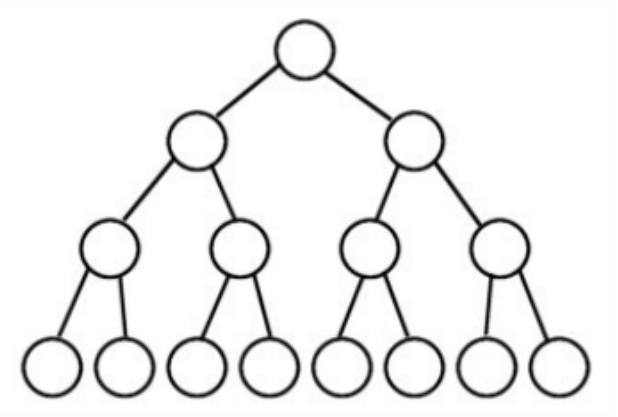
Department of Mathematics and System Analysis

Algebraic Aspects of Hidden Variable Models

Muhammad Ardiyansyah



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Muhammad Ardiyansyah

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Abstract

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Abstract

Hidden variables are random variables that we cannot observe in reality but they are important for understanding the phenomenon of our interest because they affect the observable variables. Hidden variable models aim to represent the effect of the presence of hidden variables which are theoretically thought to exist but we have no data on them. In this thesis, we focus on two hidden variable models in phylogenetics and statistics.

In phylogenetics, we seek answers to two important questions related to modeling evolution. First, we study the embedding problem in the group-based models and the strand symmetric model and its higher order generalizations. In Publication I, we provide some embeddability criteria in the group-based models equipped with certain labeling. In Publication III, we characterize the embeddability in the strand symmetric model. These results allow us to measure approximately the proportion of the set of embeddable Markov matrices within the space of Markov matrices. These results generalize the previously established embeddability results on the Jukes-Cantor and Kimura models. The second question of our interest concerns with the distinguishability of phylogenetic network models which is related to the notion of generic identifiability. In Publication II, we provide some conditions on the network topology that ensure the distinguishability of their associated phylogenetic network models under some group-based models.

The last part of this thesis is dedicated to studying the factor analysis model which is a statistical model that seeks to reduce a large number of observable variables into a fewer number of hidden variables. The factor analysis model assumes that the observed variables can be presented as a linear combination of the hidden variables together with some error terms. Moreover, the observed and the hidden variables together with the error terms are assumed to be Gaussian. We generalize the factor analysis model by dropping the Gaussianity assumption and introduce the higher order factor analysis model. In Publication IV, we provide the dimension of the higher order factor analysis model and present some conditions under which the model has positive codimension.

Keywords hidden variable model, the embedding problem, phylogenetic model, factor analysis model

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Preface

All of the research works presented henceforth was carried out while being part of the Algebra and Discrete Mathematics research group at the Department of Mathematics and System Analysis, Aalto University.

First, I am extremely grateful to my doctoral supervisor and advisor, Prof. Kaie Kubjas for her valuable advice and endless support and guidance during my doctoral study. I am also immensely thankful for her plentiful experiences in the research topics which have boosted me in conducting my research. Moreover, I would also thank her for introducing me to the exciting and interesting research problems. These research problems have opened up new and exciting subfields in mathematics and its applications which I hope to further explore in the future.

Secondly, I would like to extend my acknowledgement to all the current and former members of our research group for the perfect atmosphere in the working environment. It was a pleasure to work with all of you, to participate in several activities with you, and to discuss many different topics. I have been learning so much from every one of you which will be useful and beneficial for my future career.

I want to thank Prof. Elizabeth Gross for acting as external examiners of this dissertation and to Prof. Marta Casanellas Rius for acting as external examiner and for agreeing to act as my opponent.

Words cannot express my deepest gratitude to my beloved wife Riffa, my dear son Zachel, and of course my family for your continuous love and support. It is almost impossible to undergo this challenging journey without your presence. Like an Indonesian proverb says "*berat sama dipikul ringan sama dijinjing*", I am extremely thankful to everyone for always supporting me not only during my wonderful moments but also during my hardest times. Even Zachel and Ibund's little smiles and laughs when we play, when we read books, when we travel, and when we cook together mean a lot for me.

Lastly, I am also thankful to my friends both in Finland and Indonesia. I would also like to acknowledge the Indonesian communities in Finland who always make me feel like I am in my home country, Indonesia. There are just so many new things that I did not imagine of learning before. I learned to perform various dances and played traditional musical instruments as well. I feel like

I learn more about Indonesian culture when I am here in Helsinki which is approximately 14,000 kilometres away from my home town in Indonesia. Thanks for creating such wonderful, warm, and welcoming atmospheres and organizing a lot of fun and memorable activities.

Finally, I would like to mention an Indonesian proverb "*tak ada gading yang tak retak*". As a human being, of course I did countless mistakes with many of you. Therefore, I would like to apologize for these mistakes and I hope that we will create another wonderful moments and stories in the future. Terima kasih.

Espoo, June 30, 2023,

Muhammad Ardiyansyah

Contents

List of Publications

This thesis consists of an overview and of the following publications which are referred to in the text by their Roman numerals.

I Muhammad Ardiyansyah, Dimitra Kosta, and Kaie Kubjas. The Model-Specific Markov Embedding Problem for Symmetric Group-Based Models. *Journal of Mathematical Biology*, Volume 83, Article 33, September 2021.

II Muhammad Ardiyansyah. Distinguishing Level-2 Phylogenetic Networks Using Algebraic and Combinatorial Techniques. Submitted to *Acta Biotheoretica*, June 2022.

III Muhammad Ardiyansyah, Dimitra Kosta, and Jordi Roca-Lacostena. Embeddability of Centrosymmetric Matrices Capturing The Double-Helix Structure in Natural and Synthetic DNA. *Journal of Mathematical Biology*, Volume 86, Article 69, April 2023.

IV Muhammad Ardiyansyah and Luca Sodomaco. Dimension of Higher Order Factor Analysis Models. Accepted for publication in *Algebraic Statistics*, June 2023.

Author's Contribution

Publication I: "The Model-Specific Markov Embedding Problem for Symmetric Group-Based Models"

Some inital results in Section 4 of the paper appeared in Section 4 of the preprint arXiv:1705.09228 authored by DK and KK. The preprint was split into two based on suggestions of referees. The author derived most of results in Section 5 and 6. All co-authors contributed equally to the rest of the paper.

Publication II: "Distinguishing Level-2 Phylogenetic Networks Using Algebraic and Combinatorial Techniques"

The author did everything.

Publication III: "Embeddability of Centrosymmetric Matrices Capturing The Double-Helix Structure in Natural and Synthetic DNA"

All co-authors contributed equally to all parts of the paper.

Publication IV: "Dimension of Higher Order Factor Analysis Models"

All co-authors contributed equally to all parts of the paper.

List of Figures

Symbols

$\mathbb{R}$ the field of real numbers

$\mathbb{C}$ the field of complex numbers

$\mathbb{C}^*$ the multiplicative group of complex numbers without zero

$\mathbb{N}$ the set of natural numbers $\{1, 2, \ldots\}$

$\mathbb{Z}_{\geq 0}$ the set of nonnegative integers $\{0, 1, 2, \ldots\}$

$\mathbb{R}_{\geq 0}$ the set of nonnegative real numbers

$\mathbb{R}_{>0}$ the set of positive real numbers

$[n]$ the set $\{1, 2, \ldots, n\}$ of nonnegative integers from 1 to n

$M_n(\mathbb{K})$ the set of all $n \times n$ matrices with entries in the field $\mathbb{K}$

$GL_n(\mathbb{K})$ the set of all $n \times n$ non-singular matrices with entries in the field $\mathbb{K}$

I_n the identity matrix of size $n \times n$

$\sigma(A)$ the spectrum of $A \in M_n(\mathbb{C})$, i.e., the set of eigenvalues of A

A^T the transpose of A

A^* the conjugate transpose of A

$tr(A)$ the trace of $A \in M_n(\mathbb{C})$

$\det(A)$ the determinant of $A \in M_n(\mathbb{C})$

$\overline{z}$ the complex conjugate of $z \in \mathbb{C}$

$|z|$ the magnitude of $z \in \mathbb{C}$

$Re(z)$ the real part of $z \in \mathbb{C}$

$Im(z)$ the imaginary part of $z \in \mathbb{C}$

$\operatorname{Arg}(z)$ the principal argument of $z \in \mathbb{C}$

$f^{(j)}(x)$ the jth order derivative of the function f a the point x if it exists

B^A the space of all functions from A to B

$d_G(x,y)$ the distance between $x \in V$ and $y \in V$ in a graph $G = (V,E)$

$(\mathbb{K}^m)^{\otimes r}$ the vector space of tensors of format $\underbrace{m \times \cdots \times m}_{r \text{ times}}$ whose entries belong to the field $\mathbb{K}$

$\Delta^r(\mathbb{K}^m)$ the subspace of $(\mathbb{K}^m)^{\otimes r}$ containing all diagonal tensors

$\lfloor x \rfloor$ the largest integer that is smaller or equal to the real number x

$\lceil x \rceil$ the smallest integer that is bigger or equal to the real number x

1. Introduction

A hidden variable model is a statistical model which assumes the presence of hidden variables or factors that can possibly explain a set of observed variables in the model. These hidden variables are hypothesized to theoretically exist and they are hidden because we have no measurable data on them. Hidden variable models can explain dependencies between the observed variables. These statistical models are important because they can be used to formulate and explain many complex systems in a compact and comprehensible way. Moreover, they have been used in various disciplines, including biology, psychology, economics, social sciences, and many more. This thesis focuses on the hidden variable models in the field of phylogenetics and on the factor analysis model.

Phylogenetics is a subfield of biology that studies evolutionary relationships between extant species. The history of phylogenetics dates back to the theory of evolution proposed by Charles Darwin, the theory of species classification or taxonomy developed by Carl Linnaeus, and the theory of shared common origin introduced by Ernst Haeckel. The ultimate goal of phylogenetics is to figure out the true evolutionary relationships between a collection of species of interest and these relationships are commonly described using a special graph called a phylogenetic tree. There are numerous methods that have been used to reconstruct the true phylogenetic tree. Traditionally, phylogenetic trees were constructed by simply comparing morphological, physiological, or behavioural similarities between species. Biologists believe that these features alone cannot provide enough information to obtain the true phylogenetic tree [87, 52]. Technological advancements have made it possible to study phylogenetics using molecular data, including the DNA and protein molecules. This molecular data enables us to formulate more suitable, reliable, and rigorous mathematical models for phylogenetic studies as they provide a large number of potential characteristics available for inferring phylogenetic relationships [52, 60]. Currently the most common method for reconstructing phylogenetic trees is based on the comparison of the DNA or protein sequences of a collection of species using a phylogenetic model that encodes how these sequences are inherited from their possible ancestors and then using statistical inference to obtain a tree that best explains the data.

The most common approach in phylogenetics is to model evolution as a Markov process. This stochastic process assumes that the Markov property holds. That is, the probabilities of certain events happening in the future given the present are independent of the past. In phylogenetics, the Markov matrices in this model explain the probabilities of the occurrence of each event in the nucleotide substitution process. This Markov process is commonly modeled on a phylogenetic tree where each vertex represents a species and each edge represents a parent-child relationship of two species. Since we can only observe and take DNA samples from some subset of species in consideration, it is natural to assume the existence of some hidden variables corresponding to the extinct ancestors.

One of the most important aspects in phylogenetic modeling is to find a time-homogeneous continuous-time Markov process that realizes our set of Markov matrices encoding the nucleotide substitution process. This means that we want to decide whether the Markov process can be completely characterized in terms of instantaneous substitution rates among states in the process. If the answer is positive, then these instantaneous rates of substitution will be displayed in rate matrices and we call our Markov process embeddable. In the language of matrices, a transition matrix P in a Markov chain is said to be embeddable if there exists a rate matrix Q satisfying $P = \exp(Q)$. Any rate matrix satisfying such property is called a Markov generator of P. Given a Markov matrix, the question of finding its Markov generator is known as the embedding problem. The notion of embeddability of Markov matrices was initially introduced by Elfving [30] in 1937. An extensive list of results on the embeddability criteria of Markov matrices is given in [26, 56, 12, 20, 23, 51, 72, 13]. These results completely characterize the embeddability of Markov matrices up to order 4. Moreover, the authors of [14] provide an embeddability criteria for general Markov matrices of order n having positive and distinct eigenvalues together with an algorithm that can enumerate its Markov generators. The embedding problem, especially in the context of phylogenetics, constitutes one of the main underlying topics in this thesis.

More specifically, we are interested in finding embeddability criteria for certain nucleotide substitution models commonly used in phylogenetics. In Publication I, we consider group-based models in which the transition matrices are determined by the underlying group structure. This class of models includes the Jukes-Cantor [53] and the Kimura models [54, 55]. In Publication III, we consider a more general model that is referred to as the strand symmetric model [16] which takes into account the complimentarity of the two strands composing the DNA structure. Similar to the case of group-based models, the strand symmetric model also includes the Jukes-Cantor and the Kimura models. The main results in these two publications are derived by studying properties of matrix exponentials and logarithms.

In recent years, biologists found more evidence that phylogenetic trees cannot fully capture important biological phenomena like horizontal gene transfer or hybridization which require two fully separated branches to merge again

[1, 6, 70]. For instance, [19] and [63] suggest that these processes of reticulate evolution can possibly occur in plants and animals. Instead of modeling phylogeny on a tree, in recent years we have seen numerous methods to reconstruct phylogeny on networks which take into account these reticulate evolutionary processes [27, 57, 8, 40, 47]. Although networks can better represent biological phenomena, there is an obvious drawback, namely general networks are much more complex than trees.

This thesis also focuses on understanding how to distinguish two phylogenetic models on distinct networks. If we can distinguish phylogenetic models on networks, then the associated network parameters are generically identifiable. Generic identifiability ensures that up to a subset of measure zero, the phylogenetic model parameters can be inferred consistently. Some results on the generic identifiability of the network parameter of a phylogenetic network model using polynomial invariants known as phylogenetic invariants have been established in [40, 47, 41]. In these papers, the authors consider a class of phylogenetic networks which is called the class of level-1 networks. Roughly speaking, in each biconnected component, these networks can only contain at most one undirected cycle and at most one vertex that corresponds to reticulate evolution. To obtain these results, the authors utilize some combinatorial aspects of the networks together with some algebraic tools that include Gröbner basis computations of an ideal associated to each phylogenetic network model. In Publication II, we extend this approach to more complex networks that we refer to as level-2 networks. Unlike in level-1 networks, in level-2 networks, we allow each biconnected component to contain at most two vertices corresponding to reticulate evolution.

Another statistical model that assumes the existence of hidden variables is the factor analysis model. Given a set of observable variables, the factor analysis model seeks to find some underlying (typically fewer) hidden variables that could explain covariance between the set of observable variables. The origin of the factor analysis model dates back to the theory of human intelligence proposed by Charles Spearman [75, 74]. This model has been widely used in many fields, including social sciences, economics, and psychology. The factor analysis model is usually implemented to simplify a set of complex observed variables and hence it serves as a dimension-reduction technique in statistics. The work in [28] provides a polynomial parameterization of the factor analysis model. Moreover, the authors studied the model invariants which are polynomial equality constraints that have to be satisfied by any covariance matrix belonging to the model.

One fundamental assumption in the factor analysis model is that the random variables are assumed to be Gaussian. This assumption is rather restrictive. In Publication IV, we drop the Gaussianity assumption and we introduce the notion of higher-order factor analysis model. This higher-order factor analysis model can be described either using the moment or the cumulant tensors. We focus on studying the dimension of the higher-order factor analysis model which

is a useful information in performing model selection. This dimension will be computed using a polynomial parameterization of the higher-order factor analysis model.

This thesis is composed of the summary and four articles related to the algebraic aspects of the hidden variable models in phylogenetics and the factor analysis model. The summary part of this thesis is organized as follows. In Chapter 2, we provide the necessary mathematical preliminaries on functions of matrices, on computational algebraic geometry tools that include Gröbner basis computations, on Markov processes, and on graph theory. In Chapter 3, we present the main results of Publications I, II, and III concerning some algebraic aspects of phylogenetic models. This chapter is divided into five sections. The first two sections provide some background on the nucleotide substitution models in consideration. In the third section, we formulate the embedding problem and present the main results on the embeddability criteria for these models. In the last two sections, we explain how to associate an algebraic object to each phylogenetic network model and how to use it to distinguish phylogenetic models. Finally, in Chapter 4, we introduce the notion of higher-order factor analysis model in terms of cumulant and moment tensors and present the main result of Publication IV on the model dimension.

2. Mathematical Preliminaries

In this chapter, we present all the necessary mathematical preliminaries that are required in the subsequent chapters. We recall the general concept of functions defined on the space of matrices and some results specific to particular functions. We also present some tools from nonlinear algebra, particularly Gröbner bases and elimination theory, which are the main algebraic tools for studying the two hidden variable models presented in this thesis. Moreover, we provide some background on Markov processes and graph theory which serve as foundations for building these two hidden variable models.

2.1 Functions of matrices

In this section, we introduce the definitions and present known results in the theory of matrix functions. There are two matrix functions that are of our main interest. They are the matrix exponential and the matrix logarithm. We will need these two special matrix functions to formulate the embedding problem in Section 3.3. Almost all the results in this section can be found in most linear algebra books, such as [37], [7], and [44].

2.1.1 Jordan decomposition

We review a specific matrix decomposition of any square matrix with complex entries. This decomposition is called a Jordan decomposition of a matrix. Using a similarity transformation, this decomposition reduces any square complex matrix into a block-diagonal matrix such that each block appearing on the diagonal has a triangular form. We will mention the importance of this decomposition in the definition of a matrix function and in its relationship with the usual matrix eigendecomposition. We will see later that we can define a matrix function using its Jordan decomposition.

Definition 2.1.1. Let $A \in M_n(\mathbb{C})$. Let $\{\lambda_1, \ldots, \lambda_p\}$ be the set of eigenvalues of A.

The matrix

$$J_m(\lambda) = \begin{pmatrix} \lambda & 1 & & \\ & \lambda & \ddots & \\ & & \ddots & 1 \\ & & & \lambda \end{pmatrix} \in M_m(\mathbb{C})$$

is called an $m \times m$ *Jordan block* of the eigenvalue λ. A *Jordan canonical form* of A is a matrix $J \in M_n(\mathbb{C})$ such that there exists an invertible complex matrix Z of size n satisfying $Z^{-1}AZ = J$ and

$$J = \operatorname{diag}(J_{m_{11}}(\lambda_1), \ldots, J_{m_{1l_1}}(\lambda_1), J_{m_{21}}(\lambda_2), \ldots, J_{m_{2l_2}}(\lambda_2), \ldots, J_{m_{p1}}(\lambda_p), \ldots, J_{m_{pl_p}}(\lambda_p)),$$

where $n = m_{11} + \cdots + m_{1l_1} + \cdots + m_{p1} + \cdots + m_{pl_p}$. The factorization $A = ZJZ^{-1}$ where J is a Jordan canonical form is called a *Jordan decomposition* of A and the matrix Z is called the *Jordan transformation matrix* associated with the factorization.

Any square complex matrix in $M_n(\mathbb{C})$ can be decomposed into a Jordan canonical form and the values of λ_k appearing in the Jordan canonical form above are its eigenvalues. This Jordan canonical matrix J is unique up to the ordering of these Jordan blocks.

Recall that the *algebraic multiplicity* of an eigenvalue λ of a matrix $A \in M_n(\mathbb{C})$ is the number of times λ appears as a root of its characteristic polynomial $p_A := \det(A - \lambda I_n)$ while its *geometric multiplicity* is defined as the dimension $\dim(\operatorname{Ker}(A - \lambda I_n))$. There are the following relationships between the Jordan canonical matrix J and algebraic and geometric multiplicities of eigenvalues. The geometric multiplicity of λ_k equals the number of Jordan blocks in the Jordan matrix J with the eigenvalue λ_k and the algebraic multiplicity equals how many times λ_k is repeated along the diagonal of J. An *eigendecomposition* $A = ZJZ^{-1}$ is a special case of a Jordan decomposition of A when each of the Jordan blocks appearing in J has size one giving the diagonal structure on the matrix $J = \operatorname{diag}(\lambda_1, \lambda_2, \ldots, \lambda_p)$. In this case, A is said to be *diagonalizable*.

Example 2.1.2. We demonstrate a Jordan decomposition in the following example. Suppose that

$$A = \begin{pmatrix} 2 & 0 & 0 & 2 & 2 \\ 0 & 0 & 0 & 2 & 2 \\ 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 0 & 2 \end{pmatrix} \text{ and } Z = \begin{pmatrix} 0 & 0 & 1 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 \\ -\frac{1}{2} & 0 & 0 & \frac{1}{2} & 0 \\ 1 & 0 & 0 & 0 & 0 \\ 0 & \frac{1}{2} & 0 & 0 & \frac{1}{2} \end{pmatrix}.$$

By direct computations,

$$J := Z^{-1}AZ = \begin{pmatrix} 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 1 & 0 & 0 \\ 0 & 0 & 0 & 0 & 0 \\ 0 & 0 & 0 & 2 & 1 \\ 0 & 0 & 0 & 0 & 2 \end{pmatrix}.$$

We can see that J is a Jordan canonical form of A with a Jordan transformation matrix Z. The spectrum of A is $\{0,2\}$. The algebraic multiplicity of 0 is 3 and its geometric multiplicity is 2. So the Jordan blocks corresponding to 0 give the matrix

$$\operatorname{diag}(J_1(0), J_2(0)) = \begin{pmatrix} 0 & 0 & 0 \\ 0 & 0 & 1 \\ 0 & 0 & 0 \end{pmatrix}.$$

The algebraic multiplicity of 2 is 2 and its geometric multiplicity is 1. The Jordan block corresponding to 2 is

$$J_2(1) = \begin{pmatrix} 2 & 1 \\ 0 & 2 \end{pmatrix}.$$

Using a Jordan decomposition of a matrix, we would like to generalize the notion of a complex scalar function $f(z)$. In the following, we present the definition of a matrix function that takes a scalar function and a complex square matrix A, and then we define the output $f(A)$ to be a complex square matrix of the same size.

Suppose that $\lambda_1, \ldots, \lambda_s$ are the distinct eigenvalues of $A \in M_n(\mathbb{C})$. For each $k \in [s]$, suppose that the size of the largest Jordan block in which λ_k appears is n_k. Following the terminology in [44], a function f is *defined on the spectrum* of A if the value $f^{(j)}(\lambda_k)$, that is the jth order derivative of f evaluated at λ_k, exists for $j = 0, \ldots, n_k - 1$ and $k \in [s]$.

Definition 2.1.3 (Spectral resolution of a function)**.** Given $A \in M_n(\mathbb{C})$, let f be a scalar complex function defined on its spectrum. Let $A = ZJZ^{-1}$ be a Jordan decomposition of A. Then the matrix $f(A)$, often referred as the *the spectral resolution of f in A*, can be computed as follows:

$$f(A) := Z \cdot f(J) \cdot Z^{-1} = Z \cdot \operatorname{diag}(f(J_{m_k})) \cdot Z^{-1},$$

where

$$f(J_{m_k}) = \begin{pmatrix} f(\lambda_k) & f'(\lambda_k) & \cdots & \frac{f^{(m_k-1)}(\lambda_k)}{(m_k-1)!} \\ & f(\lambda_k) & \ddots & \vdots \\ & & \ddots & f'(\lambda_k) \\ & & & f(\lambda_k) \end{pmatrix}$$

The above definition of $f(A)$ is independent of the choice of a Jordan canonical form of A. Moreover, for a diagonalizable matrix A, the matrix $f(A)$ has the same eigenvectors as A with eigenvalues obtained by applying f to eigenvalues of A. Indeed, given an eigendecomposition $A = Z \cdot \mathrm{diag}(\lambda_1, \ldots, \lambda_p) \cdot Z^{-1}$ such that the columns of Z consist of eigenvectors of A, Definition 2.1.3 implies that

$$f(A) = Z \cdot f(\mathrm{diag}(\lambda_1, \ldots, \lambda_p)) \cdot Z^{-1} = Z \cdot \mathrm{diag}(f(\lambda_1), \ldots, f(\lambda_p)) \cdot Z^{-1}.$$

In this case, we can also see that $f(A)$ is diagonalizable as well.

Remark 2.1.4. Using the spectral resolution of a function is not the only way to define a matrix function. It is also possible to define a matrix function via the Cauchy integral and Hermite interpolation. See [44, Definition 1.4 & 1.11] for further details. It is shown in [44, Section 1.2.4] that all these three different ways to define a matrix function are equivalent but one has to be careful when using the definition via the Cauchy integral due to some analytical conditions that the function f has to possess.

Example 2.1.5. We illustrate Definition 2.1.3 for the complex scalar function $f(x) = x^n$ for a natural number $n \geq 2$. Let $A = \begin{pmatrix} 2 & 1 \\ 0 & 2 \end{pmatrix}$, which is already in a Jordan canonical form. Definition 2.1.3 gives

$$f(A) := \begin{pmatrix} f(2) & f'(2) \\ 0 & f(2) \end{pmatrix} = \begin{pmatrix} 2^n & n2^{n-1} \\ 0 & 2^n \end{pmatrix},$$

which can be verified to be A^n.

Similarly to Example 2.1.5, given a matrix $A \in M_n(\mathbb{C})$, if $f = \sum_{k=0}^{m} c_k x^k$ is a polynomial function, then the matrix $f(A)$ can be easily computed as

$$f(A) := \sum_{k=0}^{m} c_k A^k.$$

More generally, given any power series $\sum_{k=0}^{\infty} c_k x^k$, one can consider the matrix power series $\sum_{k=0}^{\infty} c_k A^k$ where $A^0 = I_n$. Therefore, it suggests that it is possible to use a series representation of a function to define its associated matrix function. But in this approach, one has to be careful whether the series converges or not.

Definition 2.1.6 (Series representation of a function)**.** Suppose that f is a complex scalar function. Let $z_0 \in \mathbb{C}$ and $A \in M_n(\mathbb{C})$. Suppose that there exists some $r > 0$ such that the series $\sum_{k=0}^{\infty} c_k (z - z_0)^k$ converges to $f(z)$ for $|z - z_0| < r$. If $|\lambda_i - z_0| < r$ for every eigenvalue λ_i of A, then the matrix series

$$\sum_{k=0}^{\infty} c_k (A - z_0 I_n)^k \text{ converges.}$$

In this case, we define $f(A) := \sum_{k=0}^{\infty} c_k (A - z_0 I_n)^k$.

In fact, it is argued in [66, Example 7.9.3] that if $|\lambda_i - z_0| < r$ for every eigenvalue λ_i of A, then the series $\sum_{k=0}^{\infty} c_k(A - z_0 I_n)^k$ always converges. The proof of this statement utilizes a Jordan form of A. Additionally, it is also shown in [66, Example 7.9.4] that any matrix function is a polynomial matrix function. Namely, given a complex scalar function f that is defined on the spectrum of $A \in M_n(\mathbb{C})$, there exists a complex polynomial function $p(z)$ such that $f(A) = p(A)$.

Example 2.1.7 (Neumann series)**.** Let us consider the function $f(z) = (1-z)^{-1}$ for $z \neq 1$. We know that the series

$$(1-z)^{-1} = \sum_{k=0}^{\infty} z^k$$

converges if and only if the magnitude of z is less than one. If every eigenvalue of A has magnitude less than one, then Definition 2.1.6 suggests that the matrix function $f(A) = (I_n - A)^{-1}$ can also be computed using the series $\sum_{k=0}^{\infty} A^k$. In fact, the series $\sum_{k=0}^{\infty} A^k$ converges if and only if every eigenvalue of A has magnitude less than one [66, Chapter 7]. Note that $I_n - A$ is non-singular if and only if 1 is not an eigenvalue of A. These facts suggest that although the Neumann series may not be convergent, the matrix $(I_n - A)^{-1}$ can still exist.

2.1.2 Matrix exponential and logarithms

In this subsection, we discuss two particular matrix functions, namely the matrix exponential and the logarithm function using definitions given in Section 2.1.1.

We start with the matrix exponentials. Definition 2.1.3 in Section 2.1.1 allows us to define the matrix exponential using the spectral resolution of the complex scalar exponential function $f(z) = e^z$, denoted by $\exp(A)$. Alternatively, as pointed out in Definition 2.1.6, we could also define $\exp(A)$ using the series representation of the function $f(z) = e^z$.

Definition 2.1.8. The exponential of a matrix $A \in M_n(\mathbb{C})$ is the matrix given by

$$\exp(A) = \sum_{k=0}^{\infty} \frac{1}{k!} A^k, \text{ where } A^0 = I_n. \tag{2.1}$$

The exponential series in (2.1) converges absolutely for any square matrix and hence it converges. It can be checked that both spectral resolution and series representation approaches to compute the matrix exponentials coincide. Consequently, we can use both spectral resolution and series representation approaches interchangeably throughout this thesis.

Example 2.1.9. We demonstrate how to compute the matrix exponential. Suppose that for some $\lambda_1, \lambda_2, \lambda_3 \in \mathbb{R}$,

$$A = \begin{pmatrix} \lambda_1 & 1 & 0 \\ 0 & \lambda_1 & 0 \\ 0 & 0 & \lambda_2 \end{pmatrix}.$$

We will now compute $\exp(tA)$ for $t \in \mathbb{R}$. Let $J_1 = \begin{pmatrix} \lambda_1 & 1 \\ 0 & \lambda_1 \end{pmatrix}$ and $J_2 = \begin{pmatrix} \lambda_2 \end{pmatrix}$. Using the series (2.1),

$$\exp(tA) = \sum_{k=0}^{\infty} \frac{t^k}{k!} A^k = \sum_{k=0}^{\infty} \frac{t^k}{k!} \begin{pmatrix} J_1^k & 0 \\ 0 & J_2^k \end{pmatrix} = \begin{pmatrix} \sum_{k=0}^{\infty} \frac{t^k}{k!} J_1^k & 0 \\ 0 & \sum_{k=0}^{\infty} \frac{t^k}{k!} J_2^k \end{pmatrix}.$$

Moreover, it can be shown that $J_1^k = \begin{pmatrix} \lambda_1^k & k\lambda_1^{k-1} \\ 0 & \lambda_1^k \end{pmatrix}$. Hence,

$$\sum_{k=0}^{\infty} \frac{t^k}{k!} J_1^k = \begin{pmatrix} e^{\lambda_1 t} & te^{\lambda_1 t} \\ 0 & e^{\lambda_1 t} \end{pmatrix} \text{ and } \sum_{k=0}^{\infty} \frac{t^k}{k!} J_2^k = e^{\lambda_2 t}.$$

Therefore, the matrix exponential $\exp(tA)$ is given by

$$\exp(tA) = \begin{pmatrix} e^{\lambda_1 t} & te^{\lambda_1 t} & 0 \\ 0 & e^{\lambda_1 t} & 0 \\ 0 & 0 & e^{\lambda_2 t} \end{pmatrix}.$$

If one uses Definition 2.1.3 to compute $\exp(tA)$, then one will arrive at the same result.

We will now mention some important properties of matrix exponentials. From the definitions of the matrix exponential, it follows that $\exp(0_n) = I_n$.

Proposition 2.1.10 ([44], Theorem 10.2)**.** *Let A and B be two commuting complex square matrices of size n. Then* $\exp(A+B) = \exp(A) \cdot \exp(B)$.

Corollary 2.1.11 ([7], Chapter 10)**.** *Let $A \in M_n(\mathbb{C})$. Then* $\exp(A)$ *is invertible and its inverse is* $\exp(-A)$.

We conclude the first part of this subsection by presenting the following well-known formula.

Proposition 2.1.12 ([7], Chapter 10)**.** *For $A \in M_n(\mathbb{C})$,*

$$\det(\exp(A)) = e^{tr(A)}.$$

The matrix exponential has been widely used in different areas in mathematics. For instance, the matrix exponential comes up when one wants to solve a homogeneous linear differential system with constant coefficients. More precisely, given a complex square matrix A of size n, the solution of a first order differential equation given in the matrix form

$$P'(t) = A \cdot P(t)$$

where $P(t) \in \mathbb{R}^n$ is of the form $P(t) = \exp(At) \cdot P(0)$. Here $P(0)$ is the initial condition. Moreover, the matrix exponential is an important object in control

theory. Namely, the matrix exponential is used whenever one wants to convert a continuous time dynamical system to a discrete time dynamical system.

In the remainder of this subsection, we will discuss matrix logarithms and how to find them. From complex analysis, we know that the complex logarithm function is a multi-valued function. This suggests that unlike in the case of matrix exponential, extra care is needed to define the matrix logarithm function. Let us start by presenting what a logarithm of a matrix is. In Chapter 3, we present the embedding problem, and in order to answer this problem, we need to study some properties of the matrix logarithm. In particular, we present some conditions known in the literature under which a (real) logarithm of a matrix exists.

Definition 2.1.13. Let $A \in M_n(\mathbb{C})$. A matrix $B \in M_n(\mathbb{C})$ is a *logarithm* of A if

$$A = \exp(B).$$

Additionally, if B is a real matrix, then B is said to be a real logarithm of A.

Definition 2.1.13 and Proposition 2.1.12 imply that if $A = \exp(B)$, then

$$\det(A) = \det(\exp(B)) = e^{tr(B)} \neq 0.$$

Thus, singular matrices cannot have logarithms.

We briefly discuss the behaviour of a matrix function under the composition of functions. Let $A \in M_n(\mathbb{C})$. Suppose that f and g are scalar complex functions such that $f(A)$ and $f(g(A))$ exist. Let $h(z) = f(g(z))$. Then it can be shown that $h(A) = f(g(A))$. See [66, Chapter 7.9] for further details.

Proposition 2.1.14. *For $A \in M_n(\mathbb{C})$, whenever the spectral resolution* $\log(A)$ *of the real logarithm function* $\log$ *in A is defined, then* $\exp(\log(A)) = A$.

The fact that the spectral resolution is consistent under the composition of functions suggests that matrix logarithms can be computed via the spectral resolution.

Example 2.1.15. We demonstrate how to compute matrix logarithms. Suppose we have the following two matrices:

$$A = \begin{pmatrix} 0.4 & -0.1 \\ 0 & 0.6 \end{pmatrix}, \qquad Z = \begin{pmatrix} 1 & -1 \\ 0 & 2 \end{pmatrix}.$$

Then an eigendecomposition of A is given by $A = Z \cdot \mathrm{diag}(0.4, 0.6) \cdot Z^{-1}$. It can be checked that the spectrum of A is $\{0.4, 0.6\}$. Then using Definition 2.1.3 for the real logarithm function log, we obtain

$$\log(A) = Z \cdot \mathrm{diag}(\log(0.4), \log(0.6)) \cdot Z^{-1} \approx \begin{pmatrix} -0.916291 & -0.202733 \\ 0 & -0.510826 \end{pmatrix}.$$

Remark 2.1.16. Just like in the scalar logarithm function case, the matrix logarithm is not unique. Namely, if $\exp(B) = A$ for some matrices $A, B \in M_n(\mathbb{C})$, then for any $k \in \mathbb{Z}$

$$\exp(B + 2\pi k i I_n) = \exp(B) \cdot \exp(2\pi k i I_n) = A \cdot I_n = A.$$

In general, it is possible that an invertible real matrix has an infinite number of real and complex logarithms.

Let f be a multi-valued complex scalar function. A single-valued complex scalar function F is a *branch* of f if for every z in the domain of f, there exists a subset U_z of the domain such that F is analytic in U_z and the value $F(z)$ is one of the values of f. In the following, we provide the definition of the branches of the logarithm function.

Definition 2.1.17. Let $k \in \mathbb{Z}$ and $z \in \mathbb{C}$. The *kth-branch of the logarithm of* z, denoted by $\log_k(z)$, is

$$\log_k(z) := \log(|z|) + (\mathrm{Arg}(z) + 2\pi k)i,$$

where $\log(|z|)$ denotes the natural logarithm of $|z|$ and $\mathrm{Arg}(z) \in (-\pi, \pi]$. When $k = 0$, then the logarithm $\log_0(z)$ is called the *principal logarithm* of z. To simplify notation, we denote the principal logarithm of z by $\log(z)$.

If we want to find all the logarithms of a matrix, then we have to consider all branches of the complex logarithm as well since each branch produces a different matrix logarithm.

Definition 2.1.18. Let $A, B \in M_n(\mathbb{C})$. The matrix B is a *primary logarithm* of A if there exists some $k \in \mathbb{Z}$ such that $B = \log_k(A)$. Otherwise, B is a *non-primary logarithm* of A. Moreover, if $B = \log_0(A)$, then it is called the *principal logarithm*, denoted by $\mathrm{Log}(A)$.

Alternatively, one could use the following approach using series representation. It is possible to define the principal logarithm of a matrix $A \in M_n(\mathbb{C})$ via Mercator's series, under the assumption that the series converges. By integrating the series representation

$$\frac{1}{1+z} = 1 - z + z^2 - z^3 + \cdots$$

between 0 and x, we obtain the Mercator's series:

$$\log(1+x) = x - \frac{x^2}{2} + \frac{x^3}{3} - \frac{x^4}{4} + \cdots$$

which converges if and only if $|x| < 1$. Then the principal logarithm can be defined via

$$\mathrm{Log}(A) = \log(I_n + A) = \sum_{k=0}^{\infty} \frac{(-1)^{k+1}}{k} A^k$$

whenever the series converges. The series converges whenever the maximum magnitude of any eigenvalue of A is less than one. In regard to this complication, it is more convenient for us to define the logarithm of a matrix using Definition 2.1.3.

Example 2.1.19. We will study the logarithm of the identity matrix I_n. Using the k-th branch logarithm function, all primary logarithms of I_n have the form

$$\log_k(I_n) = \operatorname{diag}(2\pi k i, \ldots, 2\pi k i) \text{ for } k \in \mathbb{Z}.$$

Moreover, let us mention that for $k_1, \ldots, k_n \in \mathbb{Z}$, the matrix

$$L = \operatorname{diag}(2\pi k_1 i, \ldots, 2\pi k_n i)$$

is a logarithm of I_n as well. It should be clear that L is a primary logarithm if and only if $k_1 = \cdots = k_n$. Then for any matrix $Z \in GL_n(\mathbb{C})$ and for any $k_1, \ldots, k_n \in \mathbb{Z}$,

$$\exp(Z \cdot \operatorname{diag}(2\pi k_1 i, \ldots, 2\pi k_n i) \cdot Z^{-1}) = Z \cdot \exp(\operatorname{diag}(2\pi k_1 i, \ldots, 2\pi k_n i)) \cdot Z^{-1} = I_n.$$

This will give us more logarithms of I_n which are not diagonal matrices.

Let $A \in M_n(\mathbb{C})$. Consider the matrix equation $\exp(X) = A$. According to Proposition 2.1.12, if this equation has a solution, then all eigenvalues of A are different from zero and A is invertible. Thus, the condition that A is invertible is necessary for the existence of a logarithm of A. The following result provides a condition that guarantees the existence of real logarithms of a real matrix.

Lemma 2.1.20 ([22], Theorem 1)**.** *An invertible matrix $A \in M_n(\mathbb{R})$ admits a real logarithm if and only if for every negative eigenvalue of A, its associated Jordan blocks appear an even number of times.*

In [22], the author also proves that a real square matrix admits a unique real logarithm if and only if its eigenvalues are positive and any Jordan block associated to any eigenvalue cannot appear more than once.

Among all logarithms of a matrix, the principal logarithm plays a special role since it is desired in many applications. The following theorem provides a condition that has to be satisfied by a matrix to ensure that the principal logarithm is the only logarithm of the matrix.

Theorem 2.1.21 ([44], Theorem 1.31)**.** *Let $A \in M_n(\mathbb{R})$ be a matrix with no eigenvalues in the closed negative real axis. Then A admits a unique real logarithm such that the imaginary part of every eigenvalue of this logarithm lies in the interval $(-\pi, \pi)$ and this logarithm is given by its principal logarithm.*

The following theorem enables the complete description of all logarithms of an invertible matrix.

Theorem 2.1.22 ([37], Section 8)**.** *Let $A \in GL_n(\mathbb{C})$ be an invertible matrix with eigenvalues $\lambda_1, \ldots, \lambda_p$. Suppose that $A = ZJZ^{-1}$ is a Jordan decomposition of A where*

$$J = \operatorname{diag}(J_{m_{11}}(\lambda_1), \ldots, J_{m_{1l_1}}(\lambda_1), J_{m_{21}}(\lambda_2), \ldots, J_{m_{2l_2}}(\lambda_2), \ldots, J_{m_{p1}}(\lambda_p), \ldots, J_{m_{pl_p}}(\lambda_p)).$$

Then all logarithms of A are given by

$$X = ZU \cdot \operatorname{diag}(\log_{k_{11}}(J_{m_{11}}(\lambda_1)), \ldots, \log_{k_{1l_1}}(J_{m_{1l_1}}(\lambda_1)), \ldots, \log_{k_{p1}}(J_{m_{p1}}(\lambda_p)), \ldots, \log_{k_{pl_p}}(J_{m_{pl_p}}(\lambda_p))) \cdot U^{-1}Z^{-1},$$

for $k_{11}, \ldots, k_{pl_p} \in \mathbb{Z}$ and any arbitrary non-singular matrix U that commutes with J.

Example 2.1.23. Let

$$A = \begin{pmatrix} \frac{1}{2} & \frac{1}{6} & \frac{1}{6} & \frac{1}{6} \\ \frac{1}{6} & \frac{1}{2} & \frac{1}{6} & \frac{1}{6} \\ \frac{1}{6} & \frac{1}{6} & \frac{1}{2} & \frac{1}{6} \\ \frac{1}{6} & \frac{1}{6} & \frac{1}{6} & \frac{1}{2} \end{pmatrix}.$$

In the next chapter, we will see that A is an example of a matrix whose symmetries encode some information that can be used in modelingevolution. An eigendecomposition of A is given by

$$A = Z \cdot \text{diag}(1, \frac{1}{3}, \frac{1}{3}, \frac{1}{3}) \cdot Z^{-1}$$

where

$$Z = \begin{pmatrix} 1 & 1 & 1 & 1 \\ 1 & 1 & -1 & -1 \\ 1 & -1 & 1 & -1 \\ 1 & -1 & -1 & 1 \end{pmatrix}.$$

Theorem 2.1.22 implies that all logarithms of A are given by

$$X \approx ZU \cdot \begin{pmatrix} 2\pi k_1 i & 0 & 0 & 0 \\ 0 & -1.09861 + 2\pi k_2 i & 0 & 0 \\ 0 & 0 & -1.09861 + 2\pi k_3 i & 0 \\ 0 & 0 & 0 & -1.09861 + 2\pi k_4 i \end{pmatrix} \cdot U^{-1} Z^{-1}$$

for $k_1, \ldots, k_4 \in \mathbb{Z}$ and any arbitrary non-singular matrix U that commutes with the Jordan form of A.

2.2 Tools from nonlinear algebra

In this section, we briefly discuss some nonlinear algebra tools that are going to be used to study phylogenetic networks and factor analysis models in the subsequent chapters. The most useful tools we consider here are Gröbner bases and elimination theory. Roughly speaking, finding a Gröbner basis of an ideal in a polynomial ring associated with our hidden variable models is desirable because this basis has some nice properties and behaves well in the division algorithm. Additionally, elimination theory formalizes the process of eliminating variables that one often uses in order to solve a system of polynomial equations.

Throughout this section, the field k in consideration is either $\mathbb{R}$ or $\mathbb{C}$. Most results presented in this chapter can be found in typical commutative algebra, algebraic geometry, or abstract algebra books, including [4], [21], and [35].

2.2.1 Polynomial rings, ideals and Gröbner basis

Given a field k, we can construct the *polynomial ring* $k[\bar{x}] := k[x_1, \ldots, x_n]$ with n variables $x_1, \ldots x_n$ and coefficients in k. In particular, this polynomial ring is a k-vector space with k-basis given by the set of all monomials

$$\bar{x}^a = x_1^{a_1} x_2^{a_2} \cdots x_n^{a_n}, \quad a \in \mathbb{Z}^n_{\geq 0}.$$

Any element f of the ring $k[\bar{x}]$, referred as a *polynomial*, has the form

$$f = \sum_{a \in \mathbb{Z}^n_{\geq 0}} c_a x^a, \tag{2.2}$$

where only finitely many coefficients c_a are nonzero.

In what follows, we introduce ideals which are important algebraic objects in a (polynomial) ring. In Chapter 3, we explain how algebra enters the study of phylogenetics via the use of ideals associated to phylogenetic models. These ideals will play a central role in distinguishing phylogenetic models.

Definition 2.2.1. A non-empty subset $I \subseteq k[\bar{x}]$ is an *ideal of* $k[\bar{x}]$ if the following two properties hold:

1. For any $f \in k[\bar{x}]$ and $g \in I$, the product $fg \in I$.
2. For any $g, h \in I$, the sum $g + h \in I$.

Definition 2.2.2. Given a subset $\mathscr{F} \subset k[\bar{x}]$, the *ideal generated by* the subset $\mathscr{F}$, denoted by $\langle \mathscr{F} \rangle$, is defined as the smallest ideal of $k[\bar{x}]$ containing $\mathscr{F}$. In this case, the set $\mathscr{F}$ is said to be a *generating set* or *basis* of the ideal.

Equivalently, it can be checked that the ideal generated by $\mathscr{F}$ is exactly the ideal containing all finite polynomial combinations of elements of $\mathscr{F}$, namely

$$\langle \mathscr{F} \rangle = \{ \sum_{i=1}^{n} g_i f_i : n \in \mathbb{N} \text{ and } g_i \in k[\bar{x}],\ f_i \in \mathscr{F} \text{ for } i \in [n] \}.$$

Example 2.2.3. Given an ideal $I \subseteq k[\bar{x}]$ generated by the set $\mathscr{F}$, it is possible to find other generating sets that are different than $\mathscr{F}$. For example, consider the ideal I of $\mathbb{R}[x, y, z]$ that is generated by two linear polynomials $x + 2y + 3z - 4$ and $5x + 6y + 7z - 8$. It can be easily verified that $I = \langle x - z + 2, y + 2z - 3 \rangle$. In fact, using Gaussian elimination, we can transform the system of linear equations $\begin{cases} x + 2y + 3z - 4 = 0 \\ 5x + 6y + 7z - 8 = 0 \end{cases}$ to the reduced echelon form $\begin{cases} x - z + 2 = 0 \\ y + 2z - 3 = 0 \end{cases}$.

An ideal I of $k[\bar{x}]$ is *finitely generated* if there exists a finite generating set for I. A polynomial ring over a field enjoys the following nice property.

Theorem 2.2.4 (Hilbert's Basis Theorem)**.** *Every ideal of* $k[\bar{x}]$ *is finitely generated.*

Given some ideals in a polynomial ring, the following proposition provides ways to generate more ideals in the ring.

Proposition 2.2.5 ([21], Chapter 4)**.** *If I and J are ideals in $k[\bar{x}]$, then so are their intersection $I \cap J$, their sum $I + J$, and their product IJ where*

$$I + J := \{f + g : f \in I, g \in J\} \text{ and } IJ := \{\sum_{i=1}^{s} f_i g_i : f_i \in I, g_i \in J, s \in \mathbb{N}\}.$$

Let $n = 1$. For the polynomial ring in one variable, we have the famous Euclidean algorithm. It is a well-known fact that $k[x]$ is a *principal ideal domain*, meaning that every ideal in $k[x]$ can be generated by only a single polynomial in $k[x]$. For $n > 1$, this fact is not true. In general, the polynomial ring $k[\bar{x}]$ is a *unique factorization domain*. A unique factorization domain is a domain where every element can be decomposed into a product of irreducible elements and units of the ring and this decomposition is unique up to permutations of factors and multiplication with units of the ring. More details on unique factorization domains can be found in [35, Chapter 9].

Example 2.2.6. Consider the ideals $I = \langle x^3 + 6x^2 + 12x + 8 \rangle$ and $J = \langle x^2 + x - 2 \rangle$ in $k[x]$. Note that $x^3 + 6x^2 + 12x + 8 = (x+2)^3$ and $x^2 + x - 2 = (x+2)(x-1)$. It can be checked that $I \cap J = \langle (x+2)^3(x-1) \rangle$, $I + J = \langle x + 2 \rangle$, and $IJ = \langle (x+2)^4(x-1) \rangle$.

Definition 2.2.7. Let I be an ideal of $k[\bar{x}]$.

1. The ideal I is *maximal* if I is a proper subset of $k[\bar{x}]$ and there are no other ideals contained between I and $k[\bar{x}]$.

2. The ideal I is *prime* if $fg \in I$ for some polynomials $f, g \in k[\bar{x}]$ implies that $f \in I$ or $g \in I$.

3. The ideal I is *radical* if $f^s \in I$ for some $s \in \mathbb{N}$ and some polynomial $f \in k[\bar{x}]$ implies that $f \in I$.

The following proposition provides some relations between maximal, prime, and radical ideals.

Proposition 2.2.8 ([21], Chapter 4)**.**

1. *Every maximal ideal in $k[\bar{x}]$ is prime. Moreover, every prime ideal in $k[\bar{x}]$ is radical.*

2. *Every intersection of prime ideals in $k[\bar{x}]$ is radical.*

In the rest of this subsection, we will briefly discuss the concept of a Gröbner basis of an ideal in a polynomial ring. A Gröbner basis is a special generating set of an ideal that has nice algorithmic properties. We know that for solving a linear system of equations, one can perform Gaussian elimination technique to transform the original system into a much simpler system of linear equations. Moreover, in order to compute the greatest common divisor of two polynomials, one can perform the Euclidean algorithm which works well computationally.

Finally, the simplex algorithm [24] is a very popular algorithm used to solve linear programming problems and this algorithm performs well computationally. Computing Gröbner basis is a generalization of these three well-known techniques [78]. We will also see later that every set of polynomials can be transformed into a Gröbner basis suggesting that finding a Gröbner basis of an ideal in a polynomial ring is a very natural question to ask.

We begin with the definition of a term order which is going to be used to order terms in a polynomial.

Definition 2.2.9. A *term order* on $k[\bar{x}]$ is a total order $\prec$ on the set of monomials x^a for $a \in \mathbb{Z}^n_{\geq 0}$ satisfying the following two properties:

1. If $x^a \prec x^b$ then $x^{a+c} \prec x^{b+c}$ for any $a, b, c \in \mathbb{Z}^n_{\geq 0}$.
2. We have $1 \prec x^a$ for any $a \in \mathbb{Z}^n_{\geq 0} \setminus \{0\}$.

Example 2.2.10. Let $a = (a_1, \ldots, a_n)$ and $b = (b_1, \ldots, b_n)$ be two elements in $\mathbb{Z}^n_{\geq 0}$. In the *lexicographic order*, we say that $x^b \prec x^a$ if the leftmost nonzero entry of $a - b$ is positive. The variables $x_1, \ldots, x_n$ are ordered by the lexicographic ordering as

$$x_n \prec x_{n-1} \prec \cdots \prec x_2 \prec x_1.$$

If we fix a term order $\prec$ on a polynomial ring, then the *multidegree* of f, denoted by multideg(f), is defined as the maximal $a \in \mathbb{Z}^n_{\geq 0}$ in the expansion of f such that $c_a \neq 0$. Moreover, every polynomial f has a unique *leading monomial* $\mathrm{LM}(f)$ which is defined as the largest monomial x^a occurring with nonzero coefficients in the expansion of f (see (2.2)). The *leading coefficient* of f, denoted by $\mathrm{LC}(f)$, is defined as the coefficient $c_{\mathrm{multideg}(f)}$. Finally, the *leading term* of f, denoted as $\mathrm{LT}(f)$, is defined as

$$\mathrm{LT}(f) = \mathrm{LC}(f) \cdot \mathrm{LM}(f).$$

To unify the writing, we will write the terms of a polynomial in decreasing order with respect to $\prec$.

Example 2.2.11. Using the lexicographic order for $n = 2$ in Example 2.2.10, a quadratic polynomial in $\mathbb{R}[x_1, x_2]$ is written

$$f = x_1^2 + 10x_1x_2 - 6x_1 + 5x_2^2 + 3x_2 - 19.$$

Suppose that $I \subseteq k[\bar{x}]$ is an ideal. Then the *ideal of leading terms* $\mathrm{LT}(I)$ of I is defined as follows:

$$\mathrm{LT}(I) = \langle \mathrm{LT}(f) : f \in I \rangle.$$

In general, for any ideal $I \subseteq k[\bar{x}]$, the ideal $\mathrm{LT}(I)$ and the ideal $\langle \mathrm{LT}(g) : g \in \mathscr{G} \rangle$ for a generating set $\mathscr{G}$ of I are different. Hence it motivates the following definition.

Definition 2.2.12. Fix a term order $\prec$ on $k[\bar{x}]$. Let $I \subseteq k[\bar{x}]$ be an ideal. A finite subset $\mathscr{G}$ of I is said to be a Gröbner basis of I with respect to $\prec$ if the following equality holds:

$$\mathrm{LT}(I) = \langle \mathrm{LT}(g) : g \in \mathscr{G} \rangle.$$

Alternatively, Definition 2.2.12 implies that the initial monomials of the elements of a Gröbner basis of I suffice to generate the initial ideal of I.

Proposition 2.2.13 ([21], Chapter 2)**.** *Fix a term order $\prec$ on $k[\bar{x}]$. Then every ideal I of $k[\bar{x}]$ has a Gröbner basis with respect to the term order $\prec$.*

It can be easily checked that if the set $\mathscr{G}$ is a Gröbner basis for the ideal I, then any finite subset of I containing $\mathscr{G}$ is also a Gröbner basis for I. This fact motivates the following definition to remedy the non-minimality of a Gröbner basis of an ideal.

Definition 2.2.14. Fix a term order $\prec$ on $k[\bar{x}]$. Let $I \subseteq k[\bar{x}]$ be an ideal. The set $\mathscr{G}$ is said to be a *reduced Gröbner basis* of I if the following properties hold:

1. The set $\{\mathrm{LT}(g) : g \in \mathscr{G}\}$ minimally generates $\mathrm{LT}(I)$.
2. For every $g \in \mathscr{G}$, $\mathrm{LC}(g) = 1$.
3. No trailing term of any $g \in \mathscr{G}$ lies in $\mathrm{LT}(I)$.

As a consequence of this definition, we have the following theorem.

Theorem 2.2.15 ([21], Chapter 2)**.** *For a fixed term order $\prec$ on $k[\bar{x}]$, every ideal I in $k[\bar{x}]$ admits a unique reduced Gröbner basis.*

Example 2.2.16. Let us consider the ideal $I = \langle x+2y+3z-4, 5x+6y+7z-8\rangle \subseteq \mathbb{R}[x,y,z]$ in Example 2.2.3. Let us now equip $\mathbb{R}[x,y,z]$ with the lexicographic order $z \prec y \prec x$. It can be verified that the set $\mathscr{G} = \{x-z+2, y+2z-3\}$ is a reduced Gröbner basis for I and Theorem 2.2.15 suggests that $\mathscr{G}$ is the only reduced Gröbner basis for I.

Historically, Bruno Buchberger introduced a method to compute the unique reduced Gröbner basis of an ideal I in his dissertation [11]. The name Gröbner comes from the name of Buchberger's advisor, Wolfgang Gröbner. However, Buchberger was not the first mathematician to introduce the idea of using Göbner bases. In fact, Paul Gordan in his research paper in 1900 suggested a similar idea of using Göbner bases. But Buchberger was the first mathematician to formulate a method to compute Göbner bases. This algorithm is then often referred to as the *Buchberger's algorithm*. We will recall Buchberger's algorithm shortly.

Given a fixed ideal I of $k[\bar{x}]$, a monomial x^a is a *standard monomial* if $x^a \notin \mathrm{LT}(I)$. It is known that the number of standard monomials is finite if and only if for each $i \in [n]$, there exists a natural umber n_i such that $x_i^{n_i} \in \mathrm{LT}(I)$. Let us consider the ideal $\mathrm{LT}(I) = \langle x_1^4, x_2^2\rangle \subset k[x_1,x_2]$. Here we can easily see that there are eight standard monomials. But if we consider the ideal $\mathrm{LT}(I) = \langle x_1^4, x_1x_2^2\rangle \subset k[x_1,x_2]$, then we have infinitely many standard monomials.

Let us consider the quotient ring $k[\bar{x}]/I$. It is immediate to see that the set of standard monomials forms a k-vector space basis for $k[\bar{x}]/I$. Hence the image of a polynomial f in $k[\bar{x}]/I$ can be uniquely written as a k-linear combination of

standard monomials. This linear combination expression is referred to as the *normal form* of f and this approach to compute the normal form of a polynomial f given the ideal I is called the *division algorithm*. Roughly speaking, the normal form of f is the remainder on division of f by a Gröbner basis $\mathscr{G}$ of I in the division algorithm. For more details, interested readers can check [21, Chapter 2, Section 3].

Let $\mathscr{G} \subset k[\bar{x}]$ be a subset. The goal is to decide whether $\mathscr{G}$ is a Gröbner basis for the ideal $\langle \mathscr{G} \rangle$. Let us now consider any two polynomials f and g in $\mathscr{G}$. The *S-polynomial* of f and g, denoted by $S(f,g)$, is defined as

$$S(f,g) := \frac{x^a}{\mathrm{LC}(f)} f - \frac{x^b}{\mathrm{LC}(g)} g,$$

where x^a and x^b are monomials of the smallest possible degree satisfying

$$x^a \cdot \mathrm{LM}(f) = x^b \cdot \mathrm{LM}(g).$$

It is clear that $S(f,g)$ is an element of the ideal $\langle \mathscr{G} \rangle$. We now run the polynomial division algorithm on $S(f,g)$ by $\mathscr{G}$. The resulting normal form is denoted by $\overline{S(f,g)}^{\mathscr{G}}$. If it is nonzero, then it can be expressed as a k-linear combination of monomials and none of these monomials are divisible by the leading monomial of any element of $\mathscr{G}$.

Theorem 2.2.17 (Buchberger's criterion)**.** *Fix a term order $\prec$ on $k[\bar{x}]$. Let $I \subseteq k[\bar{x}]$ be an ideal. A finite set $\mathscr{G} \subset I$ of polynomials is a Gröbner basis of I if and only if $\overline{S(f,g)}^{\mathscr{G}} = 0$ for all $f, g \in \mathscr{G}$.*

Using Buchberger's criterion, we can derive the *Buchberger's algorithm* to compute the reduced Gröbner basis $\mathscr{G}$ from any given input generating set $\mathscr{F}$ of an ideal I. Buchberger's algorithm has already been implemented in many software, for instance in `Macaulay2` [29], `Mathematica` [85], or `CoCoA` [38]. In practice, Gröbner basis computations can be used to solve systems of polynomial equations. More precisely, suppose that we want to seek the solution set of a system of polynomial equations

$$\begin{cases} f_1(x_1, \ldots, x_n) = 0 \\ \quad \vdots \\ f_m(x_1 \ldots, x_n) = 0 \end{cases}$$

It is often difficult to solve the system directly. Alternatively, we consider the ideal $I \in k[x_1, \ldots, x_n]$ that is generated by the polynomials f_i for $i \in [m]$. A Gröbner basis $\mathscr{G} = \{g_1, \ldots, g_s\}$ of I might provide an easier way to find the solution set of the original system. This Gröbner basis corresponds to the following system of polynomial equations

$$\begin{cases} g_1(x_1, \ldots, x_n) = 0 \\ \quad \vdots \\ g_s(x_1 \ldots, x_n) = 0 \end{cases}$$

We will see in the forthcoming subsection that the solution set of the original and the derived system corresponding to the Gröbner basis coincide.

Example 2.2.18. Consider the system of polynomial equations

$$\begin{cases} x+y=1 \\ x^3+y^3=2 \end{cases}$$

and the ideal $I=\langle x+y-1,\, x^3+y^3-2\rangle \subseteq k[x,y]$. A Gröbner basis of I with respect to the lexicographic order $y \prec x$ is given by the set $\mathscr{G}=\{x+y-1,\, 3y^2-3y-1\}$. The system of equations corresponding to the Gröbner basis is easier to solve. More precisely, if we solve $3y^2-3y-1=0$, then we obtain $y=\frac{3\pm\sqrt{21}}{6}$. From these values of y and using $x+y=1$, we get $x=\frac{3\mp\sqrt{21}}{6}$. Therefore, the solution set of the original system is given by the following two real points

$$\left\{\left(\frac{3-\sqrt{21}}{6},\frac{3+\sqrt{21}}{6}\right),\left(\frac{3+\sqrt{21}}{6},\frac{3-\sqrt{21}}{6}\right)\right\}.$$

In this example, we also see that an element of Gröbner basis is obtained by eliminating the variable x. We will formalize the method of eliminating variables in Section 2.2.3.

2.2.2 Algebraic varieties

In this subsection, we will recall the notion of an algebraic variety. Algebraic varieties are one of the most important objects in algebraic geometry. Furthermore, many real-life applications of mathematics require solving a system of polynomial equations. For instance, in an epidemiological model which describes how a disease spreads in a population, if one wants to find a condition when the population is at an equilibrium point, then one needs to solve a system of polynomial equations.

Definition 2.2.19. Let $\mathscr{F}\subset k[\bar{x}]$ be a finite set of polynomials. Then the *variety* of $\mathscr{F}$, denoted by $V(\mathscr{F})$, is the set of all common zeroes of the polynomials in $\mathscr{F}$:

$$V(\mathscr{F}):=\{p=(p_1,\ldots,p_n)\in k^n : f(p)=0 \text{ for } f\in\mathscr{F}\}.$$

Moreover, a subset A of k^n is said to be a *variety* if there exists a finite set $\mathscr{F}\subset k[\bar{x}]$ such that $A=V(\mathscr{F})$. If $k=\mathbb{R}$, then the variety is called a *real variety*. If $k=\mathbb{C}$, then the variety is called a *complex variety*. A subset of an algebraic variety A that is a variety is called a *subvariety* of A.

Algebraic varieties enjoy the following nice property. Let $\mathscr{F}_1\subset k[\bar{x}]$ and $\mathscr{F}_2$ be the unique reduced Gröbner basis for the ideal $\langle\mathscr{F}_1\rangle$. Then we have

$$V(\mathscr{F}_1)=V(\langle\mathscr{F}_1\rangle)=V(\langle\mathscr{F}_2\rangle)=V(\mathscr{F}_2).$$

When using the unique reduced Gröbner basis $\mathscr{G}$, we may also observe some geometric properties of a variety that are invisible from the set $\mathscr{F}$ as demonstrated in the following example.

Example 2.2.20. Let us again consider the ideal $I \subseteq \mathbb{R}[x,y,z]$ in Example 2.2.3 with the lexicographic order $z \prec y \prec x$. Using the reduced Gröbner basis $\{x-z+2, y+2z-3\}$ of I, we obtain a parameterization of $V(I)$:

$$V(I) = \{(t-2, -2t+3, t) \in \mathbb{R}^3 : t \in \mathbb{R}\}.$$

This parameterization implies that $V(I)$ is a straight line in $\mathbb{R}^3$.

We are frequently interested to enumerate the solutions of a system of polynomial equations. The set of all solutions of a system could be finite or infinite. If there are finitely many solutions, then it is possible to write them down. For instance, a nonzero complex univariate polynomial of finite degree always has finitely many solutions due to the well-known Fundamental Theorem of Algebra. If we have infinitely many solutions, then it would be desirable to obtain a parameterization of the solution set. In particular, we are interested in finding a polynomial parameterization of the solution set.

Let $A = V(f_1, \ldots, f_s) \subseteq k^n$ be a variety. Then a *polynomial parameterization* of A consists of polynomial functions $g_1, \ldots, g_n \in k[t_1, \ldots, t_m]$ such that the points that can be expressed as $x_i = g_i(t_1, \ldots, t_m)$ for every $i \in [n]$, belong to the variety A. Furthermore, it is required that the variety A is the smallest variety containing these points. We will discuss this in more detail in Section 2.2.3. An example of a parameterization of a variety can be seen in Example 2.2.20 with the real parameter t. We present parameterizations of phylogenetic models in our consideration in Chapter 3. In particular, these parameterizations are given by polynomial functions.

It is immediate that not every set is a variety. For any subset $S \subseteq k^n$, we can form the following set

$$I(S) := \{f \in k[x_1, \ldots, x_n] : f(p) = 0 \text{ for every } p \in S\}.$$

This set is a radical ideal of $k[x_1, \ldots, x_n]$. It can be checked that the variety $V(I(S))$ is the smallest variety containing the set S. This fact leads us to define the following.

Definition 2.2.21. Given $S \subseteq k^n$, the *Zariski closure* of S is the smallest variety containing S and it is equal to $V(I(S))$.

We end this subsection by presenting the notion of dimension of a variety. Let $I \subseteq k[\bar{x}]$ be an ideal. We consider a subset S of the variable set $\{x_1, \ldots, x_n\}$ with a property that no monomial with variables only in S appears in $\mathrm{LT}(I)$. We choose such a subset S with maximal cardinality among all subsets satisfying this property. Then the *dimension of* $V(I)$, denoted by $\dim(V(I))$ is defined as that maximal cardinality $|S|$. Equivalently, the dimension of a variety $V(I)$ can be defined as the supremum of the length of all chains of prime ideals of $k[\bar{x}]/I(V(I))$.

Example 2.2.22. Consider the ideal $I = \langle x+2y+3z-4, 5x+6y+7z-8\rangle \subseteq \mathbb{R}[x,y,z]$ in Example 2.2.3 with the degree lexicographic order $z \prec y \prec x$. The reduced

Gröbner basis is given by $\{x-z+2, y+2z-3\}$ and hence $\mathrm{LT}(I) = \langle x, y\rangle$. Since the variable z does not appear in $\mathrm{LT}(I)$, according to our definition, $\dim(V(I)) = 1$.

2.2.3 Elimination theory and implicitization problem

This subsection provides a tractable method for eliminating variables in a system of polynomial equations. The main result is presented in the Elimination Theorem [21, Chapter 3]. We also discuss the implicitization problem which is one application of the elimination theorem.

Definition 2.2.23. Let $I = \langle f_1, \ldots, f_s\rangle \subseteq k[\bar{x}]$ be an ideal. The lth *elimination ideal* of I, denoted by I_l, is the ideal of $k[x_{l+1}, \ldots, x_n]$ defined as

$$I_l := I \cap k[x_{l+1}, \ldots, x_n].$$

Definition 2.2.23 states that the ideal I_l consists of all implications of the system of equations after eliminating the variables $x_1, \ldots, x_l$. Surprisingly, Gröbner bases also enable us to perform this elimination procedure in a systematic way using the proper term order as presented in the following theorem.

Theorem 2.2.24 (The Elimination Theorem)**.** *Suppose that $I = \langle f_1, \ldots, f_s\rangle \subseteq k[\bar{x}]$ is an ideal. Let $\mathcal{G}$ be a Gröbner basis of I with respect to the lexicographic order $x_n \prec \cdots \prec x_2 \prec x_1$. Then $\mathcal{G} \cap k[x_{l+1}, \ldots, x_n]$ is a Gröbner basis of I_l for every $l \in [n]$.*

This method of eliminating variables will be used in the subsequent chapters to obtain some algebraic invariants associated to a phylogenetic model given a polynomial parameterization of the model. Furthermore, these algebraic invariants are the elements of the Gröbner basis of some elimination ideal associated with the phylogenetic model.

Example 2.2.25. Let us consider the ideal $I = \langle x+y+z, x^2+y^2+z^2-1, x^3+y^3+z^3-2\rangle \subseteq \mathbb{C}[x, y, z]$ with the lexicographic order $z \prec y \prec x$. A Gröbner basis of I is given by the set

$$\{f_1 := x+y+z, f_2 := 2y^2+2yz+2z^2-1, f_3 := 6z^3-3z-4\}.$$

The Elimination Theorem implies that the set $\{f_2, f_3\}$ is a Gröbner basis for the first elimination ideal I_1 and the set $\{f_3\}$ is a Gröbner basis for the second elimination ideal I_2.

The following theorem provides some relations between the partial solutions $V(I_l)$ and $\pi_l(A)$.

Theorem 2.2.26 (The Closure Theorem)**.** *Let $A = V(f_1, \ldots, f_s) \subseteq \mathbb{C}^n$ be a variety. Suppose that I is the ideal $\langle f_1, \ldots, f_s\rangle$. Let $\pi_l : \mathbb{C}^n \to \mathbb{C}^{n-l}$ be the projection map onto the last $n-l$ coordinates. Then*

1. *The Zariski closure of $\pi_l(A)$ is $V(I_l)$.*
2. *If $A \neq \emptyset$, then there exists a variety $B \subset V(I_l)$ satisfying $V(I_l) \setminus B \subseteq \pi_l(A)$.*

2.3 Markov chains

This section provides a brief overview of Markov processes. We will see in Chapter 3 that we can model an evolutionary process as a stochastic process. Many other real-life phenomena can be described using stochastic processes, including fluctuations in the stock market, epidemiology models to describe diseases like coronavirus, and growth models of some bacteria population. We will need the terminology that is going to be introduced in this section to discuss the mathematical model of an evolutionary process. Almost all the results in this chapter can be found in most probability books, for example [58], [39], and [3].

A stochastic process is one of the main objects of study in probability theory. A stochastic process, or often referred to as a random process is a collection of random variables in a common probability space and they are ordered by an index set. Stochastic processes include the well-known Markov chain, which would be the main object of interest in this section. Roughly speaking, a Markov chain is a system that can undergo changes from one state to another state under constraints given by some probabilistic rules. The Markov chain was introduced and first studied systematically by Andrey Andreyevich Markov.

In this section, we focus on the continuous-time Markov chains. Throughout the thesis, we assume that our state space S, which is the set of possible values of the random variables in a stochastic process, is finite.

Definition 2.3.1. Let $X = (X_t)_{t\geq 0}$ be a collection of random variables with a state space S. Then X is a continuous-time Markov Chain (CTMC) if the following equations are satisfied:

$$P(X_{t_n} = i_n | X_{t_1} = i_1, \ldots, X_{t_{n-1}} = i_{n-1}) = P(X_{t_n} = i_n | X_{t_{n-1}} = i_{n-1}) \tag{2.3}$$

for all $i_1, \ldots, i_n \in S$ and any time sequence $0 \leq t_1 < t_2 < \cdots < t_n$. These equations are referred to as the *Markov property*. If the Markov chain satisfies

$$P(X_{t+s} = j | X_s = i) = P(X_t = j | X_0 = i), \quad s, t \geq 0, \tag{2.4}$$

then we call it *time-homogeneous*. Otherwise, we will call the Markov chain *time-inhomogeneous*.

The Markov property (2.3) says that during the process, the probability distribution of the state at some time in the future given the current state does depend on the states occurring in the past. The Markov property in a Markov chain implies that for $0 \leq t_1 < t_2 < \cdots < t_n$,

$$P(X_{t_n} = i_n, X_{t_1} = i_1, \ldots, X_{t_{n-1}} = i_{n-1}) = P(X_{t_1} = i_1) \prod_{k=2}^{n} P(X_{t_i} = i_k | X_{t_{i-1}} = i_{k-1}).$$

As a consequence, all finite probability distributions in a Markov chain are completely specified by the initial distribution $P(X_{t_1} = i_1)$ and a set of first-order

conditional probability distributions $P(X_{t_i} = i_k | X_{t_{i-1}} = i_{k-1})$ for $i = 2, 3, \ldots$. The homogeneity property of a Markov process says that the conditional probabilities in the process are independent of the current time. If one has a time-inhomogeneous Markov chain, then it can be converted to a time-homogeneous Markov chain by by making the state space appropriately larger. Some examples of time-homogeneous Markov chains include the Poisson and the Wiener processes [58, Chapter 20].

Definition 2.3.2. Let X be a time-homogeneous CTMC. Then the *transition probability* for X is defined as follows

$$P_{ij}(t) = P(X_{t+s} = j | X_s = i) = P(X_t = j | X_0 = i), \quad s, t \geq 0.$$

We denote by $P(t) := (P_{ij}(t)) \in M_{|S|}(\mathbb{R})$ the matrix of transition probabilities for X at time t. We refer to this matrix as the *transition matrix* of X.

Remark 2.3.3. For a time-inhomogeneous CTMC, the transition probabilities are given by a function of two times: $P_{ij}(s,t) = P(X_{t+s} = j | X_s = i)$.

We now present the Chapman-Kolmogorov equation for a CTMC which is attributed to Andrei Kolmogorov and Sydney Chapman.

Proposition 2.3.4 ([39], Chapter 6)**.** *For any* $0 \leq t_1 < t_2 < t_3$*,*

$$P(t_1, t_3) = P(t_1, t_2) P(t_2, t_3).$$

Remark 2.3.5. In the case of a time-homogeneous CTMC, the corresponding Chapman-Kolmogorov equation is given by

$$P(t + s) = P(t) P(s)$$

for any $s, t \geq 0$. Therefore, the set $\{P(t) : t \geq 0\}$ is a semigroup and it is often referred to as the *transition semigroup*.

The entries of the matrix of transition probabilities in a Markov chain lie in the interval $[0, 1]$. Moreover, each row of the matrix sums to one. These properties motivate the following definition.

Definition 2.3.6. A real matrix $P \in M_n(\mathbb{R})$ is a *Markov matrix* if each of its entries is a nonnegative real number and each row sums to 1. Throughout the thesis, we might also use *transition* or *stochastic matrix* to refer to a Markov matrix.

The following proposition is a direct consequence of the Perron-Frobenius theorem [36, 71].

Proposition 2.3.7. *Let P be a Markov matrix. Then*

1. *The magnitude of any eigenvalue of P is at most one.*
2. *The vector $\mathbf{1} = (1, \ldots, 1)^T$ of ones is an eigenvector of P with eigenvalue 1.*

Remark 2.3.8. For a positive Markov matrix, the eigenvalue 1 has multiplicity 1. For a nonnegative Markov matrix, the algebraic and geometric multiplicity of the eigenvalue 1 coincide. For further details, see [66, Chapter 8].

Let X be a time-homogeneous CTMC with the state space $S = \{s_1, \ldots, s_n\}$. Let q_{ij} be the *instantaneous substitution rate* from state s_i to s_j for $i \neq j$. More precisely, for $i \neq j$,

$$q_{ij} := \lim_{\Delta t \to 0} \frac{P(X_{t+\Delta t} = j | X_t = i)}{\Delta t}.$$

We then form a matrix $Q = (q_{ij})$ such that the diagonal entries are given by $q_{ii} = -\sum_{i \neq j} q_{ij}$. The matrix Q is referred to as the *instantaneous rate matrix* of X. Every time-homogeneous CTMC is completely determined by its instantaneous rate matrix. This instantaneous rate matrix motivates the following definition.

Definition 2.3.9. A real matrix $Q \in M_n(\mathbb{R})$ is a *rate matrix* if every off-diagonal entry is nonnegative and each row sums to zero.

Remark 2.3.10. Analogous to the case of Markov matrices, it can be shown that for rate matrices, the vector $\mathbf{1}$ is an eigenvector associated with the eigenvalue 0.

Theorem 2.3.11 ([39], Chapter 6)**.** *Let X be a time-homogeneous CTMC with an instantaneous rate matrix Q. Then $P(t) = \exp(Qt)$ for every $t \in \mathbb{R}_{\geq 0}$ if and only $P(t)$ is the transition matrix from time s to $s + t$.*

Theorem 2.3.11 suggests that the transition matrix of a time-homogeneous CTMC is the exponential of its unique instantaneous rate matrix Q multiplied with some nonnegative constant. For this reason, the matrix Q is said to be the *Markov generator* of the Markov chain. Alternatively, given an instantaneous rate matrix of a CTMC, we can recover the transition matrices in the process by solving the Kolmogorov backward or forward equation [39, Chapter 6].

Example 2.3.12. Let us consider a CTMC X on the binary state space $S = \{0, 1\}$. Suppose that $a \geq 0$ and $b \geq 0$ are the instantaneous substitution rates form the state 0 to 1 and from the state 1 to 0, respectively. The instantaneous rate matrix Q of X is given by

$$\begin{pmatrix} -a & a \\ b & -b \end{pmatrix}.$$

By computing the matrix exponential of Qt for $t \geq 0$, we obtain that the transition matrices of X are given by

$$P(t) = \frac{1}{a+b} \begin{pmatrix} b + ae^{-(a+b)t} & a - ae^{-(a+b)t} \\ b - be^{-(a+b)t} & a + be^{-(a+b)t} \end{pmatrix}.$$

In Publication III, we have a great interest in studying the logarithm of certain classes of matrices with distinct and positive eigenvalues. The following result will be crucial since it can list all the real logarithms of any Markov matrix with distinct and positive eigenvalues such that every row of this logarithm sums to one.

Proposition 2.3.13 ([14]). *Let $A \in M_n(\mathbb{R})$ be a Markov matrix having distinct eigenvalues $\lambda_i \in \mathbb{R}_{>0}$ for $i \in [t]$ and $\mu_j \in \{z \in \mathbb{C} : Im(z) > 0\}$ for $j \in [s]$. Suppose that*

$$A = Z \cdot \text{diag}(1, \lambda_1, \ldots, \lambda_t, \mu_1, \overline{\mu_1}, \ldots, \mu_s, \overline{\mu_s}) \cdot Z^{-1} \in M_n(\mathbb{R})$$

be an eigendecomposition of A with $Z \in GL_n(\mathbb{C})$. Then a matrix Q is a real logarithm of A such that every row sums to zero if and only if

$$Q = Z \cdot \text{diag}(0, \log(\lambda_1), \ldots, \log(\lambda_t), \log_{k_1}(\mu_1), \overline{\log_{k_1}(\mu_1)}, \ldots, \log_{k_s}(\mu_s), \overline{\log_{k_s}(\mu_s)}) \cdot Z^{-1}$$

for some $k_1, \ldots, k_s \in \mathbb{Z}$.

2.4 Graph-theoretic terminology

In this section, we introduce all graph-theoretic terminology that is essential for discussing phylogenetic networks. In this thesis, there are two types of graphs under our consideration. Namely, they are *undirected* and *directed* graphs. Both types of graphs are important in phylogenetics. Most of the terminology here can be found in standard graph theory books such as [9]. Moreover, some of the terminology here is adapted from [40], [41], and [49].

2.4.1 Undirected and directed graphs

In mathematics, we often want to describe relationships between different objects. A graph is a mathematical object that can be used to represent relationships between different entities, such as chemical compounds in a chemical reaction network and biological entities in an evolutionary diagram. A graph G is a pair (V, E) where V is a set and $E \subseteq V \times V$. We refer to any element of V as a vertex of G and any element of E as an edge of G. We begin by presenting two types of graphs that will be used throughout the thesis. Additionally, we assume that both V and E are finite.

Definition 2.4.1. An *undirected graph* $G = (V, E)$ is a graph such that every $e \in E$ is an unordered pair (v, w) for some $v, w \in V$.

The following is some common terminology used for undirected graphs. Let $e = (v, w)$ be an edge in an undirected graph $G = (V, E)$. Then we say that both vertices v and w are *incident* to e. The *endpoints* of the e are v and w. Two vertices are said to be *adjacent* if they are the endpoints of an edge of the graph. Two edges are said to be *adjacent* if they share a common endpoint. In this thesis, we assume that our undirected graphs contain no multi-edges and self-loops. Multi-edges occur when at least two edges of the graph are incident with the same two vertices. A self-loop occurs when both endpoints of an edge coincide. Graphs with no self-loops and no multi-edges are called *simple* graphs. Finally, the *degree* of a vertex is defined as the number of edges incident to it.

Unlike in undirected graphs, every edge in a *directed graph* is assumed to have a direction.

Definition 2.4.2. A *directed graph* $G = (V, E)$, or also commonly referred to as digraph, is a graph such that every $e \in E$ is an ordered pair (v, w) for some $v, w \in V$.

Similar to undirected graphs, the following is some common terminology used for directed graphs. Let $G = (V, E)$ be a directed graph. Let $e = (v, w) \in E$ be a directed edge. Then e is *directed* from v to w. The *source* of e is the vertex v and the *target* of e is the vertex w. We say that e an *out-edge* of v. Similarly, e is said to be an *in-edge* of w. Let $u \in V$. Then the *in-degree* of u is defined as the number of in-edges of u. Similarly, the *out-degree* of u is defined as the number of out-edges of u. Finally, the *degree* of u is defined as the total sum of the in-degree and out-degree of u.

We can turn an undirected graph into a directed graph by directing each edge of the graph. Conversely, we can turn a directed graph into an undirected graph by simply forgetting the edge directions.

We will now present some graph operations that can be done to modify graphs. Let $G = (V, E)$ be a graph. The following operations can be done to directed and/or undirected graphs.

1. If we want to *delete an edge* $e \in E$ in a directed or an undirected graph, then we simply remove the edge e from the graph.

2. If we want to *delete a vertex* $v \in V$ in a directed or an undirected graph, then we delete the vertex v from the graph and additionally all edges incident to v.

3. Suppose that G is undirected. If we want to *suppress a vertex* $v \in V$ of degree two, then first we connect the vertices adjacent to v by a new edge and remove the vertex v afterwards.

4. Suppose that G is directed. If we want to *suppress a vertex* $v \in V$ of in-degree one and out-degree one, then first we connect the source of the in-edge and the target of the out-edge by a new edge and we delete the vertex v afterwards.

Let $G = (V, E)$ be an undirected or a directed graph. A *subgraph* $H = (W, F)$ of G is a graph such that $W \subseteq V$ and $F \subseteq E$ and for every $e = (v, w) \in F$, we have $v, w \in W$. Let $U \subseteq V$ be a subset. The subgraph $G|_U = (U, E|_U)$ *induced* by the subset U is defined as the graph with the vertex set U and the edge set $E|_U$ that consists of all edges of G whose both endpoints belong to U.

An *undirected path* P of an undirected graph is a sequence of vertices connected by edges such that no edges are repeated. Similarly, if G is directed, then a *directed path* P is a sequence $(v_1, \ldots, v_k)$ of vertices such that every pair (v_i, v_{i+1}) of adjacent vertices is connected by a directed edge going from v_i to v_{i+1} and

there is no repeated edges. A *directed (undirected) cycle* is a directed (undirected) path in which the first vertex is equal to the last vertex and no other vertex can occur more than once. If a directed (undirected) graph contains no directed (undirected) cycles then it is called *acyclic*.

We say that two vertices v and w in an undirected graph G are *connected* if we can find an undirected path connecting v and w. If every pair of its vertices are connected, then G is said to be *connected*. A maximal connected subgraph of G is called a *connected component* of G. We call an edge *cut-edge* if by removing this edge the graph becomes disconnected. A cut-edge is said to be *trivial* if one part of the induced partition of the vertices is a singleton. A graph G is said to be *biconnected* if it is connected and we cannot disconnect G by deleting any single vertex of G. A maximal biconnected subgraph of G is called a *biconnected component* of G.

We conclude this part by presenting another special type of graph called a *tree*.

Definition 2.4.3 ([49])**.** A *tree* is a connected graph without any (undirected) cycles.

There are some important properties of trees that are worth mentioning. If $G = (V,E)$ is a tree, then for any two vertices of G, there exists a unique undirected path connecting them. Moreover, it holds that $|V| = |E| + 1$ and adding an extra edge to G will create a cycle.

2.4.2 Rooted and binary graphs

In this section, we discuss rooted and binary graphs. Moreover, we provide some properties of rooted directed acyclic graphs.

Definition 2.4.4. A directed graph is said the be *rooted* if one of its in-degree zero vertices is declared as a root and every edge is directed away from the root.

In an undirected graph, the vertices of degree 1 are called *leaves*. In a directed graph, the vertices of in-degree 1 and out-degree 0 are called *leaves*. In the context of phylogenetics, we will see in the forthcoming chapter that the set of leaves of a graph corresponds to the set of extant species of consideration while the root of a graph corresponds to the shared common ancestors of the extant species that is hypothesized to exist. We have the following type of graphs that are determined by the degree of its vertices.

Definition 2.4.5. A (directed/undirected) graph is *binary* if every vertex that is not either a root or a leaf has degree three. The *root* in a binary graph is the distinguished vertex of in-degree zero and out-degree two.

A *split* of a set X is a partition $A|B$ of X into two disjoint non-empty subsets A and B.

Definition 2.4.6 ([80])**.** Let $T = (V,E)$ be a binary tree with its leaf set bijectively labeled by a set X. A split $A|B$ of X is *valid* if it is obtained by deleting some

edge $e \in E$ and by choosing A and B to be the two sets of leaf labels of the two connected components of $T \setminus \{e\}$. We denote by $\Sigma(T)$ the set of all valid splits of X.

Knowing the sets of splits of a leaf-labeled binary tree is useful. In fact, every leaf-labeled binary tree is uniquely determined by its set of splits $\Sigma(T)$ [80, Theorem 15.1.6]. This fact is referred to as the Splits Equivalence Theorem. Later in Section 3.4, we will use the set of valid splits of a leaf-labeled binary tree to obtain specific parameterizations of certain phylogenetic models. These parameterizations will be our main nonlinear algebra tool to study phylogenetics.

We will see in the next chapter that phylogenetics was originally modeled on a rooted tree. Moreover, we will also present that the set of splits of a tree can be used to obtain a parameterization of the associated phylogenetic model built on the tree. The set of splits of a rooted tree can be also used in obtaining a parameterization of a phylogenetic model built on a rooted directed acyclic graph *(DAG)*. This class of graphs will form the basis for rooted phylogenetic networks which we shall see in the forthcoming chapter.

Given any two vertices v and w in a rooted DAG, we say that w is a *child* of v or v is a *parent* of w if there exists a directed edge going from v to w. We say that w is a *descendant* of v or v is an *ancestor* of w if there exists a directed path going from v to w.

Lemma 2.4.7 ([49], Lemma 1.4.2)**.** *Given a rooted DAG G, every vertex of G is a descendant of the root. Moreover, any two vertices can be joined by an undirected path.*

We will complete this section by presenting the following proposition which provides a useful characterization of a rooted tree.

Proposition 2.4.8 ([49], Lemma 1.4.3)**.** *Given a rooted DAG G, G is a rooted tree if and only if G admits exactly one root and all other vertices have in-degree one.*

3. Algebraic Methods in Phylogenetics

In this chapter, we summarize Publications I, II, and III which present interplay between algebra and and hidden variable models in phylogenetics. The evolutionary histories studied in phylogenetics are usually described using a graph. We will present some mathematical models that are commonly used to model evolution. Then we state the embedding problem in the context of phylogenetics and present some criteria that can be used to check the embeddability of Markov matrices, summarizing the main results of Publications I and III. Moreover, we apply nonlinear algebra techniques to distinguish models associated to phylogenetic networks, summarizing the main results of Publication II. To keep a certain consistency, some of the notation used in this chapter differs from the notation in the related publications.

3.1 Mathematical models in phylogenetics

In this section, we present some mathematical models that could explain the evolutionary histories between a set of species on a graph using data from DNA sequence alignment. These models capture the nucleotide substitution process occurring at the DNA level. These models have been introduced, for instance, in [33] and [68].

3.1.1 Nucleotide substitution models

We start by presenting some facts about the polymer molecule *deoxyribonucleic acid (DNA)*. This molecule which is usually found in the cell nucleus and mitochondria is known to carry the hereditary genetic information which is essential for organisms to function and develop. A DNA molecule consists of two linked strands (chains) that wound around forming a shape known as a double helix. Each chain consists of deoxyribose and phosphate subunits. Each deoxyribose sugar is attached to one of four nucleotide bases: adenine (A), cytosine (C), guanine (G) or thymine (T). The Watson-Crick base pairings are the chemical bonds that pair bases in the two DNA strands. In this pairing, adenine

bonds with thymine while cytosine bonds with guanine. Moreover, nucleotides can be grouped into two groups: purines consisting of adenine and guanine, and pyrimidines consisting of cytosine and thymine. This classification is due to their chemical properties.

A mutation in the sequence bases can occur during the replication process of DNA. This is either due to mistakes when DNA is copied or as a result of environmental factors. We model this phenomenon using *nucleotide substitution models*. In these models, it is assumed that the substitution process of DNA nucleotides follows a Markov process.

We consider the following approach in studying evolution. We fix a single site of DNA sequences in our consideration. We only consider the conditional probabilities between the original and the final DNA sequences at this site. Let X and Y be the random variables at the beginning and at the end of the evolutionary process, respectively. The state space for this Markov process is the set of DNA nucleotides. We label the first, the second, the third, and the fourth row (column) of the transition matrix by the elements `A, G, C, T`, respectively. Hence we can write the transition matrix P of size four such that for any $i,j \in \{$`A, G, C, T`$\}$, the (i,j)-entry of P is given by $P_{ij} = P(Y = j|X = i)$. A *nucleotide substitution model* is a model that is determined by a subset of the set of all Markov matrices of size four. The models presented above describe the evolutionary process at a single site within a set of DNA sequences and they do not take time into consideration. If one wants to consider the evolutionary process for all sites in the DNA sequences, then it is common to additionally assume that the substitution process at every site is independent and identically distributed.

Next, we will mention some of the most common nucleotide substitution models in the literature. These models are motivated by certain biochemical properties of the DNA.

1. The *Jukes-Cantor (JC)* model, which was originally introduced in [53] in 1969, is the most elementary nucleotide substitution model. In this model, a transition matrix is assumed to have the following symmetries:

$$\begin{pmatrix} a & b & b & b \\ b & a & b & b \\ b & b & a & b \\ b & b & b & a \end{pmatrix}.$$

   These symmetries captured by the JC model suggest that it only distinguishes whether each nucleotide base remains unchanged or mutates to a different nucleotide base at the end of the process. In this model, we additionally assume that the DNA nucleotides at the root have a uniform distribution $\pi = (\pi_{\mathtt{A}}, \pi_{\mathtt{C}}, \pi_{\mathtt{G}}, \pi_{\mathtt{T}})$. Namely, it satisfies

$$\pi_{\mathtt{A}} = \pi_{\mathtt{C}} = \pi_{\mathtt{G}} = \pi_{\mathtt{T}}.$$

2. According to the groupings of nucleotides based on their chemical properties, there are two types of substitutions. The first one is the transversion which is a substitution of a purine by a pyrimidine or vice versa. The second one is the transition which is a substitution within purines or within pyrimidines. The second model in our consideration takes into account these two types of substitutions. This model is called the *Kimura* model. If we assume that all transition substitutions have the same probability and that all transversion substitutions have the same probability, then we obtain the *Kimura 2-parameter (K2P)* model [54]. A transition matrix belonging to this model is assumed to have the following symmetries:

$$\begin{pmatrix} a & b & c & c \\ b & a & c & c \\ c & c & a & b \\ c & c & b & a \end{pmatrix}.$$

If we assume different parameters for the nucleotide substitutions, namely one parameter for transitions and two parameters for transversions, then we obtain the *Kimura 3-parameter (K3P)* model [55]. In this model, a transition matrix is assumed to have the form:

$$\begin{pmatrix} a & b & c & d \\ b & a & d & c \\ c & d & a & b \\ d & c & b & a \end{pmatrix}.$$

If $b = c = d$, then the K3P model restricts to the JC model, and if $c = d$, then it restricts to the K2P model. Similar to the Jukes-Cantor model, it is common to assume that the root is uniformly distributed in both Kimura models.

3. The *strand symmetric (SS)* model [16, 86] reflects the symmetry that emerges from the complementarity between the two DNA strands. In the SS model, the transition probabilities satisfy the following conditions:

$$P_{\mathtt{AA}} = P_{\mathtt{TT}}, P_{\mathtt{AC}} = P_{\mathtt{TG}}, P_{\mathtt{AG}} = P_{\mathtt{TC}}, P_{\mathtt{AT}} = P_{\mathtt{TA}},$$

$$P_{\mathtt{CA}} = P_{\mathtt{GT}}, P_{\mathtt{CC}} = P_{\mathtt{GG}}, P_{\mathtt{CG}} = P_{\mathtt{GC}}, P_{\mathtt{CT}} = P_{\mathtt{GA}}.$$

Equivalently, a transition matrix in this model is assumed to have the following symmetries:

$$\begin{pmatrix} a & b & c & d \\ e & f & g & h \\ h & g & f & e \\ d & c & b & a \end{pmatrix}.$$

In this model, it is also assumed that the probability distribution $\pi = (\pi_A, \pi_C, \pi_G, \pi_T)$ at the root satisfies the following equalities:

$$\pi_A = \pi_T \text{ and } \pi_C = \pi_G.$$

4. Finally, we can obtain the most general nucleotide substitution model if we do not take into account any information regarding the substitution processes. This model will be referred to as the *general Markov* model. In this model, it is assumed that each substitution occurs with a different probability.

Remark 3.1.1. If one considers a two-state nucleotide substitution model, then there is the *Cavender-Farris-Neyman (CFN)* model [18, 32, 67] which assumes that the transition matrix is of the form:

$$\begin{pmatrix} a & b \\ b & a \end{pmatrix}.$$

In practice, a continuous-time Markov process specified by the nucleotide substitution process uses rate matrices that specify the instantaneous rates of change of each nucleotide along the sequence. The most common approach to model the mutation process is by a time-homogeneous CTMC.

Definition 3.1.2. The *continuous-time Jukes-Cantor (JC)* model is the time-homogeneous CTMC such that its rate matrices follow the symmetries in the JC model. In a similar fashion, we can define the *continuous-time Kimura 2-parameter (K2P)* model, the *continuous-time Kimura 3-parameter (K3P)* model, the *continuous-time strand symmetric* model, the *continuous-time general Markov* model, and the *continuous-time Cavender-Farris-Neyman (CFN)* model.

3.1.2 A phylogenetic model on phylogenetic trees

In phylogenetics, it was originally assumed that the evolutionary relationships between species can be represented by a phylogenetic tree. The idea of representing phylogenetics in terms of trees dates back to the work of Charles Darwin in *On The Origin of Species* [25] and the work of Edward Hitchcock in his book *Elementary Geology* [45]. Nowadays, it is believed that the evolutionary relationships between species are better described using a graph or a network which may contain cycles. This is due to recent findings in biology on the hybridization, horizontal gene transfer, and gene recombination processes. In this section, we start by introducing phylogenetic models on trees, and later in Section 3.4, we generalize phylogenetic models to networks.

In the rest of the thesis, we only consider binary trees. We start our discussion with the notion of phylogenetic trees. A *phylogenetic tree* $T = (V, E)$ is a rooted binary tree whose leaves are labeled by a given set of observed species. In order to build a phylogenetic model on a tree, a random variable X_v is associated to

each vertex $v \in V$. The random variable X_v has k possible states chosen from the state space S. For the DNA sequence, $S = \{\mathtt{A},\mathtt{G},\mathtt{C},\mathtt{T}\}$ and $k = 4$. Moreover, every edge $e \in E$ is labeled by a transition matrix that reflects probabilities of change of the states from a vertex to its child.

In Section 3.1, we discussed how to model evolution using a time-homogeneous continuous-time Markov process. Let $T = (V, E)$ be a phylogenetic tree with m leaves. Throughout this section, we label the leaves of T by the set $[m]$. As soon as the rate matrix $Q^{(e)}$ is specified for a fixed edge $e \in E$, by computing the matrix exponential

$$P^{(e)}(t_e) = \exp(t_e Q^{(e)}),$$

we obtain the probabilities of change from any base to any other during the evolutionary time t_e. This approach in modeling evolution using time-homogeneous CTMC is often used to approximate the biological realities in which transition rates vary over time [46]. Additionally, it is believed that in practice, modeling evolutionary processes as a time-inhomogeneous CTMC is not statistically feasible [81].

We now summarize how to compute the probability of observing a nucleotide in an organism at a site of a DNA sequence. For each $v \in V$ and $i \in S$, we want to compute the probability $P(X_v = i)$. For each non-root vertex $v \in V$, let the vertex $a(v)$ be the unique parent of v. Let $e_v \in E$ be the directed tree edge connecting $a(v)$ and v. Suppose that the transition probabilities of changes of states from $a(v)$ to v are given by a Markov matrix $P^{(e_v)} \in M_k(\mathbb{R})$. Then

$$P(X_v = j) = \sum_{i=1}^{k} P_{ij}^{(e_v)} \cdot P(X_{a(v)} = i). \tag{3.1}$$

Using (3.1), a joint probability distribution

$$p_{i_1 i_2 \ldots i_{|V|}} := P(X_{v_k} = i_k : v_k \in V \text{ for } k \in [|V|])$$

on all random variables X_v for $v \in V$ can be easily computed as well. Since our existing species correspond to the leaves of T, we need to take the marginal distribution at the leaves

$$p_{i_1 i_2 \ldots i_m} := P(X_1 = i_1, X_2 = i_2, \ldots, X_m = i_m) = \sum_{v_k \text{ is non-leaf}} \sum_{i_k \in S} p_{i_1 i_2 \ldots i_{|V|}}.$$

Using a genetic system consisting of k letters, there are k^m of these probabilities. Using this approach, the unknown parameters are the transition matrices $P^{(e)}$ for each $e \in E$ and the root distribution π. In the general Markov model, each matrix entry of $P^{(e)}$ is an independent parameter. The number of parameters will be smaller in the JC and Kimura models because some entries of the transition matrix are assumed to be equal. Equation (3.1) implies that there exists a polynomial *parameterization map* φ_T for the joint distribution of the states at the leaves. Suppose that S_T is the parameter space consisting of transition matrices for every edge and the root distribution associated to

the tree $T = (V, E)$. Moreover, we also assume that the parameter space S_T is a full-dimensional subset of $\mathbb{R}^n$ for some $n \in \mathbb{N}$. Then we have the following complex polynomial map

$$\varphi_T : S_T \to \Delta^{k^m - 1}$$

$$\theta \mapsto \mathbf{p}_T = (p_\omega)_{\omega \in S^m}$$

where Δ^{k^m-1} is the probability simplex which consists of nonnegative vectors in $\mathbb{R}^{k^m-1}$ with unit sum.

Definition 3.1.3. A *phylogenetic model on a phylogenetic tree* T, denoted by M_T, is defined as the image of the polynomial map φ_T.

That is to say, the model M_T contains all probability distributions obtained from T by varying the parameters in S_T.

3.2 The group-based model and its Fourier transform

In this section, we restrict our consideration to a special class of nucleotide substitution models with very nice properties. Namely, via a linear change of coordinates, the transition matrices belonging to these models can all be simultaneously diagonalizable. This class of models is called the group-based models. This class of models includes the CFN model, the JC, and the Kimura models mentioned in the previous section.

In the rest of this chapter, the group $\mathcal{G}$ in our consideration will be written additively and its identity element is 0. In this thesis, we assume that the group is always finite abelian.

Definition 3.2.1 (Publication I, Definition 1)**.** Given a group $\mathcal{G}$, a *labeling* of $\mathcal{G}$ is any function $L : \mathcal{G} \to \mathcal{L}$ for some finite set of labels $\mathcal{L}$.

Definition 3.2.2 (Publication I, Section 2)**.** A nucleotide substitution model is a group-based model with an underlying group $\mathcal{G}$ and a labeling L of $\mathcal{G}$ if for any edge $e \in E$ in the phylogenetic tree $T = (V, E)$ and the transition matrix $P^{(e)} = \exp(t_e Q^{(e)})$ along the edge e, the entries of the substitution rate matrix $Q^{(e)}$ along the edge e are given by

$$Q^{(e)}_{g,h} = \psi^{(e)}(h - g) \text{ for all } g, h \in \mathcal{G},$$

and for some vector $\psi^{(e)} \in \mathbb{R}^{\mathcal{G}}$ whose components sum to zero such that the following properties hold:

1. for every $g \in \mathcal{G} \setminus \{0\}$, $\psi^{(e)}(g) \geq 0$, and
2. if $L(g) = L(h)$, then $\psi(g) = \psi(h)$.

A rate matrix $Q^{(e)}$ satisfying the above three properties is called a $(\mathcal{G},L)$-rate matrix. In group-based models, we usually assume that $Q^{(e)}_{g,h} = Q^{(e)}_{h,g}$ for any $g,h \in \mathcal{G}$. That is to say, the rate matrices are assumed to be symmetric. This fact is equivalent to $\psi^{(e)}(-g) = \psi^{(e)}(g)$. Under this symmetricity assumption, by taking the matrix exponential of the rate matrix $Q^{(e)}$, the transition matrix $P^{(e)}$ satisfies

$$P^{(e)}_{g,h} = f^{(e)}(h-g)$$

for some nonnegative vector $f^{(e)} \in \mathbb{R}^{\mathcal{G}}$ with entries summing to one and $f^{(e)}(g) = f^{(e)}(-g)$ for all $g \in \mathcal{G}$.

However, in general, it is not true that the equality $\psi^{(e)}(g) = \psi^{(e)}(h)$ implies that the equality $f^{(e)}(g) = f^{(e)}(h)$ holds as well. For an instance, see Example 2 of Publication I. In Section 3.3, we introduce a class of special labeling functions called $\mathcal{G}$-compatible labelings which guarantee that the symmetries possessed by the rate matrix are preserved under the matrix exponential. In this case, we say that the transition matrix $P^{(e)}$ is a $(\mathcal{G},L)$-Markov matrix. It follows from the definition that different labelings on the same group give rise to different group-based models. In Example 1 of Publication I, one sees that the group-based models include the JC, K2P, and K3P models corresponding to three different labeling functions whose underlying group is $\mathcal{G} = \mathbb{Z}_2 \times \mathbb{Z}_2$. In these group-based models, we use the following identifications of nucleotides with the group elements of $\mathbb{Z}_2 \times \mathbb{Z}_2$: $\mathtt{A} = (0,0), \mathtt{T} = (0,1), \mathtt{C} = (1,0),$ and $\mathtt{G} = (1,1)$.

It is known that the set of characters of $\mathcal{G}$ consisting of all group homomorphism from $\mathcal{G}$ to $\mathbb{C}^*$ forms a group under multiplication and it will be referred to as the *character group* $\widehat{\mathcal{G}}$ of $\mathcal{G}$. It can also be easily seen that there is a canonical isomorphism between $\mathcal{G}$ and $\widehat{\mathcal{G}}$. The image of $g \in \mathcal{G}$ under this isomorphism is denoted by $\widehat{g} \in \widehat{\mathcal{G}}$.

Definition 3.2.3. Let $\mathcal{G}$ be a group. Then the *discrete Fourier transform* of a function $a : \mathcal{G} \to \mathbb{C}$ is the complex-valued function $\check{a} : \mathcal{G} \to \mathbb{C}$ satisfying

$$\check{a}(g) = \sum_{h \in \mathcal{G}} a(h) \cdot \widehat{g}(h).$$

In Publication II, we study group-based models that are endowed with symmetric labelings: $L(-g) = L(g)$ for all $g \in \mathcal{G}$. We call such models *symmetric group-based models*. In a symmetric group-based model, we assume that the transition matrices are real symmetric matrices in a symmetric group-based model. As a consequence, the vectors $\check{\psi}$ and $\check{f}$ are real functions.

The discrete Fourier transform is a linear map in the space $\mathbb{C}^{\mathcal{G}}$ of complex-valued functions on a group $\mathcal{G}$ to itself. Therefore, we can represent it as a matrix and we denote its matrix representation by K. For $g,h \in \mathcal{G}$, the (g,h)-entry of K is given by $K_{g,h} = \widehat{g}(h)$. Moreover, if $\mathcal{G}$ is finite abelian, then K is symmetric [61, Section 3.2]. The matrix K is invertible and $K^{-1} = \frac{1}{|\mathcal{G}|}\overline{K}^T$ [61, Corollary 3.2.2].

In a symmetric group-based model, there exist some relations between the functions $f^{(e)}$ and $\psi^{(e)}$ that specify the transition matrix $P^{(e)}$ and the rate matrix

$Q^{(e)}$ belonging to the model, respectively. More precisely, it holds that $\check{f}^{(e)}(g) = e^{\check{\psi}^{(e)}(g)}$ for all $g \in \mathscr{G}$ as a consequence of the matrix exponential. Furthermore, for any $g \in \mathscr{G}$, $\check{\psi}^{(e)}(g)$ is an eigenvalue of $Q^{(e)}$ with an eigenvector given by the column vector of the discrete Fourier matrix K indexed by the element g. Similarly, for any $g \in \mathscr{G}$, $\check{f}^{(e)}(g)$ is an eigenvalue of $P^{(e)}$ with an eigenvector given by the column vector of K indexed by the element g. More details can be found in [65, Lemma 2.2]. It implies that both matrices $Q^{(e)}$ and $P^{(e)}$ can be diagonalized via the matrix K such that the diagonal entries of the corresponding diagonal matrices are given by $\check{\psi}^{(e)} \in \mathbb{R}^{\mathscr{G}}$ and $\check{f}^{(e)} \in \mathbb{R}^{\mathscr{G}}$, respectively. For simplicity, from now on, we will drop the superscript in the rate and Markov matrices by fixing an edge of a phylogenetic tree.

Example 3.2.4. The matrix representation of the discrete Fourier transform for the group $\mathscr{G} = \mathbb{Z}_2 \times \mathbb{Z}_2$ is given by

$$K = \begin{pmatrix} 1 & 1 & 1 & 1 \\ 1 & -1 & 1 & -1 \\ 1 & 1 & -1 & -1 \\ 1 & -1 & -1 & 1 \end{pmatrix}.$$

For rate or transition matrices belonging to the K3P model, we have

$$K^{-1}.\begin{pmatrix} a & b & c & d \\ b & a & d & c \\ c & d & a & b \\ d & c & b & a \end{pmatrix}.K = \begin{pmatrix} a+b+c+d & 0 & 0 & 0 \\ 0 & a-b+c-d & 0 & 0 \\ 0 & 0 & a+b-c-d & 0 \\ 0 & 0 & 0 & a-b-c+d \end{pmatrix}$$

3.3 The embedding problem in phylogenetics and some embeddability criteria

We will formulate the embedding problem in the context of phylogenetics and we summarize the main results of Publications I and III which aim to answer the embedding problem.

In the embedding problem, we ask whether a Markov matrix P has a time-homogeneous CTMC realization. Namely, we want to decide whether a Markov matrix $P = \exp(tQ)$ for some rate matrix Q and $t \in \mathbb{R}_{\geq 0}$. It can be easily seen that tQ is a rate matrix for any $t \geq 0$ if and only if Q is a rate matrix.

Definition 3.3.1. A Markov matrix $P \in M_n(\mathbb{R})$ is said to be *embeddable* if there exists a rate matrix $Q \in M_n(\mathbb{R})$ satisfying $P = \exp(Q)$. A *Markov generator* for P is a rate matrix Q that satisfies the exponential equation.

Elfving was believed to be the first mathematician to pose the problem for finding a valid Markov generator of a given Markov matrix [30]. This problem became known as the embedding problem.

There are many applications of the embeddability property varying from phylogenetics to mathematical finance. In Section 3.1.2, in the context of phylogenetics, we have seen one approach in modeling evolution as a time-homogeneous continuous-time Markov process. A different approach appears when one only considers the conditional probabilities between the original and the final DNA sequences by considering the evolutionary process as a whole. In this case, we do not care about the mutation rates. By studying the embedding problem, one is able to connect these two approaches in modeling evolution.

We present some known embeddability criteria for Markov matrices of small sizes. The following theorem suggests that checking the embeddability of a Markov matrix of order two is equivalent to checking whether the matrix is invertible or not which is a rather easier condition to check.

Theorem 3.3.2 ([56]). *Let $P \in M_2(\mathbb{R})$ be a Markov matrix. The following statements are equivalent:*

1. *P is embeddable.*
2. *P is invertible.*
3. *$tr(P) > 1$.*

Since Markov matrices are real matrices, its complex eigenvalues occur in complex conjugate pairs.

Theorem 3.3.3 ([23]). *Let $P \in M_3(\mathbb{R})$ be a Markov matrix. Let*

$$P = Z \cdot \mathrm{diag}(1, \lambda, \overline{\lambda}) \cdot Z^{-1}$$

be an eigendecomposition of P.

1. *If $\lambda > 0$, then P is embeddable if and only if its principal logarithm is a rate matrix.*
2. *Suppose that $\lambda < 0$. If P is embeddable, then the matrix*

$$Z \cdot \mathrm{diag}(0, \log|\lambda|, \log|\lambda|) \cdot Z^{-1}$$

   is a rate matrix.

3. *If $\lambda \in \mathbb{C} \setminus \mathbb{R}$, then P is embeddable if and only if its principal logarithm is a rate matrix or the matrix*

$$Z \cdot \mathrm{diag}(0, \log_{-1}(\lambda), \log_{-1}(\overline{\lambda})) \cdot Z^{-1}$$

   is a rate matrix.

In addition to the above theorem, the following embeddability criteria appear in the literature. In [12], a characterization of the embeddability for Markov matrices of size three with repeated negative eigenvalues was presented, and in

fact, this problem is equivalent to showing whether a square root of a Markov matrix that satisfies some constraints exists or not. Moreover, more explicit embeddability criteria for this case were given in [20]. For a Markov matrix P with distinct eigenvalues, an explicit embeddability criterion was presented in [51]. This criterion utilizes an expression of the logarithm of P as a linear combination of some powers of P. Interested readers can refer to [51, Theorem 1.1] for more details. Finally, in [50], the authors presented some conditions that guarantee the existence of a Markov generator for a Markov matrix in the context of mathematical finance and credit ratings. Moreover, they also provide a method on how to search for Markov generators.

For Markov matrices or size four, the embedding problem has been completely solved in [14]. Moreover, in the same paper, the authors established a characterization of embeddable Markov matrices for larger Markov matrices with distinct eigenvalues. They also came up with an algorithm to enumerate all its Markov generators. In addition to the embeddability of general Markov matrices of size four, the embedding problem for the JC and Kimura models has been specifically studied in [72] and [13]. We summarize the embeddability criteria in Theorem 3.3.4 and 3.3.5.

Theorem 3.3.4 ([72], Corollary 3.1)**.** *Let P be a K3P Markov matrix with distinct eigenvalues $\{1, x, y, z\}$. Then P is embeddable if and only if its eigenvalues satisfy the following inequalities*

$$x, y, z > 0, \quad x \geq yz, \quad y \geq xz, \quad z \geq xy.$$

Moreover, it was proven in [72] that if a K3P Markov matrix with distinct eigenvalues is embeddable, then it has a K3P Markov generator.

Markov matrices in the K2P and JC models have repeated eigenvalues. If a Markov matrix has repeated eigenvalues, then it has infinitely many real logarithms [22]. Hence in order to conclude that a Markov matrix with repeated eigenvalues is embeddable, we need to check whether any of these real logarithms is a rate matrix. A discussion on the embeddability of K2P and JC matrices which have repeated eigenvalues was also presented in [72, 13]. The following theorem characterizes the embeddability of K2P Markov matrices.

Theorem 3.3.5 ([13], Corollary 3.9)**.** *Let P be a K2P Markov matrix having eigenvalues $1, x, y, y$.*

1. *If $y = 0$, then P is not embeddable.*
2. *If $y > 0$, then P is embeddable if and only if $y^2 \leq x$.*
3. *If $y < 0$, then P is embeddable if and only if $y^2 \leq x \leq e^{-2\pi}$.*

In [72, Section 3], the authors displayed some examples of embeddable K3P Markov matrices with no K3P Markov generators. Motivated by these examples, in Publication I, we investigate a variation of the classical embedding problem

in a general group-based model. Namely, given a Markov matrix P, we also impose that the rate matrix Q satisfying $P = \exp(Q)$ belongs to a group-based model used to model the evolution.

Definition 3.3.6 (Publication I)**.** Given a group-based model associated with a group $\mathcal{G}$ and a labeling L of $\mathcal{G}$, a $(\mathcal{G},L)$-Markov matrix P is said to be $(\mathcal{G},L)$-*embeddable* if there exists a $(\mathcal{G},L)$-rate matrix Q satisfying $P = \exp(Q)$.

We will consider the following class of labeling functions that preserves the symmetries of the entries of a rate matrix under the exponential map. Suppose that the vector x_L is the column vector whose g-th component is given by the variable $x_{L(g)}$.

Definition 3.3.7 (Publication I, Definition 2)**.** Given a group $\mathcal{G}$, let K be the matrix representation of the discrete Fourier transform for $\mathcal{G}$. The labeling L on $\mathcal{G}$ is called $\mathcal{G}$-*compatible* if for any $g_1, g_2 \in \mathcal{G}$ with $L(g_1) = L(g_2)$, the following equations hold:

$$K_{g_1,:} \cdot x_L = K_{g_2,:} \cdot x_L \text{ and } (K^{-1})_{g_1,:} \cdot x_L = (K^{-1})_{g_2,:} \cdot x_L,$$

where $K_{g,:}$ denotes the row of K labeled by $g \in \mathcal{G}$.

Given a symmetric $\mathcal{G}$-compatible labeling L, if Q is a $(\mathcal{G},L)$-rate matrix, then it can be checked that the Markov matrix obtained from the exponentiation $P = \exp(Q)$ is an $(\mathcal{G},L)$-Markov matrix.

The following theorem is the main result of Publication I. This theorem characterizes a $(\mathcal{G},L)$-embeddable transition matrix in a symmetric group-based model with a $\mathcal{G}$-compatible labeling in terms of its eigenvalues.

Theorem 3.3.8 (Publication I, Theorem 1)**.** *Let $\mathcal{G}$ be an abelian group and $L : \mathcal{G} \to \mathcal{L}$ be a symmetric $\mathcal{G}$-compatible labeling. Then a $(\mathcal{G},L)$-Markov matrix P is $(\mathcal{G},L)$-embeddable if and only if its eigenvalues $\lambda = (\lambda_g)_{g\in\mathcal{G}} \in \mathbb{R}^{\mathcal{G}}$ satisfy the following properties:*

1. $\lambda_0 = 1$,
2. $\lambda_g > 0$ *for each* $g \in \mathcal{G}$,
3. *for each nonzero* $g \in \mathcal{G}$, $\prod_{h\in\mathcal{G}} \lambda_h^{Re((K)_{g,h})} \geq 1$, *and*
4. *if* $L(g) = L(h)$, *then* $\lambda_g = \lambda_h$.

By applying the result of Theorem 3.3.8 to the JC, K2P, and K3P models, we will recover the embeddability criteria presented earlier in [72] and [13]. This is expected because the JC, K2P, and K3P models are group-based models equipped with $\mathcal{G}$-compatible labelings.

Recently, a possible genetic system consisting of eight building blocks was introduced in [48]. We refer to this genetic system as the *hachimoji DNA* system. In this DNA system, there are four additional synthetic nucleotides: `S`, `B`, `Z`,

and P together with the four standard nucleotides. The chemical bonds among these synthetic nucleotides are B S and P Z. More chemical properties of these four additional nucleotides are given in [48]. In Publication I, we propose three different group-based models for the hachimoji DNA system with the underlying group structure given by $\mathbb{Z}_2 \times \mathbb{Z}_2 \times \mathbb{Z}_2$. For these group-based models, using the main result in Theorem 3.3.8, we provide approximations of how large the set of embeddable Markov matrices in certain biologically meaningful subsets of the Markov matrices of size 8 are. This concludes the main results of Publication I.

We will now discuss the main results in Publication III which concern the embeddability of centrosymmetric matrices. It is mentioned in Section 3.1 that if one takes into account the complementarity of the two DNA strands, then one should consider the strand symmetric (SS) model. The algebraic structure of the SS model was first studied in [16]. The JC, K2P, and K3P models are special cases of the SS model. The transition matrices in the SS model belong to the class of matrices known as centrosymmetric matrices of size 4.

Definition 3.3.9. A matrix $A = (a_{ij}) \in M_n(\mathbb{R})$ is called *centrosymmetric (CS)* if for all $i, j \in [n]$,

$$a_{i,j} = a_{n+1-i,n+1-j}.$$

We have seen earlier that in the case of a group-based model, the transition matrices can be diagonalized via the discrete Fourier matrix. In the case of the SS model, it is possible to block-diagonalize an SS Markov matrix. Let us consider the matrix

$$S = \begin{pmatrix} 1 & 0 & 0 & 1 \\ 0 & 1 & 1 & 0 \\ 0 & 1 & -1 & 0 \\ 1 & 0 & 0 & -1 \end{pmatrix}. \tag{3.2}$$

We define the map $F_4 : M_4(\mathbb{R}) \to M_4(\mathbb{R})$ such that for any $P = (p_{ij}) \in M_4(\mathbb{R})$, $F(P) := S^{-1}PS$. If P is an SS matrix, then

$$F(P) := \begin{pmatrix} p_{11}+p_{14} & p_{12}+p_{13} & 0 & 0 \\ p_{21}+p_{24} & p_{22}+p_{23} & 0 & 0 \\ 0 & 0 & p_{22}-p_{23} & p_{21}-p_{24} \\ 0 & 0 & p_{12}-p_{13} & p_{11}-p_{14} \end{pmatrix}.$$

It can be seen that if P is a Markov (rate) matrix, then the upper left block matrix in $F(P)$ is a Markov (rate) matrix as well. Unlike the upper left block, the lower right block matrix does not admit such nice properties. Suppose that

$$\begin{aligned} &\lambda := p_{11}+p_{14}, \quad \mu := p_{22}+p_{23}, \\ &\alpha := p_{22}-p_{23}, \quad \alpha' := p_{21}-p_{24}, \\ &\beta := p_{11}-p_{14}, \quad \beta' := p_{12}-p_{13}, \\ &\Delta := (\alpha-\beta)^2 + 4\alpha'\beta'. \end{aligned} \tag{3.3}$$

In Publication III, we consider only SS Markov matrices whose eigenvalues are positive and pairwise different. Let $P = (p_{ij})$ be such an SS Markov matrix. By direct computations, one can check that the four distinct positive eigenvalues of P are given by:

$$1, \quad \lambda_1 := \lambda + \mu - 1, \quad \lambda_2 := \frac{(\alpha+\beta)+\sqrt{\Delta}}{2} \quad \text{and} \quad \lambda_3 = \frac{(\alpha+\beta)-\sqrt{\Delta}}{2}. \qquad (3.4)$$

The following theorem is the first main result of Publication III which enumerates all real logarithms of an SS Markov matrix having distinct positive eigenvalues. The main tool to prove this theorem is Proposition 2.3.13 that lists all possible real logarithms of a Markov matrix such that each row of this logarithm sums to one.

Theorem 3.3.10 (Publication III, Theorem 3.6)**.** *Let P be an SS Markov matrix having positive and distinct eigenvalues 1, $\lambda_1, \lambda_2, \lambda_3$ given as in* (3.4)*. Let*

$$x := \log(\lambda_1), \qquad y_k := \log(\lambda_2) + 2k\pi i, \qquad z_k := \log(\lambda_3) - 2k\pi i,$$

such that if $\Delta > 0$, then $k = 0$ and if $\Delta < 0$, then $k \in \mathbb{Z}$. Then P admits real logarithms given by

$$S \begin{pmatrix} \alpha_1 & -\alpha_1 & 0 & 0 \\ -\beta_1 & \beta_1 & 0 & 0 \\ 0 & 0 & \delta(k) & \varepsilon(k) \\ 0 & 0 & \phi(k) & \gamma(k) \end{pmatrix} S^{-1},$$

where

$$\alpha_1 = \frac{1-\lambda}{2-\lambda-\mu}x, \qquad \beta_1 = \frac{1-\mu}{2-\lambda-\mu}x,$$

$$\delta(k) = \frac{1}{2}((y_k+z_k)+(\alpha-\beta)\frac{(y_k-z_k)}{\sqrt{\Delta}}), \qquad \varepsilon(k) = \alpha'\frac{(y_k-z_k)}{\sqrt{\Delta}},$$

$$\phi(k) = \beta'\frac{(y_k-z_k)}{\sqrt{\Delta}}, \qquad \gamma(k) = \frac{1}{2}((y_k+z_k)-(\alpha-\beta)\frac{(y_k-z_k)}{\sqrt{\Delta}}).$$

Corollary 3.3.11 (Publication III, Theorem 3.6)**.** *Let P be an SS Markov matrix having positive and distinct eigenvalues $1, \lambda_1, \lambda_2, \lambda_3$ given as in* (3.4)*. Then any real logarithm $Q = (q_{ij})$ of P is an SS matrix whose entries are:*

$$q_{11} = \frac{\alpha_1+\gamma(k)}{2}, \quad q_{12} = \frac{-\alpha_1+\phi(k)}{2}, \quad q_{13} = \frac{-\alpha_1-\phi(k)}{2}, \quad q_{14} = \frac{\alpha_1-\gamma(k)}{2},$$

$$q_{21} = \frac{-\beta_1+\varepsilon(k)}{2}, \quad q_{22} = \frac{\beta_1+\delta(k)}{2}, \quad q_{23} = \frac{\beta_1-\delta(k)}{2}, \quad q_{24} = \frac{-\beta_1-\varepsilon(k)}{2}.$$

We now provide the second main result of Publication III. The theorem provides a characterization of the embeddable SS Markov matrices with positive and distinct eigenvalues.

Theorem 3.3.12 (Publication III, Theorem 3.7)**.** *An SS Markov matrix P having distinct eigenvalues $1, \lambda_1, \lambda_2, \lambda_3$ is embeddable if and only if the following inequalities are satisfied for $k = 0$ if $\Delta > 0$ or for some $k \in \mathbb{Z}$ if $\Delta < 0$:*

$$\lambda_1 > 0, \qquad (\alpha+\beta)^2 > \Delta, \qquad |\phi(k)| \leq -\alpha_1,$$

$$|\varepsilon(k)| \leq -\beta_1, \qquad \gamma(k) \leq \alpha_1, \qquad \delta(k) \leq \beta_1.$$

By applying the embeddability criteria in Theorem 3.3.12 to the case of the K3P model, which is a special case of the SS model, we recover the embeddability criteria in the K3P model presented in [72]. Moreover, these criteria allow us to approximate the size of the set of embeddable SS Markov matrices inside the space of all Markov matrices of size four having positive and distinct eigenvalues and also inside some of its biologically relevant subsets. These proportions can be observed in Table 5 and Table 7 in Publication III.

In Publication III, we also consider CS Markov matrices of order larger than 4. The biological applications of higher-order CS matrices can be justified by the existence of synthetic nucleotides. In synthetic biology, scientists aim to expand the genetic alphabet system to include synthetic unnatural base pairs which are functionally equivalent to the natural base pairs. Earlier we have seen the hachimoji DNA system consisting of eight DNA alphabets. In [64], a fully functional genetic alphabet system consisting of six letters was established. In this system, in addition to the two natural base pairs, a pair of synthetic bases `5SICS MMO2` was added to the genetic alphabet system. This unnatural base pair was shown to be biologically comparable to a natural base pair. This synthetic biological research motivates us to investigate the higher-order CS Markov matrices.

For higher-order CS matrices, we generalize the block-diagonalization procedure using the matrix S in (3.2). Namely, there exists a matrix $S_n \in M_n(\mathbb{R})$ that can block-diagonalize a CS matrix of size n. More details on the matrix S_n can be checked in Section 5.1 of Publication III.

Lemma 3.3.13 (Publication III, Lemma 5.6)**.** *Let $P \in M_n(\mathbb{R})$ be a CS matrix. Then*

$$S_n^{-1} P S_n = \operatorname{diag}(P_1, P_2),$$

where $P_1 \in M_n(\mathbb{R}^{\lceil \frac{n}{2} \rceil \times \lceil \frac{n}{2} \rceil})$ and $P_2 \in M_n(\mathbb{R}^{\lfloor \frac{n}{2} \rfloor \times \lfloor \frac{n}{2} \rfloor})$. Additionally, if P is a Markov matrix, so is P_1. Similarly, if P is a rate matrix, then so is P_1.

As a consequence, this block-diagonal representation of a CS Markov matrix allows us to provide a necessary condition on the set of embeddable matrices.

Lemma 3.3.14 (Publication III, Lemma 5.12)**.** *Let $n \geq 2$. Let $P = (p_{ij}) \in M_n(\mathbb{R})$ be an embeddable CS Markov matrix with a CS logarithm.*

1. *If n is even, then*

$$\sum_{j=1}^{\frac{n}{2}} (p_{jj} + p_{j,n-j+1}) > 1.$$

2. *If n is odd, then*

$$\sum_{j=1}^{\lfloor \frac{n}{2} \rfloor} (p_{jj} + p_{j,n-j+1}) + p_{\lfloor \frac{n}{2} \rfloor + 1, \lfloor \frac{n}{2} \rfloor + 1} > 1.$$

The previous lemma allows us to give an upper bound to the volume of the set of embeddable CS Markov matrices inside the space of CS Markov matrices. This upper bound is not difficult to compute since we only have linear inequalities defining the necessary conditions. For small values of n, these estimates can be seen in Table 8 of Publication III.

As a concrete example, in Publication III we particularly study the case of 6×6 CS Markov matrices with distinct eigenvalues and we present some embeddability criteria. Given a 6×6 CS Markov matrix, we prove in Lemma 3.3.13 that $S_6^{-1} P S_6$is a block-diagonal real matrix such that P_1 is a Markov matrix of size three. Suppose that we have the following eigendecompositions of P_1 and P_2:

$$P_1 = N_1 \cdot \mathrm{diag}(1, \lambda_1, \lambda_2) \cdot N_1^{-1} \text{ and } P_2 = N_2 \cdot \mathrm{diag}(\mu, \gamma_1, \gamma_2) \cdot N_2^{-1}$$

with $\mu \in \mathbb{R}_{>0}$ and $\lambda_i, \gamma_i \in \mathbb{C}$ and for some matrices $N_1, N_2 \in GL_3(\mathbb{C})$. We will define the matrix $Z := S_6 \cdot \mathrm{diag}(N_1, N_2)$, where S_6 is the matrix used to block-diagonalize the 6×6 CS matrices.

The following theorem in Publication III completely characterizes the embeddability of CS Markov matrices of size 6 with distinct eigenvalues based on their set of eigenvalues.

Theorem 3.3.15 (Publication III, Proposition 11-15)**.** *1. If $\lambda_i, \gamma_i \in \mathbb{R}_{>0}$ for $i \in [2]$, then P is embeddable if and only if its principal logarithm is a rate matrix.*

2. *Suppose that $\lambda_i \in \mathbb{R}_{>0}$ and $\gamma_i \in \mathbb{C} \setminus \mathbb{R}$ for $i \in [2]$. Let*

$$V := Z \cdot \mathrm{diag}(0, 0, 0, 0, 2\pi i, -2\pi i) \cdot Z^{-1}. \tag{3.5}$$

Suppose that

$$\mathscr{L} := \max_{(i,j):\ i \neq j,\ V_{i,j} > 0} \left\lceil -\frac{\mathrm{Log}(P)_{i,j}}{V_{i,j}} \right\rceil, \qquad \mathscr{U} := \min_{(i,j):\ i \neq j,\ V_{i,j} < 0} \left\lfloor -\frac{\mathrm{Log}(P)_{i,j}}{V_{i,j}} \right\rfloor.$$

Moreover, let $\mathscr{N} := \{(i,j) : i \neq j,\ V_{i,j} = 0 \text{ and } \mathrm{Log}(P)_{i,j} < 0\}$. The following statements hold:

   (a) *The matrix P is embeddable if and only if the set $\mathscr{N}$ is empty and $\mathscr{L} \leq \mathscr{U}$.*

   (b) *The set $\{Q = \mathrm{Log}(P) + kV : k \in \mathbb{Z} \text{ with } \mathscr{L} \leq k \leq \mathscr{U}\}$ is the set of Markov generators for P.*

3. *If $\lambda_i \in \mathbb{C} \setminus \mathbb{R}$ and $\gamma_i \in \mathbb{R}_{>0}$, then P is embeddable if and only if $\mathrm{Log}(P)$ or $\mathrm{Log}_{-1}(P)$ are rate matrices, where*

$$\mathrm{Log}_{-1}(P) := Z \cdot \mathrm{diag}(0, z, \bar{z}, \log(\mu), \log(\gamma_1), \log(\gamma_2)) \cdot Z^{-1} \text{ and } z := \log_{-1}(\lambda_1). \tag{3.6}$$

4. *Suppose that* $\lambda_i, \gamma_i \in \mathbb{C} \setminus \mathbb{R}$ *for* $i \in [2]$. *Let* $\mathrm{Log}_{0,0}(P)$ *denote the principal logarithm of* P *and* $\mathrm{Log}_{-1,0}(P)$ *denote the matrix in* (3.6). *Let* V *be the same matrix as in (3.5). For* $k \in \{0,-1\}$, *let us define:*

$$\mathscr{L}_k := \max_{(i,j):\ i\neq j,\ V_{i,j}>0} \left\lceil -\frac{\mathrm{Log}_{k,0}(P)_{i,j}}{V_{i,j}} \right\rceil, \qquad \mathscr{U}_k := \min_{(i,j):\ i\neq j,\ V_{i,j}<0} \left\lfloor -\frac{\mathrm{Log}_{k,0}(P)_{i,j}}{V_{i,j}} \right\rfloor.$$

Moreover, let $\mathscr{N}_k := \{(i,j) : i \neq j,\ V_{i,j} = 0 \text{ and } \mathrm{Log}_{k,0}(P)_{i,j} < 0\}$. *The following statements hold:*

   (a) P *is embeddable if and only if* $\mathscr{N}_k$ *is empty and* $\mathscr{L}_k \leq \mathscr{U}_k$ *for* $k = 0$ *or* $k = -1$.

   (b) *If* P *is embeddable, then one of its Markov generators is given by*

$$\mathrm{Log}_{k,k_2}(P) := Z \cdot \mathrm{diag}(0, \log_k(\lambda_1), \overline{\log_k(\lambda_1)}, \log(\mu), \log_{k_2}(\gamma_1), \overline{\log_{k_2}(\gamma_1)}) \cdot Z^{-1}$$

   with $k \in \{0,-1\}$ *and* $k_2 \in \mathbb{Z}$ *such that* $\mathscr{L}_k \leq k_2 \leq \mathscr{U}_k$.

5. *If the set of eigenvalues of* P *does not belong to any of the previous four cases, then* P *is not embeddable.*

By applying the previous theorem, we can approximate the proportion of the set of embeddable CS Markov matrices inside the set of all CS Markov matrices of size 6 and some of its relevant subsets. These proportions can be seen in Table 9 of Publication III. These approximate proportions of the set of embeddable CS Markov matrices of sizes four and six presented in Publication III suggest that modeling the evolutionary process as CTMC in the SS model is a strong restriction since we disregard non-embeddable matrices which are much larger in proportion compared to those that are embeddable. Hence one should take the above restrictions of modeling evolution as CTMC into consideration when working with the SS model.

3.4 The phylogenetic models on phylogenetic networks

In this section, we generalize phylogenetic models built on trees to the more general network setting. This generalization is biologically relevant due to the fact that, unlike phylogenetic trees, phylogenetic networks may be able to explain better many biological phenomena, including hybridization, horizontal gene transfer, and gene recombination [73, 69, 62]. In this section, we present a polynomial parameterization for some group-based models built on phylogenetic networks. Moreover, we show that we can apply the Fourier transformation to simplify the parameterization. We will see that in the case of trees, the parameterization will be reduced to a monomial parameterization.

We start by presenting a set of assumptions on the networks that we consider. In the rest of this chapter, we denote by X the set of taxa of our interest.

Moreover, we will always assume that our graphs/networks are binary unless otherwise stated.

Definition 3.4.1 ([40])**.** A *rooted phylogenetic network* N on X is a rooted connected simple DAG such that its leaf set is bijectively labeled by X and the other vertices are either *tree vertices* (vertices with in-degree one and out-degree two) or *reticulation vertices* (vertices with in-degree two and out-degree one).

Given a rooted phylogenetic network N, an edge directed into a reticulation vertex of N is called a *reticulation edge*. We say a leaf u of N is a *reticulation leaf* if its unique parent is a reticulation vertex of N. The set of reticulation leaves of N will be denoted by $r(N)$. The following definition provides the unrooted counterpart for phylogenetic networks.

Definition 3.4.2 ([34])**.** An *unrooted phylogenetic network* N on X is a connected undirected simple graph such that its leaf set is bijectively labeled by X and each vertex is either a tree vertex (vertex of degree three) or a leaf.

Let N be a (rooted/unrooted) phylogenetic network on X. A *(tree) cherry* is an undirected path $\{x_1, u, x_2\}$ such that x_1 and x_2 are leaves of N. Moreover, if N is rooted, then a *reticulated cherry* is an undirected path $\{x_1, u_1, u_2, x_2\}$ such that x_1 and x_2 are leaves of N and that exactly one of u_1 and u_2 is a reticulation vertex.

Definition 3.4.3 ([10, 84])**.** If every biconnected component of a rooted phylogenetic network contains at most k reticulation vertices, then the network is called a *level-k (rooted) network*. Similarly, if we can produce a phylogenetic tree by removing at most k edges in every biconnected component of an unrooted phylogenetic network and then by contracting each vertex of degree two to one of its neighbors then the network is called a *level-k (unrooted) network*.

The above definition can be used to measure how complex a phylogenetic network is in terms of the number of its reticulation vertices and also how far a phylogenetic network is from being a tree.

In this thesis, we will only consider the *semi-directed network topology* of a given phylogenetic network due to the time reversibility of the evolutionary process of the nucleotide substitution model which implies that the exact root position cannot be inferred. Given a rooted phylogenetic network N, the semi-directed topology of N can be obtained by collapsing the root of N and then by forgetting the direction of all tree edges but keeping the direction of the reticulation edges only.

In Section 3.1.2, we construct a phylogenetic model on a phylogenetic tree. We generalize this construction to the phylogenetic network setting. After that, we describe the use of discrete Fourier transform as a linear change of coordinates to simplify the polynomial parameterization of a phylogenetic model on a tree and network as well.

Let $k \in \mathbb{N}$. Let N be a level-k semi-directed network on X where $|X| = n$. We associate an $m \times m$ transition matrix $P^{(e)}$ to each edge e of N where m denotes

the cardinality of the state space of the Markov chain. In our case, $m = 4$ since we consider DNA data. Let $v_1, \ldots, v_r$ be the reticulation vertices of N where $r \geq k$. We introduce the following reticulation edge parameters. For $i \in [r]$, let e_i^0 and e_i^1 be the two edges directed into the reticulation vertex e_i. We then assign a parameter $\delta_i \in [0,1]$ to e_i^1 and $\delta_i' \in [0,1]$ to e_i^0 such that $\delta_i + \delta_i' = 1$. The parameter δ_i denotes the probability of independently keeping the edge e_i^1 but deleting e_i^0. Similarly, the parameter δ_i' denotes the probability of independently keeping the edge e_i^0 but deleting e_i^1. A vector $\sigma \in \{0,1\}^r$ of length r encodes the choices we make in keeping and/or deleting one of the two edges that are directed into a reticulation vertex. Namely, if σ_i denotes the ith component of σ, then $\sigma_i = 0$ means that e_i^0 is deleted and $\sigma_i = 1$ means that e_i^1 is deleted. Given a binary vector $\sigma \in \{0,1\}^r$, if we remove the edges according to σ, then after this process, we obtain a phylogenetic tree T_σ. For $\omega \in S^n$, let $(p_N)_\omega$ be the joint probability distribution of observing ω at the leaves where the nucleotide ω_i is observed at the leaf i. Then

$$(p_N)_\omega = \sum_{\sigma \in \{0,1\}^r} \left(\prod \delta_i^{1-\sigma_i} (1-\delta_i')_i^\sigma \right) (p_{T_\sigma})_\omega$$

where $(p_{T_n})_\omega$ denotes the joint probability of observing ω in the phylogenetic tree T_σ. We have seen in Section 3.1.2 that the joint probability $(p_{T_n})_\omega$ can be easily computed.

Let θ_N be the set of parameters on the network N consisting of the reticulation edge parameters and the entries of the transition matrices for each edge of N. Then we have the following complex polynomial map:

$$\begin{aligned} \varphi_N : \theta_N &\to \Delta^{k^m - 1} \\ \theta &\mapsto \mathbf{p}_N = (p_\omega)_{\omega \in S^n} \end{aligned} \tag{3.7}$$

Definition 3.4.4 ([40])**.** The *phylogenetic network model on a phylogenetic network N*, denoted by M_N, is defined as the image of the polynomial map φ_N.

Let T be a phylogenetic tree with n leaves. We now briefly discuss how to apply discrete Fourier transform to simplify the parameterization (3.7) of the model M_T under a group-based model. Namely, we also require that the transition matrices in the model M_T follow the symmetries determined by the group-based model. Let $\mathcal{G}$ be the underlying group for the group-based model. For each $\omega \in S^n$, we can easily compute the joint probability p_ω. To every split $A|B \in \Sigma(T)$ of the leaf set of T, we associate the variables $\{a_g^{A|B}\}_{g \in \mathcal{G}}$. The corresponding Fourier coordinate q_ω is given by the following monomial parameterization:

$$q_\omega = \begin{cases} \prod_{A|B \in \Sigma(T)} a_{\sum_{i \in A} \omega_i}^{A|B}, & \text{if } \sum_{i=1}^n \omega_i = 0 \\ 0, & \text{otherwise.} \end{cases}$$

More details on this linear change of coordinates can be found in [42, 31].

The most important property of this change of coordinates is that for trees, it reduces the polynomial parameterization of the joint probability p_ω for $\omega \in S^n$ for the model M_T into a monomial parameterization. Unlike in the trees setting, even though the Fourier transformation simplifies the complicated polynomial parameterization of the joint probability $(p_N)_\omega$ for a semi-directed network N, the resulting parameterization need not be a monomial parameterization. As an example, one can see Example 6 in Publication II. We will see in the subsequent section that we can use this Fourier transformation to distinguish two network models.

3.5 Generic identifiability and distinguishability of phylogenetic network models

In the previous section, we constructed a polynomial parameterization of a phylogenetic network model M_N. Given two phylogenetic network models, the goal of this section is to provide algebraic invariants that can be used to distinguish the two models. To achieve this goal, we briefly mention how we can associate a variety to a phylogenetic network model. Then we will define what we exactly mean by distinguishing two network models. Finally, we summarize some results of the distinguishability of certain classes of phylogenetic networks in Publication II.

Let N be a semi-directed phylogenetic network with n leaf and r reticulation vertices. We can see from the parameterization map φ_N in (3.7) that for any $\omega \in S^n$, p_ω is a homogeneous polynomial in the set of parameters θ_i and the degree of this polynomial is equal to the number of reticulation vertices plus the number of edges of the tree T_σ obtained after deleting exactly one of the two reticulation edges directed to each reticulation vertex according to $\sigma \in \{0,1\}^r$. In general, the leaf set of the tree T_σ is not equal to X.

In Publication II, we focus our study on the class of semi-directed networks called *funnel-free* networks. A semi-directed network is called funnel-free if no tree vertex and no reticulation vertex satisfy neither condition:

1. it is a parent of a reticulation vertex and as well as a child of two reticulation vertices of the network, or

2. it is a parent of two reticulation vertices of the network.

If the network is funnel-free, then the leaf set of the tree T_σ is equal to X for any $\sigma \in \{0,1\}^r$.

We consider the map φ_N in (3.7) as a complex polynomial map.

Definition 3.5.1. The *variety V_N associated with the model M_N* is defined as the Zariski closure of $\mathrm{Im}(\varphi_N)$.

Definition 3.5.2. The *vanishing ideal*, or simply just the ideal, *associated with*

the model M_N, denoted by $I_N \subseteq \mathbb{C}[p_\omega : \omega \in S^n]$, is defined as

$$I_N := \{f \in \mathbb{C}[p_\omega : \omega \in S^n] : f(v) = 0 \text{ for all } v \in V_N\}.$$

An element of the ideal I_N is called a *phylogenetic invariant* associated with the model M_N.

To compute the ideal associated with a phylogenetic model, we use the polynomial parameterization (3.7) and then the elimination theory to eliminate the model parameters corresponding to the transition matrices for each edge of the network and the reticulation edges. One approach to obtain a phylogenetic invariants is to compute a Gröbner basis of the vanishing ideal. An element of this Gröbner basis is a phylogenetic invariants associated with the model.

This notion of phylogenetic invariants for a phylogenetic tree was originally introduced in [17] and [59]. An extensive list of publications studying phylogenetic invariants includes [31, 2, 79, 76, 77, 82]. If a network is funnel-free, then it allows us to apply discrete Fourier transform to compute the phylogenetic invariants associated with the network model. If N is a tree, then the phylogenetic invariants associated with the model M_N have been well-studied. For trees with few leaves, a complete list of phylogenetic invariants has been collected in [15]. For bigger trees under group-based models, one can obtain a phylogenetic invariant by computing toric fiber products of ideals associated with some subtrees. Interested readers can check [79] for more details.

Given a family of parametric statistical models, one of the most important questions is the identifiability of the model parameters. We aim to study the identifiability of the network topology parameter for phylogenetic network models. This identifiability problem ensures that it is possible to find the unique network topology that matches the probability distribution observed in the DNA data.

We now define the notion of distinguishability of two network models.

Definition 3.5.3 ([40])**.** Let N_1 and N_2 be two distinct semi-directed networks with n leaves. If $V_{N_1} \cap V_{N_2}$ is a proper subvariety of V_{N_1} and of V_{N_2}, then N_1 and N_2 are called *distinguishable*. Otherwise, N_1 and N_2 are called *indistinguishable*.

The above definition suggests that in order to distinguish two network models associated with N_1 and N_2, we need to find two phylogenetic invariants f_1 and f_2 such that $f_1 \in I_{N_1} \setminus I_{N_2}$ and $f_2 \in I_{N_2} \setminus I_{N_1}$. This notion of distinguishability can be used to decide whether two models are generically identifiable or not.

Definition 3.5.4. Let N_1 and N_2 be two distinct semi-directed networks with n leaves. The network parameters are *generically identifiable* if the set of parameters θ in θ_{N_1} that φ_{N_1} maps into M_{N_2} has Lebesgue measure zero.

Proposition 3.5.5 ([40], Proposition 3.3)**.** *Let N_1 and N_2 be two distinct semi-directed networks with n-leaf. If N_1 and N_2 are distinguishable, then the associated network parameters are generically identifiable.*

The following theorems provide a full answer for a specific class of level-1 networks.

Theorem 3.5.6 ([41, 40, 47]). *For a fixed number of reticulation vertices, the network parameter of a phylogenetic network model under the JC, K2P, or K3P models is generically identifiable with respect to the class of models where the network parameter is an n-leaf level-1 triangle-free semi-directed network.*

The proof of the above theorem uses both combinatorial aspects of level-1 triangle-free semi-directed networks and algebraic tools that rely upon the previous results on the generic identifiability of semi-directed level-1 networks with exactly one undirected cycle with length at least 4. The case for the JC model was proven in [40] while the cases for the K2P and K3P models were proven in [47].

In Publication II, we aim to extend the results on the level-1 semi-directed networks to level-2 semi-directed networks. We present the following series of definitions before stating some of the main results of Publication II.

Definition 3.5.7 ([84]). A strict level-k rooted phylogenetic network, meaning that a network is level-k but not level-$(k-1)$, is said to be simple level-k if the set of all cut-edges of this network contains only the trivial cut-edges.

Definition 3.5.8 (Publication II, Definition 6.9). Let N_1 and N_2 be two distinct simple funnel-free level-2 semi-directed networks on X.

1. A pair (N_1, N_2) is said to be *N_1-reticulation-conflicting* (N_1-*RC*) if for every reticulation leaf x of N_1, there exists a reticulation leaf y of N_2 such that $d_{N_1}(x,y) = 3$.

2. A pair (N_1, N_2) is said to be *N_2-reticulation-conflicting* (N_2-*RC*) if for every reticulation leaf x of N_2, there exists a reticulation leaf y of N_1 such that $d_{N_2}(x,y) = 3$.

We are now ready to state the main results of Publication II on the distinguishability of level-2 semi-directed networks. We first present a result on networks with four leaves.

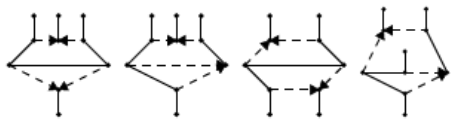

Figure 3.1. Funnel-free strict simple level-2 semi-directed networks on four leaves.

Proposition 3.5.9 (Publication II, Proposition 5.4). *Let N_1 and N_2 be two distinct 4-leaf level-2 semi-directed networks.*

(i) If N_1 is a simple funnel-free strict level-2 semi-directed network and N_2 is a level-1 network, then under the JC, K2P, or K3P model, $V_{N_1} \not\subseteq V_{N_2}$.

(ii) Suppose that N_1 and N_2 are both simple funnel-free strict level-2 semi-directed networks. If the underlying unlabelled semi-directed topology of N_1 is isomorphic to a type 1, 2, or 3 semi-directed network, and N_2 is isomorphic to a type 4 semi-directed network, then under the JC model, $V_{N_2} \not\subseteq V_{N_1}$. Here, the 4-leaf networks of type 1,2,3, and 4 can be seen in Figure (3.1). In these networks, the dashed arrows represent reticulation edges.

The main strategy to generalize the results on four leaves is to use the network restriction procedure [40, Definition 4.1] to obtain a subnetwork with four leaves and then apply Proposition 3.5.9. Given a semi-directed network N on X and $S \subseteq X$, the network $N|_S$ denotes the *restriction of N to S*. The following fact allows us to distinguish the original network using the distinguishability of certain subnetworks of the original networks.

Lemma 3.5.10 ([40])**.** *Let N_1 and N_2 be two distinct n-leaf semi-directed networks on X and $S \subseteq X$. If $V_{N|_S} \not\subseteq V_{M|_S}$, then $V_N \not\subseteq V_M$.*

The following series of theorems are some results presented in Publication II on the distinguishability of phylogenetic network models on at least five leaves that are obtained using the network restriction procedure. We start by presenting the distinguishability of two networks where each network has two reticulation leaves. Let us recall that the set of reticulation leaves of a phylogenetic network N is denoted by $r(N)$.

Theorem 3.5.11 (Publication II, Theorem 6.13)**.** *Let $n \geq 5$. Let N_1 and N_2 be two distinct n-leaf funnel-free strict simple level-2 semi-directed networks such that $|r(N_1)| = |r(N_2)| = 2$ and $r(N_1) \neq r(N_2)$. If the pair (N_1, N_2) is neither N_1-RC nor N_2-RC, then N_1 and N_2 are distinguishable.*

In the above theorem, we assume that $r(N_1) \neq r(N_2)$. If this assumption is not satisfied, then we may not be able to distinguish the networks. See Example A.2 in Appendix A of Publication II.

The following results provide a way to distinguish two networks where one of the networks has only one reticulation leaf.

Theorem 3.5.12 (Publication II, Theorem 6.14)**.** *Let $n \geq 5$. Let N_1 and N_2 be two distinct n-leaf funnel-free strict simple level-2 semi-directed networks on X. Suppose that $r(N_1) = \{a, b\}$ and $r(N_2) = \{c\}$ such that $c \notin \{a, b\}$. Let u be a purely interior reticulation vertex of N_2, a vertex whose all adjacent vertices are not leaves. If $d_{N_1}(x, y) \geq 4$ for any $x \in r(N_1)$ and $y \in r(N_2)$, then $V_{N_1} \not\subseteq V_{N_2}$. Additionally, if the pair (N_1, N_2) is not N_2-RC, $d_{N_2}(a, u) \geq 3$, and $d_{N_2}(b, u) \geq 3$, then N_1 and N_2 are distinguishable.*

Theorem 3.5.13 (Publication II, Proposition 6.15)**.** *Let $n \geq 5$. Let N_1 and N_2 be two distinct n-leaf funnel-free strict simple level-2 semi-directed networks on X. Suppose that $r(N_1) = \{a, b\}$ and $r(N_2) = \{a\}$. Let u be the purely interior reticulation vertex of N_2. If the pair (N_1, N_2) is not N_2-RC, $d_{N_2}(a, u) \geq 3$, and $d_{N_2}(b, u) \geq 3$, then $V_{N_2} \not\subseteq V_{N_1}$.*

Finally, we present a result on the distinguishability of network models where each network has only one reticulation leaf.

Theorem 3.5.14 (Publication II, Theorem 6.16)**.** *Let $n \geq 5$. Let N_1 and N_2 be two distinct n-leaf funnel-free strict simple level-2 semi-directed networks on X. Suppose that $r(N_1) = \{a\}$ and $r(N_2) = \{b\}$ such that $a \neq b$. Let u be the purely interior reticulation vertex in both networks. If the pair (N_1, N_2) is neither N_1-RC nor N_2-RC, $d_{N_1}(b,u) \geq 3$, and $d_{N_2}(a,u) \geq 3$, then N_1 and N_2 are distinguishable.*

Similarly to Theorem 3.5.11, if the assumption $r(N_1) \neq r(N_2)$ is not satisfied, then we may not be able to distinguish the networks. See Example A.4 in Appendix A of Publication II.

We conclude Publication II by studying a more general class of funnel-free strict level-2 networks. In Publication II, we refer to this class of networks as *semisimple networks*. Similar to Theorem 3.5.11, Theorem 3.5.12, Theorem 3.5.13, and Theorem 3.5.14, we prove some distinguishability results for funnel-free strict level-2 semisimple networks under certain conditions.

4. Factor Analysis Models

In this chapter, we summarize the results of Publication IV. In this publication, we focus on studying the factor analysis model that can be considered as a dimension reduction tool in statistical studies. We generalize the notion of the factor analysis model and study the model dimension and codimension. Studying the dimension and codimension of a statistical model is important because it can give us an idea of how complicated our model is. Moreover, this information is also essential if one wants to perform statistical model selection.

4.1 Moments and cumulants

We begin this section by providing some background on moment and cumulant tensors of a random vector. The moment and cumulant tensors play a significant role in statistics since many statistical calculations will require computing either moments or cumulants and they provide some useful insights on the probability distribution. If we know all moments of a probability distribution, or alternatively all cumulants, then we can possibly reconstruct a probability density function of the distribution. First studied by Chebyshev in 1961 [83], this problem of reconstructing a probability distribution from a given set of moments is often referred to as the moment problem. Moreover, both moments and cumulants can be applied to study independence between random variables. In the subsequent section, we will see that we can define a generalization of the factor analysis model in terms of moments and cumulants.

Throughout this chapter, we use the notation $(\mathbb{R}^m)^{\otimes r}$ to denote the vector space of r-dimensional tensors of format $m \times \cdots \times m$ with entries in the field $\mathbb{R}$. Moreover, $\Delta^r(\mathbb{R}^m)$ denotes the subspace of $(\mathbb{R}^m)^{\otimes r}$ containing all diagonal tensors. In what follows, we assume that all integrals or infinite sums exist and are finite. Let X be a continuous random vector in $\mathbb{R}^p$ with the probability density function $f_X(x)$ for $x \in \mathbb{R}^p$. Given a function g, the expectation of the function $g(X)$, denoted by $\mathbb{E}[g(X)]$, is defined as

$$\mathbb{E}[g(X)] := \int_{x \in \mathbb{R}^p} g(x) f_X(x) dx_1 \cdots dx_p.$$

If $\mathbb{E}[g(X)] = \pm\infty$, then we say that the expectation of $g(X)$ does not exist. If the random vector X is discrete, then to compute the expectation, we simply replace the integral with a sum over the possible discrete values.

Definition 4.1.1. Let X be a random vector in $\mathbb{R}^p$. Let $r \in \mathbb{N}$.

a. The rth-order moment tensor of X, denoted by $\mathcal{M}_X^{(r)} \in (\mathbb{R}^p)^{\otimes r}$, is the tensor whose $(j_1, \ldots, j_r)$-entry is given by

$$(\mathcal{M}_X^{(r)})_{j_1 \ldots j_r} := \mathbb{E}[X_{j_1} \cdots X_{j_r}].$$

b. The rth-order cumulant tensor of X, denoted by $\mathcal{C}_X^{(r)} \in (\mathbb{R}^p)^{\otimes r}$, is the tensor whose $(j_1, \ldots, j_r)$-entry is given by

$$(\mathcal{C}_X^{(r)})_{j_1 \ldots j_r} := \operatorname{cum}(X_{j_1}, \ldots, X_{j_r}) = \sum_{(A_1, \ldots, A_L)} (-1)^L (L-1)! \prod_{i=1}^{L} \mathbb{E}[\prod_{j \in A_i} X_j]$$

where the summation is taken over all partitions $(A_1, \ldots, A_L)$ of the set $\{j_1, \ldots, j_r\}$.

From the above definition, it can be easily seen that both moment and cumulant tensors are symmetric, meaning that its $(j_1, \ldots, j_r)$-entries will not change after any permutation of the indices $(j_1, \ldots, j_r)$. There is another way to compute moment and cumulant tensors of a random vector, namely via its generating function. Two different probability distributions can possibly have the same moments and hence the same cumulants. An example of two distributions with the same moments that are finite in all orders is given in [43].

It can be checked that the first order cumulant and moment vector of a random vector coincide and it is referred to as its mean vector. The second order cumulant matrix of a random vector is referred to as the covariance matrix. It can be shown that $\mathcal{M}_X^{(r)}$ and $\mathcal{C}_X^{(r)}$ coincide for $r \in [3]$ if the random vector X has zero mean. For $r \geq 4$, the rth-order moment and cumulant tensors are different in general. Finally, cumulants can measure how far a random variable is from being Gaussian because, for any Gaussian random variable, its rth-order cumulant vanishes for $r \geq 3$.

Let $r \in \mathbb{N}$ and $X \in \mathbb{R}^p$ be a random vector. Define the sets $\mathcal{M}_X^{(\leq r)} := \cup_{i=1}^r \mathcal{M}_X^{(i)}$ and $\mathcal{C}_X^{(\leq r)} := \cup_{i=1}^r \mathcal{C}_X^{(i)}$. The definition of cumulant tensors in Definition 4.1.1 gives rise to a polynomial map $f_r : \mathcal{M}_X^{(\leq r)} \to \mathcal{C}_X^{(\leq r)}$. Moreover, this map f_r has a polynomial inverse as well which is specified by the following relation

$$(\mathcal{M}_X^{(l)})_{j_1 \ldots j_l} = \sum_{(A_1, \ldots, A_L)} \prod_{i=1}^{L} \operatorname{cum}((X_j)_{j \in A_i})$$

where the summation is taken over all partitions $(A_1, \ldots, A_L)$ of the set $\{j_1, \ldots, j_l\}$.

4.2 The factor analysis models and its higher order generalizations

The factor analysis model was first introduced by the British statistician and psychologist, Charles Spearman in 1904. He is also well-known due to the Spearman correlation coefficient which can be used to measure the strength of a monotonic relationship between two random variables. The concept of the factor analysis model came up when Spearman studied human intelligence [75, 74]. He suspected that there are two underlying hidden variables that could explain different scores that people obtain on various tests related to intelligence. These two factors are the general intelligence referred to as the g factor, and the specific abilities intelligence referred to as the s factor.

The factor analysis model aims to estimate a model which can explain the variance and covariance between observable variables by fewer hidden variables together with some weightings. For example, in [5], the authors used factor analysis to evaluate the economic rank of some countries. In this paper, there are fifteen observed economic parameters that are used to evaluate economic performance with strong correlations between them, and the authors inferred that there are three underlying hidden factors that cause the strong correlations. Moreover, these three hidden factors provided a good analysis of the fifteen economic parameters.

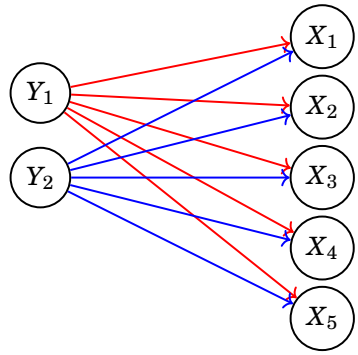

Figure 4.1. Example of a bipartite graph that encodes the interactions between the observed variables X_i's and the hidden variables Y_i's in the model $F_{5,2}$.

The factor analysis model on p observed variables and m hidden factors where $p > m$, denoted by $F_{p,m}$, is defined as follows. Let $X \in \mathbb{R}^p$ be a vector of observable random variables. The model is determined by the following equation

$$X = \Lambda Y + \varepsilon$$

where $Y \in \mathbb{R}^m$ is the vector of hidden variables, $\Lambda = (\lambda_{ij}) \in \mathbb{R}^{p \times m}$ is the coefficient matrix called *factor loadings/weightings* matrix, and $\varepsilon \in \mathbb{R}^p$ is the vector of error terms corresponding to the unique variance of each observed variable. An illustration of the model $F_{5,2}$ is presented in Figure 4.1. The red and the blue arrows in Figure 4.1 describe how the the hidden factors Y_1 and Y_2 affect the five observed variables, respectively. Moreover, the variables λ_{ij}'s encode

the weighting of how Y_j affects X_i. In summary, the factor analysis model formulates the vector of observable random variables as a linear combination of hidden factors plus some error terms.

The following are some assumptions in the factor analysis model.

1. The vectors Y and ε are assumed to be Gaussian.
2. For any pair (i,j), the variables Y_i and ε_j are independent.
3. The vectors Y and ε are centralized.
4. For any $i \neq j$, the variables Y_i and Y_j are independent. Similarly, the variables ε_i and ε_j are independent as well.

The following result provides a polynomial parameterization for the factor analysis model.

Theorem 4.2.1 ([28], Proposition 1)**.** *Every covariance matrix in the model $F_{p,m}$ can be given a parameterization as follows:*

$$F_{p,m} = \{\Sigma + \Lambda\Lambda^T \in \mathbb{R}^{p\times p} : \Lambda \in \mathbb{R}^{p\times m} \text{ and } \Sigma > 0 \text{ diagonal}\}.$$

As a consequence of this polynomial parameterization, the dimension $\dim(F_{p,m})$ of the factor analysis model $F_{p,m}$ can be computed. A dimension result is presented in [28].

Theorem 4.2.2 ([28], Theorem 2)**.** *The dimension of the model $F_{p,m}$ is*

$$\dim(F_{p,m}) = \min\left\{p(m+1) - \binom{m}{2}, \binom{p+1}{2}\right\}.$$

Since the Gaussianity of Y and ε is a rather restrictive assumption in the factor analysis model, we would like to drop this assumption. As a consequence, our probability distributions might have nonzero moments or cumulants up to certain higher order. In the classical factor analysis model, only the cumulant or moment tensors up to the second order are considered. In Publication IV, we propose the following more general model.

Definition 4.2.3 (Publication IV, Definition 2.8)**.** Let $k \in \mathbb{N}$ and $k \geq 2$. A *kth factor analysis model* is a family of random observed vectors $X \in \mathbb{R}^p$ that are correlated with a vector of random hidden variables $Y \in \mathbb{R}^m$ with $p > m$ such that

$$X = \Lambda Y + \varepsilon.$$

The model assumes the following:

1. The tensors $\mathcal{M}_Y^{(\leq k)}, \mathcal{M}_\varepsilon^{(\leq k)}, \mathcal{C}_Y^{(\leq k)}$, and $\mathcal{C}_\varepsilon^{(\leq k)}$ exist and have finite entries.
2. The vectors Y and ε are centralized.
3. For any pair (i,j), the variables Y_i and ε_j are independent.

4. For any $i \neq j$, the variables Y_i and Y_j are independent. Similarly, the variables ε_i and ε_j are independent as well.

In terms of cumulant and moment tensors, the above definition has the following counterparts.

Definition 4.2.4 (Publication IV, Definition 2.9)**.** Let $p, m, k \in \mathbb{N}$ such that $k \geq 2$ and $p > m$.

(*a*) The *kth-order cumulant factor analysis model*, denoted by $\mathscr{C}_{p,m}^{(\leq k)}$, is the subset of tuples $(\mathscr{C}^{(2)}, \ldots, \mathscr{C}^{(k)})$ of symmetric tensors that are the cumulant tensors for some random vector $X \in \mathbb{R}^p$ in the kth-order factor analysis model.

(*b*) The *kth-order moment factor analysis model*, denoted by $\mathscr{M}_{p,m}^{(\leq k)}$, is the subset of tuples $(\mathscr{M}^{(2)}, \ldots, \mathscr{M}^{(k)})$ of symmetric tensors that are the moment tensors for some random vector $X \in \mathbb{R}^p$ in the kth-order factor analysis model.

In what follows, we need the following definition of the Tucker product of a diagonal tensor with a tuple of matrices.

Definition 4.2.5. Let $\mathscr{D}^{(r)} \in \Delta^r(\mathbb{R}^m)$ be a diagonal tensor and $\Lambda = (\lambda_{ij}) \in \mathbb{R}^{p \times m}$ be a matrix. The *Tucker product* of $\mathscr{D}^{(r)}$ with the r-tuple of matrices $(\Lambda^T, \ldots, \Lambda^T)$, denoted by $\mathscr{D}^{(r)} \bullet^r \Lambda^T$, is the tensor of format $p \times \cdots \times p$ whose $(i_1 \cdots i_r)$th entry is given by

$$(\mathscr{D}^{(r)} \bullet^r \Lambda^T)_{i_1 \cdots i_r} := \mathscr{D}^{(r)} \bullet (\Lambda^T, \ldots, \Lambda^T)_{i_1 \cdots i_r} := \sum_{\ell=1}^{m} d_\ell \, \lambda_{i_1,\ell} \cdots \lambda_{i_r,\ell},$$

where for $i \in [m]$, d_i's are the diagonal entries of $\mathscr{D}^{(r)}$.

Similarly to Theorem 4.2.1, we obtain a polynomial parameterization of the model $\mathscr{C}_{p,m}^{(\leq k)}$.

Proposition 4.2.6 (Publication IV, Proposition 2.10)**.** *Let* $(\mathscr{C}^{(2)}, \ldots, \mathscr{C}^{(k)})$ *be a tuple in* $\mathscr{C}_{p,m}^{(\leq k)}$*. Then for* $2 \leq r \leq k$*,*

$$\mathscr{C}^{(r)} = \mathscr{D}^{(r)} \bullet^r \Lambda^T + \mathscr{E}^{(r)} \tag{4.1}$$

for some matrix $\Lambda \in \mathbb{R}^{p \times m}$ *and diagonal tensors* $\mathscr{D}^{(r)} \in \Delta^r(\mathbb{R}^m)$ *and* $\mathscr{E}^{(r)} \in \Delta^r(\mathbb{R}^p)$*. Moreover, if* $r = 2$*, then both matrices* $\mathscr{D}^{(2)}$ *and* $\mathscr{E}^{(2)}$ *are positive semidefinite.*

Since the moment tensors do not behave as nicely as the cumulant tensors under linear transformations of random variables, we do not expect a nice polynomial parameterization for the kth moment factor analysis model. However, the existence of an invertible polynomial map from $\mathscr{C}_X^{(r)}$ to $\mathscr{M}_X^{(r)}$ guarantees that there is a polynomial parameterization for the model $\mathscr{M}_{p,m}^{(\leq k)}$. In the polynomial parameterization in Proposition 4.2.6, we can assume without loss of generality that $\mathscr{D}^{(2)}$ is the $m \times m$ identity matrix.

The polynomial parameterization in Proposition 4.2.6 of the cumulant factor analysis model can be used to compute the dimension of the model. The following theorem is the main result of Publication IV.

Theorem 4.2.7 (Publication IV, Theorem 3.1)**.** *Let $p, m, k \in \mathbb{N}$, $k \geq 3$, and $p > m$. Then*

(a) $\dim(\mathscr{C}_{p,m}^{(\leq k)}) = \dim(\mathscr{M}_{p,m}^{(\leq k)})$, *and*

(b) $\dim(\mathscr{C}_{p,m}^{(\leq k)}) = (k-1)p + (k-2)m + \min\left\{pm - \binom{m}{2}, \binom{p+k-1}{k} - p\right\}$.

This dimension result enables us to compute the codimension of the higher-order factor analysis model which is the difference between the dimension of the codomain of the parameterization map and the dimension of the model. We conclude Publication IV by presenting some conditions on p and m that give rise to a higher-order factor analysis model with positive codimension.

Theorem 4.2.8 (Publication IV, Theorem 3.3)**.** *Let c_k be the codimension*

$$\operatorname{codim}(\mathscr{M}_{p,m}^{(\leq k)}) = \operatorname{codim}(\mathscr{C}_{p,m}^{(\leq k)})$$

of the kth-order factor analysis model. If $k \geq 3$ and $p \geq m+1$, then $c_k = h_m^{(k)}(p)/k!$, where

$$h_m^{(k)}(p) = \prod_{i=1}^{k}(p+i) - k!(k+m)p + \frac{k!}{2}[m^2 + (3-2k)m - 2]. \tag{4.2}$$

(*a*) *If $m \in [2k-3]$, then $h_m^{(k)}(p)$ has a unique positive root $p^{(k)}$. Therefore $c_k > 0$ if $p \geq \lfloor p^{(k)} \rfloor + 1$.*

(*b*) *For finitely many values of $m \geq 2k-2$, the polynomial $h_m^{(k)}(p)$ has two positive roots, the largest denoted by $p^{(k)}$. Therefore $c_k > 0$ if $p \geq \lfloor p^{(k)} \rfloor + 1$.*

(*c*) *There exists an integer $m^* \geq 2k-2$ such that $h_m^{(k)}(p)$ has no positive roots for $m \geq m^*$, in particular $c_k > 0$ for all $m \geq m^*$ and $p \geq m+1$.*

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