{P^e_x \mid e \in E(T) \text{ and } x \in \Sigma \setminus \{A\}\}]\]

and \(G' = (\mathbb{Z}_2 \times \mathbb{Z}_2)^{n-2}\) acts as a subgroup of the group \(G\) defined in Theorem 3.6. Equivalently, \(W\) is the affine GIT quotient of \(\prod_{e \in E(T)}(\mathbb{C}^4 \cap \{P^e_A = 1\})\) modulo \(G'\),

\[\prod_{e \in E(T)}(\mathbb{C}^4 \cap \{P^e_A = 1\})//G'.\]

**Proof.** Using diagram (2.7) we see that \(A(W)\) is isomorphic to \(\rho(A(V))\). By Theorem 3.6 we know that \(A(V) \simeq S^G\) and it is enough to prove \(\rho(S^G) \simeq S^{G'}\).

We first prove \(\rho(S^G) \subseteq S^{G'}\). Let \(M((P^e_A, P^e_C, P^e_G, P^e_T)_{e \in E(T)})\) be a monomial in \(S^G\). Then \(\rho(M) = M((1, P^e_C, P^e_G, P^e_T)_{e \in E(T)})\). Let \(g' = (\varepsilon_e, \delta_e)_{e \in E(T)}\) be an element in \(G'\). We have that the action of \(g\) in \(\rho(M)\) is \(g \cdot \rho(M) = M((1, \varepsilon_e P^e_C, \delta_e P^e_G, \varepsilon_e \delta_e P^e_T)_{e \in E(T)})\). Take \(g = (1, \varepsilon_e, \delta_e)_{e \in E(T)}\), which is an element of \(G\), and notice that

\[g \cdot M = g' \cdot \rho(M).\]
As $M$ is invariant by the action of $G$, it is also invariant by this element $g'$. Hence $g \cdot \rho(M) = \rho(M)$.

In order to prove the other inclusion we will use the multigrading notation introduced in \ref{grassmannian}. Let $M((P^e_C, P^e_G, P^e_T)_{e \in E(T)}) = \prod_{e \in E(T)} (P^e)^{k(e)}$ be a monomial in $S'G'$. As $S'$ is a subring of $S$, there is also a multidegree associated to $M$, namely deg($M$) = (deg($i(e)$))$_{e \in E(T)}$. Now we make $M$ multi-homogeneous: let $D$ be equal to max$_{e \in E(T)}$ deg($i(e)$) and consider the monomial $\rho := \prod_{e \in E(T)} (P^e)^{d-deg(i(e))} M$. Then $\rho$ is a monomial in $S$ invariant by the action of $G$ because $M$ was invariant by $G'$ and $\rho$ is multi-homogeneous (so that equation \ref{multigrading} holds). Moreover $\rho(N) = M$ and we are done. □

Remark 3.8. It is worth pointing out that the action of $\mathbb{Z}_2 \times \mathbb{Z}_2$ on $\Delta^3$:

$$(g, P^e) = ((\varepsilon, \delta), (1, P^e_C, P^e_G, P^e_T)) \mapsto (1, \varepsilon P^e_C, \delta P^e_G, \varepsilon \delta P^e_T).$$

is just the reflection relative to some of the axis going through the centroid of $\Delta^3$. Namely, the actions of $g_1 = (-1, 1)$, $g_2 = (1, -1)$ and $g_3 = (-1, -1)$ are the reflections relative to the $P_C$-axis, the $P_T$-axis and the $P_T$-axis, respectively. Thus, if we write $\Delta_{x, y} = \{P \in \Delta^3 | P^e_x, P^e_y \leq 0\}$ for any $x, y \in \Sigma$, then $g_1(\Delta^3_0) = \Delta^3_{C,T}$, $g_2(\Delta^3_0) = \Delta^3_{C,T}$ and $g_3(\Delta^3_0) = \Delta^3_{C,T}$.

Corollary 3.9. The Kimura variety $W$ is the geometric quotient

$$\prod_{e \in E(T)} (\mathbb{C}^4 \cap \{P^e_A = 1\})/G'$$

and coincides with the image of $\varphi_1$ (cf. Lemma \ref{grassmannian}).

Proof. By Corollary \ref{grassmannian} the variety $W$ is the categorical quotient defined by $\text{Spec}(S')G'$. As $G'$ is a finite group, the orbits of $G'$ are closed and this categorical quotient is precisely the geometric quotient $\prod_{e \in E(T)} (\mathbb{C}^4 \cap \{P^e_A = 1\})/G'$ and therefore, it coincides with the image of $\varphi_1$ (see Example 6.1 of \cite{dol}.

From this, we deduce the identifiability of the model (see subsection \ref{identifiability} and \cite{cha}). In particular, we have:

Corollary 3.10. The Kimura variety $W \subset \mathbb{C}^{4n-1}$ has dimension $3(2n-3)$ and codimension $4n-1 - 6n + 9$.

Corollary 3.11. Let $q$ be a point in the Kimura variety $W$. Then

$$|\varphi^{-1}(q)| \leq 4^{n-2}$$

and the equality holds for generic points. The same holds for $q \in W^R = \varphi((\mathbb{R}^3)_{2n-3})$ or $q \in W_\Delta = \varphi((\Delta^3)_{2n-3})$.  

Proof. By Corollary 3.9, $W$ is the image of $\varphi$ in the commutative diagram

$$
\begin{array}{c}
\prod_{e \in E(T)}(\mathbb{C}^4 \cap \{P^e_A = 1\}) \\
\downarrow \pi \\
\prod_{e \in E(T)}(\mathbb{C}^4 \cap \{P^e_A = 1\}) / G'
\end{array} \xrightarrow{\varphi} W
$$

It follows that if $q \in W$, $\varphi^{-1}(q)$ consists of one point $(1, P^e_C, P^e_G, P^e_T)_{e \in E(T)}$ and its images under the action of $G'$. Since $|G'| = 4^{n-2}$, the claim follows. Moreover, the image of a point in $\mathbb{R}^3$ (resp. $\Delta^3$) under the action of $G'$ stays in $\mathbb{R}^3$ (resp. $\Delta^3$).

Remark 3.12. Notice that the pre-images by $\varphi_1$ of the point $1_n = (1, \ldots, 1)$ are

$$
\varphi^{-1}(p) = \{(1, \varepsilon_1 P^e_C, \delta_1 P^e_G, \varepsilon_2 P^e_T)_{e \in E(T)} \mid (\varepsilon_1, \delta_1) \in G'\}.
$$

Among all of them, there is only one with biological interest: $((1,1,1,1)_{e \in E(T)})$, which represents the situation where, in probability parameters, the transition matrices of all edges are equal to the identity (no mutation occurs). In general, for any point $q \in W_+$ with real coordinates, there is one just preimage of biological interest.

Keeping the notation of subsection 2.4, it follows from Corollary 3.9, that $\varphi_+$ is injective and that it is actually a bijection onto $W_+$. This fact justifies the name biologically meaningful points given to the points of $W_+$. The following result tells us that if $q \in W_+\Delta$ has only positive coordinates, then $q$ is non-singular and $|\varphi^{-1}(q)| = 4^{n-2}$. Among all these pre-images, just the single one in $\prod_{e \in E(T)} \Delta^3_+$ has biological meaning.

Corollary 3.13. A point $q = \varphi_0(p) \in W_+\Delta$ is singular if and only if there is some $e \in E(T)$ such that $P^e \in \Delta^3 \cap \{P_C^e P_G^e P_T^e = 0\}$. In particular, no point with biological meaning is singular.

Proof. It is well-known that the singular points on $W$ are the points $\{q \in W \mid |\varphi^{-1}(q)| < 4^{n-2}\}$, i.e. those points for which at least one of their pre-images is invariant by the action of some $g \in G'$. By Remark 3.8, we know that these are precisely the points lying on an axis $P^e_x = 0$ for some $e \in E(T)$ and some $x \neq A$. As a consequence, the points in $W_+ = \varphi(\prod_{e \in E(T)} \Delta^3_+)$ are non-singular.

4. Local Complete Intersection

Given a tree with $n$ leaves, the main purpose of this section is to describe a procedure to determine a local complete intersection equal to the variety $W = W_n$ in the open set $\Delta_+$.

Notation 4.1. For every $n \in \mathbb{N}$, we will write $c(n) = 4^{n-1} - 6n + 9$. Note that in virtue of Corollary 3.10, $c(n)$ is the codimension of $W_n$. 


In Corollary 3.13 we have seen that the points of \((W_n)_+\) are non-singular. Since any regular local ring is a complete intersection, the variety \(W_n\) is a \textit{local complete intersection} at these points, i.e. the ideal \(I_W\) can be generated by \(c(n)\) polynomials in a neighborhood of these points or more precisely, a minimal system of generators for the localization of \(I_W\) in these points consists of \(c(n) = 4^n-1 - 6n + 9\) elements.

The following lemma provides a minimal system of generators for this ideal in case the tree \(T\) has \(n = 3\) leaves.

**Proposition 4.2.** Let \(T\) be a tree with 3 leaves and let \(W_3 \subset \mathbb{C}^{16}\) be the model associated to it. Then, the set of quartics

\[
\begin{align*}
&h_1 = q_{AAA}q_{ATT}q_{TCG}q_{TGC} - q_{ACC}q_{AGG}q_{AT}q_{TTA} \\
&h_2 = q_{CAA}q_{CTG}q_{TAT}q_{TGC} - q_{CAC}q_{CGT}q_{TCC}q_{TTA} \\
&h_3 = q_{AGG}q_{ATT}q_{ACC}q_{CCA} - q_{AAA}q_{ACC}q_{CGT}q_{CTG} \\
&h_4 = q_{ACC}q_{ATT}q_{AGG}q_{GGA} - q_{AAA}q_{AGC}q_{GCT}q_{GTC} \\
&h_5 = q_{CAC}q_{CTG}q_{GCT}q_{GGA} - q_{CAA}q_{CGT}q_{GAC}q_{GTC} \\
&h_6 = q_{GGA}q_{GTC}q_{ATT}q_{GCG} - q_{GGG}q_{GCT}q_{GTC}q_{TTA}
\end{align*}
\]

\(\)together with the equation \(h = q_{AAA} - 1\) is a local minimal system of generators for the ideal of \(W_3\) at the points of \((W_3)_+\). Namely, \(\{h_1, h_2, h_3, h_4, h_5, h_6, q_{AAA} - 1\}\) generate the ideal \(I_{W_3}\) in the local ring \(\mathcal{O}_{W,q}\), for any \(q \in (W_3)_+\).

**Remark 4.3.** It is worth pointing out that the minimal system of generators given in Proposition 4.2 does not depend on the point \(q \in (W_3)_+\). In the same way, the local complete intersection we will construct for an arbitrary tree of \(n\) leaves will be the same for all points in \((W_n)_+\).

**Proof of Proposition 4.2.** Let \(W' \subset \mathbb{C}^{16}\) be the variety defined by the zero set of the ideal \(\{h_1, h_2, h_3, h_4, h_5, h_6, h = q_{AAA} - 1\}\) and let \(q \in (W_3)_+\). Let \(\text{Jac}_W(q)\) be the Jacobian matrix of \(W'\) at \(q\):

\[
\begin{pmatrix}
 q_{AAA} & q_{ACC} & q_{AGG} & q_{ATT} & q_{CAC} & q_{CCA} & q_{CGT} & q_{CTG} & q_{AGA} & q_{GCT} & q_{ATC} & q_{TAT} & q_{TCG} & q_{TCC} & q_{TTA}
 h_1 & * & * & * & 0 & 0 & 0 & 0 & 0 & 0 & * & * & * & * & * \\
 h_2 & 0 & 0 & 0 & 0 & * & * & * & 0 & 0 & 0 & * & * & * & * \\
 h_3 & * & * & * & * & * & * & * & 0 & 0 & 0 & 0 & 0 & 0 & 0 \\
 h_4 & * & * & * & 0 & 0 & 0 & 0 & * & * & * & 0 & 0 & 0 & 0 \\
 h_5 & 0 & 0 & 0 & 0 & * & * & * & * & * & * & 0 & 0 & 0 & 0 \\
 h_6 & 0 & 0 & 0 & 0 & 0 & 0 & * & * & * & * & * & * & * & * \\
 h & 1 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0 & 0
\end{pmatrix}
\]

with entries

\[
\begin{align*}
(h_1, q_{AAA}) &= q_{ATT}q_{TCG}q_{TGC} \\
(h_1, q_{ACC}) &= -q_{AGG}q_{AT}q_{TTA} \\
(h_1, q_{AGG}) &= -q_{AAC}q_{AT}q_{TTA} \\
(h_1, q_{ATT}) &= q_{AAA}q_{TCG}q_{TGC} \\
(h_1, q_{CTG}) &= -q_{ACC}q_{AGG}q_{TTA} \\
(h_1, q_{TAT}) &= q_{AAA}q_{ATT}q_{TCG} \\
(h_1, q_{TTC}) &= q_{AAA}q_{AGG}q_{TAT}
\end{align*}
\]
(h_2, q_{CA}) = -q_{CGT}q_{TCG}q_{TTA} (h_2, q_{CA}) = q_{CTG}q_{TTA}q_{TGC} \\
(h_2, q_{CG}) = -q_{CA}q_{TCG}q_{TTA} (h_2, q_{CTG}) = q_{CCA}q_{TTA}q_{TGC} \\
(h_2, q_{CA}) = q_{CCA}q_{CTG}q_{TTA} (h_2, q_{CTG}) = -q_{CA}q_{CGT}q_{TTA} \\
(h_2, q_{CG}) = q_{CCA}q_{CTG}q_{TTA} (h_2, q_{TGA}) = -q_{CCA}q_{CTG}q_{TTA} \\
(h_3, q_{AAA}) = -q_{ACC}q_{CGT}q_{CTG} (h_3, q_{ACC}) = -q_{AAA}q_{CGT}q_{CTG} \\
(h_3, q_{AGG}) = q_{ATT}q_{CAC}q_{CCA} (h_3, q_{ATT}) = q_{AGG}q_{CAC}q_{CCA} \\
(h_3, q_{CA}) = q_{AGG}q_{ATT}q_{CCA} (h_3, q_{CCA}) = q_{AGG}q_{ATT}q_{CCA} \\
(h_3, q_{CG}) = -q_{AAA}q_{ACC}q_{CTG} (h_3, q_{CTG}) = -q_{AAA}q_{ACC}q_{CTG} \\
(h_4, q_{AAA}) = -q_{AGG}q_{GCT}q_{GTC} (h_4, q_{ACC}) = q_{ATT}q_{GAG}q_{GGA} \\
(h_4, q_{AGG}) = -q_{AAA}q_{GCT}q_{GTC} (h_4, q_{ATT}) = q_{AGG}q_{GAG}q_{GGA} \\
(h_4, q_{GA}) = q_{ACC}q_{ATT}q_{GGA} (h_4, q_{GCT}) = -q_{AAA}q_{AGG}q_{GTC} \\
(h_4, q_{GA}) = q_{ACC}q_{ATT}q_{GGA} (h_4, q_{GTC}) = -q_{AAA}q_{AGG}q_{GCT} \\
(h_5, q_{CA}) = q_{CTG}q_{GCT}q_{GTC} (h_5, q_{CCA}) = -q_{CTG}q_{GAG}q_{GTC} \\
(h_5, q_{CG}) = -q_{CCA}q_{GAG}q_{GTC} (h_5, q_{CTG}) = q_{CCA}q_{GAG}q_{GTC} \\
(h_5, q_{CA}) = -q_{CCA}q_{CGT}q_{GTC} (h_5, q_{CTG}) = q_{CCA}q_{CGT}q_{GTC} \\
(h_5, q_{GA}) = -q_{CCA}q_{GCT}q_{GTC} (h_5, q_{GTC}) = -q_{CCA}q_{CTG}q_{GTC} \\
(h_6, q_{GA}) = -q_{GCT}q_{GTC}q_{TTA} (h_6, q_{GCT}) = -q_{GAG}q_{GTC}q_{TTA} \\
(h_6, q_{GA}) = q_{GCT}q_{GTC}q_{TTA} (h_6, q_{GTC}) = q_{GGA}q_{GTC}q_{TTA} \\
(h_6, q_{TAT}) = q_{GGA}q_{GTC}q_{TTA} (h_6, q_{TTA}) = -q_{GAG}q_{GCT}q_{TGC} \\
(h_6, q_{TGC}) = -q_{GAG}q_{GCT}q_{TGC} \\

In general, one has that \( \text{rk}(\text{Jac}_q(W')) \leq \text{codim}_q(W') \leq 7 \). It can be seen by direct computation that the 6 \times 6-matrix obtained from \( \text{Jac}_q(W') \) by removing the last row and keeping the columns indexed by \( q_{ACC}, q_{ATT}, q_{CA}, q_{CTG}, q_{TAT}, q_{TGC} \) equals

\[
\begin{align*}
2q_{AGG}q_{GTCA}q_{CAGT} & \quad (q_{AAA}q_{AGG}q_{ATT}q_{CAC}q_{CAGT}q_{ATCG}q_{TGC}q_{TGA}) \\
q_{AAA}q_{ACC}q_{ATT}q_{ATCG}q_{TGC}q_{TTA} & \quad (q_{AAA}q_{ACC}q_{ATT}q_{CAC}q_{CAGT}q_{ATCG}q_{TGC}q_{TGA} + q_{AAA}q_{AGG}q_{ATT}q_{CAC}q_{CAGT}q_{ATCG}q_{TGC}q_{TGA} + q_{AAA}q_{ACC}q_{ATT}q_{CAGT}q_{ATCG}q_{TGC}q_{TGA} + q_{AAA}q_{ACC}q_{ATT}q_{CAGT}q_{ATCG}q_{TGC}q_{TTA} + q_{AAA}q_{ACC}q_{ATT}q_{CAGT}q_{ATCG}q_{TGC}q_{TTA} + q_{AAA}q_{AGG}q_{ATT}q_{CAGT}q_{ATCG}q_{TGC}q_{TTA})
\end{align*}
\]

which is clearly positive in \( \Delta_+ \). Therefore, \( \text{rk}(\text{Jac}_q(W')) = 7 \) and so, \( q \) is a non-singular point of \( W' \) and \( W' \) is a local complete intersection at \( q \). Therefore, \( W' \subset \mathbb{C}^{16} \) is a subvariety of dimension 9 containing \( W_3 \), which has also dimension 9 and is non-singular at \( q \). It follows that \( W' \) and \( W_3 \) coincide in a neighborhood of \( q \) and we are done. \( \square \)

Remark 4.4. For future reference, it is worth noting that the matrix \( J' \) obtained from \( \text{Jac}_q(W') \) by removing the columns \( q_{AAA}, q_{CCA}, q_{GGA}, q_{TTA} \) and the last row has maximal rank equal to 6.
Next, we want to describe a procedure to give a minimal system of generators for the ideal of $W_n$ around any point $q \in (W_n)_+$. Some of these generators are determined recursively from subtrees of $T$, while the remaining are easily inferred from some matrices to be defined later.

First we describe how these generators are to be constructed by induction on the number of leaves. Then, we will prove that the whole set of these polynomials generate a complete intersection which equals the variety $W_n$ in a neighborhood of any $q \in (W_n)_+$. The generators of this local complete intersection ideal will not depend on the point $q$, as we pointed out in Remark 4.3.

**Generators of degree 4.** As above, write $R = \mathbb{C}[Q_n]$ for the ring of polynomials in the unknowns $Q_n$. Following the idea of Chang [Cha96], write $v_1, \ldots, v_n$ for the leaves of $T$. By reordering the leaves, we may assume that $v_{n-1}$ and $v_n$ form a cherry, i.e. are joined to a node $m$. Take the tree $T'$ with leaves $L(T') = L(T) \cup \{m\} - \{v_{n-1}, v_n\}$, interior nodes $N(T') = N(T) - \{m\}$ and edges $E(T') = E(T) - \{[m, v_{n-1}], [m, v_n]\}$, where $[m, n]$ is the edge containing the nodes $m$ and $n$ (see figure 4). In virtue of Corollaries 3.9 and 3.10, the variety $W_{n-1}$ associated to $T'$ is the image of the polynomial map in (2.4)

$$\varphi_{n-1} : (\mathbb{C}^3)^{2n-5} \longrightarrow \mathbb{C}^{4n-2}$$

and has dimension $3(2n - 5)$.

Assume that we have constructed a local complete intersection $\{g_1, \ldots, g_{c(n-1)}\}$ at the points of $(W_{n-1})_+$ (equivalently, $\{g_1, \ldots, g_{c(n-1)}\}$ generate the localization of the ideal $I_{T'}$ at the points of $(W_{n-1})_+$). The map $j_{n-1} : Q_{n-1} \rightarrow Q_n$ defined by $q_{x_1 \ldots x_{n-1}} \mapsto q_{x_1 \ldots x_{n-1} A}$ induces a ring homomorphism

$$\psi_{n-1} : \mathbb{C}[Q_{n-1}] \rightarrow R.$$

Write

$$J(n-1) = \{f_1^{(n-1)}, \ldots, f_{c(n-1)}^{(n-1)}\} \subset R$$

for the set of polynomials being the image by $\psi_{n-1}$ of the generators $\{g_i\}$.

Analogously, let $T''$ be the tree with 3 leaves determined by the vertices $v_1, v_{n-1}$ and $v_n$. The variety $W_3 \subset \mathbb{C}^{16}$ is the image of

$$\varphi_3 : (\mathbb{C}^4)^3 \longrightarrow \mathbb{C}^{16}$$

and has dimension 9. A complete system of generators $\{h_1, \ldots, h_6\}$ of the ideal $I_{T''} \subset \mathbb{C}[Q_3]$ is given by Lemma 4.2 As above, the map $j_3 : Q_3 \rightarrow Q_n$ defined by $q_{xyz} \mapsto q_{xA \ldots Ayz}$ induces a ring homomorphism

$$\psi_3 : \mathbb{C}[Q_3] \rightarrow R.$$

Write

$$J(3) = \{f_1^{(3)}, \ldots, f_6^{(3)}\} \subset R$$
for the set of polynomials being the image by $\psi_{n-1}$ of $\{h_1, \ldots, h_k\}$. The polynomials in $J(3)$ and $J(n - 1)$ are quartics, but we still need to construct an extra set of polynomials of degree 2.

**Generators of degree 2.** Now, for each letter $z \in \Sigma$, write $M(z)$ for the $4 \times 4^{n-3}$ matrix with rows indexed by the couples $\{xy \mid x + y = z\}$, columns indexed by $\{x_1 \ldots x_{n-2} \mid \sum_{i=1}^{n-2} x_i = z\}$ and whose $(xy, x_1 \ldots x_{n-2})$-entry is precisely $q_{x_1 \ldots x_{n-2}xy}$:

$$M(z) = \begin{pmatrix} \ddots & \cdots & \ddots \\ \cdot & \cdots & q_{x_1 \ldots x_{n-2}xy} & \cdots \\ \ddots & \cdots & \ddots \end{pmatrix}$$

For each of these matrices, take the set of the $3(4^{n-3} - 1)$ $2 \times 2$-minors containing $q_{zA \ldots AzA}$: we obtain polynomials $F_i(z) \in R$ of the form

$$q_{x_1 \ldots x_{n-2}xy}q_{zA \ldots AzA} - q_{x_1 \ldots x_{n-2}zA}q_{zA \ldots Ax} = 0$$

for $i = 1, \ldots, 3(4^{n-3} - 1)$. We get a total of $12(4^{n-3} - 1)$ polynomials. For each letter $z \in \Sigma$, write $K(z) = \{F_i(z)\}$ for this set of polynomials and

$$K = \bigcup_{z \in \Sigma} K(z).$$

**Theorem 4.5.** At each point $q \in (W_n)_+$, the ideal generated by the set

$$J(3) \cup J(n - 1) \cup K$$

together with the equation $q_{A \ldots A} = 1$ is a local complete intersection that defines the Kimura variety $W_n$ in a neighborhood of $q$.

**Proof.** First of all, direct computation shows that the number of polynomials being considered equals the codimension of the variety $W$, i.e.

$$|J(3)| + |J(n - 1)| + |K| + 1 = 6 + (4^{n-2} - 6(n - 1) + 8) + 12(4^{n-3} - 1) + 1 = 4^{n-1} - 6n + 9.$$  

By [SS05] we know that the ideal $\wp = (q_{A \ldots A} - 1, J(3), J(n - 1), K)$ is contained in $I_{W_n}$ and so, $W_n$ is contained in the variety $W'$ defined by $\wp$. We claim that $q$ is non-singular in $W'$. From this, we deduce that $W'$ is a local complete intersection at $q$, and as we did in the proof of Lemma 4.2 we conclude that it is equal to $W_n$ in a neighborhood of $q$.

Now we prove that $q$ is a smooth point of $W'$. Write $Q_0 = \{q_{AA \ldots AAA}, q_{CA \ldots ACA}, q_{GA \ldots AGA}, q_{TA \ldots ATA}\}$, and notice that

$$Q_0 = j_3(Q_3) \cap j_n-1(Q_{n-1}).$$
By reordering the rows and columns if necessary, we may assume that the jacobian matrix of $W'$ at $q$ has the form
\[
Jac_q(W') = \begin{pmatrix}
B & 0 & 0 \\
0 & J' & 0 \\
* & * & D
\end{pmatrix}
\]
where the $c(n - 1) \times 4^n - 2$-matrix $B$ equals the jacobian matrix $Jac_{q_{n-1}}(W_{n-1})$, $J'$ is the $6 \times 12$-matrix of Lemma 4.4. In this way, the columns of the submatrix $B$ are indexed by the unknowns in $Q_{n-1}$ while the rows are indexed by the equations $\{q_{A...A} - 1\} \cup J(n - 1)$. Similarly, the columns of $J'$ are indexed by $Q_3 \setminus \{q_{AAA}, q_{CCA}, q_{GGA}, q_{TTA}\}$ while the rows are indexed by $J(3)$. The columns of the matrix $D$ are indexed by the remaining unknowns while its rows are indexed by the equations $\{F_i^{(z)}\}_{z \in \Sigma}$. Each of these equations has the form (4.1) and so, its partial derivative relative to the unknown $q_{x_1...x_{n-2}xy}$ is equal to $q_{xA...xA}$. Therefore, by reordering rows and columns if necessary we may assume that the matrix $D$ is a diagonal matrix (and all its entries are strictly positive because $q \in (W_n)_+)$.

In virtue of 4.4 we know that $\text{rank}(J') = 6$ and, by induction hypothesis, $B$ has maximal rank equal to $c(n - 1) = 4^n - 2 - 6n + 15$. It follows that the matrix $Jac_q(W')$ has maximal rank equal to
\[
\text{rank}(Jac_q(W')) = 4^n - 2 - 6n + 15 + 6 + 12(4^{n-3} - 1) = 4^{n-1} - 6n + 9,
\]
and we are done. □

Remark 4.6. The set of quadrics $K$ contains the information of invariants coming from the splits of the tree (see [SS05] and [Eri05]). Although in theory a tree can
be reconstructed from its splits (see [Eri05] Theorem 19.14), the variety defined by $K$ is much bigger than $W$ because it has codimension $12(4^n - 3) - 1$.

**Remark 4.7.** In [CFS07], we studied a phylogenetic reconstruction method (already introduced in [CGS05]) which was based on a set of generators of the ideal associated to the Kimura model. The simulation studies performed there showed that it is actually a very competitive and highly efficient method. In the case of 4-leaved trees and for the Kimura 3-parameter model, a minimal system of generators for the corresponding ideal consists of 8002 polynomials of degrees 2, 3 and 4. Because of the results of this paper, it is enough to deal with the 48 invariants listed in the following example (or in general, the codimension of the variety $W_n$). This leads to a substantial improvement in the efficiency and effectiveness of the method. Simulations studies on this variant of the method can be seen on the webpage [http://www.ma1.upc.edu/~jfernandez/ci.html](http://www.ma1.upc.edu/~jfernandez/ci.html) and the reader should contrast them to [CFS07]. Moreover, the fact that we provide the smallest set of local generators in Theorem 4.5 gives some hope for the generalization of phylogenetic reconstruction methods based on algebraic geometry to trees with a large number of leaves.

**Example 4.8.** Let $T$ be the unrooted 4-leaved tree of figure 2(b). The above procedure gives rise to the following 48 invariants: 36 quadrics

\[
\begin{align*}
&\text{AAAAAQATTATCGAGTAC} - \text{AAAAAQAGGGAQTTAA}, \\
&\text{ACCAQAAGGGAQTTAA} - \text{ACCAQAAGGGAQTTAA}, \\
&\text{AGGAGGTTAATCGAGTAC} - \text{AGGAGGTTAATCGAGTAC}, \\
&\text{AGGAGGTTAATCGAGTAC} - \text{AGGAGGTTAATCGAGTAC},
\end{align*}
\]

and 12 quartics

\[
\begin{align*}
&\text{AAAAAQATTATCGAGTAC} - \text{AAAAAQAGGGAQTTAA}, \\
&\text{ACCAQAAGGGAQTTAA} - \text{ACCAQAAGGGAQTTAA}, \\
&\text{AGGAGGTTAATCGAGTAC} - \text{AGGAGGTTAATCGAGTAC}, \\
&\text{AGGAGGTTAATCGAGTAC} - \text{AGGAGGTTAATCGAGTAC},
\end{align*}
\]
$qCACAqGCTGATGqGAAqGCTAqGAGAqGTCAq$
$qGGAAqGCTCATATATCGAqGAGAqGCTATTTAAq$
$qAAAAqATTTqACGTqTAGCq$
$qCACAqCATGqTAATTqACGTqTAGCq$
$qAACCAqATTTqACGTqCAACAq$
$qAACCAqATTTqGAAGqGAGAq$
$qAAAAqAAGGqGACTqGATCq$
$qCACAqCATGqGACTqGAGAqGAGAqGATCq$
$qCACAqCATGqGACTqGAGAqGAGAqGATCq$
$qAACCAqATTTqGAAGqGAGAqGACTqGATCq$
$qCACAqCATGqGACTqGAGAqGAGAqGATCq$
$qAACCAqATTTqGAAGqGAGAqGACTqGATCq$
$qAACCAqATTTqGAAGqGAGAqGACTqGATCq$
$qAACCAqATTTqGAAGqGAGAqGACTqGATCq$

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