

# The Moran model with selection (and mutation): Fixation probabilities, ancestral lines, and an alternative particle representation

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# 1 Introduction

Evolution is a complex phenomenon that is influenced by several processes such as random reproduction (drift), mutation, selection, recombination and migration. A major topic of population genetics research is to understand how these processes lead to changes in the genetic structure of a population, see e.g. [15] for an overview. In this thesis, we are particularly interested in the interplay of drift, mutation and (fertility) selection.

The classical models of population genetics are the *Wright-Fisher model* and the *Moran model*. Both are devoted to populations of fixed size  $N$  that evolve forward in time (i.e. from the past to the present). The Wright-Fisher model considers non-overlapping generations, whereas, in the Moran model, generations do overlap. As a customary concept in population genetics, the diffusion limit, which assumes infinite population size, weak selection and mutation, is often taken as a starting point to analyse population development, see e.g. [11, 16] for an introduction.

In line with the historical perspective of evolutionary research, modern approaches aim at tracing back the ancestry of a sample of individuals taken from a present population. The result is the so-called *genealogy*. Classical tasks are to ask, when the *most recent common ancestor* of a sample lived, and which type he had.

The central tool to describe the genealogy of a finite sample in the absence of selection is the *Kingman coalescent* [27, 28], which was formulated by Kingman in the year 1982. Here, individuals are represented by lines, which merge simultaneously whenever two individuals originate from the same ancestor. In 1997, Krone and Neuhauser [32, 37] incorporated selection and introduced the *ancestral selection graph (ASG)* as an extension of the Kingman coalescent. By including so-called *virtual branches* in addition to the *real branches* that define the true genealogy, the ASG copes with selection. See [1] for an overview of the area.

Generically, even the ancestry of an entire population that evolves at constant size over a long time span without recombination will eventually coalesce backwards in time into a single ancestral line. The individuals that constitute this ancestral line are unique and – say at time  $t$  – termed the *common ancestor at time  $t$* . Their type composition may differ substantially from the distribution at present time. This mirrors the fact that the ancestral line consists of those individuals that are successful in the long run; thus, its type distribution is expected to be biased towards the favourable types [17, Sec. 1.2].

This thesis is devoted to the ancestral line in the Moran model in continuous time with two types, mutation, and selection. The two types differ in their reproduction rates, i.e. one type can be regarded as the ‘fit’ type, the other one as the ‘unfit’ type.

We are particularly interested in the stationary distribution of the type process along the ancestral line, to be called the *ancestral type distribution*. Thereby, we build on previous works by Fearnhead [17] and Taylor [43]. They consider different augmentations; each of them is termed *common ancestor process*.

Fearnhead’s approach is based on a simplified version of the ASG, which we call *pruned ASG*. In addition to a single real branch, it only consists of virtual branches

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of the less ‘fit’ type. Fearnhead calculates the ancestral type distribution in terms of the coefficients of a series expansion that is related to the number of (‘unfit’) virtual branches. The coefficients are denoted  $a_n$ ,  $n \geq 1$ , and termed *Fearnhead’s coefficients*. Fearnhead gives a suggestive interpretation of the  $a_n$  via an algorithm to simulate from the stationary distribution of the common ancestor process.

Taylor starts from an approach of Barton, Etheridge and Sturm [3], namely, from the *structured coalescent* that relies on a description of the full population, and uses a backward-forward construction. He characterises the ancestral type distribution in terms of the fixation probability of the offspring of all ‘fit’ individuals (regardless of the offspring’s types). This fixation probability is calculated via a boundary value problem. The fixation probability may be reformulated as a series expansion in the frequency of ‘unfit’ individuals, where the coefficients contain the  $a_n$ .

Both approaches rely strongly on analytical tools; in particular, they employ the diffusion limit from the very beginning. The results only have partial interpretations in terms of the graphical representation of the model (i.e., the representation that makes all individual lineages and their interactions explicit). The aim of this thesis is to complement the approaches of Fearnhead and Taylor by starting from the graphical representation for a population of finite size  $N$  and staying with the resulting discrete setting all the way through, performing the diffusion limit only at the very end. This way, we extend previous results and gain new insight into the underlying particle picture.

The pivotal quantity is the fixation probability of the offspring of the ‘fit’ individuals. As a main achievement, we obtain the fixation probability in the finite-size model in closed form. Additionally, the fixation probability may be represented as an equivalent to the series expansion in the diffusion limit, namely, with the help of a discrete analogue to Fearnhead’s coefficients (which are denoted by  $a_n^N$ ,  $1 \leq n \leq N - 1$ ).

According to Taylor, the representation of the fixation probability is suggestive: It is a decomposition into a neutral part and additional terms due to offspring created by selective events. In contribution to make this interpretation more precise, we use couplings between Moran models with and without selection, and calculate fixation probabilities within these settings.

We furthermore aim at drawing a more fine-grained picture, which is – even for the case of a vanishing mutation rate – still missing. Let us further comment on this: In the Moran model with selection, fixation probabilities follow a classical result of Kimura [26], which is based on type frequencies in the diffusion limit. Short and elegant standard arguments today start from the discrete setting, use a first-step or martingale approach, and arrive at the fixation probability in the form of a (normalised) series expansion in the reproduction rate of the favourable type [11, Thm. 6.1]. It is easily seen to converge to Kimura’s result in the diffusion limit. Recently, Mano [35] obtained the fixation probabilities with the help of the ASG, based on methods of duality (again in the diffusion limit); his argument is nicely recapitulated in [39].

What is still missing is a derivation within the framework of a full particle representation. To this end, we introduce an alternative particle system, which we call the *labelled Moran model*, and which is particularly well-suited for finite populations under selection.

In the new model, individuals are labelled  $1, 2, \dots, N$ . Neutral resampling events may take place between arbitrary labels, whereas selective events only occur in the direction of increasing labels. With the help of elementary methods, we not only recover fixation probabilities, but also obtain detailed insight into the nature of the selective events that play a role in the fixation process forward in time. The number of these selective events establishes a link to the common ancestor process in the special case without mutation: Its distribution corresponds to the discrete version of Fearnhead's coefficients.

A first step towards an understanding of Fearnhead's coefficients in the Moran model with selection and mutation is provided by a new urn model that is reminiscent of *Hoppe's urn* [21]: We have balls of two types and weights that correspond to neutral and selective reproduction rates. By incorporating additional markings to some of the balls, we may characterise the discrete coefficient  $a_1^N$  for  $N = 3$  and  $N = 4$  as an explicit probability in this new model.

The thesis is organised as follows. In Ch. 2, we start with a brief survey of our setting in the thesis, i.e. we recapitulate the Moran model, genealogical processes and Fearnhead's and Taylor's concepts concerning the common ancestor process. In Ch. 3, we build on the framework of Fearnhead and Taylor and start from a finite population. We calculate fixation probabilities and re-derive Taylor's boundary value problem and the ancestral type distribution. The derivation will be elementary and, at the same time, it will provide a direct interpretation of the differential equation. To formalise the additional terms in the fixation probability that go back to selective events, we go on with a coupling approach in Ch. 4. In Ch. 5, we restrict ourselves to a Moran model with selection. We introduce the labelled Moran model and show that it has the same empirical type distribution as the original Moran model, provided the initial values are chosen appropriately. Within this setting, we characterise *defining events* and point out the relevance of selective defining events on the fixation process. A simulation algorithm finally enlightens the representation given by Fearnhead and Taylor. Ch. 6 presents – in the spirit of an outlook – modifications of Hoppe's urn model and the corresponding genealogical interpretations in line with the *killed coalescent*. At least for  $N = 3$  and  $N = 4$ , this urn model provides new tentative insights concerning the structure of  $a_1^N$ . Ch. 7 summarises and discusses the results.





## 2 Preliminaries

In this chapter, we introduce the basic framework of the thesis. In Sec. 2.1, we start with a short outline of the Moran model [36], which describes population development forward in time. Sec. 2.2 devotes to the backward point of view. The main concepts, as the Kingman coalescent and the ASG, are recapitulated. In Sec. 2.3, we introduce the common ancestor type process and briefly summarise Fearnhead's and Taylor's approaches.

We stick to the presentation in the nicely written review paper of Baake and Bialowons [1] throughout this chapter. In addition, we use the letters  $t$  and  $\tau$  to indicate forward and backward time, respectively.

### 2.1 The Moran model with selection and mutation

We consider a haploid population of fixed size  $N \in \mathbb{N}$  in which each individual is characterised by a type  $i \in S = \{0, 1\}$ . If an individual reproduces, its single offspring inherits the parent's type and replaces a randomly chosen individual, maybe its own parent. This way, the replaced individual dies and the population size remains constant.

Individuals of type 1 reproduce at rate 1, whereas individuals of type 0 reproduce at rate  $1 + s_N$ ,  $s_N \geq 0$ . Accordingly, type-0 individuals are termed 'fit', type-1 individuals are 'unfit'. In line with a central idea of the ASG (see Sec. 2.2.2), we decompose reproduction events into neutral and selective ones. Neutral ones occur at rate 1 and happen to all individuals, whereas selective events occur at rate  $s_N$  and are reserved for type-0 individuals.

Mutation occurs independently of reproduction. An individual of type  $i$  mutates to type  $j$  at rate  $u_N \nu_j$ ,  $u_N \geq 0$ ,  $0 \leq \nu_j \leq 1$ ,  $\nu_0 + \nu_1 = 1$ . This is to be understood in the sense that every individual, regardless of its type, mutates at rate  $u_N$  and the new type is  $j$  with probability  $\nu_j$ . Note that this includes the possibility of 'empty' or 'silent' mutations, i.e. mutations from type  $i$  to type  $i$  (cf. [1, Sec. 6]).

The Moran model has a well-known graphical representation as an interacting particle system (cf. Fig. 1). The vertical lines represent the  $N$  individuals, with time running from top to bottom in the figure. Reproduction events are represented by arrows with the reproducing individual at the base and the offspring at the tip. Mutation events are marked by bullets.

We are now interested in the process  $(Z_t^N)_{t \geq 0}$  (or simply  $Z^N$ ), where  $Z_t^N$  is the number of individuals of type 0 at time  $t$ . Note that this is the *type frequency representation*, which contains less information than the *particle (or graphical) representation* in Fig. 1 that makes interactions between individuals explicit. When the number of type-0 individuals is  $k$ , it increases by one at rate  $\lambda_k^N$  and decreases by one at rate  $\mu_k^N$ , where

$$\lambda_k^N = \frac{k(N-k)}{N}(1 + s_N) + (N-k)u_N\nu_0 \quad \text{and} \quad \mu_k^N = \frac{k(N-k)}{N} + ku_N\nu_1. \quad (1)$$

Thus  $Z^N$  is a birth-death process with birth rates  $\lambda_k^N$  and death rates  $\mu_k^N$ . For  $u_N > 0$

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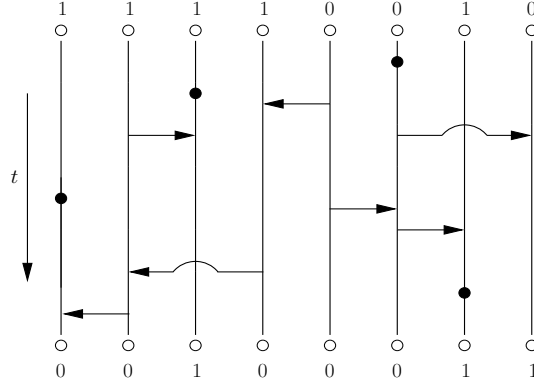


Figure 1: A realisation of the Moran model for  $N = 8$ . The types ( $0 = \text{fit}$ ,  $1 = \text{unfit}$ ) are indicated for the initial population (top) and the final one (bottom).

and  $0 < \nu_0, \nu_1 < 1$  its stationary distribution is  $(\pi_Z^N(k))_{0 \leq k \leq N}$  with

$$\pi_Z^N(k) = C_N \prod_{i=1}^k \frac{\lambda_{i-1}^N}{\mu_i^N}, \quad 0 \leq k \leq N, \quad (2)$$

where  $C_N$  is a normalising constant (cf. [10, p. 19], [6, p. 345]). As usual, an empty product is understood as 1. Up to a constant  $\tilde{C}_N$ , an alternative expression for the stationary distribution is

$$\pi_Z^N(k) = \tilde{C}_N \frac{(1 + s_N)^k}{k!(N-k)!} \binom{Nu_N \nu_0}{(k)} (Nu_N \nu_1)_{(N-k)}, \quad 0 \leq k \leq N, \quad (3)$$

where  $y_{(i)} := y(y+1) \cdots (y+i-1)$  for  $y \in \mathbb{R}$ ,  $i \in \mathbb{N}$ , cf. [11, Thm. 8.4], [45, Sec. 3.1].

To arrive at a diffusion, we consider the usual rescaling

$$(X_t^N)_{t \geq 0} := \frac{1}{N} (Z_{Nt}^N)_{t \geq 0},$$

and assume that  $\lim_{N \rightarrow \infty} Nu_N = \theta$ ,  $0 \leq \theta < \infty$ , and  $\lim_{N \rightarrow \infty} Ns_N = \sigma$ ,  $0 \leq \sigma < \infty$ . As  $N \rightarrow \infty$ , we obtain the well-known diffusion limit

$$(X_t)_{t \geq 0} := \lim_{N \rightarrow \infty} (X_t^N)_{t \geq 0}.$$

Given  $x \in [0, 1]$ , a sequence  $(k_N)_{N \in \mathbb{N}}$  with  $k_N \in \{0, \dots, N\}$  and  $\lim_{N \rightarrow \infty} k_N/N = x$ ,  $(X_t)_{t \geq 0}$  is characterised by the drift coefficient

$$a(x) = \lim_{N \rightarrow \infty} (\lambda_{k_N}^N - \mu_{k_N}^N) = (1-x)\theta\nu_0 - x\theta\nu_1 + (1-x)x\sigma \quad (4)$$

and the diffusion coefficient

$$b(x) = \lim_{N \rightarrow \infty} \frac{1}{N} (\lambda_{k_N}^N + \mu_{k_N}^N) = 2x(1-x). \quad (5)$$

Hence, the infinitesimal generator  $A$  of the diffusion is defined by

$$Af(x) = (1-x)x \frac{\partial^2}{\partial x^2} f(x) + [(1-x)\theta\nu_0 - x\theta\nu_1 + (1-x)x\sigma] \frac{\partial}{\partial x} f(x), \quad f \in \mathcal{C}^2([0, 1]). \quad (6)$$

The stationary density  $\pi_X$  – known as Wright’s formula – is given by

$$\pi_X(x) = C(1-x)^{\theta\nu_1-1} x^{\theta\nu_0-1} \exp(\sigma x), \quad (7)$$

where  $C$  is a normalising constant. See [11, Ch. 7, 8] or [16, Ch. 4, 5] for reviews of diffusion processes in population genetics and [25, Ch. 15] for a general survey of diffusion theory.

In contrast to our approach starting from the Moran model, in [3, 17, 43] the diffusion limit of the *Wright-Fisher model* is chosen as the basis for the studies. This is, however, of minor importance, since both diffusion limits differ only by a rescaling of time by a factor of 2, which manifests itself in a diffusion coefficient in the Moran model, which is twice the one in the Wright-Fisher model (cf. [11, Ch. 7], [16, Ch. 5] or [25, Ch. 15]). The reason for the factor 2 is the fact that reproduction events occur due to the sampling of ordered pairs of individuals in the Moran model, but occur due to unordered pairs in the Wright-Fisher model (cf. [1, Sec. 6], [10, p. 23] and [36]), see also Sec. 2.2.1.

Note that the particle representation of the Moran model does not carry over to the  $N \rightarrow \infty$  limit. A way to retain a particle representation even in the diffusion limit is provided by the *lookdown* construction of Donnelly and Kurtz [7, 8, 9], see [12, Ch. 5] for an overview. Let us briefly recall the central ideas for the case without selection.

In the so-called  $N$ -particle lookdown process [7] the individuals (of a population of size  $N$ ) are randomly assigned the levels  $1, \dots, N$  of the particle representation. We say that the initial ordering is *exchangeable* [12, Ch. 5.2]. Neutral arrows (which as usually lead to a replacement of the individual at the tip) occur at rate  $2/N$ , but we only allow for arrows that point from a lower to a higher level. This reproduction mechanism guarantees that an individual may only be replaced by an individual of a lower level. But note that the levels are not inheritable. Independently of reproduction, each individual mutates according to the mutation process of the Moran model. As a main result in [7], the  $N$ -particle lookdown process has the same empirical type distribution as the Moran model at any time  $t$ , provided both processes start with the same exchangeable initial type distribution. In particular, the  $N$ -particle lookdown process coincides with the first  $N$  levels of the  $(N+k)$ -particle lookdown process,  $k \geq 0$ . Hence, the construction carries over to the  $N \rightarrow \infty$  limit.

An extension to the  $N$ -particle lookdown process is the so-called modified lookdown [8]: On the occasion of neutral arrows from level  $i$  to  $j$ ,  $j > i$ , the individual at level  $N$  is removed, the descendant of the individual at level  $i$  moves to level  $j$ , and the individual that occupies level  $k$ ,  $j \leq k \leq N-1$ , is shifted to level  $k+1$ . In the corresponding  $N \rightarrow \infty$  limit we only have reproduction events, since the removal takes place ‘at infinity’ [12, Ch. 5.3].

The modified lookdown can be used to incorporate fecundity and viability selection [9], see e.g. [2] for a recent approach in this area.

## 2.2 Genealogical processes

One of the central topics in population genetics is the backward point of view, i.e. to infer information about past development from a present population. A classical task is to ask for stochastic processes to obtain *genealogies* of a finite sample from the population. A genealogy is represented by a *genealogical tree*, i.e. by the branches that represent the joint history of the sample individuals, together with the types along these branches [1, Sec. 7].

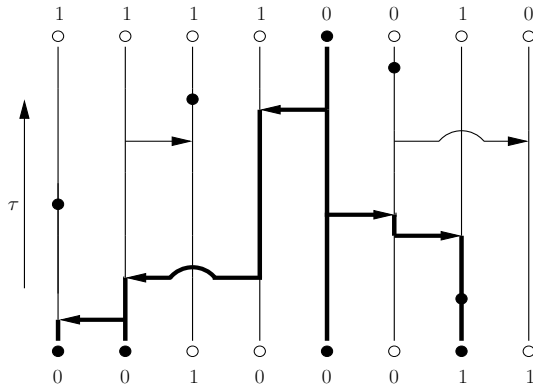


Figure 2: The genealogy of the four sample individuals that are marked black at the bottom is represented by fat lines. The MRCA (marked black at the top) is of type 0, one sample individual mutates to type 1.

For a given realisation of the Moran model we extract the corresponding genealogy of a given sample in the following way, see also Fig. 2 and [1, Sec. 7]: We start from the sample of individuals and follow their lines in the graphical representation of the Moran model backwards in time. Whenever we meet the tip of an arrow, we continue to trace back the lineage at its origin. If an arrow joins two sample lines, we say that these lines merge. The corresponding event is termed *coalescence* and the involved individuals go back to the same *ancestor*. At each coalescence event the number of sample lines is decreased by one, until a single line (the root of the genealogical tree) remains. The corresponding individual is called *most recent common ancestor (MRCA)* of the sample.

In this context, the question arises as to how construct genealogies without concrete realisations of the Moran model, i.e. random genealogies. Thereby, we assume that the population is stationary and evolves according to  $(X_t)_{t \geq 0}$ . In the absence of selection, i.e.  $\sigma = 0$ , the fundamental tool to construct so-called *neutral genealogies* is the *Kingman coalescent*; as the name suggests, it was introduced by Kingman [27, 28]. Its achievement lies in the independence of mutation and reproduction processes. The incorporation of selection compromises this independence. Krone and Neuhauser finally disposed of

this obstacle by constructing the equivalent to the Kingman coalescent, the *ancestral selection graph* (ASG) [32, 37].

In the following sections we give an introduction to neutral genealogies and the ancestral selection graph. We follow the presentation in [1, Sec. 7,8].

### 2.2.1 Neutral genealogies

In the absence of selection, reproduction rates are independent of the types. As a consequence, we may superimpose the mutation process on the reproduction model. Therefore, the construction of genealogies results from two steps.

First, we construct the genealogical tree without types, which is known as Kingman coalescent, in the sense of a death process. We start from the sample individuals (of unknown types) and work backwards in time by simultaneously merging two randomly chosen lines: If there are  $n$  lines, we merge two randomly chosen ones at rate  $n(n-1)$ . (Remember that a coalescence event corresponds to a reproduction event forward in time, in which one of the sample individuals reproduces and its offspring replaces another sample individual.) At some time, the number of lines almost surely decreases to 1 (cf. [38, p. 112] and [4, Prop. 5.1]) and the MRCA of the sample is reached. Note that concerning the death process it is irrelevant, whether the population evolves according to  $X^N$  (with  $s_N = 0$ ) or  $X$ .

In a second step, we insert the types. We choose the type of the MRCA according to (7) (or according to (2), if we consider  $X^N$  instead). We superimpose the mutation process forward in time along the branches of the reproduction model: On each line, mutations occur independently at rates  $\theta\nu_i$  (or  $Nu_N\nu_i$ ),  $i \in S$ . If a line splits into two (due to a coalescence event), the descendants inherit the type of their parent, i.e. of the line prior to the splitting point.

Note that the neutral genealogies of the Moran model and the Wright-Fisher model coincide up to a factor of 2: In the Wright-Fisher model, the probability that two individuals go back to the same ancestor of the previous generation is  $1/N$ . The corresponding Kingman coalescent evolves only at death rate  $\binom{n}{2}$ , if there are  $n$  lines in the graph.

### 2.2.2 Ancestral selection graph

The basic idea in the construction of the genealogical process with selection ( $\sigma > 0$ ) is to isolate the reproduction process from the mutation process. In this context, the central tool is the decomposition of reproduction events into neutral and selective reproductions (cf. Sec. 2.1).

First, we construct a *branching-coalescing graph* by means of a birth-death process. We begin with the description for  $X^N$  (with  $s_N > 0$ ) and switch over to the diffusion limit  $X$  afterwards. The idea is to work with what we call the *extended Moran model without types*, i.e. we ignore the types and assume that all individuals are capable of neutral and selective reproductions. That is, selective arrows appear on every line, and when we later superimpose the types, they are only incorporated, if they originate from type 0.

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For a realisation of the extended Moran model without types, we start from a sample and trace the sample lines backwards in time. If we meet the tip of a neutral arrow, we proceed as in Sec. 2.2.1: A neutral arrow between two lines in the branching-coalescing graph gives rise to a coalescence event and decreases the number of lines by one. If we meet the tip of a selective arrow, it is, due to the unknown types, unclear, if the selective reproduction really occurs. (Note that we restrict ourselves to selective arrows between a line in the graph and a line outside the graph (at rate  $n(N - n)s_N$  if there are  $n$  lines), since the rate to observe a selective arrow that joins two lines in the graph (at rate  $n(n - 1)s_N$  if there are  $n$  lines) tends to 0 in the diffusion limit.) There are two possibilities: The *incoming branch*, i.e. the line at the origin of the selective arrow, is the parent, or the *continuing branch*, i.e. the line at its tip, is the parent (cf. Fig. 3). We consider both possibilities and trace back both incoming and continuing branch. The result is an increase in the number of lines by one, and the corresponding event is termed *branching*.

After a finite time, the number of branches in the graph reduces to 1 and we stop the described procedure. The corresponding individual is termed *ultimate ancestor (UA)*, the resulting graph is called *ASG without types*, see Fig. 3.

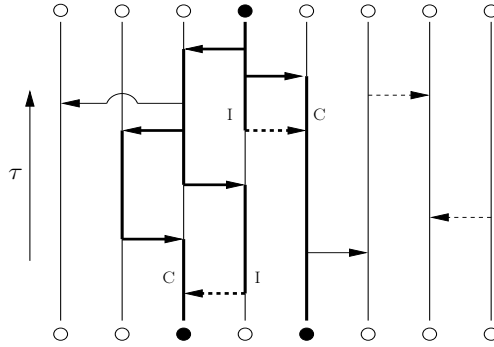


Figure 3: The ASG without types for a given collection of neutral (solid) and selective (dashed) arrows. The ASG is represented by fat lines, sample individuals and the UA are marked black at the bottom and the top, respectively. Neutral arrows imply coalescence events, selective arrows branching events. Incoming (I) and continuing (C) branch are indicated for each branching.

The stochastic process to obtain realisations of the ASG without types, which is based on the diffusion  $X$ , is given as follows: If the branching-coalescing graph consists of  $n$  lines, two randomly chosen ones merge at rate  $n(n - 1)$  (cf. Sec. 2.2.1) and a randomly chosen line splits into two at rate  $\lim_{N \rightarrow \infty} n(N - n)s_N = n\sigma$ . The underlying birth-death process almost surely reaches 1 in finite time [32, Thm. 3.2] and the construction is finished then. As a brief explanation note that coalescence events occur at a quadratic rate, while branching events only occur at a linear rate. A realisation is given in Fig. 4, left.

In the second step, we insert the types to obtain the corresponding *ASG with types*

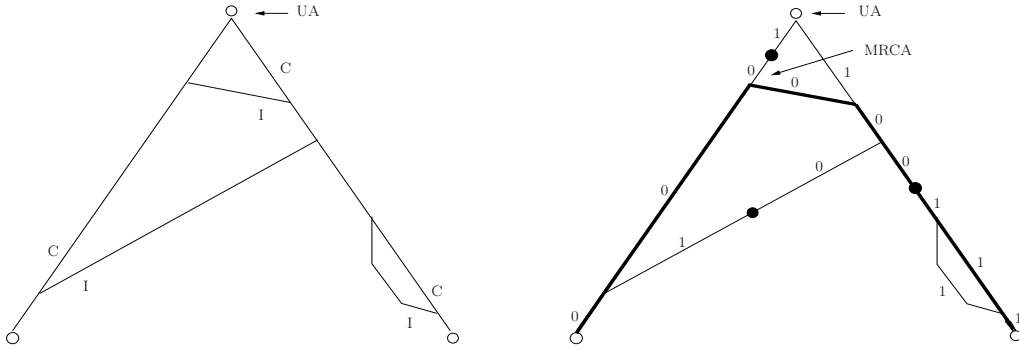


Figure 4: Left: The ASG without types of a sample of size 2. Right: The ASG with types (bold and thin lines) is obtained by running the mutation process down the graph. Running backwards in time yields the embedded genealogy (bold) and decomposes the lines of the ASG into real (bold) and virtual (thin) branches.

(cf. Fig. 4). We choose the type of the UA according to (7). (Note that the type of the UA is indeed distributed according to Wright’s formula, see [9, Thm. 8.2] for an explanation in the context of the *ancestral influence graph* of Donnelly and Kurtz [9, Sec. 8], or our reasoning in Sec. 2.2.3.) We superimpose the mutation process forward in time, i.e. mutations occur at rate  $\theta\nu_i$ ,  $i \in S$ , independently on disjoint lines. At a coalescence event the descendants inherit the parent’s type (cf. Sec. 2.2.1). At a branching event we determine the descendant’s type as follows, see also Fig. 5: The selective arrow is only used, if the incoming branch is of type 0. Then, the incoming branch is the parent (the so-called *parental line*) and the descendant inherits the type 0. Otherwise, if the incoming branch is of the less fit type, the selective arrow is not used and the continuing branch is the parental one. Then, the descendant inherits the type of the continuing branch. In [42, Alg. 3.1] an algorithm to simulate the ASG with types is given.

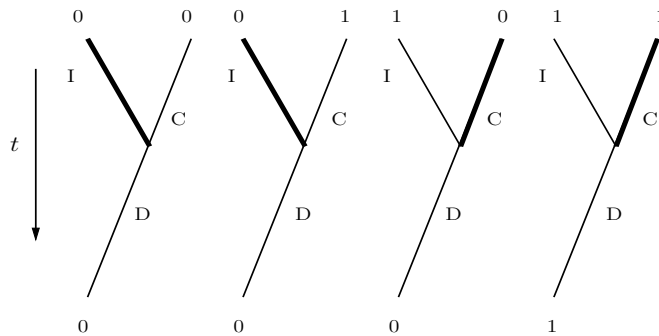


Figure 5: For every type composition of incoming (I) and continuing (C) branches, the parental line is marked bold. The descendant (D) inherits the type of the parental line.

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In the final step, we extract the embedded genealogy (cf. Fig. 4). We start from the sample of individuals and trace back their lines in the ASG with types. Every time we meet a selective arrow, we continue to trace back only the parental line, which we have determined in the second step. The number of lines almost surely decreases to 1 (by means of coalescence events), the corresponding individual is the MRCA of the sample. It may be younger than the UA of the sample (as in Fig. 4).

The lines of the genealogy are termed *real*, the lines outside *virtual*. Real branches are the ancestral lines of the sample, whereas every branching event gives rise to a new virtual branch.

### 2.2.3 Conditional ancestral selection graph

Krone and Neuhauser [32, 37] extract the genealogy of a sample of fixed size in a procedure that is subdivided into three steps. There are two crucial points to note: First, within the construction of the ASG without types the graph is traced backwards in time until the UA is reached, although we are only interested in the MRCA. Second, the type-decomposition of the sample follows from the choice of the UA's type and the mutation process. Thus, the construction does not state the stochastic process to obtain genealogies of samples of known types.

As an enhancement of the construction in [32, 37], the *conditional ASG* [40] is a tool to generate the ASG with types with embedded genealogy of a sample of known types backwards in time. In particular, the conditional ASG does not stop as soon as the number of branches reduces to 1: Due to branching events, this number will almost surely increase again.

One considers the process  $(\mathbf{R}_\tau, \mathbf{V}_\tau)_{\tau \geq 0} := ((R_{0,\tau}, R_{1,\tau}), (V_{0,\tau}, V_{1,\tau}))_{\tau \geq 0}$ , where  $R_{i,\tau}$ ,  $i \in S$ , is the number of real type- $i$  branches and  $V_{i,\tau}$ ,  $i \in S$ , is the number of virtual type- $i$  branches, both at time  $\tau$  before the present. Since the sample at time  $\tau = 0$  consists only of real lines,  $\mathbf{V}_0 = (0, 0)$ , whereas  $\mathbf{R}_0$  is drawn from the stationary density (7).

Let  $(\mathbf{r}, \mathbf{v}) = ((r_0, r_1), (v_0, v_1))$  be a realisation of  $(\mathbf{R}_\tau, \mathbf{V}_\tau)_{\tau \geq 0}$  at some time  $\tau$ . The probability to observe the marginal type configuration of  $(\mathbf{r}, \mathbf{v})$ , when there are  $r + v := r_0 + r_1 + v_0 + v_1$  lineages in the ASG at time  $\tau$ , is termed  $q(\mathbf{r}, \mathbf{v})$ . The probability  $q(\mathbf{r}, \mathbf{v})$  is time-independent and follows [40, Thm. 2.3]

$$q(\mathbf{r}, \mathbf{v}) = \frac{(r + v)!}{r_0! r_1! v_0! v_1!} p(\mathbf{r} + \mathbf{v}), \quad (8)$$

where

$$p(\mathbf{n}) := \mathbb{E}_{\pi_X} (X^{n_0} (1 - X)^{n_1}) = \int_0^1 x^{n_0} (1 - x)^{n_1} \pi_X(x) dx \quad (9)$$

for an *unordered* sample  $\mathbf{n} = (n_0, n_1)$  of size  $n = n_0 + n_1$  with  $n_i$  individuals of type  $i$ ,  $i \in S$ .  $p(\mathbf{n})$  is the probability of drawing an *ordered* sample that has the same marginal type distribution as  $\mathbf{n}$  from a population in stationarity. (Note that we understand ordered samples of size  $n$  as tuples  $\mathbf{c} = (c_1, \dots, c_n)$ , where  $c_j$  denotes the type of the  $j$ th



sample individual,  $1 \leq j \leq n$ . The corresponding unordered sample is  $(\#_0(\mathbf{c}), \#_1(\mathbf{c}))$ , where  $\#_i(\mathbf{c})$ ,  $i \in S$ , is the number of type- $i$  individuals in the ordered sample  $\mathbf{c}$ .) The probability to draw ordered samples from a population in stationarity is invariant under permutations of the individuals, and thus  $p$  only depends on the marginal type distribution of the samples. This property, which is known as *exchangeability*, is caused by the absence of spatial information in the Moran model, and justifies our slightly unusual notation in (9) (cf. [1, Sec. 9]).

From (8) and (9) we can conclude that it is reasonable to choose the type of the UA according to Wright's density in the second step of the construction in Sec. 2.2.2. In contrast, the state at the time of the MRCA may contain virtual branches. Thus, in general, the type of the MRCA has not the same distribution as a sample of size 1 in equilibrium [40, Sec. 2].

The transition rates of the conditional ASG can be read off from the following recursion for the stationary sampling distribution  $q$  [40, Thm. 2.5] (see also [42, Prop. 3.1, Alg. 3.2] to simulate the conditional ASG):

$$\begin{aligned}
 (r + v - 1 + \theta + \sigma)q(\mathbf{r}, \mathbf{v}) &= \sum_{\substack{i \in S: \\ r_i, v_i > 0}} [(r_i - 1)q(\mathbf{r} - \mathbf{e}_i, \mathbf{v}) \\
 &\quad + (v_i - 1 + 2r_i)q(\mathbf{r}, \mathbf{v} - \mathbf{e}_i)] \\
 + \theta \sum_{\substack{i, j \in S: \\ i \neq j, r_j, v_j > 0}} &\left[ \frac{\nu_j(r_i + 1)}{r + v} q(\mathbf{r} + \mathbf{e}_i - \mathbf{e}_j, \mathbf{v}) \right. \\
 &\quad \left. + \frac{\nu_j(v_i + 1)}{r + v} q(\mathbf{r}, \mathbf{v} + \mathbf{e}_i - \mathbf{e}_j) \right] \\
 + \sigma \left[ \sum_{i \in S} \frac{(r_i + v_i)(v_i + 1)}{(r + v)(r + v + 1)} q(\mathbf{r}, \mathbf{v} + \mathbf{e}_i) \right. \\
 &\quad \left. + \frac{2(r_0 + v_0)(v_1 + 1)}{(r + v)(r + v + 1)} q(\mathbf{r}, \mathbf{v} + \mathbf{e}_1) \right], \tag{10}
 \end{aligned}$$

where  $\mathbf{e}_0 := (1, 0)$  and  $\mathbf{e}_1 := (0, 1)$ . Note that (10) enhances the recursions given in [32, Thm. 5.2] and [17, Sec. 1.3] and is obtained by a first-step decomposition backwards in time. Here, the second-last row goes back to a branching event, in which both incoming and continuing branch are of the same type, whereas the last row represents a branching with different types of incoming and continuing branches.

As it can be seen from the rate of the transition  $(\mathbf{r}, \mathbf{v}) \rightarrow (\mathbf{r} + \mathbf{e}_i - \mathbf{e}_j, \mathbf{v})$  (third row), which is a mutation of a real branch, the virtual branches are indispensable in the construction of the embedded genealogy. Thus, the process of real branches backwards in time alone would not be Markovian (cf. [1, Sec. 10]).

## 2.3 The common ancestor type process

As in Sec. 2.2, we assume that the population is stationary and evolves according to the diffusion process  $(X_t)_{t \geq 0}$ . Then, at any time  $t$ , there almost surely exists a unique

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individual that is, at some time  $s > t$ , ancestral to the whole population (cf. Fig. 6). (One way to see this is via [32, Thm. 3.2, Corollary 3.4], which shows that the expected time to the UA in the ASG remains bounded even if the sample size tends to infinity.) We say that the descendants of this individual become fixed and call it the *common ancestor at time  $t$* . The lineage of these distinguished individuals over time defines the so-called *ancestral line*. Denoting the type of the common ancestor at time  $t$  by  $I_t$ ,  $I_t \in S$ , we term  $(I_t)_{t \geq 0}$  the *common ancestor type process* or *CAT process* for short. Of particular importance is its stationary type distribution  $\alpha = (\alpha_i)_{i \in S}$ , to which we will refer as the *ancestral type distribution*. Unfortunately, the CAT process is not Markovian. But two approaches are available that augment  $(I_t)_{t \geq 0}$  by a second component to obtain a Markov process. They go back to Fearnhead [17] and Taylor [43]; we will recapitulate them below.

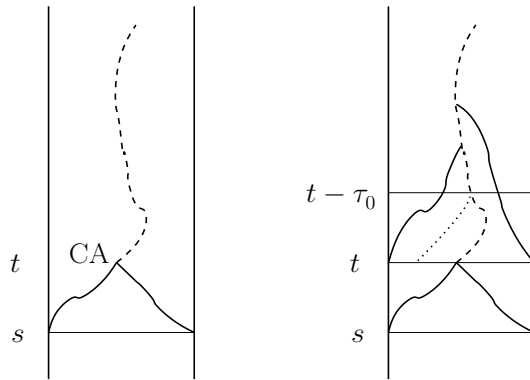


Figure 6: Left: The common ancestor at time  $t$  (CA) is the individual whose progeny will eventually fix in the population (at time  $s > t$ ). Right: If we pick an arbitrary individual at time  $t$ , there exists a minimal time  $\tau_0$  so that the individual's line of ancestors (dotted) corresponds to the ancestral line (dashed) up to time  $t - \tau_0$ . (Figure by Anton Wakolbinger.)

### 2.3.1 Fearnhead's approach

The conditional ASG  $(\mathbf{R}_\tau, \mathbf{V}_\tau)_{\tau \geq 0}$  of Sec. 2.2.3 yields a tool to trace the ancestry of a single individual backwards in time. Since there is only one real branch,  $\mathbf{R}_\tau = \mathbf{e}_{J_\tau}$ , where  $J_\tau$  is the type of the individual's ancestor at time  $\tau$  before the present (that is, at forward time  $t - \tau$ ). We write  $(J_\tau, \mathbf{V}_\tau)_{\tau \geq 0} := (\mathbf{e}_{J_\tau}, \mathbf{V}_\tau)_{\tau \geq 0}$  for short. Note that there is a minimal time  $\tau_0$  so that, for all  $\tau \geq \tau_0$ ,  $J_\tau = I_{t-\tau}$  (see also Fig. 6), provided the underlying process  $(X_t)_{t \geq 0}$  is extended to  $(-\infty, \infty)$ .

Following [1, Sec. 9], we define (with the probability  $p$  as in (9))

$$p(j \mid \mathbf{n}) := \frac{p(\mathbf{n} + \mathbf{e}_j)}{p(\mathbf{n})} \quad (11)$$

### 2.3 The common ancestor type process

as the conditional probability to draw an individual of type  $j$ ,  $j \in S$ , from a population in equilibrium, provided a configuration  $\mathbf{n}$  has already been taken from this population.

With this notation at hand, and with the help of (8) and (11), the transitions and rates of  $(J_\tau, \mathbf{V}_\tau)_{\tau \geq 0}$  can be obtained from recursion (10), cf. [17, Sec. 1.4] and [1, Sec. 10]. See Fig. 7 for an illustration of the transitions.

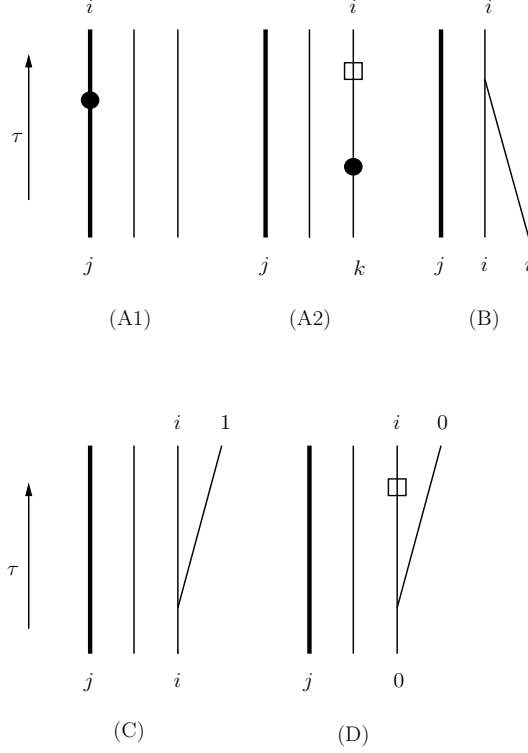


Figure 7: Transitions of  $(J_\tau, \mathbf{V}_\tau)_{\tau \geq 0}$  out of  $(j, \mathbf{v})$ . The single ancestral line is illustrated by the bold line, virtual lines by thin ones. Types are noted at the bottom and the top, respectively. Boxes indicate lines that may be removed.

(A1) Mutation of the real branch, i.e.  $(j, \mathbf{v}) \rightarrow (i, \mathbf{v})$ , occurs at rate

$$\theta \nu_j \frac{p(\mathbf{v} + \mathbf{e}_i)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

(A2) Mutation of a virtual branch, i.e.  $(j, \mathbf{v}) \rightarrow (j, \mathbf{v} - \mathbf{e}_k + \mathbf{e}_i)$ , occurs at rate

$$v_k \theta \nu_k \frac{p(\mathbf{v} + \mathbf{e}_j - \mathbf{e}_k)}{p(\mathbf{v} + \mathbf{e}_j)} p(i | \mathbf{v} + \mathbf{e}_j - \mathbf{e}_k).$$

(B) Coalescence of two branches of type  $i$ , i.e.  $(j, \mathbf{v}) \rightarrow (j, \mathbf{v} - \mathbf{e}_i)$ , occurs at rate

$$(v_i + \delta_{ji} - 1)(v_i + \delta_{ji}) \frac{p(\mathbf{v} + \mathbf{e}_j - \mathbf{e}_i)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

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- (C) Branching, where the incoming branch is of type 1 and the continuing branch of type  $i$  (thus, the continuing branch is parental), i.e.  $(j, \mathbf{v}) \rightarrow (j, \mathbf{v} + \mathbf{e}_1)$ , occurs at rate

$$\sigma(v_i + \delta_{ji}) \frac{p(\mathbf{v} + \mathbf{e}_j + \mathbf{e}_1)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

- (D) Branching, where the incoming branch is of type 0 and the continuing branch of type  $i$  (thus, the incoming branch is parental), i.e.  $(j, \mathbf{v}) \rightarrow (j, \mathbf{v} + \mathbf{e}_i)$ , occurs at rate

$$\sigma(v_0 + \delta_{j0}) p(i \mid \mathbf{v} + \mathbf{e}_j).$$

In [1, Sec. 10] an alternative approach towards the rates of  $(J_\tau, \mathbf{V}_\tau)_{\tau \geq 0}$  is given with the help of a time-reversal of continuous-time Markov chains with respect to the (stationary) measure  $p$ . (Given a Markov chain in continuous time on a discrete state space  $E$  that is characterised by a generator  $Q = (Q_{ij})_{i,j \in E}$  with stationary distribution  $\pi = (\pi_i)_{i \in E}$ , its time-reversal (with respect to  $\pi$ ) is defined as a continuous-time Markov chain with generator  $\hat{Q} = (\hat{Q}_{ij})_{i,j \in E}$ ,  $\hat{Q}_{ij} = \pi_j Q_{ji} / \pi_i$  and stationary distribution  $\pi$  [38, Thm. 3.7.1].)

Now, the construction of the conditional ASG of a single individual may be simplified according to the framework in [17]. To this end, let us note the following: First, the new type of the mutated virtual branch in transition (A2) complies with a random draw from the stationary population, provided the sample  $\mathbf{v} + \mathbf{e}_j - \mathbf{e}_k$  has already been taken from this population. Second, the type of the continuing branch in transition (D) is drawn from the stationary distribution, given the configuration  $\mathbf{v} + \mathbf{e}_j$  has previously been chosen; see the conditional probabilities in (A2) and (D).

Following [17, Thm. 1] and the arguments in [1, Sec. 11], sample individuals that are randomly chosen from stationarity, conditional on the remaining sample, may be removed from the conditional ASG of sample size 1 without compromising the Markov property. The process of the lines after removal is still given by the transitions (A1)-(D):

Transition (D) leads to a removal of the continuing branch and thus becomes an ‘empty’ event. We only observe branchings due to (C), that is, we only attach virtual branches of type 1 to the graph. (This constraint was already analysed by Slade [41, Thm. 2.4] in the conditional ASG of arbitrary sample size. A reduced version of recursion (10) is given, in which the branching rate is minimised to attachments of virtual branches of type 1 only.) As soon as a virtual branch mutates (due to (A2)), we remove it, and the remaining set of virtual branches contains only unfit ones. We refer to the resulting construction as the *pruned ASG*. It is described by the process  $(J_\tau, V_\tau)_{\tau \geq 0}$ , where  $V_\tau$  (with values in  $\mathbb{N}_0$ ) is the number of virtual branches (of type 1). Note that  $(J_\tau, V_\tau)_{\tau \geq 0}$  is termed *common ancestor process (CAP)* in [17].

Let  $\mathbf{v} = (0, v)$ ,  $v \in \mathbb{N}_0$ . The transitions and rates of  $(J_\tau, V_\tau)_{\tau \geq 0}$  can be read off from (A1)-(D) (cf. [17, Sec. 2], [1, Sec. 11]):

( $\tilde{A}$ ) Mutation of the real branch, i.e.  $(j, v) \rightarrow (i, v)$ , occurs at rate

$$\theta \nu_j \frac{p(\mathbf{v} + \mathbf{e}_i)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

( $\tilde{B}$ ) Coalescence or removal of a virtual branch due to mutation, i.e.  $(j, v) \rightarrow (j, v - 1)$ , occurs at rate

$$((v + \delta_{j1} - 1)(v + \delta_{j1}) + v\theta\nu_1) \frac{p(\mathbf{v} + \mathbf{e}_j - \mathbf{e}_1)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

( $\tilde{C}$ ) Branching to an incoming branch of type 1, i.e.  $(j, v) \rightarrow (j, v + 1)$ , occurs at rate

$$(v + 1)\sigma \frac{p(\mathbf{v} + \mathbf{e}_j + \mathbf{e}_1)}{p(\mathbf{v} + \mathbf{e}_j)}.$$

(Wakeley [46] mimics the approaches of Fearnhead [17] and Slade [41] to simplify the conditional ASG for samples of arbitrary size. Similarly, the real lines are augmented only by unfit virtual ones.)

Fearnhead provides a representation of the stationary distribution of the pruned ASG, which we denote by  $\pi_F$ . This stationary distribution is expressed in terms of constants  $\rho_1^{(k)}, \dots, \rho_{k+1}^{(k)}$  defined by the following *backward* recursion:

$$\rho_{k+1}^{(k)} = 0 \quad \text{and} \quad \rho_{i-1}^{(k)} = \frac{\sigma}{i + \sigma + \theta - (i + \theta\nu_1)\rho_i^{(k)}}, \quad k \in \mathbb{N}, 2 \leq i \leq k + 1. \quad (12)$$

The limit  $\rho_i := \lim_{k \rightarrow \infty} \rho_i^{(k)}$  exists and satisfies  $0 \leq \rho_i \leq 1$  for all  $i \geq 1$  [17, Lemma 1]. The stationary distribution of the pruned ASG is given by [17, Thm. 3]

$$\pi_F(j, v) = \begin{cases} (\prod_{i=1}^v \rho_i) \mathbb{E}_{\pi_X}(X(1-X)^v), & \text{if } j = 0, \\ (\prod_{i=1}^v \rho_i) (1 - \rho_{v+1}) \mathbb{E}_{\pi_X}((1-X)^{v+1}), & \text{if } j = 1 \end{cases} \quad (13)$$

for all  $v \in \mathbb{N}_0$ . Fearnhead proves this result by straightforward verification of the stationarity condition; the calculation is somewhat cumbersome and does not yield insight into the connection with the graphical representation of the pruned ASG.

An interpretation is given by way of a *rule for simulating from  $\pi_F$*  [17, Remark 3]: One simultaneously chooses individuals from a population in stationarity and denotes them as real respectively virtual branches. If we have already picked  $v - 1$ ,  $v \geq 1$ , virtual branches and the following draw is a type-0 individual, we term it the real branch. If the draw is a type-1 individual, it is called the real branch with probability  $1 - \rho_v$  and virtual branch with probability  $\rho_v$ , respectively. This way, one draws individuals until one obtains the real branch. The corresponding realisation  $(j, v)$  represents the type  $j$  of the real branch and the number  $v$  of ‘unsuccessful’ draws from stationarity (which

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coincides with the number of unfit virtual lines). In particular,  $\prod_{i=1}^v \rho_i$  is the probability to obtain at least  $v$  unfit virtual lines, given that the first  $v$  draws are type-1 individuals.

Marginalising over the number of virtual branches results in the stationary type distribution of the ancestral line, namely,

$$\alpha_j = \sum_{v \geq 0} \pi_F(j, v). \quad (14)$$

Reversing the direction of time in the pruned ASG yields an augmentation of the CAT process (for  $\tau \geq \tau_0$ ) by a collection of unfit virtual branches. Obviously, the time-reversal has the same stationary distribution (13) as the pruned ASG. Using [38, Thm. 3.7.1], we determine the transitions and the corresponding rates with the help of  $(\tilde{A})$ - $(\tilde{C})$  and (13). (See also [17, Corollary 2] for the mutation process on the real branch.)

( $\hat{A}$ ) Mutation of the real branch, i.e.  $(j, v) \rightarrow (i, v)$ , occurs at rate

$$\theta \nu_i \frac{1 - \rho_{v+1} \delta_{i1}}{1 - \rho_{v+1} \delta_{j1}}.$$

( $\hat{B}$ ) Removal of a virtual line, i.e.  $(j, v) \rightarrow (j, v - 1)$ , occurs at rate

$$\frac{v\sigma}{\rho_v} \frac{1 - \rho_v \delta_{j1}}{1 - \rho_{v+1} \delta_{j1}}.$$

( $\hat{C}$ ) Branching to a virtual line, i.e.  $(j, v) \rightarrow (j, v + 1)$ , occurs at rate

$$((v + \delta_{j1})(v + 1 + \delta_{j1}) + (v + 1)\theta \nu_1) \frac{\rho_{v+1}(1 - \rho_{v+2} \delta_{j1})}{1 - \rho_{v+1} \delta_{j1}}.$$

### 2.3.2 Taylor's approach

We start to recapitulate the *structured coalescent process*, which is based on the work of Kaplan et al. [24], and which is further investigated in [3] (for a brief survey see also [15, Ch. 5.5]). One considers the process  $(\mathbf{R}_\tau, \tilde{X}_\tau)_{\tau \geq 0}$ , where, as in Sec. 2.2.3,  $\mathbf{R}_\tau$  is the ancestral type composition of a sample with configuration  $\mathbf{R}_0$ , and  $\tilde{X}_\tau$  is the frequency of type-0 individuals, in both cases at time  $\tau$  before the present. Since the diffusion  $(X_t)_{t \geq 0}$  of the Moran model forward in time possesses a reversible stationary measure,  $(X_t)_{t \geq 0}$  and  $(\tilde{X}_\tau)_{\tau \geq 0}$  share the same dynamics according to the infinitesimal generator  $A$  as in (6), cf. [3, Sec. 2]. (Following [11, Ch. 7.5],  $A$  is self-adjoint with respect to  $\pi_X$ .) Then, the structured coalescent is characterised by the following generator  $G$  [3, Lemma 3.1], [15, Thm. 5.14]:

$$Gf(\mathbf{r}, x) = \frac{r_0(r_0 - 1)}{x} (f(\mathbf{r} - \mathbf{e}_0, x) - f(\mathbf{r}, x)) \quad (15)$$

$$+ \frac{r_1(r_1 - 1)}{1 - x} (f(\mathbf{r} - \mathbf{e}_1, x) - f(\mathbf{r}, x)) \quad (16)$$

$$+ r_0\theta\nu_0 \frac{1 - x}{x} (f(\mathbf{r} - \mathbf{e}_0 + \mathbf{e}_1, x) - f(\mathbf{r}, x)) \quad (17)$$

$$+ r_1\theta\nu_1 \frac{x}{1 - x} (f(\mathbf{r} + \mathbf{e}_0 - \mathbf{e}_1, x) - f(\mathbf{r}, x)) \quad (18)$$

$$+ Af(\mathbf{r}, x),$$

where  $f(\mathbf{r}, \cdot) \in \mathcal{C}^2([0, 1])$  for all  $\mathbf{r} \in \mathbb{N}_0^2 \setminus \{(0, 0)\}$  and  $Af(\mathbf{r}, x)$  is understood as acting on the second component  $x$  of  $f(\mathbf{r}, x)$  for a given  $\mathbf{r}$ . Terms (15) and (16) represent coalescence events within the  $r_i$  individuals of type  $i$ ,  $i \in S$ , terms (17) and (18) go back to mutations. In the diffusion of the Moran model, coalescence events occur twice as fast as in the Wright-Fisher diffusion (cf. Sec. 2.2.1), which is considered in [3, 15, 43]. Hence, the factors  $r_i(r_i - 1)$  are replaced by  $\binom{r_i}{2}$ ,  $i \in S$ , in [3, 15, 43]. See [3, Sec. 4] for existence and uniqueness of the structured coalescent process.

Obviously, the construction in [3] is also valid for samples of size 1. One obtains the so-called *structured retrospective process* [43, Sec. 2] that we denote by  $(J_\tau, \tilde{X}_\tau)_{\tau \geq 0}$ , where  $J_\tau$  is as usual the ancestor's type at time  $\tau$ . As in Fearnhead's approach, there is a minimal time  $\tau_0$  so that  $J_\tau = I_{t-\tau}$  for all  $\tau \geq \tau_0$ . Changes in the sample are only caused by mutations, thus, the generator  $G$  simplifies to [43, Sec. 2]

$$Gf(0, x) = \theta\nu_0 \frac{1 - x}{x} (f(1, x) - f(0, x)) + Af(0, x),$$

$$Gf(1, x) = \theta\nu_1 \frac{x}{1 - x} (f(0, x) - f(1, x)) + Af(1, x),$$

where  $f$  is twice continuously differentiable on  $(\{0\} \times (0, 1]) \cup (\{1\} \times [0, 1))$  and has compact support. Existence and uniqueness of the structured retrospective process are proven in [43, Prop. 2.2].

Time-reversal (for  $\tau \geq \tau_0$ ) with respect to the stationary distribution of the structured retrospective process yields the process  $(I_t, X_t)_{t \geq 0}$ , see [43, Prop. 2.7] for existence and uniqueness. Thus, the augmentation of the CAT process relies on a description of the *full* population forward in time (in the diffusion limit of the Moran model as  $N \rightarrow \infty$ ). In [43] this process is termed *common ancestor process (CAP)*, but keep in mind that it is  $(J_\tau, V_\tau)$  that is called CAP in [17].

In order to derive the stationary distribution of  $(I_t, X_t)_{t \geq 0}$ , which we denote by  $\pi_T$ , define  $h(x)$  as the probability that the common ancestor at a given time is of type 0, provided that the frequency of type-0 individuals at this time is  $x$ . Obviously  $h(0) = 0$ ,  $h(1) = 1$ . Since the process is time-homogeneous,  $h$  is independent of time. The marginal distributions of  $\pi_T$  are  $\alpha$  (with respect to the first variable) and  $\pi_X$  (with

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respect to the second variable).  $\pi_T$  may then be written as the product of the marginal density  $\pi_X(x)$  and the conditional probability  $h(x)$  (cf. [43, Sec. 2.1]):

$$\begin{aligned}\pi_T(0, x) dx &= h(x)\pi_X(x)dx, \\ \pi_T(1, x) dx &= (1 - h(x))\pi_X(x)dx.\end{aligned}$$

Since  $\pi_X$  is well-known (cf. (7)), it remains to specify  $h$ . Taylor uses a backward-forward construction within diffusion theory to derive a boundary value problem for  $h$ , namely:

$$\begin{aligned}\frac{1}{2}b(x)h''(x) + a(x)h'(x) - \left(\theta\nu_1\frac{x}{1-x} + \theta\nu_0\frac{1-x}{x}\right)h(x) + \theta\nu_1\frac{x}{1-x} &= 0, \\ h(0) = 0, h(1) &= 1.\end{aligned}\tag{19}$$

He also shows that (19) has a unique solution. The stationary distribution of  $(I_t, X_t)_{t \geq 0}$  is thus determined in a unique way as well. The function  $h$  is smooth in  $(0, 1)$  and its derivative  $h'$  can be continuously extended to  $[0, 1]$  (cf. [43, Lemma 2.3, Prop. 2.4]).

In the neutral case (i.e., without selection,  $\sigma = 0$ ), all individuals reproduce at the same rate, independently of their types. For reasons of symmetry, the common ancestor thus is a uniform random draw from the population; consequently,  $h(x) = x$ . In the presence of selection, Taylor determines the solution of the boundary value problem via a series expansion in  $\sigma$  (cf. [43, Sec. 4] and see Sec. 3.4), which yields

$$h(x) = x + \sigma x^{-\theta\nu_0} (1-x)^{-\theta\nu_1} \exp(-\sigma x) \int_0^x (\tilde{x} - p) p^{\theta\nu_0} (1-p)^{\theta\nu_1} \exp(\sigma p) dp \tag{20}$$

$$\text{with } \tilde{x} = \frac{\int_0^1 p^{\theta\nu_0+1} (1-p)^{\theta\nu_1} \exp(\sigma p) dp}{\int_0^1 p^{\theta\nu_0} (1-p)^{\theta\nu_1} \exp(\sigma p) dp} = \frac{\mathbb{E}_{\pi_X}(X^2(1-X))}{\mathbb{E}_{\pi_X}(X(1-X))}.\tag{21}$$

Notice that  $\tilde{x}$  is the conditional probability to obtain a fit individual, provided the population is in equilibrium, a sample containing an individual of each type has already been taken and we pick one additional individual [43, Sec. 4].

The stationary type distribution of the ancestral line now follows via marginalisation:

$$\alpha_0 = \int_0^1 h(x)\pi_X(x)dx \quad \text{and} \quad \alpha_1 = \int_0^1 (1 - h(x))\pi_X(x)dx.\tag{22}$$

Following [43, Sec. 2.1], we define  $\psi(x) := h(x) - x$  and write

$$h(x) = x + \psi(x).\tag{23}$$

Since  $h(x)$  is the conditional probability that the common ancestor is fit,  $\psi(x)$  is the part of this probability that is due to selective reproduction. Substituting (23) into (19) leads to a boundary value problem for  $\psi$ :

$$\begin{aligned}\frac{1}{2}b(x)\psi''(x) + a(x)\psi'(x) - \left(\theta\nu_1\frac{x}{1-x} + \theta\nu_0\frac{1-x}{x}\right)\psi(x) + \sigma x(1-x) &= 0, \\ \psi(0) = \psi(1) &= 0.\end{aligned}\tag{24}$$



Here, the smooth inhomogeneous term is more favourable as compared to the divergent inhomogeneous term in (19). Note that Taylor actually derives the boundary value problems (19) and (24) for the more general case of frequency-dependent selection, but restricts himself to frequency-independence to derive solution (20).

The work of Taylor now establishes a link to Fearnhead's framework by an alternative representation of  $h$  respectively  $\psi$  (cf. [43, Sec. 4.1]):

$$h(x) = x + x \sum_{n \geq 1} a_n (1-x)^n \quad \text{respectively} \quad \psi(x) = x \sum_{n \geq 1} a_n (1-x)^n \quad (25)$$

$$\text{with } a_n := \prod_{i=1}^n \rho_i. \quad (26)$$

The  $a_n$ , to which we refer as *Fearnhead's coefficients*, can be shown to follow the second-order *forward* recursion [43, Sec. 4.1]

$$\begin{aligned} (2 + \theta\nu_1) a_2 - (2 + \sigma + \theta) a_1 + \sigma &= 0, \\ (n + \theta\nu_1) a_n - (n + \sigma + \theta) a_{n-1} + \sigma a_{n-2} &= 0, \quad n \geq 3. \end{aligned} \quad (27)$$

Indeed,  $h(x)$  given as in (25) solves the boundary problem (19) and therefore equals (20) [43, Lemma 4.1].

The forward recursion (27) is greatly preferable to the backward recursion (12), which can only be solved approximately with initial value  $\rho_n \approx 0$  for some large  $n$ . What is still missing is the initial value,  $a_1$ . To calculate it, Taylor defines (cf. [43, Sec. 4.1])

$$v(x) := \frac{h(x) - x}{x} = \frac{\psi(x)}{x} = \sum_{n \geq 1} a_n (1-x)^n \quad (28)$$

and uses

$$a_n = \frac{(-1)^n}{n!} v^{(n)}(1). \quad (29)$$

(Note the missing factor of  $1/n$  in equation (28) in [43].) This way, a straightforward (but lengthy) calculation (that includes a differentiation of expression (20)) yields

$$a_1 = -v'(1) = -\psi'(1) = \frac{\sigma}{1 + \theta\nu_1} (1 - \tilde{x}). \quad (30)$$



### 3 The common ancestor process in the discrete setting

Our focus is on the stationary type distribution  $(\alpha_i)_{i \in S}$  of the CAT process. We have seen so far that it corresponds to the marginal distribution of both  $\pi_T$  and  $\pi_F$ , with respect to the first variable. Our aim now is to establish a closer connection between the properties of the ancestral type distribution and the graphical representation of the Moran model.

We define  $h^N$  as the equivalent of  $h$  in the case of finite population size  $N$ , that is,  $h_k^N$  is the probability that one of the  $k$  fit individuals is the common ancestor given  $Z_0^N = k$ . Obviously  $h_0^N = 0$ ,  $h_N^N = 1$ . Equivalently, we introduce the new function  $\psi_k^N := h_k^N - k/N$  as the part of  $h_k^N$  that goes back to selective reproductions (in comparison to the neutral case). We therefore speak of  $\psi^N$  (as well as of  $\psi$ ) as the ‘extra’ absorption probability.

In a first step (Sec. 3.1), we concentrate on a Moran model of finite size and trace the descendants of the initially ‘fit’ individuals forward in time. Decomposition according to what can happen after the first step gives difference equations for  $h^N$  and  $\psi^N$ , which turn into the differential equations for  $h$  and  $\psi$  in the diffusion limit (Sec. 3.3). Coming along with this approach, we solve the difference equations and obtain the fixation probability in the finite-size model in closed form (Sec. 3.2). In a second step, we solve the differential equation analytically (Sec. 3.4) and derive the coefficients of the ancestral type distribution within the discrete setting and within the framework of the diffusion limit (Sec. 3.5-3.6).

Most of our results are based on joint work with Thiemo Hustedt [23] and published in [31].

#### 3.1 Difference equations for $h^N$ and $\psi^N$

Note that the main contents of this section are, though in a less sophisticated way, already described in [29].

**Equation for  $h^N$ .** Since it is essential to make the connection with the graphical representation explicit, we start from a population of finite size  $N$ , rather than from the diffusion limit. Namely, we look at a new Markov process  $(\mathbf{M}_t, Z_t^N)_{t \geq 0}$  with the natural filtration  $(\mathcal{F}_t^N)_{t \geq 0}$ , where  $\mathcal{F}_t^N := \sigma((\mathbf{M}_s, Z_s^N) \mid 0 \leq s \leq t)$ .  $Z_t^N$  is the number of fit individuals as before and  $\mathbf{M}_t = (M_{0,t}, M_{1,t})$  holds the number of descendants of types 0 and 1 at time  $t$  of an unordered sample with composition  $\mathbf{M}_0 = (M_{0,0}, M_{1,0})$  collected at time 0. More precisely, we start with a  $\mathcal{F}_0^N$ -measurable state  $(\mathbf{M}_0, Z_0^N) = (\mathbf{m}, k)$  (this means that  $\mathbf{M}_0$  must be independent of the future evolution; but note that it need not be a random sample) and observe the population evolve in forward time. At time  $t$ , count the type-0 descendants and the type-1 descendants of our initial sample  $\mathbf{M}_0$  and summarise the results in the unordered sample  $\mathbf{M}_t$ . Together with  $Z_t^N$ , this gives the current state  $(\mathbf{M}_t, Z_t^N)$  (cf. Fig. 8).

As soon as the initial sample is ancestral to all  $N$  individuals, it clearly will be

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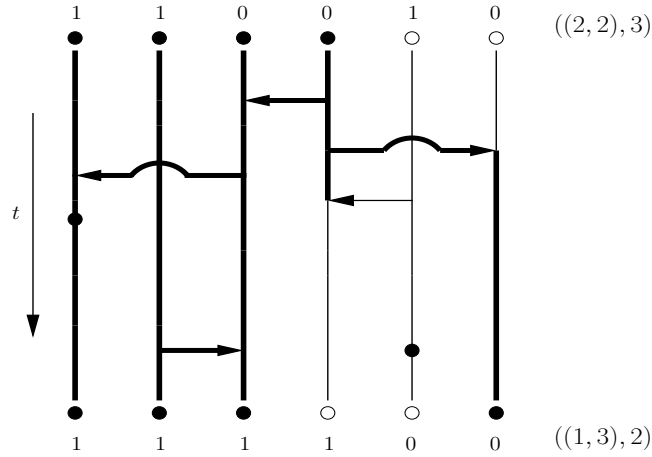


Figure 8: The process  $(\mathbf{M}_t, Z_t^N)_{t \geq 0}$ . The initial sample  $\mathbf{M}_0 = (2, 2)$  in a population of size  $N = 6$  (whose number of type-0 individuals is  $Z_0^N = 3$ ) is marked black at the top. Fat lines represent their descendants. At the later time (bottom), the descendants consist of one type-0 individual and three type-1 individuals, the entire population has two individuals of type 0. The initial and final states of the process are noted at the right.

ancestral to all  $N$  individuals at all later times. Therefore

$$\mathcal{A}_N := \{(\mathbf{m}, k) : k \in \{0, \dots, N\}, m_0 \leq k, |\mathbf{m}| = N\},$$

where  $|\mathbf{m}| = m_0 + m_1$  for a sample  $\mathbf{m} = (m_0, m_1)$ , is a closed (or invariant) set of the Markov chain. (Given a Markov chain  $(Y(t))_{t \geq 0}$  in continuous time on a discrete state space  $E$ , a non-empty subset  $\mathcal{A} \subseteq E$  is called closed (or invariant) provided that  $\mathbb{P}(Y(s) = j \mid Y(t) = i) = 0 \forall s > t, i \in \mathcal{A}, j \notin \mathcal{A}$  (cf. [38, Ch. 3.2]).)

From now on we restrict ourselves to the initial value  $(\mathbf{M}_0, Z_0^N) = ((k, 0), k)$ , i.e. the population consists of  $k$  fit individuals and the initial sample contains them all. Our aim is to calculate the probability of absorption in  $\mathcal{A}_N$ , which will also give us the fixation probability  $h_k^N$  of the descendants of the type-0 individuals. In other words,  $h_k^N$  is the probability that the common ancestor at time 0 belongs to our fit sample  $\mathbf{M}_0$ . It is important to note that, given absorption in  $\mathcal{A}^N$ , the common ancestor is a random draw from the initial sample. Therefore,

$$\mathbb{P}(\text{a specific type-0 individual will fix} \mid Z_0^N = k) = \frac{h_k^N}{k}. \quad (31)$$

Likewise,

$$\mathbb{P}(\text{a specific type-1 individual will fix} \mid Z_0^N = k) = \frac{1 - h_k^N}{N - k}. \quad (32)$$

We will now calculate the absorption probabilities with the help of ‘first-step analysis’ (cf. [38, Thm. 3.3.1] and [11, Thm. 7.5], see also [11, Sec. 6.1.1] for an approach in the case without mutation). Let us recall the method for convenience.

**Lemma 1** ('first-step analysis'). Assume that  $(Y(t))_{t \geq 0}$  is a Markov chain in continuous time on a discrete state space  $E$ ,  $\mathcal{A} \subseteq E$  is a closed set and  $T_x$ ,  $x \in E$ , is the waiting time to leave the state  $x$ . Then for all  $y \in E$ ,

$$\begin{aligned} \mathbb{P}(Y \text{ absorbs in } \mathcal{A} \mid Y(0) = y) &= \sum_{z \in E: z \neq y} \mathbb{P}(Y(T_y) = z \mid Y(0) = y) \\ &\quad \times \mathbb{P}(Y \text{ absorbs in } \mathcal{A} \mid Y(0) = z). \end{aligned}$$

So let us decompose the event 'absorption in  $\mathcal{A}_N$ ' according to the first step away from the initial state. Below we analyse all possible transitions (which are illustrated in Fig. 9), state the transition rates and calculate absorption probabilities, based upon the new state. We assume throughout that  $0 < k < N$ .

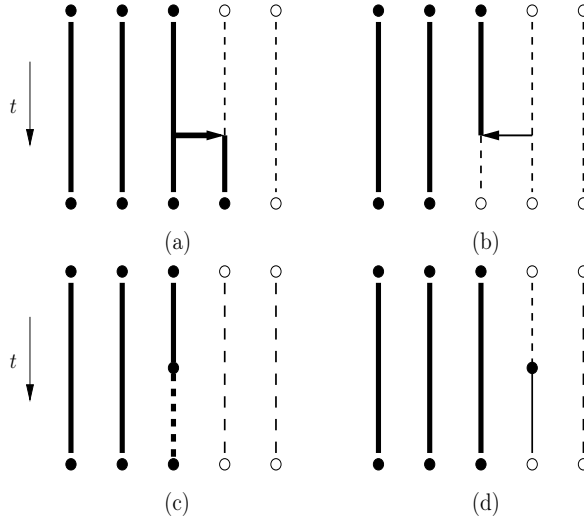


Figure 9: Transitions out of  $((k, 0), k)$ . Solid lines represent type-0 individuals, dashed ones type-1 individuals. Descendants of type-0 individuals (marked black at the top) are represented by bold lines.

(a)  $((k, 0), k) \rightarrow ((k + 1, 0), k + 1)$ :

One of the  $k$  sample individuals of type 0 reproduces and replaces a type-1 individual. We distinguish according to the kind of the reproduction event.

(a1) Neutral reproduction rate:  $\frac{k(N-k)}{N}$ .

(a2) Selective reproduction rate:  $\frac{k(N-k)}{N} s_N$ .

In both cases, the result is a sample containing all  $k + 1$  fit individuals. Now  $(\mathbf{M}_t, Z_t^N)$  starts afresh in the new state  $((k + 1, 0), k + 1)$ , with absorption probability  $h_{k+1}^N$ .

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(b)  $((k, 0), k) \rightarrow ((k-1, 0), k-1)$  :

A type-1 individual reproduces and replaces a (sample) individual of type 0. This occurs at rate  $k(N-k)/N$  and leads to a sample that consists of all  $k-1$  fit individuals. The absorption probability, if we start in the new state, is  $h_{k-1}^N$ .

(c)  $((k, 0), k) \rightarrow ((k-1, 1), k-1)$ :

This transition describes a mutation of a type-0 individual to type 1 and occurs at rate  $ku_N\nu_1$ . The new sample contains all  $k-1$  fit individuals, plus a single unfit one. Starting now from  $((k-1, 1), k-1)$ , the absorption probability has two contributions: First, by definition, with probability  $h_{k-1}^N$ , one of the  $k-1$  fit individuals will be the common ancestor. In addition, by (32), the single unfit individual has fixation probability  $(1 - h_{k-1}^N)/(N - (k-1))$ , so the probability to absorb in  $\mathcal{A}_N$ , when starting from the new state, is

$$\begin{aligned} & \mathbb{P}(\text{absorption in } \mathcal{A}_N \mid (\mathbf{M}_0, Z_0^N) = ((k-1, 1), k-1)) \\ &= h_{k-1}^N + \frac{1 - h_{k-1}^N}{N - (k-1)}. \end{aligned}$$

(d)  $((k, 0), k) \rightarrow ((k, 0), k+1)$ :

This is a mutation from type 1 to type 0, which occurs at rate  $(N-k)u_N\nu_0$ . We then have  $k+1$  fit individuals in the population altogether, but the new sample contains only  $k$  of them. Arguing as in (c) and this time using (31), we get

$$\begin{aligned} & \mathbb{P}(\text{absorption in } \mathcal{A}_N \mid (\mathbf{M}_0, Z_0^N) = ((k, 0), k+1)) \\ &= h_{k+1}^N - \frac{h_{k+1}^N}{k+1}. \end{aligned}$$

Note that, in steps (c) and (d) (and already in (31) and (32)), we have used the permutation invariance of the fit (respectively unfit) lines to express the absorption probabilities as a function of  $k$  (the number of fit individuals in the population) alone. This way, we need not cope with the full state space of  $(\mathbf{M}_t, Z_t^N)$ . Taking together the first-step principle with the results of (a)-(d), we obtain the linear system of equations for  $h^N$  (with the rates  $\lambda_k^N$  and  $\mu_k^N$  as in (1)):

$$(\lambda_k^N + \mu_k^N) h_k^N = \lambda_k^N h_{k+1}^N + \mu_k^N h_{k-1}^N + ku_N\nu_1 \frac{1 - h_{k-1}^N}{N - (k-1)} - (N-k)u_N\nu_0 \frac{h_{k+1}^N}{k+1}, \quad (33)$$

$0 < k < N$ , which is complemented by the boundary conditions  $h_0^N = 0$ ,  $h_N^N = 1$ .

**Equation for  $\psi^N$ .** As before, we consider  $(\mathbf{M}_t, Z_t^N)_{t \geq 0}$  with start in  $((k, 0), k)$ , where  $\psi^N$  is the part of absorption probability in  $\mathcal{A}_N$  that goes back to selective reproductions. Substituting  $h_k^N = \psi_k^N + k/N$  in (33) yields the following difference equation for  $\psi^N$ :

$$\begin{aligned} (\lambda_k^N + \mu_k^N) \psi_k^N &= \lambda_k^N \psi_{k+1}^N + \mu_k^N \psi_{k-1}^N + \frac{k(N-k)}{N^2} s_N \\ &\quad - ku_N\nu_1 \frac{\psi_{k-1}^N}{N - (k-1)} - (N-k)u_N\nu_0 \frac{\psi_{k+1}^N}{k+1} \end{aligned} \quad (34)$$

( $0 < k < N$ ), together with the boundary conditions  $\psi_0^N = \psi_N^N = 0$ . It has a nice interpretation, which is completely analogous to that of  $h^N$  except in case (a2): If one of the fit sample individuals reproduces via a selective reproduction event, the extra absorption probability is  $\psi_{k+1}^N + 1/N$  (rather than  $h_{k+1}^N$ ). Here,  $1/N$  is the neutral fixation probability of the individual just created via the selective event;  $\psi_{k+1}^N$  is the extra absorption probability of all  $k + 1$  type-0 individuals present after the event. The neutral contribution gives rise to the  $k(N - k)s_N/N^2$  term on the right-hand side of (34).

### 3.2 Solution of the difference equation

In this section, we derive an explicit expression for the fixation probabilities  $h_k^N$ , that is, a solution of the difference equation (33), or equivalently, (34). Although the calculations only involve standard techniques, we perform them here explicitly since this yields additional insight. Since there is no danger of confusion, we omit the subscript (or superscript)  $N$  for economy of notation.

The following lemma specifies the extra absorption probabilities  $\psi_k$  in terms of a recursion.

**Lemma 2.** *Let  $k \geq 1$ . Then*

$$\psi_{N-k} = \frac{k(N-k)}{\mu_{N-k}} \left( \frac{\mu_{N-1}}{N-1} \psi_{N-1} + \frac{\lambda_{N-k+1}}{(k-1)(N-k+1)} \psi_{N-k+1} - \frac{s(k-1)}{N^2} \right). \quad (35)$$

**Remark 1.** *The quantity  $\lambda_k/(k(N-k)) = (1+s)/N + u\nu_0/k$  is well-defined for all  $1 \leq k \leq N$ , and  $k(N-k)/\mu_k = (N-k)/(\frac{N-k}{N} + u\nu_1)$  is well-defined even for  $k = 0$ .*

*Proof of Lemma 2.* Let  $1 < i < N - 1$ . Set  $k = i$  in (34) and divide by  $i(N - i)$  to obtain

$$\begin{aligned} \left( \frac{\lambda_i}{i(N-i)} + \frac{\mu_i}{i(N-i)} \right) \psi_i &= \left( \frac{1+s}{N} + \frac{u\nu_0}{i+1} \right) \psi_{i+1} + \left( \frac{1}{N} + \frac{u\nu_1}{N-(i-1)} \right) \psi_{i-1} + \frac{s}{N^2} \\ &= \frac{\lambda_{i+1}}{(i+1)(N-i-1)} \psi_{i+1} + \frac{\mu_{i-1}}{(i-1)(N-i+1)} \psi_{i-1} + \frac{s}{N^2}. \end{aligned} \quad (36)$$

Together with

$$\left( \frac{\lambda_1}{N-1} + \frac{\mu_1}{N-1} \right) \psi_1 = \frac{\lambda_2}{2(N-2)} \psi_2 + \frac{s}{N^2}, \quad (37)$$

$$\left( \frac{\lambda_{N-1}}{N-1} + \frac{\mu_{N-1}}{N-1} \right) \psi_{N-1} = \frac{\mu_{N-2}}{2(N-2)} \psi_{N-2} + \frac{s}{N^2}, \quad (38)$$

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and the boundary conditions  $\psi_0 = \psi_N = 0$ , we obtain a new linear system of equations for the vector  $\psi = (\psi_k)_{0 \leq k \leq N}$ . Summation over the last  $k$  equations yields

$$\begin{aligned} \sum_{i=N-k+1}^{N-1} \left( \frac{\lambda_i}{i(N-i)} + \frac{\mu_i}{i(N-i)} \right) \psi_i &= \sum_{i=N-k+1}^{N-2} \frac{\lambda_{i+1}}{(i+1)(N-i-1)} \psi_{i+1} \\ &+ \sum_{i=N-k+1}^{N-1} \frac{\mu_{i-1}}{(i-1)(N-i+1)} \psi_{i-1} + \frac{s(k-1)}{N^2}, \end{aligned}$$

which proves the assertion.  $\square$

Lemma 2 allows for an explicit solution for  $\psi$ .

**Theorem 1.** For  $1 \leq \ell, n \leq N-1$ , let

$$\chi_\ell^n := \prod_{i=\ell}^n \frac{\lambda_i}{\mu_i} \quad \text{and} \quad K := \sum_{n=0}^{N-1} \chi_1^n. \quad (39)$$

The solution of recursion (35) is then given by

$$\psi_{N-k} = \frac{k(N-k)}{\mu_{N-k}} \sum_{n=N-k}^{N-1} \chi_{N-k+1}^n \left( \frac{\mu_{N-1}}{N-1} \psi_{N-1} - \frac{s(N-1-n)}{N^2} \right) \quad (40)$$

with

$$\psi_{N-1} = \frac{1}{K} \frac{N-1}{\mu_{N-1}} \frac{s}{N^2} \sum_{n=0}^{N-2} (N-1-n) \chi_1^n. \quad (41)$$

An alternative representation is given by

$$\psi_{N-k} = \frac{1}{K} \frac{k(N-k)}{\mu_{N-k}} \frac{s}{N^2} \sum_{\ell=0}^{N-k-1} \sum_{n=N-k}^{N-1} (n-\ell) \chi_1^\ell \chi_{N-k+1}^n. \quad (42)$$

*Proof.* We first prove (40) by induction over  $k$ . For  $k=1$ , (40) is easily checked to be true. Inserting the induction hypothesis for some  $k-1 \geq 0$  into recursion (35) yields

$$\begin{aligned} \psi_{N-k} &= \frac{k(N-k)}{\mu_{N-k}} \left[ \frac{\mu_{N-1}}{N-1} \psi_{N-1} \right. \\ &\quad \left. + \frac{\lambda_{N-k+1}}{\mu_{N-k+1}} \sum_{n=N-k+1}^{N-1} \chi_{N-k+2}^n \left( \frac{\mu_{N-1}}{N-1} \psi_{N-1} - \frac{s(N-1-n)}{N^2} \right) - \frac{s(k-1)}{N^2} \right], \end{aligned}$$



which immediately leads to (40). For  $k = N$ , (40) gives (41), since  $\psi_0 = 0$  and  $k(N - k)/\mu_{N-k}$  is well-defined by Remark 1. We now check (42) by inserting (41) into (40) and then use the expression for  $K$  as in (39):

$$\begin{aligned}\psi_{N-k} &= \frac{1}{K} \frac{k(N-k)}{\mu_{N-k}} \frac{s}{N^2} \sum_{n=N-k}^{N-1} \chi_{N-k+1}^n \left[ \sum_{\ell=0}^{N-1} (N-1-\ell) \chi_1^\ell - \sum_{\ell=0}^{N-1} (N-1-n) \chi_1^\ell \right] \\ &= \frac{1}{K} \frac{k(N-k)}{\mu_{N-k}} \frac{s}{N^2} \sum_{\ell=0}^{N-1} \sum_{n=N-k}^{N-1} (n-\ell) \chi_1^\ell \chi_{N-k+1}^n.\end{aligned}$$

Then we split the first sum according to whether  $\ell \leq N - k - 1$  or  $\ell \geq N - k$ , and use  $\chi_1^\ell = \chi_1^{N-k} \chi_{N-k+1}^\ell$  in the latter case:

$$\begin{aligned}\psi_{N-k} &= \frac{1}{K} \frac{k(N-k)}{\mu_{N-k}} \frac{s}{N^2} \left[ \sum_{\ell=0}^{N-k-1} \sum_{n=N-k}^{N-1} (n-\ell) \chi_1^\ell \chi_{N-k+1}^n \right. \\ &\quad \left. + \chi_1^{N-k} \sum_{\ell=N-k}^{N-1} \sum_{n=N-k}^{N-1} (n-\ell) \chi_{N-k+1}^\ell \chi_{N-k+1}^n \right].\end{aligned}$$

The first sum is the right-hand side of (42) and the second sum disappears due to symmetry.  $\square$

Let us note that the fixation probabilities thus obtained have been well-known for the case with selection in the absence of mutation (see, e.g., [11, Thm. 6.1] and Ch. 5), but, to the best of our knowledge, have not yet appeared in the literature for the case with mutation. Note also that our calculations in Sec. 3.1 and Sec. 3.2 do not require  $u_N > 0$  or  $0 < \nu_0, \nu_1 < 1$ .

### 3.3 Differential equations for $h$ and $\psi$

In Sec. 3.1 we have presented a difference equation for the conditional probability  $h^N$  (cf. (33)). To derive the boundary value problem for  $h$  from this difference equation, it remains to prove that  $\lim_{N \rightarrow \infty} h_{k_N}^N = h(x)$ , with  $x \in [0, 1]$ ,  $0 < k_N < N$ ,  $\lim_{N \rightarrow \infty} k_N/N = x$  and  $h$  as given as in (20).

Since  $h_k^N = k/N + \psi_k^N$  and  $h(x) = x + \psi(x)$ , respectively, it suffices to show the corresponding convergence of the  $\psi_k^N$ . For ease of exposition we assume in Lemma 3 and Thm. 2 that the process is stationary.

**Lemma 3.** *Let  $\tilde{x}$  be as in (21). Then*

$$\lim_{N \rightarrow \infty} N \psi_{N-1}^N = \frac{\sigma}{1 + \theta \nu_1} (1 - \tilde{x}).$$

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*Proof.* Since the stationary distribution  $\pi_Z^N$  of  $(Z_t^N)_{t \geq 0}$  (cf. (2)) satisfies

$$\prod_{i=1}^{n-1} \frac{\lambda_i^N}{\mu_i^N} = \frac{\pi_Z^N(n) \mu_n^N}{C_N \lambda_0^N} \quad (43)$$

for  $1 \leq n \leq N$ , equation (41) leads to

$$\begin{aligned} N\psi_{N-1}^N &= \frac{Ns_N}{1 + Nu_N\nu_1} \frac{\sum_{n=1}^N \pi_Z^N(n) \mu_n^N \frac{N-n}{N}}{\sum_{n=1}^N \pi_Z^N(n) \mu_n^N} \\ &= \frac{Ns_N}{1 + Nu_N\nu_1} \frac{\sum_{n=1}^N \pi_Z^N(n) \frac{n(N-n)^2}{N^3} \left(1 + \frac{Nu_N\nu_1}{N-n}\right)}{\sum_{n=1}^N \pi_Z^N(n) \frac{n(N-n)}{N^2} \left(1 + \frac{Nu_N\nu_1}{N-n}\right)}, \end{aligned}$$

where we have used (1) in the last step. The stationary distribution of the rescaled process  $(X_t^N)_{t \geq 0}$  is given by  $(\pi_X^N(i/N))_{0 \leq i \leq N}$ , where  $\pi_X^N(i/N) = \pi_Z^N(i)$ . Besides, the sequence of processes  $(X_t^N)_{t \geq 0}$  converges to  $(X_t)_{t \geq 0}$  in distribution, hence

$$\begin{aligned} \lim_{N \rightarrow \infty} N\psi_{N-1}^N &= \lim_{N \rightarrow \infty} \frac{Ns_N}{1 + Nu_N\nu_1} \frac{\mathbb{E}_{\pi_X^N} \left( X^N (1 - X^N)^2 \left(1 + \frac{u_N\nu_1}{1 - X^N}\right) \right)}{\mathbb{E}_{\pi_X^N} \left( X^N (1 - X^N) \left(1 + \frac{u_N\nu_1}{1 - X^N}\right) \right)} \\ &= \frac{\sigma}{1 + \theta\nu_1} \frac{\mathbb{E}_{\pi_X} (X(1 - X)^2)}{\mathbb{E}_{\pi_X} (X(1 - X))} = \frac{\sigma}{1 + \theta\nu_1} (1 - \tilde{x}), \end{aligned}$$

as claimed.  $\square$

**Remark 2.** The proof gives an alternative way to obtain the initial value  $a_1$  (cf. (30)) of recursion (27).

**Theorem 2.** For a given  $x \in [0, 1]$ , let  $(k_N)_{N \in \mathbb{N}}$  be a sequence with  $0 < k_N < N$  and  $\lim_{N \rightarrow \infty} k_N/N = x$ . Then

$$\lim_{N \rightarrow \infty} \psi_{k_N}^N = \psi(x),$$

where  $\psi$  is the solution of the boundary value problem (24).

*Proof.* Using first Theorem 1, then (43) and finally (1), we obtain

$$\begin{aligned} \psi_k^N &= \frac{k(N-k)}{\mu_k^N} \sum_{n=1}^{N-k} \left( \prod_{i=k+1}^{N-n} \frac{\lambda_i^N}{\mu_i^N} \right) \left( \frac{\mu_{N-1}^N}{N-1} \psi_{N-1}^N - \frac{s_N(n-1)}{N^2} \right) \\ &= \frac{k(N-k)}{\mu_k^N} (\mu_{k+1}^N \pi_Z^N(k+1))^{-1} \sum_{n=0}^{N-k-1} \mu_{N-n}^N \pi_Z^N(N-n) \left( \frac{\mu_{N-1}^N}{N-1} \psi_{N-1}^N - \frac{s_N n}{N^2} \right) \\ &= \left( 1 + \mathcal{O}\left(\frac{1}{N}\right) \right) \left( \frac{k+1}{N} \frac{N-k-1}{N} \pi_Z^N(k+1) \right)^{-1} \\ &\quad \times \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N-n) \frac{N-n}{N} \frac{n}{N} \left( 1 + \frac{Nu_N\nu_1}{n} \right) \left( (1 + Nu_N\nu_1) N\psi_{N-1}^N - Ns_N \frac{n}{N} \right), \end{aligned}$$

where  $\mathcal{O}(1/N)$  denotes the Landau symbol ‘big O’ as  $N \rightarrow \infty$ . In order to analyse the convergence of this expression, define

$$\begin{aligned} S_1^N(k) &:= \frac{k+1}{N} \frac{N-k-1}{N} \pi_Z^N(k+1), \\ S_2^N(k) &:= \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N-n) \frac{N-n}{N} \frac{n}{N} \left( (1 + Nu_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N} \right) \\ &= \int_0^1 T_k^N(y) dy, \\ S_3^N(k) &:= \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N-n) \frac{N-n}{N} u_N \nu_1 \left( (1 + Nu_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N} \right) \\ &= \int_0^1 \tilde{T}_k^N(y) dy \end{aligned}$$

with step functions  $T_k^N : [0, 1] \rightarrow \mathbb{R}$ ,  $\tilde{T}_k^N : [0, 1] \rightarrow \mathbb{R}$  given by

$$\begin{aligned} T_k^N(y) &:= \begin{cases} \mathbb{1}_{\{n \leq N-k-1\}} \pi_Z^N(N-n) \frac{N-n}{N} \frac{n}{N} \left( (1 + Nu_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N} \right), \\ \text{if } \frac{n}{N} \leq y < \frac{n+1}{N}, n \in \{0, \dots, N-1\}, \\ 0, \text{ if } y = 1, \end{cases} \\ \tilde{T}_k^N(y) &:= \begin{cases} \mathbb{1}_{\{n \leq N-k-1\}} \pi_Z^N(N-n) \frac{N-n}{N} u_N \nu_1 \left( (1 + Nu_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N} \right), \\ \text{if } \frac{n}{N} \leq y < \frac{n+1}{N}, n \in \{0, \dots, N-1\}, \\ 0, \text{ if } y = 1. \end{cases} \end{aligned}$$

Consider now a sequence  $(k_N)_{N \in \mathbb{N}}$  as in the assumptions. Then  $\lim_{N \rightarrow \infty} \pi_Z^N(k_N) = \pi_X(x)$  (cf. [11, p. 319]), and due to Lemma 3

$$\begin{aligned} \lim_{N \rightarrow \infty} S_1^N(k_N) &= x(1-x)\pi_X(x), \\ \lim_{N \rightarrow \infty} T_{k_N}^N(k_N) &= \mathbb{1}_{\{y \leq 1-x\}} \pi_X(1-y)(1-y)y(\sigma(1-\tilde{x}) - \sigma y), \\ \lim_{N \rightarrow \infty} \tilde{T}_{k_N}^N(k_N) &= 0. \end{aligned}$$

Since  $T_k^N$  and  $\tilde{T}_k^N$  are bounded, we have

$$\begin{aligned} \lim_{N \rightarrow \infty} S_2^N(k_N) &= \int_0^{1-x} \pi_X(1-y)(1-y)y(\sigma(1-\tilde{x}) - \sigma y) dy, \\ \lim_{N \rightarrow \infty} S_3^N(k_N) &= 0, \end{aligned}$$

thus

$$\lim_{N \rightarrow \infty} \psi_{k_N}^N = (x(1-x)\pi_X(x))^{-1} \int_0^{1-x} \pi_X(1-y)(1-y)y(\sigma(1-\tilde{x}) - \sigma y) dy.$$

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Substituting on the right-hand side yields

$$\begin{aligned}
\lim_{N \rightarrow \infty} \psi_{k_N}^N &= (x(1-x)\pi_X(x))^{-1} \sigma \int_x^1 \pi_X(y)y(1-y)(y-\tilde{x})dy \\
&= (x(1-x)\pi_X(x))^{-1} \sigma \left[ \int_0^1 \pi_X(y)y(1-y)(y-\tilde{x})dy \right. \\
&\quad \left. + \int_0^x \pi_X(y)y(1-y)(\tilde{x}-y)dy \right] \\
&= (x(1-x)\pi_X(x))^{-1} \sigma \int_0^x \pi_X(y)y(1-y)(\tilde{x}-y)dy = \psi(x),
\end{aligned}$$

where the second-last equality goes back to the definition of  $\tilde{x}$  in (21), and the last is caused by (20), (23) and (7).  $\square$

**Remark 3.** A proof of the convergence of the  $\psi_k^N$  that also includes the non-stationary case is given in [23, Lemma 3.18, Thm. 3.19], but it is quite lengthy and cumbersome. The simplicity of our approach (which requires the population to be stationary) is based on the substitution of the products  $\prod_{i=\ell}^n \lambda_i^N / \mu_i^N$  of Thm. 1 by the stationary distribution  $\pi_Z^N$ .

**Remark 4.** The case without mutation is particularly simple:  $h_k^N$  corresponds to the fixation probability of type 0 given that there are initially  $k$  type-0 individuals in the population. It is well-known that (cf. [11, Thm. 6.1])

$$h_k^N = \frac{\sum_{i=N-k}^{N-1} (1+s_N)^i}{\sum_{i=0}^{N-1} (1+s_N)^i},$$

which becomes with the help of the geometric series

$$h_k^N = \frac{(1+s_N)^{N-k} - (1+s_N)^N}{1 - (1+s_N)^N} = \frac{\left(1 + \frac{Ns_N}{N}\right)^{N\frac{N-k}{N}} - \left(1 + \frac{Ns_N}{N}\right)^N}{1 - \left(1 + \frac{Ns_N}{N}\right)^N}.$$

We then perform the  $N \rightarrow \infty$  limit for a sequence  $(k_N)_{N \in \mathbb{N}}$  with  $0 \leq k_N \leq N$  and  $\lim_{N \rightarrow \infty} k_N/N = x$ , and obtain

$$\lim_{N \rightarrow \infty} h_{k_N}^N = \frac{\exp(\sigma(1-x)) - \exp(\sigma)}{1 - \exp(\sigma)} = \frac{1 - \exp(-\sigma x)}{1 - \exp(-\sigma)} = h(x),$$

where the last equality is a classical result of Kimura [26]. In Ch. 5 a detailed investigation will follow.

**Differential equations for  $h$  and  $\psi$ .** Rearranging difference equation (33) results in

$$\begin{aligned}
&\frac{1}{2} \frac{1}{N} (\lambda_k^N + \mu_k^N) N^2 (h_{k+1}^N - 2h_k^N + h_{k-1}^N) \\
&+ \frac{1}{2} (\lambda_k^N - \mu_k^N) (N (h_{k+1}^N - h_k^N) - N (h_{k-1}^N - h_k^N)) \\
&+ \frac{k}{N} \frac{N}{N - (k-1)} Nu_N \nu_1 (1 - h_{k-1}^N) - \frac{N-k}{N} \frac{N}{k+1} Nu_N \nu_0 h_{k+1}^N = 0.
\end{aligned} \tag{44}$$

Let us consider a sequence  $(k_N)_{N \in \mathbb{N}}$  with  $0 < k_N < N$  and  $\lim_{N \rightarrow \infty} k_N/N = x$ . According to Thm. 2 (in the stationary case) and Remarks 3 and 4 (in the non-stationary case), the probabilities  $h_{k_N}^N$  converge to  $h(x)$  as  $N \rightarrow \infty$ . Then, equation (44), with  $k$  replaced by  $k_N$ , together with (4) and (5) leads to Taylor's boundary value problem (19), but now with a plausible interpretation attached to it.

Equivalently, we start from the difference equation for  $\psi^N$  (cf. (34)) and perform  $N \rightarrow \infty$  in the same way as for  $h$ . We obtain Taylor's boundary value problem (24) and now have an interpretation in terms of the graphical representation to go with it.

### 3.4 The solution of the differential equation

As a little detour, let us further deal with the boundary value problem (19). To solve it, Taylor assumes that  $h$  can be expanded in a power series in  $\sigma$ . This yields a recursive series of boundary value problems (for the various powers of  $\sigma$ ), which are solved by elementary methods and combined into a solution of  $h$  (cf. [43, Sec. 4]).

However, the calculations are slightly long-winded. In what follows we show that the boundary value problem (19) (or equivalently (24)) may be solved in a direct and elementary way, without the need for a series expansion. Defining

$$c(x) := -\theta\nu_1 \frac{x}{1-x} - \theta\nu_0 \frac{1-x}{x}$$

and remembering the drift coefficient  $a(x)$  (cf. (4)) and the diffusion coefficient  $b(x)$  (cf. (5)), differential equation (24) reads

$$\frac{1}{2}b(x)\psi''(x) + a(x)\psi'(x) + c(x)\psi(x) = -\sigma x(1-x)$$

or, equivalently,

$$\psi''(x) + 2\frac{a(x)}{b(x)}\psi'(x) + 2\frac{c(x)}{b(x)}\psi(x) = -\sigma. \quad (45)$$

Since the alternating sum of the coefficients' derivatives is

$$2\frac{c(x)}{b(x)} - \frac{d}{dx}2\frac{a(x)}{b(x)} + 0 = 0, \quad (46)$$

(45) is an exact differential equation (for the concept of exactness, see [18, Ch. 3.11] or [5, Ch. 2.6]). Solving it corresponds to solving its primitive

$$\psi'(x) + 2\frac{a(x)}{b(x)}\psi(x) = -\sigma(x - \tilde{x}). \quad (47)$$

The constant  $\tilde{x}$  plays the role of an integration constant and will be determined by the initial conditions later. (Obviously, (45) is recovered by differentiating (47) and observing (46).) As usual, we consider the homogeneous equation

$$\varphi'(x) + 2\frac{a(x)}{b(x)}\varphi(x) = \varphi'(x) + \left(\sigma - \frac{\theta\nu_1}{1-x} + \frac{\theta\nu_0}{x}\right)\varphi(x) = 0$$

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first. According to [11, Ch. 7.4] and [16, Ch. 4.3], its solution  $\varphi_1$  is given by

$$\varphi_1(x) = \exp\left(\int^x -2\frac{a(z)}{b(z)}dz\right) = \kappa(1-x)^{-\theta\nu_1}x^{-\theta\nu_0}\exp(-\sigma x) = \frac{2C\kappa}{b(x)\pi_X(x)}.$$

(Note the link to the stationary distribution (if it exists) provided by the last expression (cf. [11, Thm. 7.8] and [16, Ch. 4.5]).) Of course the same expression is obtained via separation of variables. Again we will deal with the constant  $\kappa$  later.

Variation of parameters yields the solution  $\varphi_2$  of the inhomogeneous equation (47):

$$\varphi_2(x) = \varphi_1(x) \int_{\beta}^x \frac{-\sigma(p - \tilde{x})}{\varphi_1(p)} dp = \sigma\varphi_1(x) \int_{\beta}^x \frac{\tilde{x} - p}{\varphi_1(p)} dp. \quad (48)$$

Finally, it remains to specify the constants of integration  $\tilde{x}$ ,  $\kappa$  and the constant  $\beta$  to comply with  $\varphi_2(0) = \varphi_2(1) = 0$ . We observe that the factor  $\kappa$  cancels in (48), thus its choice is arbitrary.  $\varphi_1(x)$  diverges for  $x \rightarrow 0$  and  $x \rightarrow 1$ , so the choice of  $\beta$  and  $\tilde{x}$  has to guarantee  $B(0) = B(1) = 0$ , where  $B(x) = \int_{\beta}^x \frac{\tilde{x} - p}{\varphi_1(p)} dp$ . Hence  $\beta = 0$  and

$$\tilde{x} \int_0^1 \frac{1}{\varphi_1(p)} dp = \int_0^1 \frac{p}{\varphi_1(p)} dp \Leftrightarrow \tilde{x} = \frac{\int_0^1 \frac{p}{\varphi_1(p)} dp}{\int_0^1 \frac{1}{\varphi_1(p)} dp}.$$

For the sake of completeness, l'Hôpital's rule can be used to check that  $\varphi_2(0) = \varphi_2(1) = 0$ . The result indeed coincides with Taylor's (cf. (20), (21)).

We close this section with a brief consideration of the initial value  $a_1$  of the recursions (27). Since, by (30),  $a_1 = -\psi'(1)$ , it may be obtained by analysing the limit  $x \rightarrow 1$  of (47). In the quotient  $a(x)\psi(x)/b(x)$ , numerator and denominator disappear as  $x \rightarrow 1$ . According to l'Hôpital's rule we get

$$\lim_{x \rightarrow 1} \frac{a(x)\psi(x)}{b(x)} = \lim_{x \rightarrow 1} \frac{(-\theta\nu_0 - \theta\nu_1 + \sigma(1 - 2x))\psi(x) + a(x)\psi'(x)}{2(1 - 2x)} = \frac{1}{2}\theta\nu_1\psi'(1),$$

therefore the limit  $x \rightarrow 1$  of (47) yields

$$-\psi'(1)(1 + \theta\nu_1) = \sigma(1 - \tilde{x}).$$

Thus, we obtain  $a_1$  without the need to differentiate expression (20).

### 3.5 Fearnhead's coefficients

Let us now turn to the ancestral type distribution and Fearnhead's coefficients that characterise it. To this end, we start from the linear system of equations for  $\psi^N = (\psi_k^N)_{0 \leq k \leq N}$  in (36)-(38). Let

$$\tilde{\psi}_k^N := \frac{\psi_k^N}{k(N - k)} \quad (49)$$

for  $1 \leq k \leq N-1$ . In terms of these new variables, (38) reads

$$-\mu_{N-1}^N \tilde{\psi}_{N-1}^N + \mu_{N-2}^N \tilde{\psi}_{N-2}^N - \lambda_{N-1}^N \tilde{\psi}_{N-1}^N + \frac{s_N}{N^2} = 0. \quad (50)$$

We now perform linear combinations of (36) and (38) (again expressed in terms of the  $\tilde{\psi}_{N-k}^N$ ) to obtain

$$\begin{aligned} & \sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} (\lambda_{N-k}^N + \mu_{N-k}^N) \tilde{\psi}_{N-k}^N \\ &= \sum_{k=2}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} \lambda_{N-k+1}^N \tilde{\psi}_{N-k+1}^N + \sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} \mu_{N-k-1}^N \tilde{\psi}_{N-k-1}^N \quad (51) \\ &+ \frac{s_N}{N^2} \sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} \end{aligned}$$

for  $3 \leq n \leq N-1$ . Noting that the last sum disappears as a consequence of the binomial theorem, rearranging turns (51) into

$$\sum_{k=0}^{n-1} (-1)^{n-k-1} \binom{n-1}{k} \mu_{N-k-1}^N \tilde{\psi}_{N-k-1}^N + \sum_{k=1}^{n-1} (-1)^{n-k} \binom{n-1}{k} \lambda_{N-k}^N \tilde{\psi}_{N-k}^N = 0. \quad (52)$$

On the basis of equations (50) and (52) for  $(\tilde{\psi}_k^N)_{1 \leq k \leq N-1}$  we will now establish a discrete version of Fearnhead's coefficients, and a corresponding discrete version of recursion (27) and initial value (30). Motivated by the limiting expression (25), we choose the ansatz

$$h_{N-k}^N = \frac{N-k}{N} + (N-k) \sum_{i=1}^k a_i^N \frac{k_{[i]}}{N_{[i+1]}} \quad \text{respectively} \quad \psi_{N-k}^N = (N-k) \sum_{i=1}^k a_i^N \frac{k_{[i]}}{N_{[i+1]}}, \quad (53)$$

where we adopt the usual notation

$$y_{[j]} := y(y-1) \dots (y-j+1) \quad (54)$$

for  $y \in \mathbb{R}$ ,  $j \in \mathbb{N}$ . Again we omit the upper (and lower) population size index  $N$  (except for the one of the  $a_n^N$ ) in the following theorem.

**Theorem 3.** *The  $a_n^N$ ,  $1 \leq n \leq N-1$ , satisfy the following relations:  $a_1^N = N\psi_{N-1}$ ,*

$$(N-2) \left[ \left( \frac{2}{N} + uv_1 \right) a_2^N - \left( \frac{2}{N} + \frac{N-1}{N}s + u \right) a_1^N + \frac{N-1}{N}s \right] = 0, \quad (55)$$

and, for  $3 \leq n \leq N-1$ :

$$(N-n) \left[ \left( \frac{n}{N} + uv_1 \right) a_n^N - \left( \frac{n}{N} + \frac{N-(n-1)}{N}s + u \right) a_{n-1}^N + \frac{N-(n-1)}{N}sa_{n-2}^N \right] = 0. \quad (56)$$

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*Proof.* At first we note that the initial value  $a_1^N$  follows directly from (53) for  $k = 1$ . Then, we remark that, by (49) and (53),

$$\tilde{\psi}_{N-k} = \frac{1}{k} \sum_{i=1}^k a_i^N \frac{k^{[i]}}{N^{[i+1]}} \quad (57)$$

for  $1 \leq k \leq N - 1$ . To prove (55), we insert (57) into (50) and write the resulting equality as

$$\mu_{N-2} a_2^N - (\mu_{N-1} - \mu_{N-2} + \lambda_{N-1})(N-2)a_1^N + \frac{(N-1)(N-2)}{N} s = 0,$$

which is easily checked to coincide with (55).

To prove (56), we express (52) in terms of the  $a_n^N$  via (57). The first sum of (52) becomes

$$\begin{aligned} \sum_{k=0}^{n-1} (-1)^{n-k-1} \binom{n-1}{k} \mu_{N-k-1} \tilde{\psi}_{N-k-1} &= \sum_{k=0}^{n-1} (-1)^{n-k-1} \binom{n-1}{k} \mu_{N-k-1} \sum_{i=1}^{k+1} a_i^N \frac{k^{[i-1]}}{N^{[i+1]}} \\ &= \sum_{i=1}^n a_i^N \sum_{k=i}^n (-1)^{n-k} \binom{n-1}{k-1} \frac{(k-1)^{[i-1]}}{N^{[i+1]}} \mu_{N-k}. \end{aligned}$$

Analogously, the second sum of (52) turns into

$$\sum_{k=1}^{n-1} (-1)^{n-k} \binom{n-1}{k} \lambda_{N-k} \tilde{\psi}_{N-k} = \sum_{i=1}^{n-1} a_i^N \sum_{k=i}^{n-1} (-1)^{n-k} \binom{n-1}{k} \frac{(k-1)^{[i-1]}}{N^{[i+1]}} \lambda_{N-k}.$$

Multiplying with  $N!$ , (52) is thus reformulated as

$$\sum_{i=1}^n a_i^N (N-i-1)_{[n-i]} (A_{\mu,i}^n + A_{\lambda,i}^n) = 0, \quad (58)$$

where

$$A_{\mu,i}^n := \sum_{k=i}^n (-1)^{n-k} \binom{n-1}{k-1} (k-1)_{[i-1]} \mu_{N-k}, \quad (59)$$

$$A_{\lambda,i}^n := \sum_{k=i}^{n-1} (-1)^{n-k} \binom{n-1}{k} (k-1)_{[i-1]} \lambda_{N-k}. \quad (60)$$

It remains to evaluate the  $A_{\mu,i}^n$  and the  $A_{\lambda,i}^n$  for  $1 \leq i \leq n$ . First, we note that

$$\binom{n-1}{k-1} (k-1)_{[i-1]} = \frac{(n-1)!}{(n-i)!} \binom{n-i}{k-i}$$



for  $i \leq k \leq n$  and apply this to the right-hand side of (59). This results in

$$A_{\mu,i}^n = \frac{(n-1)!}{(n-i)!} \sum_{k=i}^n (-1)^{n-k} \binom{n-i}{k-i} \mu_{N-k} = \frac{(n-1)!}{(n-i)!} \sum_{k=0}^{n-i} (-1)^k \binom{n-i}{k} \mu_{N-n+k},$$

where the sum corresponds to the  $(n-i)$ th difference quotient of the mapping

$$\mu : \{0, \dots, N\} \rightarrow \mathbb{R}_{\geq 0}, \quad k \mapsto \mu_k = -\frac{k^2}{N} + k(1 + u\nu_1)$$

taken at  $N-n$ . Since  $\mu$  is a quadratic function, we conclude that  $A_{\mu,i}^n = 0$  for all  $1 \leq i \leq n-3$ . In particular, in the second difference quotient (i.e.  $i = n-2$ ) the linear terms cancel each other and  $A_{\mu,n-2}^n$  simplifies to

$$\begin{aligned} A_{\mu,n-2}^n &= \frac{(n-1)!}{2} [\mu_{N-n} - 2\mu_{N-n+1} + \mu_{N-n+2}] \\ &= \frac{(n-1)!}{2} [-(N-n)^2 + 2(N-n+1)^2 - (N-n+2)^2] = -\frac{(n-1)!}{N}. \end{aligned}$$

For the remaining quantities  $A_{\mu,n-1}^n$  and  $A_{\mu,n}^n$  we have

$$A_{\mu,n-1}^n = (n-1)! (\mu_{N-n} - \mu_{N-n+1}) = (n-1)! \left( \frac{1}{N} (N-2n+1) - u\nu_1 \right)$$

and

$$A_{\mu,n}^n = (n-1)! \mu_{N-n} = (n-1)! (N-n) \left( \frac{n}{N} + u\nu_1 \right).$$

We now calculate the  $A_{\lambda,i}^n$ . Since

$$\binom{n-1}{k} (k-1)_{[i-1]} = \frac{1}{k} \frac{(n-1)!}{(n-1-i)!} \binom{n-1-i}{k-i}$$

for  $i \leq k \leq n-1$ , we obtain that

$$\begin{aligned} A_{\lambda,i}^n &= \frac{(n-1)!}{(n-1-i)!} \sum_{k=i}^{n-1} (-1)^{n-k} \binom{n-1-i}{k-i} \frac{\lambda_{N-k}}{k} \\ &= \frac{(n-1)!}{(n-1-i)!} \sum_{k=0}^{n-1-i} (-1)^{k+1} \binom{n-1-i}{k} \frac{\lambda_{N-(n-1-k)}}{n-1-k}, \end{aligned}$$

where the sum now coincides with the  $(n-1-i)$ th difference quotient of the affine function

$$\lambda : \{0, \dots, N-1\} \rightarrow \mathbb{R}_{\geq 0}, \quad k \mapsto \frac{\lambda_k}{N-k} = \frac{k}{N}(1+s) + u\nu_0$$

taken at  $N-(n-1)$ . Consequently,  $A_{\lambda,i}^n = 0$  for all  $1 \leq i \leq n-3$ , and in  $A_{\lambda,n-2}^n$  (more precisely in the first difference quotient of  $\lambda$  at  $N-(n-1)$ ) the constant terms cancel

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each other. Thus,

$$\begin{aligned} A_{\lambda, n-2}^n &= (n-1)! \left[ -\frac{\lambda_{N-(n-1)}}{n-1} + \frac{\lambda_{N-(n-2)}}{n-2} \right] \\ &= (n-1)! \frac{1+s}{N} [N - (n-2) - (N - (n-1))] = (n-1)! \frac{1+s}{N} \end{aligned}$$

and so

$$A_{\lambda, n-1}^n = -(n-1)! \frac{\lambda_{N-(n-1)}}{n-1} = -(n-1)! \left[ \frac{N - (n-1)}{N} (1+s) + u\nu_0 \right].$$

Combining (58) with the results for  $A_{\mu, i}^n$  and  $A_{\lambda, i}^n$  yields the assertion (56).  $\square$

It will not come as a surprise now that the discrete recursions of the  $a_n^N$  obtained in Thm. 3 lead to Fearnhead's coefficients  $a_n$  in the limit  $N \rightarrow \infty$ . According to Sec. 3.3,  $\psi_{k_N}^N$  converges to  $\psi(x)$  for any given sequence  $(k_N)_{N \in \mathbb{N}}$  with  $0 < k_N < N$  and  $\lim_{N \rightarrow \infty} k_N/N = x$ . Comparing (53) with (25), we obtain

$$\lim_{N \rightarrow \infty} a_n^N = a_n \quad (61)$$

for all  $n \geq 1$ . The recursions (27) of Fearnhead's coefficients then follow directly from the recursions in Thm. 3 in the limit  $N \rightarrow \infty$ .

### 3.6 Derivatives of $\psi$

In this section, we turn to the derivatives  $\psi^{(n)}(1)$ ,  $n \geq 1$ , which are also closely linked to Fearnhead's coefficients (via (28) and (29)). More precisely, we will derive a system of equations for the  $\psi^{(n)}(1)$ . To this end, we deal with the difference quotients of the mapping  $\psi^N$  first. Again we omit the index  $N$  in the following lemma.

**Lemma 4.**  $\psi = (\psi_k)_{0 \leq k \leq N}$  satisfies the following relations:

$$-2\psi_{N-1} + \psi_{N-2} = \frac{2(N-2)}{\mu_{N-2}} \frac{1}{N} \left[ \left( s + \frac{N u \nu_0}{N-1} \right) \psi_{N-1} - \frac{s}{N} \right] \quad (62)$$

and, for  $n \geq 3$ :

$$\sum_{i=1}^n (-1)^i \binom{n}{i} \psi_{N-i} = \frac{n(N-n)}{\mu_{N-n}} \frac{1}{N} \sum_{i=1}^{n-1} (-1)^{i-1} \binom{n-1}{i} \left( s + \frac{N u \nu_0}{N-i} \right) \psi_{N-i}. \quad (63)$$

*Proof.* At first we note that

$$\frac{\lambda_{N-k+1}}{(k-1)(N-k+1)} = \frac{1}{N} + \frac{1}{N} \left( s + \frac{N u \nu_0}{N-k+1} \right) \quad (64)$$

and

$$\frac{\mu_{N-k}}{N-k} + \frac{n-k}{N} = \frac{\mu_{N-n}}{N-n} \quad (65)$$

for  $1 \leq k \leq n$ . Then, we apply this to the left-hand side of (62) after insertion of Lemma 2 for  $k = 2$ :

$$\begin{aligned} -2\psi_{N-1} + \psi_{N-2} &= \frac{2(N-2)}{\mu_{N-2}} \left[ -\frac{\mu_{N-2}}{N-2} \psi_{N-1} + \frac{\mu_{N-1}}{N-1} \psi_{N-1} + \frac{1}{N} \psi_{N-1} \right. \\ &\quad \left. + \frac{1}{N} \left( s + \frac{Nuv_0}{N-1} \right) \psi_{N-1} - \frac{s}{N^2} \right] \\ &= \frac{2(N-2)}{\mu_{N-2}} \frac{1}{N} \left[ \left( s + \frac{Nuv_0}{N-1} \right) \psi_{N-1} - \frac{s}{N} \right], \end{aligned}$$

which is the right-hand side of (62).

To prove (63), we express  $\psi_{N-n}$  via Lemma 2 for  $k = n$ . Due to (64), the left-hand side of (63) becomes

$$\begin{aligned} \sum_{i=1}^n (-1)^i \binom{n}{i} \psi_{N-i} &= \frac{n(N-n)}{\mu_{N-n}} \left[ \sum_{i=1}^{n-1} (-1)^i \binom{n}{i} \frac{\mu_{N-n}}{n(N-n)} \psi_{N-i} + (-1)^n \frac{\mu_{N-1}}{N-1} \psi_{N-1} \right. \\ &\quad \left. + (-1)^n \frac{1}{N} \psi_{N-n+1} + (-1)^n \frac{1}{N} \left( s + \frac{Nuv_0}{N-n+1} \right) \psi_{N-n+1} \right. \\ &\quad \left. - (-1)^n \frac{(n-1)s}{N^2} \right]. \end{aligned}$$

Then we use (65) to split the first sum and obtain

$$\begin{aligned} &= \frac{n(N-n)}{\mu_{N-n}} \left[ \frac{1}{N} \sum_{i=1}^{n-2} (-1)^i \binom{n-1}{i} \psi_{N-i} + \sum_{i=1}^{n-1} (-1)^i \frac{(n-1)!}{i!(n-i)!} \frac{\mu_{N-i}}{N-i} \psi_{N-i} \right. \\ &\quad \left. + (-1)^n \frac{\mu_{N-1}}{N-1} \psi_{N-1} + (-1)^n \frac{1}{N} \left( s + \frac{Nuv_0}{N-n+1} \right) \psi_{N-n+1} - (-1)^n \frac{(n-1)s}{N^2} \right]. \end{aligned}$$

Finally, we substitute each factor  $\mu_{N-i} \psi_{N-i} / (N-i)$  in the second sum according to Lemma 2. Rearranging (again with the help of (64)) results in

$$\begin{aligned} &= \frac{n(N-n)}{\mu_{N-n}} \left[ \frac{1}{N} \sum_{i=1}^{n-2} (-1)^i \binom{n-1}{i} \psi_{N-i} + \sum_{i=1}^n (-1)^i \binom{n-1}{i-1} \frac{\mu_{N-1}}{N-1} \psi_{N-1} \right. \\ &\quad \left. + \frac{1}{N} \sum_{i=2}^{n-1} (-1)^i \binom{n-1}{i-1} \psi_{N-i+1} + \frac{1}{N} \sum_{i=2}^n (-1)^i \binom{n-1}{i-1} \left( s + \frac{Nuv_0}{N-i+1} \right) \psi_{N-i+1} \right. \\ &\quad \left. - \frac{s}{N^2} \sum_{i=2}^n (-1)^i \binom{n-1}{i-1} (i-1) \right]. \end{aligned}$$

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The first and third sum cancel each other and the second and fifth sum disappear as a consequence of the binomial theorem. The remaining fourth sum equals the right-hand side of (63), which proves the assertion.  $\square$

**Theorem 4.** *The  $\psi^{(n)}(1)$ ,  $n \geq 1$ , satisfy the following relations:*

$$\psi^{(2)}(1) = \frac{2}{2 + \theta\nu_1} [ -(\sigma + \theta\nu_0) \psi'(1) - \sigma ], \quad (66)$$

$$\psi^{(3)}(1) = \frac{3}{3 + \theta\nu_1} [ -(\sigma + \theta\nu_0) \psi^{(2)}(1) + 2\theta\nu_0 \psi'(1) ] \quad (67)$$

and, for  $n \geq 4$ :

$$\psi^{(n)}(1) = \frac{n}{n + \theta\nu_1} [ -(n-1 + \sigma + \theta) \psi^{(n-1)}(1) - (n-1)\sigma \psi^{(n-2)}(1) ]. \quad (68)$$

*Proof.* Since

$$\frac{n(N-n)}{\mu_{N-n}^N} \frac{1}{N} = \frac{n}{n + Nu_N \nu_1} \quad (69)$$

for  $n \geq 1$ , and  $\psi^{(2)}(1) = \lim_{N \rightarrow \infty} N^2 [ -2\psi_{N-1}^N + \psi_{N-2}^N ]$  and  $-\psi'(1) = \lim_{N \rightarrow \infty} N\psi_{N-1}^N$ , (62) of Lemma 4 immediately yields the right-hand side of (66).

To prove (67) and (68), we express the difference quotients of  $\psi^N$  after insertion of (63) as

$$\begin{aligned} \sum_{i=1}^n (-1)^i \binom{n}{i} \psi_{N-i}^N &= \frac{n(N-n)}{\mu_{N-n}^N} \frac{1}{N} \left[ \left( s_N + \frac{Nu_N \nu_0}{N - (n-1)} \right) \sum_{i=1}^{n-1} (-1)^{i-1} \binom{n-1}{i} \psi_{N-i}^N \right. \\ &\quad \left. + \sum_{i=1}^{n-2} (-1)^{i-1} \binom{n-1}{i} \left( \frac{Nu_N \nu_0}{N-i} - \frac{Nu_N \nu_0}{N - (n-1)} \right) \psi_{N-i}^N \right] \\ &= \frac{n(N-n)}{\mu_{N-n}^N} \frac{1}{N} \left[ \left( s_N + \frac{Nu_N \nu_0}{N - (n-1)} \right) \sum_{i=1}^{n-1} (-1)^{i-1} \binom{n-1}{i} \psi_{N-i}^N \right. \\ &\quad \left. - \frac{n-1}{N - (n-1)} \sum_{i=1}^{n-2} (-1)^{i-1} \binom{n-2}{i} \frac{Nu_N \nu_0}{N-i} \psi_{N-i}^N \right]. \quad (70) \end{aligned}$$

For  $n = 3$ , the last equality directly yields

$$\begin{aligned} \psi^{(3)}(1) &= \lim_{N \rightarrow \infty} N^3 [ -3\psi_{N-1}^N + 3\psi_{N-2}^N - \psi_{N-3}^N ] \\ &= \lim_{N \rightarrow \infty} N^3 \frac{3(N-3)}{\mu_{N-3}^N} \frac{1}{N} \left[ \left( s_N + \frac{Nu_N \nu_0}{N-2} \right) (2\psi_{N-1}^N - \psi_{N-2}^N) \right. \\ &\quad \left. - \frac{2Nu_N \nu_0}{(N-1)(N-2)} \psi_{N-1}^N \right], \end{aligned}$$

which corresponds to the right-hand side of (67). For  $n \geq 4$ , we rearrange (70) to obtain

$$\begin{aligned}
 \sum_{i=1}^n (-1)^i \binom{n}{i} \psi_{N-i}^N &= \frac{n(N-n)}{\mu_{N-n}^N} \frac{1}{N} \left[ \left( s_N + \frac{Nu_N \nu_0}{N-(n-1)} \right) \sum_{i=1}^{n-1} (-1)^{i-1} \binom{n-1}{i} \psi_{N-i}^N \right. \\
 &\quad - \frac{n-1}{N-(n-1)} \sum_{i=1}^{n-2} (-1)^{i-1} \binom{n-2}{i} \left( s_N + \frac{Nu_N \nu_0}{N-i} \right) \psi_{N-i}^N \\
 &\quad \left. - \frac{(n-1)s_N}{N-(n-1)} \sum_{i=1}^{n-2} (-1)^i \binom{n-2}{i} \psi_{N-i}^N \right] \\
 &= \frac{n(N-n)}{\mu_{N-n}^N} \frac{1}{N} \left[ \left( s_N + \frac{Nu_N \nu_0}{N-(n-1)} \right) \sum_{i=1}^{n-1} (-1)^{i-1} \binom{n-1}{i} \psi_{N-i}^N \right. \\
 &\quad - \frac{n-1}{N-(n-1)} \frac{N\mu_{N-n+1}^N}{(n-1)(N-n+1)} \sum_{i=1}^{n-1} (-1)^i \binom{n-1}{i} \psi_{N-i}^N \\
 &\quad \left. - \frac{(n-1)s_N}{N-(n-1)} \sum_{i=1}^{n-2} (-1)^i \binom{n-2}{i} \psi_{N-i}^N \right],
 \end{aligned}$$

where in the last equality the second sum is replaced by (63) of Lemma 4 (with  $n$  replaced by  $n-1$ ). Together with (69), it follows that

$$\begin{aligned}
 \psi^{(n)}(1) &= \lim_{N \rightarrow \infty} N^n \sum_{i=1}^n (-1)^i \binom{n}{i} \psi_{N-i}^N \\
 &= \frac{n}{n + \theta \nu_1} \left[ -(\sigma + \theta \nu_0 + n - 1 + \theta \nu_1) \psi^{(n-1)}(1) - (n-1)\sigma \psi^{(n-2)}(1) \right],
 \end{aligned}$$

which proves assertion (68) of Thm. 4.  $\square$

Thm. 4 provides a further characterisation of  $\psi^{(n)}(1)$ .

**Proposition 1.** *Let  $n \geq 3$ . Then*

$$\psi^{(n)}(1) = \frac{n}{n + \theta \nu_1} \left[ -\sigma \psi^{(n-1)}(1) + \theta \nu_0 \sum_{i=1}^{n-1} (-1)^{n-i} \frac{(n-1)!}{i!} \psi^{(i)}(1) \right]. \quad (71)$$

*Proof.* We prove the proposition by induction over  $n$ . According to (67) of Thm. 4, the assertion is true for  $n = 3$ . For some  $n \geq 4$ , we insert the induction hypothesis for  $n-1$  into the right-hand side of (68):

$$\begin{aligned}
 \psi^{(n)}(1) &= \frac{n}{n + \theta \nu_1} \left[ (n-1)\sigma \psi^{(n-2)}(1) - (n-1)\theta \nu_0 \sum_{i=1}^{n-2} (-1)^{n-1-i} \frac{(n-2)!}{i!} \psi^{(i)}(1) \right. \\
 &\quad \left. - (\sigma + \theta \nu_0) \psi^{(n-1)}(1) - (n-1)\sigma \psi^{(n-2)}(1) \right],
 \end{aligned}$$

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where the first and the last summand cancel each other. Rearranging of the remaining summands results in

$$\psi^{(n)}(1) = \frac{n}{n + \theta\nu_1} \left[ -\sigma\psi^{(n-1)}(1) + \theta\nu_0 \sum_{i=1}^{n-1} (-1)^{n-i} \frac{(n-1)!}{i!} \psi^{(i)}(1) \right],$$

which is the right-hand side of (71). □

Note that one may derive the recursions (27) of Fearnhead's coefficients alternatively from the derivatives  $\psi^{(n)}(1)$ , see [29] and [23, Sec. 3.3.5]. The idea is to reformulate the representation of Fearnhead's  $a_n$  in (29) as  $a_1 = -\psi'(1)$  and

$$a_n = \frac{(-1)^n}{n!} \sum_{i=0}^n (-1)^{n-i} \frac{n!}{i!} \psi^{(i)}(1) = \frac{(-1)^n}{n!} \psi^{(n)}(1) + a_{n-1}$$

for  $n \geq 2$ , and then use Thm. 4 and Prop. 1, respectively.

## 4 A coupling approach

In this chapter, we work with couplings to give alternative interpretations of the quantities  $\psi_k^N = h_k^N - k/N$  and  $h_k^N - h_{k-1}^N$ . First, we start with a short introduction to coupling theory (Sec. 4.1). Then, we consider applications in the Moran model without mutation (Sec. 4.2) and go on with an extension to the case with mutation (Sec. 4.3). Since we restrict ourselves to populations of finite size  $N$ , we omit the upper and lower index  $N$  throughout this chapter to make our notation less complicated.

### 4.1 The coupling method

Coupling methods are applied in many areas of probability theory and are a useful tool in the field of interacting particle systems. A short overview can be found in [4, Ch. 13], an elaborate survey in [34]. [33] introduces the use of coupling methods in interacting particle systems. We begin with a formal definition (cf. [4, Def. 13.1], [34, Ch. 1.1], [44]).

**Definition 1.** (a) Let  $P$  and  $P'$  be two probability measures on a measurable space  $(\Omega, \mathcal{F})$ . By a coupling of  $P$  and  $P'$  we mean any probability measure  $\hat{P}$  on  $(\Omega, \mathcal{F}) \times (\Omega, \mathcal{F})$  such that  $P$  and  $P'$  are the marginal distributions of  $\hat{P}$ , i.e.

$$\hat{P}(A \times \Omega) = P(A) \quad \text{and} \quad \hat{P}(\Omega \times A) = P'(A)$$

for all  $A \in \mathcal{F}$ .

(b) Let  $(Y_t)_{t \geq 0}$  and  $(Y'_t)_{t \geq 0}$  be two Markov processes on a measurable space  $(\Omega, \mathcal{F})$ . A process  $(\hat{Y}_t, \hat{Y}'_t)_{t \geq 0}$  on  $(\Omega, \mathcal{F}) \times (\Omega, \mathcal{F})$  is called a coupling of  $(Y_t)_{t \geq 0}$  and  $(Y'_t)_{t \geq 0}$  if

$$\hat{Y} \stackrel{D}{=} Y \quad \text{and} \quad \hat{Y}' \stackrel{D}{=} Y'.$$

Now we describe how to couple certain classes of interacting particle systems, whereas we restrict ourselves to the state space  $\{0, 1\}^{\mathcal{S}}$ ,  $\mathcal{S}$  countable (see [20, Ch. 6, 10] and [34, Ch. 5.3] for surveys). An *interacting particle system* is a (continuous-time) Markov process  $(\Gamma_t)_{t \geq 0}$  on  $\{0, 1\}^{\mathcal{S}}$  with  $\Gamma_t = (\Gamma_t(j))_{j \in \mathcal{S}}$ . We interpret  $\mathcal{S}$  as a set of positions. On each of these positions sits a particle, the one at position  $j$ ,  $j \in \mathcal{S}$ , is of type  $\Gamma_t(j) \in \{0, 1\}$  at time  $t$ .

We assume that  $(\Gamma_t)_{t \geq 0}$  is characterised by the so-called *flip rate function*  $c : \mathcal{S} \times \{0, 1\}^{\mathcal{S}} \rightarrow \mathbb{R}_{\geq 0}$  [34, Ch. 5.3].  $c$  is independent of time and defined as follows:

$$\begin{aligned} \mathbb{P}(\Gamma_{t+h}(j) \neq \gamma(j) \mid \Gamma_t = \gamma) &= c(j, \gamma)h + o(h), \\ \mathbb{P}(\Gamma_{t+h}(j) \neq \gamma(j), \Gamma_{t+h}(i) \neq \gamma(i) \mid \Gamma_t = \gamma) &= o(h), \quad i, j \in \mathcal{S}, i \neq j, \end{aligned}$$

where  $o(h)$  denotes the Landau symbol ‘little o’ as  $h \rightarrow 0$ .  $c(j, \gamma)$  indicates the rate at which a particle at position  $j$  changes its type (from 0 to 1 or from 1 to 0), given the configuration of the interacting particle system is  $\gamma = (\gamma(j))_{j \in \mathcal{S}} \in \{0, 1\}^{\mathcal{S}}$ . The rate for

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a so-called flip at position  $j$  depends on the current configuration. Almost surely we do not observe more than one flip at the same time. Classical examples are the contact process and the voter model (cf. [20, Ch. 6, 10]).

Note that we omit more general interacting particle systems that are characterised by the so-called *speed* (or *rate*) *function*  $c : \{0, 1\}^{\mathcal{S}} \times \{0, 1\}^{\mathcal{S}} \rightarrow \mathbb{R}_{\geq 0}$ , where  $c(\gamma, \gamma')$ ,  $\gamma, \gamma' \in \{0, 1\}^{\mathcal{S}}$ , is the rate at which  $(\Gamma_t)_{t \geq 0}$ , when in configuration  $\gamma$ , jumps to  $\gamma'$ . As an example see the exclusion model in [20, Ch. 10].

The next definition introduces the *Vasershtein* or *basic coupling* of interacting particle systems that are characterised by flip rate functions (cf. [34, Ch. 5.3]).

**Definition 2.** Let  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  be interacting particle systems on  $\{0, 1\}^{\mathcal{S}}$  with flip rate functions  $c$  and  $c'$ , respectively. Let  $c_{\min}(j, \gamma, \gamma') := \min(c(j, \gamma), c'(j, \gamma'))$ , and define the operator  $C_j : \{0, 1\}^{\mathcal{S}} \rightarrow \{0, 1\}^{\mathcal{S}}$ ,  $\gamma \mapsto C_j(\gamma)$ , as follows:

$$C_j(\gamma)(i) = \begin{cases} 1 - \gamma(i), & \text{if } i = j, \\ \gamma(i), & \text{if } i \neq j, \end{cases}$$

i.e., the operator  $C_j$  flips the type of a configuration at position  $j$ . Let  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  be a Markov process on  $\{0, 1\}^{\mathcal{S}} \times \{0, 1\}^{\mathcal{S}}$  with the following transition rates:

(i) If  $\gamma(j) \neq \gamma'(j)$ :

$$(\gamma, \gamma') \rightarrow (C_j(\gamma), \gamma') \quad \text{at rate } c(j, \gamma),$$

$$(\gamma, \gamma') \rightarrow (\gamma, C_j(\gamma')) \quad \text{at rate } c'(j, \gamma').$$

(ii) If  $\gamma(j) = \gamma'(j)$ :

$$(\gamma, \gamma') \rightarrow (C_j(\gamma), C_j(\gamma')) \quad \text{at rate } c_{\min}(j, \gamma, \gamma'), \quad (72)$$

$$(\gamma, \gamma') \rightarrow (C_j(\gamma), \gamma') \quad \text{at rate } c(j, \gamma) - c_{\min}(j, \gamma, \gamma'), \quad (73)$$

$$(\gamma, \gamma') \rightarrow (\gamma, C_j(\gamma')) \quad \text{at rate } c'(j, \gamma') - c_{\min}(j, \gamma, \gamma'). \quad (74)$$

Then, the Markov process  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  is a coupling of  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$ .

We understand  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  as an interacting particle system, where now

$$\{(j, k) \mid j \in \mathcal{S}, k \in \{1, 2\}\}$$

is the set of positions. The individual that occupies position  $(j, 1)$  is understood to be of type  $\hat{\Gamma}_t(j)$  at time  $t$ , the individual that occupies position  $(j, 2)$  of type  $\hat{\Gamma}'_t(j)$ . With regard to our applications in the next sections, note the following observation (cf. [34, Ch. 5.3]):

**Remark 5.** Assume that  $\gamma \geq \gamma'$ , i.e.  $\gamma(j) \geq \gamma'(j)$  for all  $j \in \mathcal{S}$ , and

$$(a) \quad c(j, \gamma) \geq c'(j, \gamma') = c_{\min}(j, \gamma, \gamma'), \text{ if } \gamma(j) = \gamma'(j) = 0,$$

$$(b) \quad c'(j, \gamma') \geq c(j, \gamma) = c_{\min}(j, \gamma, \gamma'), \text{ if } \gamma(j) = \gamma'(j) = 1.$$

As a consequence, in the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  we only observe transitions (72) and (73) in case (a) and transitions (72) and (74) in case (b). In general,  $\hat{\Gamma}_t \geq \hat{\Gamma}'_t$  for all  $t \geq 0$  if  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ .



## 4.2 Coupling in a Moran model with selection

The Moran model, as described in Sec. 2.1, is an interacting particle system. Here, the set of positions is  $\mathcal{S} = \{1, \dots, N\}$ , and particles can be considered individuals. Each of the  $N$  positions is occupied by an individual, which is of type 0 or type 1. As already known, these individuals mutate and reproduce (and the offspring, which inherits the parent's type, replaces an individual at a randomly chosen position). Reproductions that lead to a replacement of an individual of a different type and 'non-empty' mutations, i.e. mutations that change the type, are visible as flips in the configuration.

For the moment, we forgo the effect of mutation, thus  $u = 0$ . For a given configuration  $\gamma \in \{0, 1\}^{\mathcal{S}}$ , define  $\|\gamma\|$  as the number of type-0 individuals in configuration  $\gamma$ . Let  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  be interacting particle systems on  $\{0, 1\}^{\mathcal{S}}$ , which are defined by flip rate functions

$$c(j, \gamma) = \begin{cases} \frac{N - \|\gamma\|}{N}, & \text{if } \gamma(j) = 0, \\ \frac{\|\gamma\|}{N}, & \text{if } \gamma(j) = 1 \end{cases} \quad (75)$$

and

$$c'(j, \gamma) = \begin{cases} \frac{N - \|\gamma\|}{N}, & \text{if } \gamma(j) = 0, \\ \frac{\|\gamma\|}{N}(1 + s), & \text{if } \gamma(j) = 1, \end{cases} \quad (76)$$

respectively. Obviously,  $(\Gamma_t)_{t \geq 0}$  describes the evolution of  $N$  individuals according to a Moran model with solely neutral reproductions,  $(\Gamma'_t)_{t \geq 0}$  according to a Moran model with both neutral and selective reproductions. Since there is no mutation, one of the two types almost surely becomes fixed in the population. Therefore, the absorbing states of both  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  are  $\mathbf{1}$  and  $\mathbf{0}$ , where  $\mathbf{1} = (1, \dots, 1)$  and  $\mathbf{0} = (0, \dots, 0)$  are vectors of length  $N$ .

Let  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  be the Vasershtein coupling (cf. Def. 2) of  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  under the assumption that  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ , i.e.  $\|\hat{\Gamma}_0\| \leq \|\hat{\Gamma}'_0\|$ . For configurations  $\gamma, \gamma' \in \{0, 1\}^{\mathcal{S}}$  with  $\gamma \geq \gamma'$ , we have  $c_{\min}(j, \gamma, \gamma') = c'(j, \gamma')$  for  $j \in \mathcal{S}$  with  $\gamma(j) = \gamma'(j) = 0$  and  $c_{\min}(j, \gamma, \gamma') = c(j, \gamma)$  for  $j \in \mathcal{S}$  with  $\gamma(j) = \gamma'(j) = 1$ . According to Remark 5,  $\hat{\Gamma}_t \geq \hat{\Gamma}'_t$  for all  $t \geq 0$ . Graphically, the Vasershtein coupling can be understood as follows:

- (R1) Individuals at positions  $(j, 1)$  and  $(j, 2)$  with different types *reproduce independently*. By this we will mean that the individuals at  $(j, 1)$  and  $(j, 2)$  reproduce at times of independent Poisson processes with rate 1 and rate  $1 + s$ , respectively. The offspring replace, independently of each other, individuals at randomly chosen positions in  $\mathcal{S} \times \{1\}$  and  $\mathcal{S} \times \{2\}$ , respectively.
- (R2) Individuals at positions  $(j, 1)$  and  $(j, 2)$  that are of the same type *reproduce together* at rate 1. By this we will mean that the individuals at  $(j, 1)$  and  $(j, 2)$  reproduce at times of the same rate one Poisson process and replace individuals at positions  $(k, 1)$  and  $(k, 2)$ , respectively, where  $k$  is a random draw from  $\mathcal{S}$ .

#### 4 A coupling approach

(R3) If both individuals at positions  $(j, 1)$  and  $(j, 2)$  are of type 0, the individual that sits at  $(j, 2)$  *reproduces additionally* at rate  $s$ . This will mean that only the individual at  $(j, 2)$  reproduces at times of a rate  $s$  Poisson process and the offspring replaces an individual at a randomly chosen position in  $\mathcal{S} \times \{2\}$ , whereas the individual at  $(j, 1)$  does not reproduce.

We observe three absorbing states:

$$\mathcal{A}_{0,0} := (\mathbf{0}, \mathbf{0}), \quad \mathcal{A}_{1,0} := (\mathbf{1}, \mathbf{0}) \text{ and } \mathcal{A}_{1,1} := (\mathbf{1}, \mathbf{1}). \quad (77)$$

Now we can build a bridge to the extra absorption probability  $\psi_k = h_k - k/N$  (cf. Sec. 2.3.2). According to the method of coupling (cf. Def. 1),  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  have the same absorption probabilities as  $(\hat{\Gamma}_t)_{t \geq 0}$  and  $(\hat{\Gamma}'_t)_{t \geq 0}$ , respectively, which are independent of the other component. Let  $\ell \leq k$ . Then

$$\begin{aligned} \frac{\ell}{N} &= \mathbb{P}(\Gamma \text{ absorbs in } \mathbf{0} \mid \|\Gamma_0\| = \ell) \\ &= \mathbb{P}(\hat{\Gamma} \text{ absorbs in } \mathbf{0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) \end{aligned} \quad (78)$$

and

$$\begin{aligned} h_k &= \mathbb{P}(\Gamma' \text{ absorbs in } \mathbf{0} \mid \|\Gamma'_0\| = k) \\ &= \mathbb{P}(\hat{\Gamma}' \text{ absorbs in } \mathbf{0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \cup \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0). \end{aligned} \quad (79)$$

For the absorption probabilities in the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  we obtain

$$\begin{aligned} \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= \frac{\ell}{N}, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= h_k - \frac{\ell}{N}, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= 1 - \frac{\ell}{N} - \left( h_k - \frac{\ell}{N} \right) \\ &= 1 - h_k. \end{aligned} \quad (80)$$

For  $k = \ell$ , (80) yields

$$\mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 = \hat{\Gamma}'_0) = h_k - \frac{k}{N} = \psi_k.$$

This way, we obtain an alternative possibility to characterise the quantity  $\psi_k$  as the absorption probability in  $\mathcal{A}_{1,0}$  for the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$ , given  $\|\hat{\Gamma}_0\| = \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 =$

$\hat{\Gamma}'_0$ . We thus formalise the characterisation of  $\psi_k$  as ‘extra’ absorption probability in Sec. 2.3.2.

Additionally, our reasoning allows for a further use of the method of ‘first-step analysis’ (cf. Lemma 1). Since a detailed application of the method is given in Sec. 3.1, we just go briefly through it at this point. Again we consider the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  with  $\|\hat{\Gamma}_0\| = \|\hat{\Gamma}'_0\| = k$  and  $\hat{\Gamma}_0 = \hat{\Gamma}'_0$ . According to the rates given in Def. 2 and (75) and (76), we only observe transition (72) for  $j \in \mathcal{S}$  with  $\hat{\Gamma}_0(j) = \hat{\Gamma}'_0(j) = 0$ ; there are  $k$  of these positions. In contrast, for  $j \in \mathcal{S}$  with  $\hat{\Gamma}_0(j) = \hat{\Gamma}'_0(j) = 1$ , transitions (72) and (74) are possible; there are  $N - k$  of these positions. Thus

$$\begin{aligned} \left( \frac{k(N-k)}{N} (1+s) + \frac{k(N-k)}{N} \right) \psi_k &= \frac{k(N-k)}{N} \psi_{k-1} + \frac{k(N-k)}{N} \psi_{k+1} \\ &\quad + \frac{k(N-k)}{N} s \left( h_{k+1} - \frac{k}{N} \right). \end{aligned}$$

Due to (1), this difference equation corresponds to (34) for  $u = 0$ . The coupling between two Moran models with and without selection, respectively, permits to formalise our heuristic first-step interpretation of (34) in Sec. 2.3.2.

At the end, we briefly consider the coupling between two copies of the Moran model with selection. Therefore, let  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  be two interacting particle systems that are characterised by the same flip rate function  $c'$  as in (76). We consider the corresponding Vasershtein coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  (cf. Def. 2) with initial configuration  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ . Again, for  $\gamma, \gamma' \in \{0, 1\}^{\mathcal{S}}$  with  $\gamma \geq \gamma'$ , we obtain that  $c_{\min}(j, \gamma, \gamma') = c'(j, \gamma')$  for  $j$  with  $\gamma(j) = \gamma'(j) = 0$  and  $c_{\min}(j, \gamma, \gamma') = c(j, \gamma)$  for  $j$  with  $\gamma(j) = \gamma'(j) = 1$ . Therefore,  $\hat{\Gamma}_t \geq \hat{\Gamma}'_t$  for all  $t \geq 0$  (cf. Remark 5). We interpret the coupling in the following sense:

(R1) Individuals at positions  $(j, 1)$  and  $(j, 2)$  with different types reproduce independently.

(R2) If both individuals at positions  $(j, 1)$  and  $(j, 2)$  are of type 1, they reproduce together at rate 1, if they are both of type 0, they reproduce together at rate  $1 + s$ .

$(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  absorbs almost surely in  $\mathcal{A}_{0,0}$ ,  $\mathcal{A}_{1,0}$  or  $\mathcal{A}_{1,1}$  (cf. (77)). Mimicking our calculations in (78) and (79) by replacing  $\ell/N$  by  $h_\ell$ ,  $\ell \leq k$ , we obtain the following absorption probabilities:

$$\begin{aligned} \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= h_\ell, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= h_k - h_\ell, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= 1 - h_k. \end{aligned} \tag{81}$$

For  $k \geq 1$ ,  $\ell = k - 1$ , (81) becomes

$$\mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = k - 1, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) = h_k - h_{k-1},$$

which gives an interpretation of the difference  $h_k - h_{k-1}$  as an absorption probability in the Vasershtein coupling. A detailed analysis of this quantity will follow in Ch. 5.

### 4.3 Coupling in a Moran model with selection and mutation

This section deals with the coupling of Moran models with mutation. We consider interacting particle systems  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  on  $\{0, 1\}^{\mathcal{S}}$  with flip rate functions

$$c(j, \gamma) = \begin{cases} \frac{N - \|\gamma\|}{N} + u\nu_1, & \text{if } \gamma(j) = 0, \\ \frac{\|\gamma\|}{N} + u\nu_0, & \text{if } \gamma(j) = 1 \end{cases}$$

and

$$c'(j, \gamma) = \begin{cases} \frac{N - \|\gamma\|}{N} + u\nu_1, & \text{if } \gamma(j) = 0, \\ \frac{\|\gamma\|}{N}(1 + s) + u\nu_0, & \text{if } \gamma(j) = 1 \end{cases} \quad (82)$$

and  $u > 0$ . Here,  $(\Gamma_t)_{t \geq 0}$  corresponds to a Moran model with mutation but without selection,  $(\Gamma'_t)_{t \geq 0}$  to a Moran model with mutation and selection. The offspring of the type-0 or the type-1 individuals will almost surely become fixed in the population (cf. Sec. 2.3). The closed sets (cf. Sec. 3.1) of  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  are

$$\begin{aligned} \mathcal{A}_0 &:= \{\gamma \in \{0, 1\}^{\mathcal{S}} \mid \text{individual at position } j \text{ is descendant of type-0 individual } \forall j \in \mathcal{S}\}, \\ \mathcal{A}_1 &:= \{\gamma \in \{0, 1\}^{\mathcal{S}} \mid \text{individual at position } j \text{ is descendant of type-1 individual } \forall j \in \mathcal{S}\}. \end{aligned}$$

We define the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  between these two interacting particle systems in the natural way:

- (R1) Individuals at positions  $(j, 1)$  and  $(j, 2)$  that are of different types reproduce independently.
- (R2) Individuals at  $(j, 1)$  and  $(j, 2)$  that are of the same type reproduce together at rate 1.
- (R3) If both individuals at  $(j, 1)$  and  $(j, 2)$  are of type 0, the individual that occupies  $(j, 2)$  reproduces additionally at rate  $s$ .
- (M) Individuals at  $(j, 1)$  and  $(j, 2)$  *mutate together* to type  $i$  at rate  $u\nu_i$ ,  $i \in S$ . By this we will mean that mutations to type  $i$  occur on  $j \times \{1, 2\}$  at times of the same rate  $u\nu_i$  Poisson process  $\mathcal{P}_{ji}$ , independently of the individual's types on  $j \times \{1, 2\}$  at time  $t^-$ ,  $t \in \mathcal{P}_{ji}$ . Empty mutations are included.

Although  $\hat{\Gamma}_t \geq \hat{\Gamma}'_t$  for all  $t \geq 0$ , if  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ , the coupling almost surely absorbs in one of the four sets  $\mathcal{A}_{i,j} := \mathcal{A}_i \times \mathcal{A}_j$ ,  $i, j \in \{0, 1\}$ . To calculate the absorption probabilities, we

### 4.3 Coupling in a Moran model with selection and mutation

use the method of coupling (cf. Def. 1) in the same way as in Sec. 4.2:

$$\begin{aligned}\frac{\ell}{N} &= \mathbb{P}(\Gamma \text{ absorbs in } \mathcal{A}_0 \mid \|\Gamma_0\| = \ell) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \cup \mathcal{A}_{0,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k),\end{aligned}$$

$$\begin{aligned}\frac{N - \ell}{N} &= \mathbb{P}(\Gamma \text{ absorbs in } \mathcal{A}_1 \mid \|\Gamma_0\| = \ell) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \cup \mathcal{A}_{1,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k),\end{aligned}$$

$$\begin{aligned}h_k &= \mathbb{P}(\Gamma' \text{ absorbs in } \mathcal{A}_0 \mid \|\Gamma'_0\| = k) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \cup \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k),\end{aligned}$$

$$\begin{aligned}1 - h_k &= \mathbb{P}(\Gamma' \text{ absorbs in } \mathcal{A}_1 \mid \|\Gamma'_0\| = k) \\ &= \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,1} \cup \mathcal{A}_{1,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k).\end{aligned}$$

These calculations yield

$$\begin{aligned}&\mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k) \\ &- \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k) = h_k - \frac{\ell}{N}.\end{aligned}$$

For  $k = \ell$ , we therefore obtain a formal interpretation of the quantity  $\psi_k = h_k - k/N$  (in comparison to the one of Sec. 2.3.2), namely, as the difference of two absorption probabilities in the coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$ .

We close this section with a brief consideration of a coupling between two versions of the Moran model with selection and mutation. Let  $(\Gamma_t)_{t \geq 0}$  and  $(\Gamma'_t)_{t \geq 0}$  be two interacting particle systems on  $\{0, 1\}^S$  with flip rate function  $c'$  as in (82). Here, a slightly different coupling  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  is useful, which complies with  $\hat{\Gamma}_t \geq \hat{\Gamma}'_t$  for all  $t \geq 0$ , if  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ .

- (R1) Individuals at positions  $(j, 1)$  and  $(j, 2)$  with different types reproduce together at rate 1.
- (R2) If both individuals at  $(j, 1)$  and  $(j, 2)$  are of type 1, they reproduce together at rate 1, if they are both of type 0, they reproduce together at rate  $1 + s$ .
- (R3) If both individuals at  $(j, 1)$  and  $(j, 2)$  are of different types, the type-0 individual at  $(j, 2)$  reproduces additionally at rate  $s$ .
- (M) Individuals at positions  $(j, 1)$  and  $(j, 2)$  mutate together to type  $i$  at rate  $u\nu_i$ ,  $i \in S$ .

#### 4 A coupling approach

According to this law for coupling, and under the assumption that  $\hat{\Gamma}_0 \geq \hat{\Gamma}'_0$ , a descendant of a type-0 individual at position  $(j, 1)$  implies that the individual at  $(j, 2)$  is also a descendant of a type-0 individual. Therefore,

$$\mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,1} \mid \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) = 0.$$

Note that this is not the case if we allow for independent reproductions at positions  $(j, 1)$  and  $(j, 2)$  with different types. Likewise, this property fails in our previous coupling between Moran models with and without selection due to the selective advantage of type-0 individuals in  $\mathcal{S} \times \{2\}$ . In particular, this is independent of the reproduction mechanism of individuals at positions  $(j, 1)$  and  $(j, 2)$  with different types.

Analogously to our previous calculations, the absorption probabilities of  $(\hat{\Gamma}_t, \hat{\Gamma}'_t)_{t \geq 0}$  are

$$\begin{aligned} \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{0,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= h_\ell, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,0} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= h_k - h_\ell, \\ \mathbb{P}((\hat{\Gamma}, \hat{\Gamma}') \text{ absorbs in } \mathcal{A}_{1,1} \mid \|\hat{\Gamma}_0\| = \ell, \|\hat{\Gamma}'_0\| = k, \hat{\Gamma}_0 \geq \hat{\Gamma}'_0) &= 1 - h_k. \end{aligned} \quad (83)$$

For  $k \geq 1$ ,  $\ell = k - 1$ , (83) includes a new point of view concerning the differences  $h_k - h_{k-1}$ .

## 5 Fixation in the Moran model with selection

In this chapter, we reanalyse the process of fixation in the Moran model with selection, i.e. we focus on a vanishing mutation rate ( $u_N = 0 = \theta$ ). As in Sec. 2.1,  $Z_t^N$  is the number of individuals of type 0 at time  $t$ . Remember that  $Z^N$  is a birth-death process with birth rates  $\lambda_i^N$  and death rates  $\mu_i^N$  when in state  $i$ , where now

$$\lambda_i^N = (1 + s_N)i \frac{N-i}{N} \quad \text{and} \quad \mu_i^N = (N-i) \frac{i}{N}. \quad (84)$$

The absorbing states are 0 and  $N$ , thus, one of the two types, 0 or 1, will almost surely become fixed in the population in finite time. Let  $T_k^N := \min\{t \geq 0 \mid Z_t^N = k\}$ ,  $0 \leq k \leq N$ , be the first hitting time of  $Z^N$ . The *fixation probability* of type 0 given that there are initially  $k$  type-0 individuals is well-known to be (cf. [11, Thm. 6.1])

$$h_k^N = \mathbb{P}(T_N^N < T_0^N \mid Z_0^N = k) = \frac{\sum_{i=N-k}^{N-1} (1 + s_N)^i}{\sum_{i=0}^{N-1} (1 + s_N)^i}. \quad (85)$$

The corresponding fixation probability of type-0 individuals in the diffusion  $(X_t)_{t \geq 0}$  follows a classical result of Kimura [26], which is based on type frequencies in the diffusion limit (see also [16, Ch. 5.3] and [25, Ch. 15]): Define the first-passage time  $T_x := \inf\{t \geq 0 \mid X_t = x\}$  for  $x \in [0, 1]$  and let  $\sigma > 0$ . Then

$$h(x) = \mathbb{P}(T_1 < T_0 \mid X_0 = x) = \frac{1 - \exp(-\sigma x)}{1 - \exp(-\sigma)}. \quad (86)$$

The standard derivations of (85) and (86) are based on the type frequency representation, without a connection to the particle picture. They may also be deduced directly from Thm. 1 and (20) by inserting  $u_N = 0$  and  $\theta = 0$ , respectively (see also Remark 4). In Sec. 4.2, we already established a term-by-term interpretation of the right-hand side of (85), i.e. of  $h_i^N - h_{i-1}^N$ , with the help of a coupling between two copies of the Moran model with selection.

To obtain further insights, we first characterise fixation probabilities by way of a *reflection principle* (Sec. 5.1). Sec. 5.2 introduces an alternative particle system, which we call the *labelled Moran model*. Here, the main idea is that each of the  $N$  individuals is characterised by a different reproductive behaviour, which we indicate by a label. With the help of a coupling argument, we obtain the probability that an individual of a particular label becomes fixed (Sec. 5.2.1). This way, we also identify reproduction events that affect the fixation probability of a given label; they are termed *defining events* (Sec. 5.2.2). The number of *selective defining events* turns out as the pivotal quantity characterising fixation probabilities. In Sec. 5.3, we pass to the diffusion limit. Sec. 5.4 establishes a connection to Fearnhead's coefficients, both in the discrete setting and in the diffusion limit, and points out the particle representation behind (53) and (25). We continue with a simulation algorithm that generates the label that becomes fixed together with the targets of the selective defining events (Sec. 5.5). Most of the results of this chapter are published in [30].

## 5.1 Reflection principle

In [11, Sec. 6.1.1] equation (85) is proven in two ways, namely, using a first-step approach and a martingale argument, respectively. Both approaches rely on the process  $(Z_t^N)_{t \geq 0}$ , without reference to an underlying particle representation. As a warm up exercise, we complement this by an approach based on the particle picture, which we call the *reflection principle*.

**Definition 3.** *Let a graphical realisation of the Moran model be given, with  $Z_0^N = k$ ,  $1 \leq k \leq N - 1$ , and fixation of type 0. Now interchange the types (i.e., replace all 0 individuals by 1 individuals and vice versa), without otherwise changing the graphical realisation. This results in a graphical realisation of the Moran model with  $Z_0^N = N - k$  in which type 1 becomes fixed, and is called the *reflected realisation*.*

Put differently, in the case  $Z_0^N = k$ ,  $1 \leq k \leq N - 1$ , reflection transforms a realisation in which the (offspring of the)  $k$  fit individuals become fixed (altogether, this happens with probability  $h_k^N$ ) into a realisation in which the (offspring of the) unfit individuals become fixed (which happens with probability  $1 - h_{N-k}^N$ ), and vice versa. This operation does not change the graphical structure, but the weights of the realisations are different since a different weight is attached to some of the arrows. To make the situation tractable, we will work with what we will call the *reduced Moran model*: Starting from the original Moran model, we remove those selective arrows that appear between individuals that are both of the fit type. Obviously, this does not affect the process  $Z^N$  (in particular, it does not change the fixation probabilities), but it changes the graphical representation. See Fig. 10 for an illustration of the reflection principle in the reduced Moran model.

Let now  $\Omega^N$  be the set of graphical realisations of the reduced Moran model for  $t \in [0, T]$ , where  $T := \min\{T_0^N, T_N^N\}$ . Let  $P_k^N$  be the probability measure on  $\Omega^N$ , given  $Z_0^N = k$ ,  $1 \leq k \leq N - 1$ . For a realisation  $\omega \in \Omega^N$  with  $Z_0^N = k$ ,  $1 \leq k \leq N - 1$ , in which 0 becomes fixed (cf. Fig. 10, left), we define  $\bar{\omega} \in \Omega^N$  as the corresponding reflected realisation (cf. Fig. 10, right).

Our strategy will now be to deduce the fixation probabilities by comparing the weights of  $\omega$  and  $\bar{\omega}$ . To assess the relative weights of  $P_k^N(\omega)d\omega$  and  $P_{N-k}^N(\bar{\omega})d\bar{\omega}$ , note first that, since the fit individuals become fixed in  $\omega$ , all  $N - k$  type-1 individuals have to be replaced by fit ones, i.e. by arrows that appear at rate  $(1 + s_N)/N$ . In  $\bar{\omega}$ , the corresponding arrows point from unfit to fit individuals and thus occur only at rate  $1/N$ , cf. the dashed arrows in Fig. 10. Second, we have to take into account so-called *new descendants* defined as follows:

**Definition 4.** *A descendant of a type- $i$  individual,  $i \in S$ , that originates by replacing an individual of a different type ( $j \neq i$ ) is termed a *new descendant of type  $i$* .*

**Remark 6.** *The total number of new descendants of type  $i$  is almost surely finite.*

Every new descendant of a type-1 individual goes back to an arrow that occurs at rate  $1/N$ . If the fit individuals go to fixation (as in  $\omega$ ), all new descendants of type 1



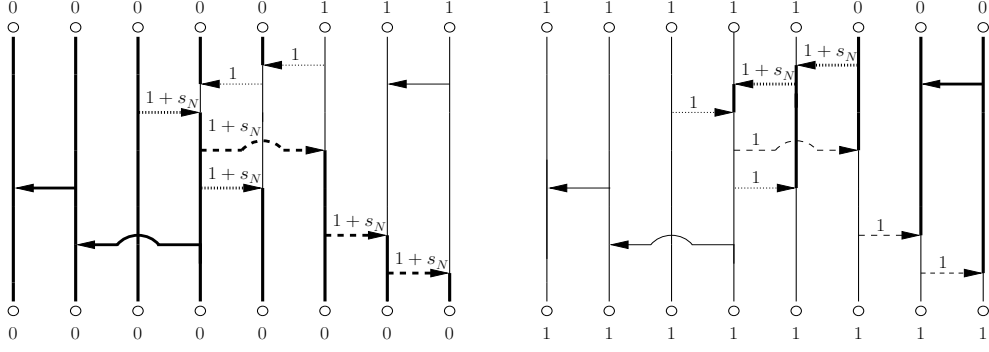


Figure 10: Reflection principle in the reduced Moran model:  $N = 8$ ,  $k = 5$ , realisations  $\omega$  (left) and  $\bar{\omega}$  (right). Bold lines represent type-0 individuals, thin ones type-1 individuals; likewise, arrows emanating from type-0 (type-1) individuals are bold (thin). Interchange of types transforms the realisation on the left into the realisation on the right and vice versa, such that the respective other type becomes fixed. Arrows that are marked by 1 respectively  $1 + s_N$  appear at rate  $1/N$  respectively  $(1 + s_N)/N$ ; unmarked arrows appear at rate  $1/N$ . Dashed arrows represent the elimination of individuals (except new descendants) of the type that eventually gets lost in the population. The births of their new descendants and their replacements are represented by dotted arrows. Altogether,  $P_3^8(\bar{\omega})d\omega = (1 + s_N)^{-3}(1 + s_N)^2(1 + s_N)^{-2}P_5^8(\omega)d\omega$ .

must eventually be replaced by arrows that emanate from fit individuals and therefore occur at rate  $(1 + s_N)/N$ . In  $\bar{\omega}$  this situation corresponds to new descendants of type 0 that originate from arrows at rate  $(1 + s_N)/N$  and are eventually eliminated by arrows that emanate from type-1 individuals. See the thin and bold dotted arrows in Fig. 10, which always appear in pairs.

Let now  $D_1^N(\omega)$  be the number of new descendants of type 1 in  $\omega$ . Then  $D_1^N(\omega) < \infty$  almost surely and we obtain for the measure of  $\bar{\omega}$

$$\begin{aligned} P_{N-k}^N(\bar{\omega})d\omega &= (1 + s_N)^{-(N-k)}(1 + s_N)^{D_1^N(\omega)}(1 + s_N)^{-D_1^N(\omega)}P_k^N(\omega)d\omega \\ &= (1 + s_N)^{-(N-k)}P_k^N(\omega)d\omega. \end{aligned}$$

Note that the effects of creating new descendants and their replacement cancel each other, so that the relative weights of  $P_{N-k}^N(\bar{\omega})d\omega$  and  $P_k^N(\omega)d\omega$  do not depend on  $\omega$ . Since reflection provides a one-to-one correspondence between realisations with fixation of type 0 and of type 1, respectively, we obtain the system of equations

$$1 - h_{N-k}^N = (1 + s_N)^{-(N-k)}h_k^N, \quad 1 \leq k \leq N - 1, \quad (87)$$

which is supplemented by  $h_0^N = 0$  and  $h_N^N = 1$ , and which is solved by (85).

## 5.2 Labelled Moran model

In this section, we introduce a new particle model, which we call the *labelled Moran model*, and which has the same empirical type distribution as the original Moran model with selection, provided the initial conditions are chosen appropriately. As before, we consider a population of fixed size  $N$  in continuous time, but now every individual is assigned a label  $i \in \{1, \dots, N\}$  with different reproductive behaviour to be specified below. (Be aware that *label 1* (in the labelled Moran model) and *type 1* (in the original Moran model) are not identical: As it will become clear below, the reproductive behaviour of label 1 is completely different from the one of type 1.) As in the original Moran model, birth events are represented by arrows; they lead to a single offspring, which inherits the parent's label, and replaces an individual as explained below. Again we distinguish between neutral (at rate  $1/N$ ) and selective (at rate  $s_N/N$ ) events. Neutral arrows appear as before, at rate  $1/N$  per ordered pair of lines, irrespective of their labels. But we only allow for selective arrows emanating from a label  $i$  and pointing to a label  $j$ ,  $j > i$  (at rate  $s_N/N$  per ordered pair of lines with such labels). Equivalently, we may take together both types of arrows, such that an arrow points from a label  $i$  to a label  $j \leq i$  at rate  $1/N$  and to label  $j > i$  at rate  $(1 + s_N)/N$ . We will make use of both points of view. The idea is to imply a graduation in fitness in the sense that label 1 is the fittest label, label 2 the second-fittest label and so forth, whereas label  $N$  finally is the less-fittest label. That is, each label behaves as 'unfit' towards lower labels and as 'fit' towards higher labels.

We now fix an initial population that contains all  $N$  labels so that initially position  $i$  in the graphical representation is occupied by label  $i$ ,  $1 \leq i \leq N$ . Note that this spatial ordering does not influence the reproductive behaviour of the labels.

An example is given in Fig. 11, where neutral arrows are marked by 1 and selective arrows by  $s_N$ .

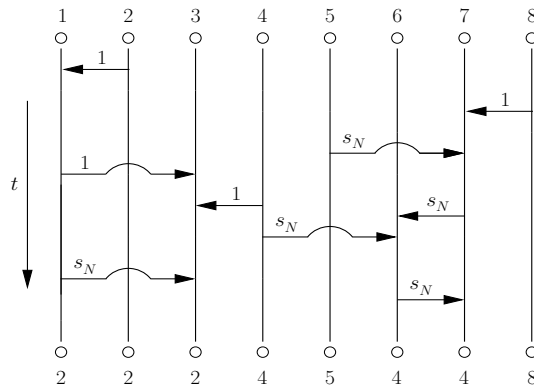


Figure 11: A realisation of the labelled Moran model for  $N = 8$ . The labels are indicated for the initial population (top) and a later one (bottom).

### 5.2.1 Ancestors and fixation probabilities

Since there is no mutation in the labelled Moran model, one of the  $N$  labels will eventually take over in the population, i.e. one label will become fixed. We denote this label by  $I^N$  and term it the ancestor. Its distribution is given in Thm. 5.

**Theorem 5.**  $I^N$  is distributed according to

$$\eta_i^N := \mathbb{P}(I^N = i) = (1 + s_N)^{N-i} \eta_N^N = h_i^N - h_{i-1}^N, \quad 1 \leq i \leq N, \quad (88)$$

with

$$\eta_N^N := \mathbb{P}(I^N = N) = \frac{1}{\sum_{i=0}^{N-1} (1 + s_N)^i} = 1 - h_{N-1}^N. \quad (89)$$

We will give two proofs of Thm. 5. The first provides an intuitive explanation and is based on the type frequencies. In the second proof, we will use an alternative approach in the spirit of the reflection principle of Sec. 5.1, which provides more insight into the particle representation. This proof is somewhat more complicated, but it permits us to classify reproduction events into those that have an effect on the fixation probability of a given label and those that do not; this will become important later on. In analogy with Def. 4 we understand *new descendants of labels in  $\mathcal{N}$* ,  $\mathcal{N} \subseteq \{1, \dots, N\}$ , as descendants of individuals with labels in  $\mathcal{N}$  that originate by replacing an individual of a label in the complement of  $\mathcal{N}$ .

*First proof of Thm. 5.* For a given  $i$ , let  $\bar{Z}_t^{N,i}$  be the number of individuals with labels in  $\{1, \dots, i\}$  at time  $t$ . Like  $Z^N$ , the process  $\bar{Z}^{N,i} = (\bar{Z}_t^{N,i})_{t \geq 0}$  is a birth-death process with both rates  $\lambda_j^N$  and  $\mu_j^N$  of (84). This is because every individual with label in  $\{1, \dots, i\}$  sends arrows into the set with labels in  $\{i+1, \dots, N\}$  at rate  $(1 + s_N)/N$ ; in the opposite direction, the rate is  $1/N$  per pair of individuals; and arrows within the label classes do not matter. Since  $\bar{Z}_0^{N,i} = i$ , the processes  $\bar{Z}^{N,i}$  and  $Z^N$  thus have the same law provided  $Z_0^N = i$ . As a consequence,  $\mathbb{P}(I^N \leq i) = \sum_{j=1}^i \eta_j^N = h_i^N$ ,  $1 \leq i \leq N$ , which, together with (85), immediately yields the assertions of Thm. 5.  $\square$

*Second proof of Thm. 5.* This proof aims at a direct calculation of  $\eta_i^N$ ,  $1 \leq i \leq N-1$ , as a function of  $\eta_N^N$ . Let a realisation of the labelled Moran model be given in which label  $N$  becomes fixed (this happens with probability  $\eta_N^N$ , still to be calculated). The basic idea now is to move every arrow by way of a cyclic permutation of the arrows' positions, while keeping the initial ordering of the labels. More precisely, in the graphical representation we move every arrow  $i$  positions to the right (or, equivalently,  $N-i$  positions to the left). That is, we shift an arrow that appears at time  $t$  with base at position  $k$  and tip at position  $\ell$ , such that it becomes an arrow that emanates from position  $(k+i) \bmod N$  and points to position  $(\ell+i) \bmod N$ , again at time  $t$ . We thus obtain what we will call the *permuted realisation of order  $i$* . In this realisation, label  $i$  is fixed, as illustrated in Fig. 12 for a labelled Moran model of size  $N = 8$ .

## 5 Fixation in the Moran model with selection

Throughout, we keep the original meaning of the labels: Between every ordered pair of lines with labels  $(i, j)$ , arrows appear at rate  $(1 + s_N)/N$  if  $j > i$ , and at rate  $1/N$  otherwise. Since, in the permuted realisation, we change the *position* of each arrow, it now may affect a different pair of *labels*, which may change the arrow's rate. As a result, the permuted realisation has a different weight than the original one; this will now be used to calculate  $\eta_i^N$ . (Mathematically, the following may be seen as a *coupling argument*.)

So, let  $\Omega^N$  be the set of realisations of the labelled Moran model for  $t \in [0, T]$ , where  $T$  now is the time at which one of the labels is fixed. Let  $P^N$  be the probability measure on  $\Omega^N$ . For a realisation  $\omega_N \in \Omega^N$  in which label  $N$  becomes fixed (cf. Fig. 12, left), we define  $\omega_i \in \Omega^N$  as the corresponding permuted realisation of order  $i$  (cf. Fig. 12, right). We will now calculate  $\eta_i^N$  by assessing the weight of the measure of  $\omega_i$  relative to that of  $\omega_N$ . Below we run briefly through the cases to analyse the change of weight on the various types of arrows.

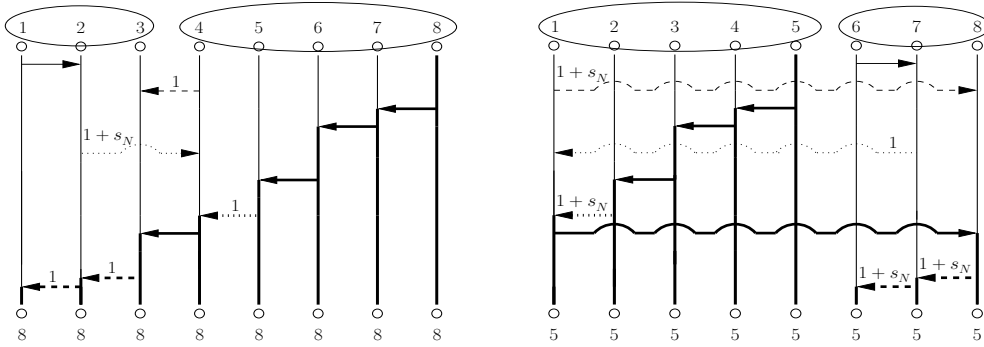


Figure 12: Cyclic permutation. Realisations  $\omega_8$  (left) and  $\omega_5$  (right) in a labelled Moran model of size  $N = 8$ . A shift of every arrow of  $i = 5$  positions to the right transforms  $\omega_8$ , in which label  $N = 8$  becomes fixed, into  $\omega_5$  (with fixation of label  $i = 5$ ). Descendants of the label that becomes fixed are marked bold in both cases. The label sets  $\{1, \dots, N - i\}$  and  $\{N - i + 1, \dots, N\}$  (left), and  $\{1, \dots, i\}$  and  $\{i + 1, \dots, N\}$  (right), respectively, are encircled at the top. Arrows within these sets (solid) appear at the same rates in  $\omega_8$  and  $\omega_5$ . Arrows between these label sets are dotted or dashed, their rates differ between  $\omega_8$  and  $\omega_5$ . Arrows that are marked by 1 respectively  $1 + s_N$  appear at rate  $1/N$  respectively  $(1 + s_N)/N$ . There is exactly one new descendant of the labels in  $\{1, \dots, N - i\}$  (left) and  $\{i + 1, \dots, N\}$  (right), respectively. It originates from the respective first dotted arrow, and is replaced via the second dotted arrow. Replacements of the labels  $1, \dots, N - i$  (left) and of  $i + 1, \dots, N$  (right), respectively, (except their new descendants) are represented by dashed arrows. Altogether,  $P^8(\omega_5)d\omega_8 = (1 + s_N)^{-1}(1 + s_N)(1 + s_N)^3 P^8(\omega_8)d\omega_8$ .

- (a) Arrows in  $\omega_N$  that point from and to labels within the sets  $\{1, \dots, N - i\}$  or  $\{N - i + 1, \dots, N\}$ , respectively, turn into arrows within the sets  $\{i + 1, \dots, N\}$  or

$\{1, \dots, i\}$ , respectively, under the permutation. Such arrows retain their ‘direction’ (with respect to the labels) and thus appear at identical rates in  $\omega_N$  and  $\omega_i$  (cf. the solid arrows in Fig. 12).

- (b) Arrows in  $\omega_N$  that emanate from the set of labels  $\{1, \dots, N - i\}$  and point to the set of labels  $\{N - i + 1, \dots, N\}$  occur at rate  $(1 + s_N)/N$  and create new descendants of labels in  $\{1, \dots, N - i\}$ . Since label  $N$  becomes fixed, every such new descendant is eventually eliminated by an arrow at rate  $1/N$ , see also the dotted arrows in Fig. 12, left. The corresponding situation in  $\omega_i$  concerns new descendants of labels in  $\{i + 1, \dots, N\}$ , which result from neutral arrows and finally are replaced at rate  $(1 + s_N)/N$  each (cf. the dotted arrows in Fig. 12, right).
- (c) It remains to deal with the replacement of the labels  $1, \dots, N - i$  (except for their new descendants) in  $\omega_N$ . Exactly  $N - i$  neutral arrows are responsible for this, they transform into arrows at rate  $(1 + s_N)/N$  through the permutation (cf. the dashed arrows in Fig. 12).

Let now  $D_{\leq i}^N(\omega_N)$  be the number of new descendants of labels in  $\{1, \dots, i\}$  in  $\omega_N$  ( $D_{\leq i}^N(\omega_N) < \infty$  almost surely). Then, (a)-(c) yield for the measure of  $\omega_i$

$$\begin{aligned} P^N(\omega_i)d\omega_N &= (1 + s_N)^{D_{\leq i}^N(\omega_N)}(1 + s_N)^{-D_{\leq i}^N(\omega_N)}(1 + s_N)^{N-i}P^N(\omega_N)d\omega_N \\ &= (1 + s_N)^{N-i}P^N(\omega_N)d\omega_N. \end{aligned}$$

As in the reflection principle, the effects of the new descendants cancel each other and so the relative weights of  $P^N(\omega_i)d\omega_N$  and  $P^N(\omega_N)d\omega_N$  are independent of the particular choice of  $\omega_N$ . Since the cyclic permutation yields a one-to-one correspondence between realisations that lead to fixation of label  $N$  and label  $i$ , respectively, we obtain

$$\eta_i^N = (1 + s_N)^{N-i}\eta_N^N, \quad 1 \leq i \leq N. \quad (90)$$

Finally, the normalisation

$$1 = \sum_{i=1}^N \eta_i^N = \eta_N^N \sum_{i=1}^N (1 + s_N)^{N-i}$$

yields the assertion of Thm. 5. □

### 5.2.2 Defining events

We are now ready to investigate the effect of selection in more depth. The second proof of Thm. 5 allows us to identify the reproduction events that affect the distribution of  $I^N$  (i.e. that are responsible for the factor  $(1 + s_N)^{N-I^N}$  in (88)) with the dashed arrows in Fig. 12 (cf. case (c)), whereas dotted arrows appear pairwise and their effects cancel each other (cf. case (b)). It suggests itself to term these reproductions *defining events*:

**Definition 5.** A defining event is an arrow that emanates from the set of labels  $\{1, \dots, I^N\}$  and targets individuals with labels in the set  $\{I^N + 1, \dots, N\}$  that are not new descendants of labels in  $\{I^N + 1, \dots, N\}$ .

Loosely speaking, a defining event occurs every time the descendants of  $\{1, \dots, I^N\}$  ‘advance to the right’. In particular, for every  $j \in \{I^N + 1, \dots, N\}$ , the first arrow that emanates from a label in  $\{1, \dots, I^N\}$  and hits the individual at position  $j$  is a defining event. Altogether, there will be  $N - I^N$  defining events until fixation. It is important to note that they need not be reproduction events of the ancestral label  $I^N$  itself. See Fig. 13 for an illustration.

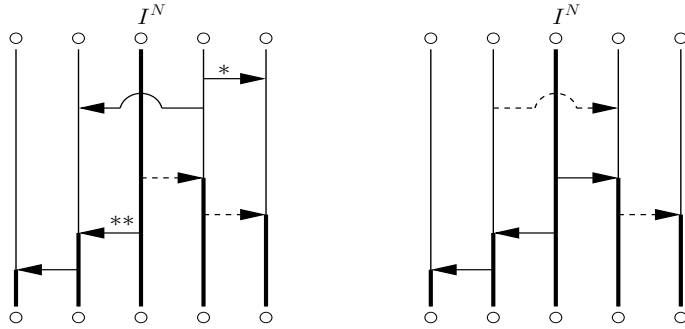


Figure 13: Defining events. Descendants of the ancestor  $I^N$  are marked bold, defining events are represented by dashed arrows. Left: Both defining events are reproduction events of  $I^N$ . The arrows that are indicated by  $*$  and  $**$  are not defining events: The first one ( $*$ ) concerns only labels within  $\{I^N + 1, \dots, N\}$ , the second one ( $**$ ) targets a new descendant of  $\{I^N + 1, \dots, N\}$ . Right: Only the second defining event is a reproduction event of  $I^N$ , the first one of a label less than  $I^N$ .

It is clear that all defining events appear at rate  $(1 + s_N)/N$ . Decomposing these arrows into neutral and selective ones reveals that each defining event is either a selective (probability  $s_N/(1 + s_N)$ ) or a neutral (probability  $1/(1 + s_N)$ ) reproduction event, independently of the other defining events and of  $I^N$ . Let  $V_i^N$ ,  $I^N + 1 \leq i \leq N$ , be the corresponding family of Bernoulli random variables that indicate whether the respective defining event is selective. Let  $Y^N$  denote the *number of selective defining events*, that is,

$$Y^N := \sum_{i=I^N+1}^N V_i^N \tag{91}$$

with the independent and identically distributed (i.i.d.) Bernoulli variables  $V_i^N$  just defined.  $Y^N$  will turn out as a pivotal quantity for everything to follow. Let us now characterise its distribution, and the dependence between  $I^N$  and  $Y^N$ . This will also provide us with an alternative representation of the fixation probabilities  $h_i^N$ .

It is clear from (91) that, given  $I^N = i$ ,  $Y^N$  follows a binomial distribution with parameters  $N - i$  and  $s_N/(1 + s_N)$ . Thus, for  $0 \leq n \leq N - i$ , we obtain (via (88))

$$\mathbb{P}(Y^N = n, I^N = i) = \binom{N-i}{n} \left(\frac{s_N}{1+s_N}\right)^n \left(\frac{1}{1+s_N}\right)^{N-i-n} \eta_i^N = \binom{N-i}{n} s_N^n \eta_N^N \quad (92)$$

and thus

$$\mathbb{P}(Y^N = n) = \sum_{i=1}^N \binom{N-i}{n} s_N^n \eta_N^N = \sum_{i=0}^{N-1} \binom{i}{n} s_N^n \eta_N^N = \binom{N}{n+1} s_N^n \eta_N^N, \quad (93)$$

where the last equality is caused by the well-known identity

$$\sum_{i=0}^k \binom{i}{\ell} = \binom{k+1}{\ell+1}, \quad \ell, k \in \mathbb{N}_0, 0 \leq \ell \leq k. \quad (94)$$

In particular,  $\mathbb{P}(Y^N = 0) = N\eta_N^N$ . Obviously,  $\mathbb{P}(Y^N = n)$ ,  $n \geq 1$ , may also be expressed recursively as

$$\mathbb{P}(Y^N = n) = s_N \frac{N-n}{n+1} \mathbb{P}(Y^N = n-1). \quad (95)$$

Furthermore, equations (92) and (93) immediately yield the conditional probability

$$\mathbb{P}(I^N = i \mid Y^N = n) = \frac{\binom{N-i}{n}}{\binom{N}{n+1}}. \quad (96)$$

The intuitive content of this important fact will become clear in Sec. 5.5. Another interesting characterisation is the following:

**Proposition 2.** *Let  $W^N := Y^N + 1$ . Then  $W^N$  follows the binomial distribution with parameters  $N$  and  $s_N/(1 + s_N)$ , conditioned to be positive.*

*Proof.* Let  $\mathcal{W}$  be a random variable distributed according to  $\text{Bin}(N, s_N/(1 + s_N))$ . Then

$$\mathbb{P}(\mathcal{W} > 0) = 1 - \frac{1}{(1+s_N)^N} = \left(1 - \frac{1}{1+s_N}\right) \sum_{i=0}^{N-1} \frac{1}{(1+s_N)^i}$$

by the geometric series. For  $n \geq 1$ , therefore,

$$\mathbb{P}(\mathcal{W} = n \mid \mathcal{W} > 0) = \frac{\binom{N}{n} s_N^n (1+s_N)^{-N}}{\frac{s_N}{1+s_N} \sum_{i=0}^{N-1} (1+s_N)^{-i}} = \frac{\binom{N}{n} s_N^{n-1}}{\sum_{i=0}^{N-1} (1+s_N)^i} = \mathbb{P}(Y^N = n-1),$$

where the last step is caused by (93). This proves the claim.  $\square$

## 5 Fixation in the Moran model with selection

Note that label  $N$  is obviously not capable of selective reproduction events and its fixation implies the absence of (selective) defining events. Its fixation probability  $\eta_N^N$  coincides with the fixation probability of any label  $i$ ,  $1 \leq i \leq N$ , in the absence of any selective defining events:  $\mathbb{P}(Y^N = 0, I^N = i) = \eta_N^N$ , cf. (92). For this reason, we term  $\eta_N^N$  the *basic fixation probability* of every label  $i$ ,  $1 \leq i \leq N$ , and express all relevant quantities in terms of  $\eta_N^N$ . Note that, for  $s_N > 0$ ,  $\eta_N^N$  is different from the *neutral fixation probability*,  $\eta_i^N = 1/N$ , that applies to every label  $i$ ,  $1 \leq i \leq N$ , in the case  $s_N = 0$ .

Decomposition according to the number of selective defining events yields a further alternative representation of the fixation probability  $h_i^N$  (cf. (85)) of the Moran model. Consider

$$\mathbb{P}(I^N \leq i \mid Y^N = n) = \frac{1}{\binom{N}{n+1}} \sum_{j=1}^i \binom{N-j}{n} = \frac{1}{\binom{N}{n+1}} \sum_{j=N-i}^{N-1} \binom{j}{n} = \frac{\binom{N}{n+1} - \binom{N-i}{n+1}}{\binom{N}{n+1}}, \quad (97)$$

where we have used (96) and (94). This leads us to the following series expansion in  $s_N$ :

$$\begin{aligned} h_i^N &= \mathbb{P}(I^N \leq i) = \sum_{n=0}^{N-1} \mathbb{P}(I^N \leq i \mid Y^N = n) \mathbb{P}(Y^N = n) \\ &= \sum_{n=0}^{N-1} \left[ \binom{N}{n+1} - \binom{N-i}{n+1} \right] s_N^n \eta_N^N, \end{aligned} \quad (98)$$

which, together with (97) and (93), also provides an alternative representation of the extra absorption probability  $\psi_i^N = h_i^N - i/N$ :

$$\begin{aligned} \psi_i^N &= \sum_{n=1}^{N-1} \mathbb{P}(I^N \leq i \mid Y^N = n) \mathbb{P}(Y^N = n) - \mathbb{P}(I^N \leq i \mid Y^N = 0) \mathbb{P}(Y^N \geq 1) \\ &= \sum_{n=1}^{N-1} \left[ \binom{N-1}{n} - \binom{N-i-1}{n} \right] \frac{N-i}{n+1} s_N^n \eta_N^N. \end{aligned} \quad (99)$$

### 5.3 Diffusion limit of the labelled Moran model

In this section we analyse the number of selective defining events in the diffusion limit. First of all we recapitulate from (92) and (88) that

$$\mathbb{P}(Y^N = n, I^N \leq i) = \frac{1}{N} \sum_{j=1}^i \binom{N-j}{n} \left( \frac{s_N}{1+s_N} \right)^n \left( \frac{1}{1+s_N} \right)^{N-j-n} N(h_j^N - h_{j-1}^N).$$

For a sequence  $(i_N)_{N \in \mathbb{N}}$  with  $i_N \in \{1, \dots, N\}$ ,  $\lim_{N \rightarrow \infty} i_N/N = x$ , and  $x \in [0, 1]$ , this yields

$$\lim_{N \rightarrow \infty} \mathbb{P}(Y^N = n, I^N/N \leq i_N/N) = \int_0^x \frac{(\sigma(1-y))^n}{n!} \exp(-\sigma(1-y)) h'(y) dy, \quad (100)$$



where we have used the convergence of the binomial to the Poisson distribution. Thus, the sequence of random variables  $(Y^N, I^N/N)_{N \in \mathbb{N}}$  converges in distribution to a pair  $(Y^\infty, I^\infty)$  of random variables with values in  $\mathbb{N}_0 \times [0, 1]$  and distribution function (100). Marginalisation with respect to the second variable implies that  $I^\infty$  has distribution function  $h$ . As a consequence,  $Y^\infty$  follows a Poisson distribution with parameter  $\sigma(1 - I^\infty)$  in the sense of a two-stage random experiment. That is, given  $I^\infty = x$ ,  $Y^\infty$  is Poisson-distributed with parameter  $\sigma(1 - x)$ . (Of course this can also be checked directly by taking the  $N \rightarrow \infty$  limit in (93).)

Let us now consider  $\mathbb{P}(Y^\infty \geq n) = \lim_{N \rightarrow \infty} \mathbb{P}(Y^N \geq n)$ . Obviously,  $\mathbb{P}(Y^\infty \geq 0) = 1$  and

$$\begin{aligned} \mathbb{P}(Y^\infty = 0) &= \lim_{N \rightarrow \infty} \mathbb{P}(Y^N = 0) = \lim_{N \rightarrow \infty} N \eta_N^N = \lim_{N \rightarrow \infty} \left[ \frac{1}{N} \sum_{i=0}^{N-1} \left( 1 + \frac{N s_N}{N} \right)^{\frac{i}{N} N} \right]^{-1} \\ &= \left[ \int_0^1 \exp(\sigma p) dp \right]^{-1} = \frac{\sigma}{\exp(\sigma) - 1}, \end{aligned} \quad (101)$$

where we have used (93) and (89). According to (95) we have the recursion

$$\mathbb{P}(Y^\infty = n) = \frac{\sigma}{n+1} \mathbb{P}(Y^\infty = n-1) \quad (102)$$

for  $n \geq 1$  and iteratively via (102) and (101)

$$\mathbb{P}(Y^\infty = n) = \frac{\sigma^n}{(n+1)!} \mathbb{P}(Y^\infty = 0) = \frac{\sigma^{n+1}}{(n+1)! (\exp(\sigma) - 1)} \quad (103)$$

for  $n \geq 0$ . With an argument analogous to that in Prop. 2, one obtains from (103) that  $W^\infty := Y^\infty + 1$  follows a Poisson distribution with parameter  $\sigma$ , conditioned to be positive, i.e.

$$\mathbb{P}(W^\infty = n) = \frac{\frac{\sigma^n}{n!} \exp(-\sigma)}{1 - \exp(-\sigma)}$$

for  $n \geq 1$ .

We now aim at expressing  $h$  in terms of a decomposition according to the values of  $Y^\infty$  (in analogy with (98)). We recapitulate equation (97) to obtain

$$\mathbb{P}(I^\infty \leq x \mid Y^\infty = n) = \lim_{N \rightarrow \infty} \mathbb{P}(I^N \leq i_N \mid Y^N = n) = 1 - \lim_{N \rightarrow \infty} \frac{\binom{N-i_N}{n+1}}{\binom{N}{n+1}} = 1 - (1-x)^{n+1} \quad (104)$$

for a sequence  $(i_N)_{N \in \mathbb{N}}$  as at the beginning of the section. Then, the equivalent to (98) is a series expansion in  $\sigma$ :

$$\begin{aligned} h(x) &= \mathbb{P}(I^\infty \leq x) = \sum_{n \geq 0} \mathbb{P}(I^\infty \leq x \mid Y^\infty = n) \mathbb{P}(Y^\infty = n) \\ &= \frac{1}{\exp(\sigma) - 1} \sum_{n \geq 1} \frac{1}{n!} (1 - (1-x)^n) \sigma^n. \end{aligned} \quad (105)$$

## 5 Fixation in the Moran model with selection

Note that this can also be derived directly from (86) by a Taylor expansion around the point  $x = 1$ . The equivalent to (99) reads (via (104) and (103))

$$\begin{aligned}\psi(x) &= \sum_{n \geq 1} \mathbb{P}(I^\infty \leq x \mid Y^\infty = n) \mathbb{P}(Y^\infty = n) - \mathbb{P}(I^\infty \leq x \mid Y^\infty = 0) \mathbb{P}(Y^\infty \geq 1) \\ &= \frac{1}{\exp(\sigma) - 1} \sum_{n \geq 2} \frac{1}{n!} (1 - x - (1 - x)^n) \sigma^n.\end{aligned}$$

For the sake of completeness, we briefly recapitulate how to derive (103)-(105) directly (i.e. without taking the  $N \rightarrow \infty$  limit) from the fact that  $Y^\infty$  is Poisson-distributed with parameter  $\sigma(1 - I^\infty)$ . For the distribution of  $Y^\infty$  we have

$$\begin{aligned}\mathbb{P}(Y^\infty = n) &= \int_0^1 \frac{\sigma^n (1 - y)^n}{n!} \exp(-\sigma(1 - y)) h'(y) dy = \frac{\sigma^{n+1}}{n!(\exp(\sigma) - 1)} \int_0^1 (1 - y)^n dy \\ &= \frac{\sigma^{n+1}}{(n + 1)!(\exp(\sigma) - 1)}.\end{aligned}$$

For the conditional probability we then obtain

$$\begin{aligned}\mathbb{P}(I^\infty \leq x \mid Y^\infty = n) &= \frac{\int_0^x \frac{\sigma^n (1 - y)^n}{n!} \exp(-\sigma(1 - y)) h'(y) dy}{\frac{\sigma^{n+1}}{(n + 1)!(\exp(\sigma) - 1)}} \\ &= (n + 1) \int_0^x (1 - y)^n dy = 1 - (1 - x)^{n+1}.\end{aligned}$$

Combining both results leads to the representation in (105).

Finally note that (105) may also be expressed as

$$h(x) = \sum_{n \geq 1} (1 - (1 - x)^n) \mathbb{P}(W^\infty = n) = \mathbb{E}(1 - (1 - x)^{W^\infty}). \quad (106)$$

Interestingly, but not unsurprisingly, this coincides with a representation given by Pokalyuk and Pfaffelhuber in their proof of Kimura's fixation probability (86) [39, Lemma 2.2]. They follow an argument of Mano [35] that establishes a connection between the ASG at stationarity and the fixation probability. A key here is the insight that the number of lines in the ASG at stationarity follows a Poisson distribution with parameter  $\sigma$ , conditioned to be positive – which coincides with the distribution of  $W^\infty$ .

### 5.4 Fearnhead's coefficients in the labelled Moran model

Our study of the labelled Moran model continues our work concerning the coefficients  $a_n^N$ ,  $1 \leq n \leq N - 1$ , (cf. Thm. 3) and  $a_n$ ,  $n \geq 1$ , (cf. (27), (30)) of the representations (53) and (25), respectively. Although the framework of Fearnhead and Taylor only handles the case with stationarity, we have proven the results of Thm. 1 and Thm. 3

and the convergence of  $h^N$  to  $h$  (cf. Remark 4) even for  $u = 0$  respectively  $\theta = 0$ . As a consequence, equations (53), (25), (27) and (30) are also valid in this case. It is the purpose of this section to arrive at a thorough understanding of representations (53) and (25). In particular, we aim at a probabilistic understanding of the coefficients  $a_n^N$  and  $a_n$ .

We begin to point out the connection to Fearnhead's coefficients in the discrete setting for  $u = 0$ . With the definition  $a_0^N := 1$ , the recursion of the  $a_n^N$  simplifies to

$$a_n^N - a_{n+1}^N = s_N \frac{N-n}{n+1} (a_{n-1}^N - a_n^N) \quad (107)$$

for  $1 \leq n \leq N-2$ . According to Thm. 3 we obtain that

$$1 - a_1^N = 1 - N\psi_{N-1}^N = N(1 - h_{N-1}^N) = N\eta_N^N = \mathbb{P}(Y^N = 0),$$

where the second-last equality goes back to (89) of Thm. 5 and the last one to (93). Hence, the  $a_n^N - a_{n+1}^N$ ,  $0 \leq n \leq N-2$ , have the same initial value (at  $n = 0$ ) and follow the same recursion (compare (107) with (95)) as the  $\mathbb{P}(Y^N = n)$ , so they agree. As a result

$$a_n^N - a_{n+1}^N = \mathbb{P}(Y^N = n)$$

for  $0 \leq n \leq N-2$ . Via normalisation, we further have

$$\mathbb{P}(Y^N = N-1) = 1 - \sum_{n=0}^{N-2} (a_n^N - a_{n+1}^N) = a_{N-1}^N$$

and thus

$$a_n^N = \mathbb{P}(Y^N \geq n) \quad (108)$$

for  $0 \leq n \leq N-1$ . In particular, we may reformulate (98) as

$$h_i^N = \sum_{n=0}^{N-1} \frac{\binom{N}{n+1} - \binom{N-i}{n+1}}{\binom{N}{n+1}} (a_n^N - a_{n+1}^N), \quad (109)$$

where  $a_N^N := 0$ .

An analogue argumentation also holds in the diffusion limit for  $\theta = 0$ . With the definition  $a_0 := 1$  and the help of (61), we obtain that

$$\mathbb{P}(Y^\infty \geq n) = \lim_{N \rightarrow \infty} \mathbb{P}(Y^N \geq n) = \lim_{N \rightarrow \infty} a_n^N = a_n \quad (110)$$

for  $n \geq 0$ . We conclude that

$$a_n - a_{n+1} = \mathbb{P}(Y^\infty = n) \quad \text{and} \quad \rho_{n+1} = \frac{a_{n+1}}{a_n} = \mathbb{P}(Y^\infty \geq n+1 \mid Y^\infty \geq n)$$

for  $n \geq 0$  and  $\rho_{n+1}$  as in (26). (Alternatively, this follows by mimicking our approach in the discrete setting.) In particular, (105) may be represented as

$$h(x) = \sum_{n \geq 0} (1 - (1-x)^{n+1}) (a_n - a_{n+1}), \quad (111)$$

which is the  $N \rightarrow \infty$  limit of (109).

For  $u = 0$  respectively  $\theta = 0$  our reasoning yields a probabilistic meaning of Fearnhead's coefficients by way of the distribution of the number of selective defining events. Additionally, a probabilistic interpretation of the structure of the fixation probability is established by decomposing according to this quantity (compare (53) and (25) with (109) and (111), respectively). We will come back to this in the next section.

### 5.5 The targets of selective defining events and construction of the ancestral line

We have, so far, been concerned with the ancestral label and with the number of selective defining events, but have not investigated the *targets* of these events yet. This will now be done. We begin with another definition, see also Fig. 14.

**Definition 6.** Let  $Y^N$  take the value  $n$ . We then denote by  $J_1^N, \dots, J_n^N$ , with  $J_1^N < \dots < J_n^N$ , the (random) positions that are hit by the  $n$  selective defining events.

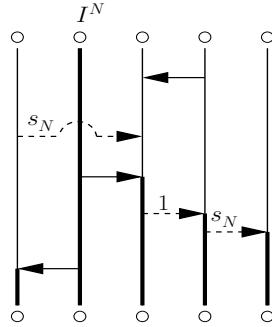


Figure 14: Targets of selective defining events. The descendants of the ancestor  $I^N$  are marked bold, dashed arrows represent defining events. Here,  $N = 5$ ,  $I^N = 2$ ,  $Y^N = 2$ ,  $J_1^N = 3$ ,  $J_2^N = 5$ . Note that the first selective defining event hits position 3, which is occupied by an individual of label 4 at that time.

In terms of the family  $V_i^N$ ,  $I^N + 1 \leq i \leq N$ , of i.i.d. Bernoulli random variables (cf. Sec. 5.2.2), the  $J_1^N, \dots, J_{Y^N}^N$  are characterised as

$$\{J_1^N, \dots, J_{Y^N}^N\} = \{i \in \{I^N + 1, \dots, N\} \mid V_i^N = 1\}.$$

(For  $Y^N = 0$  we have the empty set.) Given that  $Y^N = n$ , the  $n$ -tuple  $(J_1^N, \dots, J_n^N)$  is uniformly distributed (without replacement) on the set of positions  $\{I^N + 1, \dots, N\}$ :

$$\mathbb{P}(J_1^N = j_1, \dots, J_n^N = j_n \mid I^N = i, Y^N = n) = \frac{1}{\binom{N-i}{n}},$$

## 5.5 The targets of selective defining events and construction of the ancestral line

which implies (via (96))

$$\mathbb{P}(I^N = i, J_1^N = j_1, \dots, J_n^N = j_n \mid Y^N = n) = \frac{1}{\binom{N}{n+1}}. \quad (112)$$

Hence, the  $(n+1)$ -tuples  $(I^N, J_1^N, \dots, J_n^N)$  are sampled from  $\{1, \dots, N\}$  uniformly without replacement. Note that  $W^N$  of Prop. 2 is the size of this tuple. Note also that the tuples do not contain any further information about the appearance of arrows in the particle picture, i.e. we do not learn which label ‘sends’ the arrows.

It is instructive to formulate a simulation algorithm (or a construction rule) for these tuples.

**Algorithm 1.** *First draw the size of the tuple  $(I^N, J_1^N, \dots, J_n^N)$ , that is, a realisation  $n+1$  of  $W^N$  (according to (93) respectively Prop. 2). Then simulate  $(I^N, J_1^N, \dots, J_n^N)$  in the following way:*

**Step 0:** *Generate a random number  $U^{(0)}$  that is uniformly distributed on  $\{1, \dots, N\}$ . Define  $\mathcal{I}^{(0)} := U^{(0)}$ . If  $n > 0$ , continue with step 1, otherwise stop.*

**Step 1:** *Generate (independently of  $U^{(0)}$ ) a second random number  $U^{(1)}$  that is uniformly distributed on  $\{1, \dots, N\} \setminus \{U^{(0)}\}$ .*

(a) *If  $U^{(1)} > \mathcal{I}^{(0)}$ , define  $\mathcal{I}^{(1)} := \mathcal{I}^{(0)}$ ,  $\mathcal{J}_1^{(1)} := U^{(1)}$ .*

(b) *If  $U^{(1)} < \mathcal{I}^{(0)}$ , define  $\mathcal{I}^{(1)} := U^{(1)}$ ,  $\mathcal{J}_1^{(1)} := \mathcal{I}^{(0)}$ .*

*If  $n > 1$ , continue with step 2, otherwise stop.*

**Step k:** *Generate (independently of  $U^{(0)}, \dots, U^{(k-1)}$ ) a random number  $U^{(k)}$  that is uniformly distributed on  $\{1, \dots, N\} \setminus \{U^{(0)}, \dots, U^{(k-1)}\}$ .*

(a) *If  $U^{(k)} > \mathcal{I}^{(k-1)}$ , define  $\mathcal{I}^{(k)} := \mathcal{I}^{(k-1)}$  and assign the variables  $U^{(k)}, \mathcal{J}_1^{(k-1)}, \dots, \mathcal{J}_{k-1}^{(k-1)}$  to  $\mathcal{J}_1^{(k)}, \dots, \mathcal{J}_k^{(k)}$ , such that  $\mathcal{J}_1^{(k)} < \dots < \mathcal{J}_k^{(k)}$ .*

(b) *If  $U^{(k)} < \mathcal{I}^{(k-1)}$ , define  $\mathcal{I}^{(k)} := U^{(k)}$ ,  $\mathcal{J}_1^{(k)} := \mathcal{I}^{(k-1)}$  and  $\mathcal{J}_\ell^{(k)} := \mathcal{J}_{\ell-1}^{(k-1)}$  for  $2 \leq \ell \leq k$ .*

*If  $n > k$ , continue with step  $k+1$ , otherwise stop.*

The simulation algorithm first produces a vector  $(U^{(0)}, \dots, U^{(n)})$  uniformly distributed on the set of *unordered*  $(n+1)$ -tuples and then turns it into the vector  $(\mathcal{I}^{(n)}, \mathcal{J}_1^{(n)}, \dots, \mathcal{J}_n^{(n)})$ , which is uniformly distributed on the set of *ordered*  $(n+1)$ -tuples. The latter therefore has the same distribution as the vector of random variables  $(I^N, J_1^N, \dots, J_n^N)$ , given  $Y^N = n$  (cf. (112)). The interesting point now is that we may interpret the algorithm as a procedure for the construction of the ancestral line. It successively adds selective arrows to realisations of the labelled Moran model, such that they coincide with additional selective defining events. Obviously,  $n+1$  is the number of steps.

## 5 Fixation in the Moran model with selection

In step 0, we randomly choose one of the  $N$  labels. This represents the label that becomes fixed, i.e. the ancestor, in a particle representation with no selective defining events (cf. Fig. 15, left). (Actually, this coincides with the neutral situation,  $s_N = 0$ .) In the following steps selective arrows are added one by one, where the point to note is that each of them may or may not move the ancestor ‘to the left’, depending on whether (a) (cf. Figs. 15 and 16, middle) or (b) (cf. Figs. 15 and 16, right) applies. For instance, consider step  $k$ , that is, a realisation with  $k - 1$  selective defining events is augmented by a further one. In case (a), we add a selective defining event that does not change the label that becomes fixed (i.e. the ancestor remains the same) and that targets the newly chosen position  $U^{(k)}$ . In contrast, in case (b) the additional selective defining event emanates from position  $U^{(k)}$  and hits the ancestor of the previous step. The result is a shifting of the ancestor ‘to the left’, i.e. to position  $U^{(k)}$ . To avoid misunderstandings, we would like to emphasise that the details of the genealogies in Figs. 15 and 16 are for the purpose of illustration only; the only thing we really construct is the sequence of tuples  $(\mathcal{I}^{(n)}, \mathcal{J}_1^{(n)}, \dots, \mathcal{J}_n^{(n)})$ .

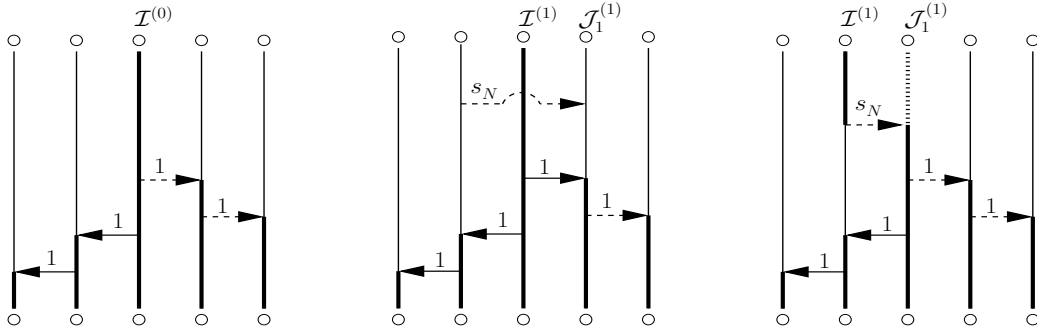


Figure 15: Steps 0 and 1 of Algorithm 1 and corresponding genealogical interpretations. Left: step 0 (no selective defining events), middle: step 1 (a), right: step 1 (b) (each with one selective defining event). Defining events are represented by dashed arrows. The ancestor ( $\mathcal{I}^{(0)}$  and  $\mathcal{I}^{(1)}$ , respectively) and the target of a selective defining event ( $\mathcal{J}_1^{(1)}$ , if present) are indicated at the top. Bold lines represent the genealogy of the entire population at the bottom. In the middle, the ancestral line is unaffected by the selective defining event. On the right, it is shifted to a lower label; the previous ancestor (which is no longer the true ancestor) is represented by the dotted line.

We now have everything at hand to provide a genealogical interpretation for the fixation probabilities  $h_i^N$  respectively  $h(x)$  (cf. (53) respectively (25)). We have seen that the tuples  $(\mathcal{I}^{(n)}, \mathcal{J}_1^{(n)}, \dots, \mathcal{J}_n^{(n)})$  are constructed such that

$$\mathbb{P}(\mathcal{I}^{(n)} = i, \mathcal{J}_1^{(n)} = j_1, \dots, \mathcal{J}_n^{(n)} = j_n) = \mathbb{P}(I^N = i, J_1^N = j_1, \dots, J_n^N = j_n \mid Y^N = n)$$

for all  $n \geq 0$  and  $j_1, \dots, j_n \in \{i + 1, \dots, N\}$ , and, via marginalisation,

$$\mathbb{P}(\mathcal{I}^{(n)} = i) = \mathbb{P}(I^N = i \mid Y^N = n).$$

## 5.5 The targets of selective defining events and construction of the ancestral line

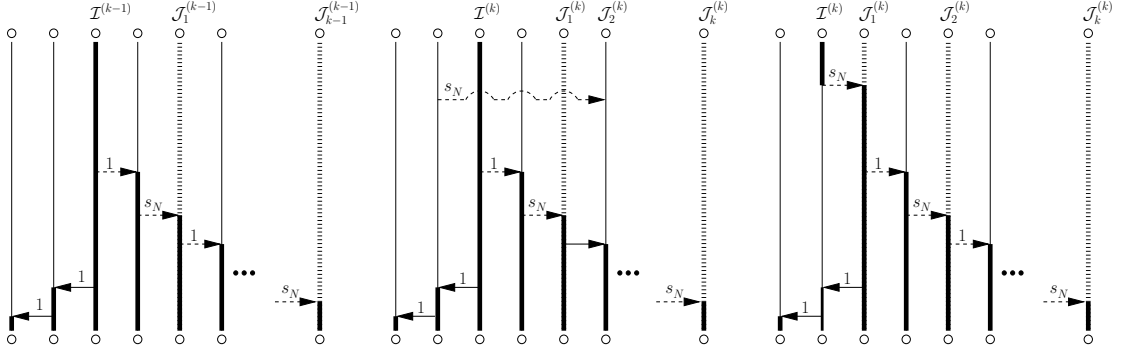


Figure 16: Step  $k$  of Algorithm 1 and corresponding genealogical interpretations. Situation after step  $k - 1$  (left) and its modification according to step  $k$  (a) (middle) and step  $k$  (b) (right). As in Fig. 15, dashed arrows represent defining events, ancestors ( $\mathcal{I}^{(k-1)}$  and  $\mathcal{I}^{(k)}$ , respectively) and targets of selective defining events ( $\mathcal{J}_1^{(k-1)}, \dots, \mathcal{J}_{k-1}^{(k-1)}$  and  $\mathcal{J}_1^{(k)}, \dots, \mathcal{J}_k^{(k)}$ , respectively) are indicated at the top. Bold lines represent the genealogy of the population at the bottom, dotted lines correspond to lines that have been ancestors in previous steps of Algorithm 1. Each selective defining event that goes back to case (b) gives rise to a new dotted line.

We reformulate the decomposition of (98) to obtain

$$\begin{aligned}
 h_i^N &= \mathbb{P}(I^N \leq i \mid Y^N = 0) \mathbb{P}(Y^N \geq 0) \\
 &+ \sum_{n=1}^{N-1} [\mathbb{P}(I^N \leq i \mid Y^N = n) - \mathbb{P}(I^N \leq i \mid Y^N = n-1)] \mathbb{P}(Y^N \geq n) \\
 &= \mathbb{P}(\mathcal{I}^{(0)} \leq i) \mathbb{P}(Y^N \geq 0) + \sum_{n=1}^{N-1} [\mathbb{P}(\mathcal{I}^{(n)} \leq i) - \mathbb{P}(\mathcal{I}^{(n-1)} \leq i)] \mathbb{P}(Y^N \geq n) \\
 &= \mathbb{P}(\mathcal{I}^{(0)} \leq i) \mathbb{P}(Y^N \geq 0) + \sum_{n=1}^{N-1} \mathbb{P}(\mathcal{I}^{(n)} \leq i, \mathcal{I}^{(n-1)} > i) \mathbb{P}(Y^N \geq n), \tag{113}
 \end{aligned}$$

where the last equality results from the fact that the ancestor's label is non-increasing in  $n$ . In (113), the fixation probability  $h_i^N$  is thus decomposed according to the first step in the algorithm in which the ancestor has a label in  $\{1, \dots, i\}$ . The probability that this event takes place in step  $n$ ,  $1 \leq n \leq N - 1$ , may, in view of the simulation algorithm, be expressed explicitly as

$$\begin{aligned}
 \mathbb{P}(\mathcal{I}^{(n)} \leq i, \mathcal{I}^{(n-1)} > i) &= \mathbb{P}(\mathcal{I}^{(n)} \leq i, \mathcal{I}^{(0)}, \mathcal{I}^{(1)}, \dots, \mathcal{I}^{(n-1)} > i) \\
 &= \mathbb{P}(U^{(n)} \leq i, U^{(0)}, U^{(1)}, \dots, U^{(n-1)} > i) \\
 &= \frac{i}{N} \prod_{j=0}^{n-1} \frac{N-i-j}{N-j-1} = \frac{i(N-i)_{[n]}}{N_{[n+1]}}, \tag{114}
 \end{aligned}$$

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where  $y_{[j]}$  is defined as in (54). Together with (108) this yields

$$h_i^N = \frac{i}{N} + i \sum_{n=1}^{N-1} a_n^N \frac{(N-i)_{[n]}}{N_{[n+1]}},$$

which is the representation (53). We thus have found a term-by-term interpretation.

In particular, (114) implies that

$$\lim_{N \rightarrow \infty} \mathbb{P}(\mathcal{I}^{(n)} \leq i_N, \mathcal{I}^{(n-1)} > i_N) = x(1-x)^n$$

for a sequence  $(i_N)_{N \in \mathbb{N}}$  with  $i_N \in \{1, \dots, N\}$  and  $\lim_{N \rightarrow \infty} i_N/N = x$ ,  $x \in [0, 1]$ . Thus, in the diffusion limit, and with  $a_n = \lim_{N \rightarrow \infty} \mathbb{P}(Y^N \geq n)$  of (110), (113) turns into

$$h(x) = \lim_{N \rightarrow \infty} h_{i_N}^N = x + \sum_{n \geq 1} x(1-x)^n a_n,$$

which is the representation (25), and which is easily checked to coincide with (105) (as obtained by the direct approach of Sec. 5.3).

**Remark 7.** *Algorithm 1 yields a further interpretation of the extra absorption probability  $\psi_i^N = h_i^N - i/N$ ,  $1 \leq i \leq N$ , as the probability that it takes at least one step until the ancestor has a label in  $\{1, \dots, i\}$ , see (113).*



## 6 Hoppe urns and coalescents with killing

Originally, the *coalescent with killing* and *Hoppe's urn model* are based on a neutral Wright-Fisher diffusion with an infinitely-many-alleles model of mutation (cf. [11, Ch. 1.3], [16, Ch. 9.5, 10.5], [21, 22]). This mutation mechanism postulates that mutations create new types, which have never been in the population before, at a fixed rate. Within this setting the coalescent with killing and Hoppe's urn serve as tools to prove the Ewens sampling formula, see e.g. [11, Ch. 1.3] and [19].

In Sec. 6.1, we carry over, and recapitulate in passing, the concepts of the coalescent with killing and Hoppe's urn to the neutral Moran model of size  $N$  with two types and mutation. We consider the usual rescaling, i.e. the mutation rates are  $Nu_N\nu_i$ ,  $i \in S$ , and assume that the population is stationary. (Note that in the cited literature the factors  $\nu_i$  are omitted.) This way, we obtain a genealogical construction, which we term *neutral coalescent with killing*. As a tentative enhancement to incorporate selection, we first introduce an extended version of the neutral coalescent with killing, which we call *non-neutral coalescent with killing*, and second the corresponding non-neutral urn model (Sec. 6.2). In both the neutral and the non-neutral case the type distributions according to our models yield the stationary distribution  $(\pi_Z^N(k))_{0 \leq k \leq N}$  (cf. (2)). As a further outlook, we modify the non-neutral urn model so that it provides us (at least for the cases  $N = 3$  and  $N = 4$ ) with an interpretation of the discrete Fearnhead coefficient  $a_1^N$  given as in Thm. 3 (Sec. 6.2.1). Note that we omit the population size index  $N$  in Sec. 6.1 and Sec. 6.2 and reintroduce it in Sec. 6.2.1.

### 6.1 The neutral case

**Neutral coalescent with killing.** Let us first point out the construction, to which we refer as the neutral coalescent with killing, and which simplifies the neutral genealogy of Sec. 2.2.1 so that it contains only non-mutant ancestors of a sample from the population. The idea is that it is not necessary to know the full genealogy if one is only interested in the types of a random sample. Instead, it suffices to trace back each sample line (by merging lines in the way described in Sec. 2.2) up to the first mutation on that line backwards in time. That is, we stop to further trace back a line and remove it from the graph as soon as the first mutation occurs on it. Then, the mutation determines the type of the line prior to the mutation event (in backward time), and all its descendants in the sample are of the same type (cf. Fig. 17).

The number of non-mutant lines in the neutral coalescent with killing of a sample of size  $n$  evolves according to a death process with rate

$$d_k = k(k-1) + kNu$$

when in state  $k$ ,  $1 \leq k \leq n$ . At jump times, say from  $k$  to  $k-1$ , two randomly chosen lines merge because of a coalescence event with probability

$$\frac{k(k-1)}{d_k} = \frac{k-1}{k-1+Nu}.$$

6 Hoppe urns and coalescents with killing

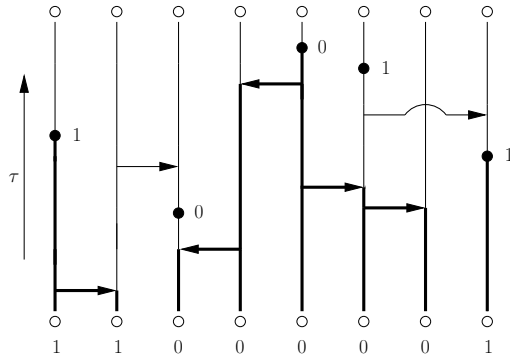


Figure 17: Neutral coalescent with killing of the entire population. Non-mutant ancestors are represented by fat lines, mutations by bullets. The type that a mutation creates is noted on the right of the corresponding bullet. Each fat line stops (possibly after a certain number of coalescence events) at the first mutation backwards in time.

With probability

$$\frac{kNu}{d_k} = \frac{Nu}{k - 1 + Nu}$$

a randomly chosen line is removed from the graph on account of a mutation event (we say that the line is killed). It then obtains type  $i$  with probability  $\nu_i$ ,  $i \in S$ , and all its descendants in the sample inherit the type  $i$ . The absorbing state is 0, whereas the last jump is a mutation event.

Note that in [16, Ch. 10.5] the jumps of the death process are termed defining events, contrary to our use of the term in Sec. 5.2.2.

**A neutral urn model.** The associated urn model due to Hoppe [22, Sec. 2] models the discrete-time jump process of the neutral coalescent with killing forward in time, and thus generates type compositions of samples (cf. [11, Thm. 1.9]). The urn contains one *mutation ball* with mass  $Nu$  and several *type- $i$  balls* with mass 1,  $i \in S$ . At each time step a ball is drawn with replacement and with probability proportional to its mass. If the mutation ball is chosen, a ball with mass 1 is added to the urn, it is a type- $i$  ball with probability  $\nu_i$ ,  $i \in S$ . If a type- $i$  ball is chosen, we add one further type- $i$  ball. At the beginning (at time 0), the urn contains only the single mutation ball and balls with mass 1 are added successively for  $n$  time steps. The number  $n$  corresponds to the sample size, type- $i$  balls represent individuals of type  $i$ ,  $i \in S$ . A draw of the mutation ball (with probability  $Nu/(k + Nu)$  at time  $k + 1$ ) represents the appearance of a new non-mutant ancestor due to a mutation in the neutral coalescent with killing forward in time, a draw of a type- $i$  ball (with probability  $k/(k + Nu)$  at time  $k + 1$ ) the reproduction event of a type- $i$  individual.

Let now  $\pi_{\mathbb{Z}}^N(k | n)$ ,  $0 \leq k \leq n$ , be the probability to obtain a sample that consists of

$k$  type-0 individuals from the neutral coalescent with killing (in a population of size  $N$ ), given the sample size is  $n$ ; our choice of notation will become clear below.  $\pi_Z^N(k | n)$  coincides with the probability that the corresponding urn contains  $k$  type-0 balls after the  $n$ 'th draw. Given the number of type- $i$  balls after the  $k$ 'th draw is  $k_i$ , we add a further type- $i$  ball at time  $k + 1$  with probability

$$\frac{k_i + Nu\nu_i}{k + Nu}$$

and thus

$$\begin{aligned} \pi_Z^N(k | n) &= \binom{n}{k} \frac{\prod_{i=0}^{k-1} (i + Nu\nu_0) \prod_{i=0}^{n-k-1} (i + Nu\nu_1)}{\prod_{i=0}^{n-1} (i + Nu)} \\ &= \binom{n}{k} \frac{(Nu\nu_0)_{(k)} (Nu\nu_1)_{(n-k)}}{(Nu)_{(n)}}. \end{aligned}$$

As a result, the type composition of a sample of size  $n$  follows a beta-binomial distribution with parameters  $Nu\nu_0$ ,  $Nu\nu_1$  and  $n$  (see also [45, Sec. 2]). If we consider the entire population instead, i.e. replace  $n$  by  $N$ , we obtain the stationary distribution  $\pi_Z^N(k)$ ,  $0 \leq k \leq N$ , given as in (3).

## 6.2 Outlook: The non-neutral case

**Non-neutral coalescent with killing.** Here, we deal with the question how to extend the neutral coalescent with killing to work also with selection. In order to approach this problem, let us tentatively consider a death process that evolves at rate

$$d_k = (1 + s)k(k - 1) + kNu$$

when in state  $k$ ,  $0 \leq k \leq n$ . It is assumed that, as before, the number of non-mutant lines in a sample of size  $n$  decreases due to mutation and reproduction events, but now *every* individual is capable of selective reproductions at rate  $Ns$ . At jump times, for instance from  $k$  to  $k - 1$ , with probability

$$\frac{kNu}{d_k} = \frac{Nu}{(1 + s)(k - 1) + Nu}$$

a randomly chosen line is killed because of a mutation event. With probability

$$\frac{k(k - 1)}{d_k} = \frac{k - 1}{(1 + s)(k - 1) + Nu}$$

two randomly chosen lines merge by reasons of a neutral reproduction, and with probability

$$\frac{sk(k - 1)}{d_k} = \frac{s(k - 1)}{(1 + s)(k - 1) + Nu}$$

by reasons of a selective reproduction. Every time a line is removed in occurrence of a mutation event, its type is determined in the usual way: With probability  $\nu_i$ ,  $i \in S$ , the mutation creates a type- $i$  individual and all its descendants are also of type  $i$ . As a consequence, the types that are involved in neutral and selective reproduction events are clarified. In our construction type-1 individuals may also give rise to selective reproduction events and we term the corresponding realisations for obvious reasons *invalid*. On the contrary, realisations in which type-1 individuals are only involved in neutral reproductions are called *valid*.

**A non-neutral urn model.** We modify the urn model of Sec. 6.1 by incorporating a selective advantage of the type-0 balls. The urn now contains one mutation ball with mass  $Nu$  and a certain number of *type- $i$  pairs*. Each type- $i$  pair consists of one *neutral type- $i$  ball* with mass 1 and one *selective type- $i$  ball* with mass  $s$ ,  $i \in S$ . As before, a ball is drawn from the urn with replacement and with probability proportional to its mass. If we draw the mutation ball, we add a type- $i$  pair to the urn, i.e. a neutral and a selective type- $i$  ball, which are both of type  $i$  with probability  $\nu_i$ ,  $i \in S$ . If we choose a type- $i$  ball instead (independently of its status as neutral or selective), we add a type- $i$  pair. Again, the urn model starts with the single mutation ball and at each time step we add two balls, one with mass 1 and one with mass  $s$ , until the urn contains  $2n + 1$  balls (i.e. we draw  $n$  times).

The urn model simulates the jump process of the non-neutral coalescent with killing forward in time. As before, the draw of the mutation ball (with probability  $Nu/((1 + s)k + Nu)$  at time  $k + 1$ ) represents the occurrence of a mutation event and thus the appearance of a new non-mutant ancestor. But now a type- $i$  pair symbolises a single type- $i$  individual. The choice of a neutral or a selective type- $i$  ball (with probabilities  $k/((1 + s)k + Nu)$  or  $sk/((1 + s)k + Nu)$  at time  $k + 1$ , respectively) corresponds to a neutral or a selective reproduction of a type- $i$  individual, respectively. If at least one selective type-1 ball is chosen within  $n$  steps, the corresponding realisation is termed *invalid*, otherwise we term it *valid*.

From now on let  $n = N$ , i.e. the non-neutral coalescent with killing starts from the entire population of unknown type composition. We then define  $\pi_{\mathbb{Z}}^N(k, \text{valid} | N)$ ,  $0 \leq k \leq N$ , as the probability to obtain a valid realisation that leads to  $k$  type-0 individuals. Again, our choice of notation will become clear below. In the associated urn model,  $\pi_{\mathbb{Z}}^N(k, \text{valid} | N)$  is the probability of a valid realisation that results in  $k$  type-0 pairs within  $N$  draws. Given the urn contains  $k_i$  type- $i$  pairs after the  $k$ 'th draw, we add a further type-0 pair at time  $k + 1$  with probability

$$\frac{(1 + s)k_0 + Nu\nu_0}{(1 + s)k + Nu}, \quad (115)$$

and a type-1 pair, indeed without creating an invalid realisation, with probability

$$\frac{k_1 + Nu\nu_1}{(1 + s)k + Nu}. \quad (116)$$

Altogether our reasoning yields

$$\begin{aligned}\pi_Z^N(k, \text{valid} \mid N) &= \binom{N}{k} \frac{\prod_{i=0}^{k-1} ((1+s)i + Nu\nu_0) \prod_{i=0}^{N-k-1} (i + Nu\nu_1)}{\prod_{i=0}^{N-1} ((1+s)i + Nu)} \\ &= \binom{N}{k} \frac{(1+s)^k \left(\frac{Nu\nu_0}{1+s}\right)_{(k)} (Nu\nu_1)_{(N-k)}}{(1+s)^N \left(\frac{Nu}{1+s}\right)_{(N)}}.\end{aligned}\tag{117}$$

Note that we allow for selective type-1 balls (and thus for selective reproductions of type-1 individuals) to guarantee independence of the ordering in which type-0 and type-1 balls are added to the urn. Finally,

$$\frac{\pi_Z^N(k, \text{valid} \mid N)}{\sum_{i=0}^N \pi_Z^N(i, \text{valid} \mid N)},$$

$0 \leq k \leq N$ , is the probability to obtain  $k$  type-0 pairs within  $N$  draws, given the realisation is valid. Surprisingly, it corresponds to the stationary distribution  $\pi_Z^N(k)$ ,  $0 \leq k \leq N$ , of the Moran model with selection and mutation (cf. (3)).

### 6.2.1 The discrete Fearnhead coefficient $a_1^N$

In this section, we turn towards the discrete Fearnhead coefficient  $a_1^N$ . Let us recall from Thm. 3 that  $a_1^N = N\psi_{N-1}^N$ . According to a slight (but useful) modification of (41) in Thm. 1, we obtain

$$a_1^N = s_N \frac{N-1}{N} \frac{\sum_{i=1}^{N-1} (N-i) \phi_i^{N, N-1}}{\sum_{i=1}^N \phi_i^{N, N}},\tag{118}$$

where

$$\phi_\ell^{N, k} := \left( \prod_{i=1}^{\ell-1} \lambda_i^N \right) \left( \prod_{i=\ell}^{k-1} \mu_i^N \right)\tag{119}$$

for  $1 \leq \ell \leq k \leq N$ . We now aim at specifying  $a_1^N$  in terms of an urn model that is closely linked to the non-neutral one. We include the index  $N$  (except for the one of the rates  $s$  and  $u$ , i.e. we write  $s$  and  $u$  instead of  $s_N$  and  $u_N$ , respectively) to avoid confusion when we deal with the cases  $N = 3$  and  $N = 4$  later on.

Again, the urn contains one mutation ball with mass  $Nu$  and a certain number of type- $i$  pairs, each consisting of a neutral type- $i$  ball with mass 1 and a selective type- $i$  ball with mass  $s$ . But now some of the type- $i$  pairs are *marked* (in comparison to the remaining *unmarked* type- $i$  pairs). The marking of a type- $i$  pair is understood as markings of the type- $i$  balls that constitute the marked type- $i$  pair. At the beginning, the urn contains the single mutation ball, an unmarked type-0 pair and an unmarked type-1 pair. At each time step a ball is drawn with replacement and with probability proportional to its mass. On account of the draw of the mutation ball we add a marked type- $i$  pair to the urn; the specific type follows the probability measure  $(\nu_i)_{i \in S}$ . If we draw a marked or unmarked type- $i$  ball (either neutral or selective), we add a marked or unmarked type- $i$

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pair, respectively. The status as marked or unmarked is thus inheritable. Note that the marking of a ball indicates that it originates (directly or indirectly) from a draw of the mutation ball.

Altogether, we draw  $N - 1$  times. The concepts of valid and invalid realisations are understood as before: If we draw at least one (marked or unmarked) selective type-1 ball, we term the realisation valid, otherwise invalid.

Let  $\zeta^N(n_0, n_{0m}, n_1)$ ,  $n_0, n_{0m}, n_1 \in \mathbb{N}_0$ , be the probability to obtain a valid realisation that altogether contains  $n_0 + 1$  unmarked type-0 pairs,  $n_{0m}$  marked type-0 pairs and  $n_1 + 1$  (marked or unmarked) type-1 pairs, i.e.  $n_0 + n_{0m} + n_1 = N - 1$ . Since the initial configuration of balls contains each an unmarked type-0 pair and a type-1 pair,  $n_0$  and  $n_1$  denote how often unmarked type-0 pairs and type-1 pairs are added, respectively.

Therefore, we mimic our reasoning in (115)-(117): We consider the urn after the  $k$ 'th draw and assume that it consists of  $k_0 + 1$  and  $k_{0m}$  unmarked and marked type-0 pairs, respectively, and  $k_1 + 1$  type-1 pairs, i.e.  $k_0 + k_{0m} + k_1 = k$ . With probability

$$\frac{(1+s)(k_0+1)}{(1+s)(k+2)+Nu} \quad (120)$$

the next draw gives rise to an increase in the number of unmarked type-0 pairs by one, with probability

$$\frac{(1+s)k_{0m}+Nu\nu_0}{(1+s)(k+2)+Nu} \quad (121)$$

in the number of marked type-0 pairs. Summation gives

$$\frac{(1+s)(k_0+k_{0m}+1)+Nu\nu_0}{(1+s)(k+2)+Nu}, \quad (122)$$

which is the probability to increase the number of both kinds of type-0 pairs by one within the next draw. Finally, we add a type-1 pair without creating an invalid realisation with probability

$$\frac{k_1+1+Nu\nu_1}{(1+s)(k+2)+Nu}. \quad (123)$$

Combining (120), (121) and (123) yields

$$\begin{aligned} & \zeta^N(n_0, n_{0m}, n_1) \\ &= \binom{N-1}{n_0, n_{0m}, n_1} \frac{\prod_{i=0}^{n_0-1} (1+s)(i+1) \prod_{i=0}^{n_{0m}-1} ((1+s)i+Nu\nu_0) \prod_{i=0}^{n_1-1} (i+1+Nu\nu_1)}{\prod_{i=0}^{N-2} ((1+s)(i+2)+Nu)} \\ &= \frac{1}{m_{cum}^N} \frac{(N-1)!}{n_{0m}!n_1!} (1+s)^{n_0} \prod_{i=0}^{n_{0m}-1} ((1+s)i+Nu\nu_0) \prod_{i=1}^{n_1} (i+Nu\nu_1), \end{aligned} \quad (124)$$

where

$$m_{cum}^N = \prod_{i=1}^{N-1} ((1+s)(i+1)+Nu)$$

is the cumulative mass of balls in the urn, taken over all (discrete) time steps. A combination of (122) and (123) results in

$$\begin{aligned}
 & \mathbb{P}(n_0 + n_{0m} = n, \text{ valid realisation}) \\
 &= \frac{1}{m_{cum}^N} \binom{N-1}{n} \prod_{i=1}^n ((1+s)i + N\nu_0) \prod_{i=1}^{N-1-n} (i + N\nu_1) \\
 &= \frac{1}{m_{cum}^N} \binom{N-1}{n} \prod_{i=1}^n \frac{N\lambda_i^N}{N-i} \prod_{i=1}^{N-1-n} \frac{N\mu_{N-i}^N}{N-i} = \frac{1}{m_{cum}^N} \frac{N^{N-1}}{(N-1)!} \phi_{n+1}^{N,N}
 \end{aligned}$$

for  $0 \leq n \leq N-1$ , where we have used (119) in the last equality. The probability to obtain a valid realisation then follows via marginalisation:

$$\mathbb{P}(\text{valid realisation}) = \frac{1}{m_{cum}^N} \frac{N^{N-1}}{(N-1)!} \sum_{i=1}^N \phi_i^{N,N}. \quad (125)$$

We now define  $Y_s^N$  as the number of unmarked type-0 balls with mass  $s$  that are *drawn* in the urn model. For the cases  $N=3$  and  $N=4$  we explicitly calculate the conditional probability

$$\mathbb{P}(Y_s^N \geq 1 \mid \text{valid realisation}).$$

We will use (124) and the fact that every time an unmarked type-0 ball is drawn, it has mass 1 with probability  $1/(1+s)$  and mass  $s$  with probability  $s/(1+s)$ . Given the urn contains  $n_0 + 1$  unmarked type-0 pairs,  $Y_s^N$  follows a binomial distribution with parameters  $n_0$  and  $s/(1+s)$ .

Our results for the conditional probability will coincide with the corresponding expression in (118) and thus provide a tentative interpretation of  $a_1^N$  for  $N=3$  and  $N=4$ .

$N=3$ : We start with

$$\begin{aligned}
 & \mathbb{P}(Y_s^3 \geq 1, \text{ valid realisation}) \\
 &= [\zeta^3(1, 1, 0) + \zeta^3(1, 0, 1)] \frac{s}{1+s} + \zeta^3(2, 0, 0) \frac{2s + s^2}{(1+s)^2}
 \end{aligned}$$

and calculate the expression on the right-hand side with the help of the following decomposition. Its choice will become clear in what follows.

(a)

$$\zeta^3(1, 0, 1) \frac{s}{1+s} + \zeta^3(2, 0, 0) \frac{s}{(1+s)^2} = \frac{2s}{m_{cum}^3} (2 + 3\nu_1) = \frac{6s}{m_{cum}^3} \mu_1^3.$$

(b)

$$\zeta^3(1, 1, 0) \frac{s}{1+s} + \zeta^3(2, 0, 0) \frac{s + s^2}{(1+s)^2} = \frac{2s}{m_{cum}^3} (3\nu_0 + 1 + s) = \frac{3s}{m_{cum}^3} \lambda_1^3.$$

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Taking together the results of (a) and (b) with (125), we obtain

$$\mathbb{P}(Y_s^3 \geq 1 \mid \text{valid realisation}) = s \frac{2}{9} \frac{6\mu_1^3 + 3\lambda_1^3}{\phi_1^{3,3} + \phi_2^{3,3} + \phi_3^{3,3}} = a_1^3,$$

where the last equality is caused by (118).

$N = 4$ : First we consider

$$\begin{aligned} \mathbb{P}(Y_s^4 \geq 1, \text{valid realisation}) &= [\zeta^4(1, 1, 1) + \zeta^4(1, 2, 0) + \zeta^4(1, 0, 2)] \frac{s}{1+s} \\ &\quad + [\zeta^4(2, 1, 0) + \zeta^4(2, 0, 1)] \frac{2s + s^2}{(1+s)^2} \\ &\quad + \zeta^4(3, 0, 0) \frac{3s + 3s^2 + s^3}{(1+s)^3} \end{aligned}$$

and decompose the right-hand side according to (a)-(c) below.

(a)

$$\begin{aligned} &\zeta^4(1, 0, 2) \frac{s}{1+s} + \zeta^4(2, 0, 1) \frac{s}{(1+s)^2} + \zeta^4(3, 0, 0) \frac{s}{(1+s)^3} \\ &= \frac{6s}{m_{cum}^4} \left[ \frac{1}{2} (1 + 4u\nu_1)(2 + 4u\nu_1) + (1 + 4u\nu_1) + 1 \right] \\ &= \frac{6s}{m_{cum}^4} (3 + 4u\nu_1)(1 + 2u\nu_1) = \frac{24s}{m_{cum}^4} \mu_1^4 \mu_2^4. \end{aligned}$$

(b)

$$\begin{aligned} &\zeta^4(1, 1, 1) \frac{s}{1+s} + \zeta^4(2, 1, 0) \frac{s}{(1+s)^2} + \zeta^4(2, 0, 1) \frac{s + s^2}{(1+s)^2} + \zeta^4(3, 0, 0) \frac{s + s^2}{(1+s)^3} \\ &= \frac{6s}{m_{cum}^4} [4u\nu_0(1 + 4u\nu_1) + 4u\nu_0 + (1+s)(1 + 4u\nu_1) + (1+s)] \\ &= \frac{6s}{m_{cum}^4} (1 + s + 4u\nu_0)(2 + 4u\nu_1) = \frac{16s}{m_{cum}^4} \lambda_1^4 \mu_2^4. \end{aligned}$$

(c)

$$\begin{aligned} &\zeta^4(1, 2, 0) \frac{s}{1+s} + \zeta^4(2, 1, 0) \frac{s + s^2}{(1+s)^2} + \zeta^4(3, 0, 0) \frac{s + 2s^2 + s^3}{(1+s)^3} \\ &= \frac{6s}{m_{cum}^4} \left[ \frac{1}{2} 4u\nu_0(1 + s + 4u\nu_0) + (1+s)4u\nu_0 + (1 + 2s + s^2) \right] \\ &= \frac{6s}{m_{cum}^4} (1 + s + 4u\nu_0)(1 + s + 2u\nu_0) = \frac{8s}{m_{cum}^4} \lambda_1^4 \lambda_2^4. \end{aligned}$$



With the help of (a)-(c) and (125) we finally have

$$\mathbb{P}(Y_s^4 \geq 1 \mid \text{valid realisation}) = s \frac{3\mu_1^4\mu_2^4 + 2\lambda_1^4\mu_2^4 + \lambda_1^4\lambda_2^4}{4\phi_1^{4,4} + \phi_2^{4,4} + \phi_3^{4,4} + \phi_4^{4,4}} = a_1^4.$$

We close this section with a generalisation of the sum in (a). Thm. 6 allows for an explicit representation in terms of the urn model, which provides us with a nice interpretation for arbitrary  $N$ .

**Theorem 6.** *Let  $N \geq 1$ . Then*

$$\begin{aligned} \mathbb{P}(Y_s^N = 1, n_{0m} = 0, \text{valid realisation}) &= \sum_{i=1}^{N-1} \zeta^N(i, 0, N-1-i) \frac{s}{(1+s)^i} \\ &= \frac{sN^{N-2}}{m_{cum}^N} \frac{N-1}{(N-2)!} \phi_1^{N, N-1}. \end{aligned} \quad (126)$$

To check the assertion of Thm. 6 we first need the following identity.

**Lemma 5.** *Let  $N \geq 1$  and  $z \in \mathbb{R}_{\geq 0}$ . Then*

$$\sum_{i=1}^N \prod_{j=1}^{N-i} \left(1 + \frac{z}{j}\right) = N \prod_{j=2}^N \left(1 + \frac{z}{j}\right).$$

*Proof.* We prove the assertion by induction over  $N$ . For  $N = 1$  it is obviously true. For some  $N > 1$  we split the sum on the left-hand side according to whether  $i = 1$  or  $i \geq 2$  and insert the induction hypothesis into the second sum. This way, we obtain

$$\begin{aligned} \sum_{i=1}^N \prod_{j=1}^{N-i} \left(1 + \frac{z}{j}\right) &= \prod_{j=1}^{N-1} \left(1 + \frac{z}{j}\right) + (N-1) \prod_{j=2}^{N-1} \left(1 + \frac{z}{j}\right) \\ &= (N+z) \prod_{j=2}^{N-1} \left(1 + \frac{z}{j}\right) = N \prod_{j=2}^N \left(1 + \frac{z}{j}\right), \end{aligned}$$

as claimed.  $\square$

*Proof of Thm. 6.* First we replace  $\zeta^N(i, 0, N-1-i)$  by the right-hand side of (124) and reformulate the resulting expression with the help of Lemma 5 for  $N-1$  and  $z = Nu\nu_1$ :

$$\begin{aligned} \sum_{i=1}^{N-1} \zeta^N(i, 0, N-1-i) \frac{s}{(1+s)^i} &= \frac{s(N-1)!}{m_{cum}^N} \sum_{i=1}^{N-1} \frac{1}{(N-1-i)!} \prod_{j=1}^{N-1-i} (j + Nu\nu_1) \\ &= \frac{s(N-1)!}{m_{cum}^N} (N-1) \prod_{j=2}^{N-1} \left(1 + \frac{Nu\nu_1}{j}\right). \end{aligned}$$

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Then we use the identity  $N\mu_{N-j}^N/(j(N-j)) = 1 + N\nu\nu_1/j$  and obtain

$$= \frac{sN^{N-2}}{m_{cum}^N} \frac{N-1}{(N-2)!} \prod_{j=2}^{N-1} \mu_{N-j}^N = \frac{sN^{N-2}}{m_{cum}^N} \frac{N-1}{(N-2)!} \phi_1^{N,N-1},$$

where the last equality is caused by (119). □

Finally, dividing (126) by (125) yields

$$\mathbb{P}(Y_s^N = 1, n_{0m} = 0 \mid \text{valid realisation}) = s \frac{N-1}{N} \frac{(N-1)\phi_1^{N,N-1}}{\sum_{i=1}^N \phi_i^{N,N}},$$

which is the first summand (i.e.  $i = 1$ ) of  $a_1^N$  in (118).

Altogether, the framework of our urn model is a first (tentative) approach to handle Fearnhead's coefficients in a Moran model with selection *and* mutation. As a beginning, we have found a meaning of  $a_1^3$  and  $a_1^4$ , which obviously remains to be cleared up properly. Moreover, the complete understanding of the urn model of arbitrary size  $N$ , and its genealogical interpretation are left for future research.

## 7 Summary and discussion

### 7.1 Summary

In this thesis, we have reanalysed the stationary distribution of the common ancestor process in the Moran model with selection and mutation. Thereby, the works by Fearnhead [17] and Taylor [43] establish important results with the help of diffusion theory and analytical tools, but the particle representation can only be partially recovered behind the continuous limit. We have therefore made a first step towards complementing the picture by attacking the problem from the discrete (finite-population) side.

The pivotal quantity considered here is the fixation probability of the offspring of all type-0 individuals, regardless of the types of the offspring. Starting from the particle picture and using elementary arguments of first-step analysis, we obtained a difference equation for these fixation probabilities. In the limit  $N \rightarrow \infty$ , the equation turns into the (second-order ODE) boundary problem obtained via diffusion theory by Taylor [43], but now with an intuitive interpretation attached to it.

We have given the solution of the difference equation in closed form; the resulting fixation probabilities provide a generalisation of the well-known finite-population fixation probabilities in the case with selection only (note that they do not require the population to be stationary). As a little detour, we also revisited the limiting continuous boundary value problem and solved it via elementary methods, without the need of the series expansion employed previously.

The fixation probabilities are intimately related with the stationary type distribution on the ancestral line and can thus be used for an alternative derivation of the recursions that characterise Fearnhead's coefficients. Fearnhead obtained these recursions by guessing and direct (but technical) verification of the stationarity condition; Taylor derived them in a constructive way by inserting the ansatz (28) into the boundary value problem (24) and performing a somewhat tedious differentiation exercise. Here, we have taken a third route that relies on the difference equation (36) and stays entirely within the discrete setting. This way, we also obtained the discrete analogue to Fearnhead's coefficients.

The fixation probability may be decomposed into a neutral part plus the extra absorption probability. A coupling approach enlightens this representation: We formalised the extra absorption probability in terms of fixation probabilities in a coupling between Moran models. In addition, we have found interpretations for the  $h_k^N - h_l^N$  as fixation probabilities in couplings.

The intriguing representation (25), which was also shown to have a discrete analogue in terms of the  $a_n^N$ , carries over to the case without mutation (which turns the stationary Markov chain into an absorbing one). Indeed, it was our motivation to reanalyse the process of fixation in a Moran model with selection by means of the labelled Moran model. Here, the basic idea was that selective events – in contrast to neutral ones – only occur between ordered pairs with increasing labels. With the help of cyclic permutations we calculated  $\eta_i^N$ , i.e. the fixation probability of label  $i$ . These  $\eta_i^N$  coincide with the individual terms in the series expansion (85).

## 7 Summary and discussion

Likewise, the selective defining events established themselves as a main tool: Fearnhead's  $a_n^N$  and  $a_n$  were shown to reduce to the  $\mathbb{P}(Y^N \geq n)$  and  $\mathbb{P}(Y^\infty \geq n)$ , respectively. Additionally, we obtained a term-by-term interpretation of the fixation probability (53) (respectively (25) in the diffusion limit) as a decomposition according to the number of selective defining events. These may successively shift the ancestral line to the left, thus placing more weight on the 'fit' individuals.

As an outlook, we finally developed a modification of Hoppe's urn model for a Moran model with selection and mutation, which is also closely connected to a genealogical interpretation in the sense of the killed coalescent. Its type distribution was shown to coincide with the stationary distribution (2). As the main benefit of the new urn model, we started in addition to the mutation ball with certain other balls and marked those that (directly or indirectly) originated from the mutation ball. For  $N = 3$  and  $N = 4$  we pointed out a connection between the coefficient  $a_1^N$  and the number of marked balls.

### 7.2 Discussion

More than fifteen years after the discovery of the ancestral selection graph by Neuhauser and Krone [32, 37], ancestral processes with selection and mutation constitute an active area of research, see, e.g., the recent contributions [13, 14, 45]. Still, the ASG remains a challenge: Despite the elegance and intuitive appeal of the concept, it is difficult to handle when it comes to concrete applications. Indeed, only very few properties of genealogical processes in mutation-selection balance could be described explicitly until today (see [46, 48] for an example in the context of the conditional ASG). Even the special case of a single ancestral line (emerging from a sample of size one) is not yet fully understood.

In this contribution we started from the discrete setting. Altogether, the finite-population results contain more information than those obtained within the diffusion limit. First, because they are not restricted to weak selection, and second, because they are more directly related to the underlying particle picture. Both motivations also underlie, for example, the work by Vogl and Clemente [45], who considered small mutation rates and derived discrete equivalents to results that already have been obtained for the diffusion process.

But even the Moran model with selection still is the subject of state-of-the-art research, particularly when it comes to ancestral processes and genealogies. See for a first example the recent work by Pokalyuk and Pfaffelhuber [39], who reanalysed the process of fixation under strong selection with the help of an ASG. As a second example, one considers particle representations via the lookdown construction [7, 8, 9]; a recent approach that investigates viability selection is introduced by Bah, Pardoux and Sow [2].

Indeed, the labelled Moran model is reminiscent of the  $N$ -particle lookdown process. It is interesting to compare both constructions: In the lookdown with fertility selection, *neutral* arrows only point in one direction (from lower to higher levels), whereas *selective* arrows may appear between arbitrary levels. In contrast, the labelled Moran model contains *neutral arrows in all directions*, but *selective arrows* only occur from lower to

higher labels. Also, the labelled Moran model deliberately dispenses with exchangeability, which is an essential ingredient of the lookdown. It may be conceived that it is possible to transform the labelled Moran model into a lookdown (by way of random permutations), but this remains to be elucidated.

We certainly do not advertise the labelled Moran model as a general-purpose tool; in particular, due to its arbitrary neutral arrows, it does not allow the construction of a sequence of models with increasing  $N$  on the same probability space. However, it turned out to be particularly useful for the purpose considered in this thesis. It enlightens the representations (53) and (25), and provides us with interpretations of Fearnhead's  $a_n^N$  and  $a_n$ . The observation that our  $W^\infty$ , that is, 1 plus the number of selective defining events in the diffusion limit, has the same distribution as the number of branches in the ASG fits nicely into this context, but still requires some further thought.

Needless to say, the next challenge will be to extend the results to the case with mutation. It is known already that the coefficients  $a_n$  may be interpreted as the probabilities that there are  $n$  virtual branches in the pruned ASG at stationarity. But the probabilistic content of the recursions (27) remains to be decoded.

For the cases  $N = 3$  and  $N = 4$  our urn model permits a tentative interpretation of  $a_1^N$ ; and the decompositions (a)-(b) and (a)-(c) are a first tentative step towards a term-by-term interpretation. Clearly, these decompositions still remain to be clarified. The next steps will be first to check, if it is even possible to generalise the interpretation of  $a_1^N$  for  $N \geq 5$ , and second to elucidate the genealogical content. A further obvious task will be to identify the remaining coefficients  $a_2^N, \dots, a_{N-1}^N$ .

It would also be desirable to generalise the results to finite type spaces, for example, in the spirit of Etheridge and Griffiths [13]. They considered a multi-type Moran model with selection and mutation and derived the corresponding dual process, which was interpreted as a variant of the conditional ASG of a population of finite size. They also started to represent fixation probabilities in a multi-type Moran model with selection via this dual. Afterwards, a passage to the limit yielded the dual of the diffusion with weak selection. In [14], this approach was imitated for a Moran model with a more general reproduction mechanism, namely, with multiple offspring and survival depending on the type and the brood-size.

Altogether, the thesis is a first step towards a better understanding of the particle picture related to the common ancestor process. Given the tools and insights that have become available, it seems to be possible, although not straightforward, to delve further into this approach.



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