The common ancestor process revisited

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Abstract. We consider the Moran model in continuous time with two types, mutation, and selection. We concentrate on the ancestral line and its stationary type distribution. Building on work by Fearnhead (J. Appl. Prob. 39 (2002), 38-54) and Taylor (Electron. J. Probab. 12 (2007), 808-847), we characterise this distribution via the fixation probability of the offspring of all individuals of favourable type (regardless of the offsprings' types). We concentrate on a finite population and stay with the resulting discrete setting all the way through. This way, we extend previous results and gain new insight into the underlying particle picture.

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1 Introduction

Understanding the interplay of random reproduction, mutation, and selection is a major topic of population genetics research. 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. Generically, in populations that evolve at constant size over a long time span without recombination, the ancestral lines will eventually coalesce backwards in time into a single line of descent. This ancestral line is of special interest. In particular, its 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.

This article is devoted to the ancestral line in a classical model of population genetics, namely, the Moran model in continuous time with two types, mutation, and selection (i.e., one type is 'fitter' than the other). We are particularly interested in the stationary distribution of this type process, to be called the *ancestral type distribution*. We build on previous work by Fearnhead [9] and Taylor [20]. Fearnhead's approach is based on the *ancestral selection graph*, or ASG for short [14, 16]. The ASG is an extension of Kingman's coalescent [12, 13], which is the central tool to describe the genealogy of a finite sample in the *absence* of selection. The ASG copes with selection by including so-called *virtual branches* in addition to the *real branches* that define the true genealogy. Fearnhead calculates the ancestral type distribution in terms of the coefficients of a series expansion that is related to the number of virtual branches.

Taylor uses diffusion theory and a backward-forward construction that relies on a description of the *full* population. He characterises the ancestral type distribution in terms of the fixation probability of the offspring of all 'fit' individuals (regardless of the offsprings' types). This fixation probability is calculated via a boundary value problem.

Both approaches rely strongly on analytical tools; in particular, they employ the diffusion limit (which assumes infinite population size, weak selection and mutation) 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 individual lineages and their interactions explicit). The aim of this article is to complement these approaches by starting from the graphical representation for a population of finite size and staying with the resulting discrete setting all the way through, performing the diffusion limit only at the very end. This will result in an extension of the results to arbitrary selection strength, as well as a number of new insights, such as an intuitive explanation of Taylor's boundary value problem in terms of the particle picture, and an alternative derivation of the ancestral type distribution.

The paper is organised as follows. We start with a short outline of the Moran model (Section 2). In Section 3, we introduce the common ancestor type process and briefly recapitulate Taylor's and Fearnhead's approaches. 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 a difference equation, which turns into Taylor's diffusion equation in the limit. We solve this difference equation and obtain the fixation probability in the finite-size model in closed form. In Section 5, we derive the coefficients of the ancestral type distribution within the discrete setting. Section 6 summarises and discusses the results.

2 The Moran model with mutation and selection

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 $1 + s_N$, $s_N \geqslant 0$. Accordingly, type-0 individuals are termed 'fit', type-1 individuals are 'unfit'. In line with a central idea of the ASG, we will 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 $u_N \nu_j$, $u_N \geqslant 0$, $0 < \nu_j < 1$, $\nu_0 + \nu_1 = 1$. 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 ν_j. Note that this includes the possibility of 'silent' mutations, i.e. mutations from type i to type i.

The Moran model has a well-known graphical representation as an interacting particle system (cf. Fig. 1). The N 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.
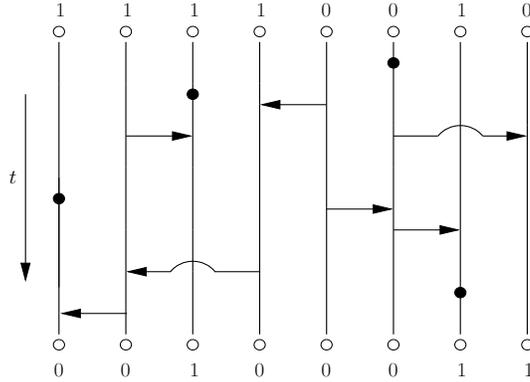

Figure 1: The Moran model. The types ($0 =$ fit, $1 =$ unfit) are indicated for the initial population (top) and the final one (bottom).

We are now interested in the process $\left(Z_t^N\right)_{t \geqslant 0}$, where Z_t^N is the number of individuals of type 0 at time t. When the number of type-0 individuals is k, it increases by one at rate λ_k^N and decreases by one at rate μ_k^N, where

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

Thus $\left(Z_t^N\right)_{t \geqslant 0}$ is a birth-death process with birth rates λ_k^N and death rates μ_k^N. For $u_N > 0$ and $0 < \nu_0, \nu_1 < 1$ its stationary distribution is $\left(\pi_Z^N(k)\right)_{0 \leqslant k \leqslant N}$ with

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

where C_N is a normalising constant (cf. [4, p. 19]). (As usual, an empty product is understood as 1.)

To arrive at a diffusion, we consider the usual rescaling

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

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

$$(X_t)_{t\geqslant 0} := \lim_{N\to\infty}\left(X_t^N\right)_{t\geqslant 0}.$$

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

$$a(x) = \lim_{N\to\infty}(\lambda_{k_N}^N - \mu_{k_N}^N) = (1-x)\theta\nu_0 - x\theta\nu_1 + (1-x)x\sigma \tag{3}$$

and the diffusion coefficient

$$b(x) = \lim_{N\to\infty}\frac{1}{N}\left(\lambda_{k_N}^N + \mu_{k_N}^N\right) = 2x(1-x). \tag{4}$$

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

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

The stationary density π_X – known as Wright's formula – is given by

$$\pi_X(x) = C(1-x)^{\theta\nu_1-1}x^{\theta\nu_0-1}\exp(\sigma x), \tag{5}$$

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

In contrast to our approach starting from the Moran model, [9] and [20] choose the diffusion limit of the *Wright-Fisher model* as the basis for the common ancestor process. This is, however, of minor importance, since both diffusion limits differ only by a rescaling of time by a factor of 2 (cf. [5, Ch. 7], [8, Ch. 5] or [11, Ch. 15]).

3 The common ancestor type process

Assume that the population is stationary and evolves according to the diffusion process $(X_t)_{t\geqslant 0}$. Then, at any time t, there almost surely exists a unique individual that is, at some time $s > t$, ancestral to the whole population; cf. Fig. 2. (One way to see this is via [14, Thm. 3.2, Corollary 3.4], which shows that the expected time to the ultimate ancestor in the ASG remains bounded 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 $(I_t)_{t\geqslant 0}$ 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 $(I_t)_{t\geqslant 0}$ by a second component to obtain a Markov process. They go back to Fearnhead [9] and Taylor [20]; we will recapitulate them below.
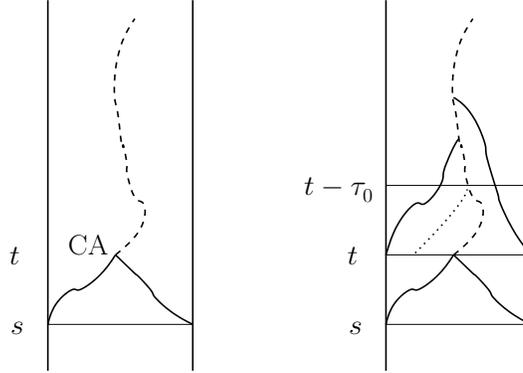
Figure 2: Left: The common ancestor at time t (CA) is the individual whose progeny will eventually fix in the population (at time $s > t$). Right: If we pick an arbitrary individual at time t, there exists a minimal time τ_0 so that the individual's line of ancestors (dotted) corresponds to the ancestral line (dashed) up to time $t - \tau_0$.

3.1 Taylor's approach

For ease of exposition, we start with Taylor's approach [20]. It relies on a description of the *full* population forward in time (in the diffusion limit of the Moran model as $N \to \infty$) and builds on the so-called *structured coalescent* [2]. The process is $(I_t, X_t)_{t \geq 0}$, with states (i, x), $i \in S$ and $x \in [0, 1]$. In [20] this process is termed *common ancestor process (CAP)*.

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. Denote the (stationary) distribution of $(I_t, X_t)_{t \geq 0}$ by π_T. Its marginal distributions are α (with respect to the first variable) and π_X (with respect to the second variable). π_T may then be written as the product of the marginal density $\pi_X(x)$ and the conditional probability $h(x)$ (cf. [20]):

$$\pi_T(0, x)\, dx = h(x)\pi_X(x)dx,$$
$$\pi_T(1, x)\, dx = (1 - h(x))\, \pi_X(x)dx.$$

Since π_X is well-known (5), it remains to specify h. Taylor uses a backward-forward construction within diffusion theory to derive a boundary value problem for h, namely:

$$\frac{1}{2}b(x)h''(x) + a(x)h'(x) - \left(\theta\nu_1 \frac{x}{1-x} + \theta\nu_0 \frac{1-x}{x}\right)h(x) + \theta\nu_1 \frac{x}{1-x} = 0, \tag{6}$$
$$h(0) = 0, h(1) = 1.$$

Taylor shows that (6) 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. [20, Lemma 2.3, Prop. 2.4]).

5

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 σ (cf. [20, Sec. 4] and see below), which yields

$$h(x) = x + \sigma x^{-\theta \nu_0} (1 - x)^{-\theta \nu_1} \exp(-\sigma x) \int_0^x (\tilde{x} - p) \, p^{\theta \nu_0} (1 - p)^{\theta \nu_1} \exp(\sigma p) dp \quad (7)$$

$$\text{with } \tilde{x} = \frac{\int_0^1 p^{\theta \nu_0 + 1} (1 - p)^{\theta \nu_1} \exp(\sigma p) dp}{\int_0^1 p^{\theta \nu_0} (1 - p)^{\theta \nu_1} \exp(\sigma p) dp} = \frac{\mathbb{E}_{\pi_X}(X^2(1 - X))}{\mathbb{E}_{\pi_X}(X(1 - X))}. \quad (8)$$

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

$$\alpha_0 = \int_0^1 h(x)\pi_X(x)dx \quad \text{and} \quad \alpha_1 = \int_0^1 (1 - h(x)) \, \pi_X(x)dx. \quad (9)$$

Following [20], we define $\psi(x) := h(x) - x$ and write

$$h(x) = x + \psi(x). \quad (10)$$

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 (10) into (6) leads to a boundary value problem for ψ:

$$\frac{1}{2}b(x)\psi''(x) + a(x)\psi'(x) - \left(\theta \nu_1 \frac{x}{1 - x} + \theta \nu_0 \frac{1 - x}{x} \right) \psi(x) + \sigma x (1 - x) = 0,$$
$$\psi(0) = \psi(1) = 0. \quad (11)$$

Here, the smooth inhomogeneous term is more favourable as compared to the divergent inhomogeneous term in (6). Note that Taylor actually derives the boundary value problems (6) and (11) for the more general case of frequency-dependent selection, but restricts himself to frequency-independence to derive solution (7).

3.2 Fearnhead's approach

We can only give a brief introduction to Fearnhead's approach [9] here. On the basis of the ASG, the ancestry of a randomly chosen individual from the present (stationary) population is traced backwards in time. More precisely, one considers the process $(J_\tau)_{\tau \geqslant 0}$ with values in S, where J_τ is the type of the individual's ancestor at time τ before the present (that is, at forward time $t - \tau$). Obviously, there is a minimal time τ_0 so that, for all $\tau \geqslant \tau_0$, $J_\tau = I_{t-\tau}$ (see also Fig. 2), provided the underlying process $(X_t)_{t \geqslant 0}$ is extended to $(-\infty, \infty)$.

To make the process Markov, the true ancestor (known as the *real* branch) is augmented by a collection of *virtual* branches (see [1, 14, 16, 19] for the details). Following [9, Thm. 1], certain virtual branches may be removed (without compromising the Markov

6

property) and the remaining set of virtual branches contains only unfit ones. We will refer to the resulting construction as the *pruned ASG*. It is described by the process $(J_\tau, V_\tau)_{\tau \geqslant 0}$, where V_τ (with values in $\mathbb{N}_0$) is the number of virtual branches (of type 1). $(J_\tau, V_\tau)_{\tau \geqslant 0}$ is termed *common ancestor process* in [9] (but keep in mind that it is (I_t, X_t) that is called CAP in [20]). Reversing the direction of time in the pruned ASG yields an alternative augmentation of the CAT process (for $\tau \geqslant \tau_0$).

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

$$\rho_{k+1}^{(k)} = 0 \quad \text{and} \quad \rho_{j-1}^{(k)} = \frac{\sigma}{j + \sigma + \theta - (j + \theta\nu_1)\rho_j^{(k)}}, \quad k \in \mathbb{N}_0, 0 \leqslant j \leqslant k+1. \tag{12}$$

The limit $\rho_j := \lim_{k\to\infty} \rho_j^{(k)}$ exists (cf. [9, Lemma 1]) and the stationary distribution of the pruned ASG is given by (cf. [9, Thm. 3])

$$\pi_F(i, n) = \begin{cases} a_n \mathbb{E}_{\pi_X}(X(1-X)^n), & \text{if } i = 0, \\ (a_n - a_{n+1})\mathbb{E}_{\pi_X}((1-X)^{n+1}), & \text{if } i = 1, \end{cases}$$

$$\text{with} \quad a_n := \prod_{j=1}^n \rho_j$$

for all $n \in \mathbb{N}_0$. 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. Marginalising over the number of virtual branches results in the stationary type distribution of the ancestral line, namely,

$$\alpha_i = \sum_{n \geqslant 0} \pi_F(i, n). \tag{13}$$

Furthermore, this reasoning points to an alternative representation of h respectively ψ (cf. [20]):

$$h(x) = x + x \sum_{n \geqslant 1} a_n(1-x)^n \quad \text{respectively} \quad \psi(x) = x \sum_{n \geqslant 1} a_n(1-x)^n. \tag{14}$$

The a_n, to which we will refer as *Fearnhead's coefficients*, can be shown [20] to follow the second-order *forward* recursion

$$\begin{aligned} &(2 + \theta\nu_1)\, a_2 - (2 + \sigma + \theta)\, a_1 + \sigma = 0, \\ &(n + \theta\nu_1)\, a_n - (n + \sigma + \theta)\, a_{n-1} + \sigma a_{n-2} = 0, \quad n \geqslant 3. \end{aligned} \tag{15}$$

Indeed, (14) solves the boundary problem (6) and therefore equals (7) (cf. [20, Lemma 4.1]).

The forward recursion (15) 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. [20, Sec. 4.1])

$$v(x) := \frac{h(x) - x}{x} = \frac{\psi(x)}{x} = \sum_{n \geqslant 1} a_n (1 - x)^n \tag{16}$$

and uses[1]

$$a_n = \frac{(-1)^n}{n!} v^{(n)}(1). \tag{17}$$

This way a straightforward (but lengthy) calculation (that includes a differentiation of expression (7)) yields

$$a_1 = -v'(1) = -\psi'(1) = \frac{\sigma}{1 + \theta \nu_1} (1 - \tilde{x}). \tag{18}$$

4 Discrete approach

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 π_T and π_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. In a first step we re-derive the differential equations for h and ψ in a direct way, on the basis of the particle picture for a finite population. This derivation will be elementary and, at the same time, it will provide a direct interpretation of the resulting differential equations.

4.1 Difference and differential equations for h and ψ

Equations for h. 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 $(\boldsymbol{M}_t, Z_t^N)_{t \geqslant 0}$ where Z_t^N is the number of fit individuals as before and $\boldsymbol{M}_t = (M_0, M_1)_t$ holds the number of descendants of types 0 and 1 at time t of an unordered sample with composition $\boldsymbol{M}_0 = (M_0, M_1)_0$ collected at time 0. More precisely, we start with an arbitrary state $(\boldsymbol{M}_0, Z_0^N) = (\boldsymbol{m}, k)$ (where $\boldsymbol{M}_0$ need not to 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 $\boldsymbol{M}_0$ and summarise the results in the unordered sample $\boldsymbol{M}_t$. Together with Z_t^N, this gives the current state $(\boldsymbol{M}_t, Z_t^N)$ (cf. Fig. 3).

As soon as the initial sample is ancestral to all N individuals, it clearly will be ancestral to all N individuals at all later times. Therefore

$$\mathcal{A}_N := \{(\boldsymbol{m}, k) : k \in \{0, \ldots, N\}, m_0 \leqslant k, |\boldsymbol{m}| = N\},$$

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[1]Note the missing factor of $1/n$ in his equation (28).
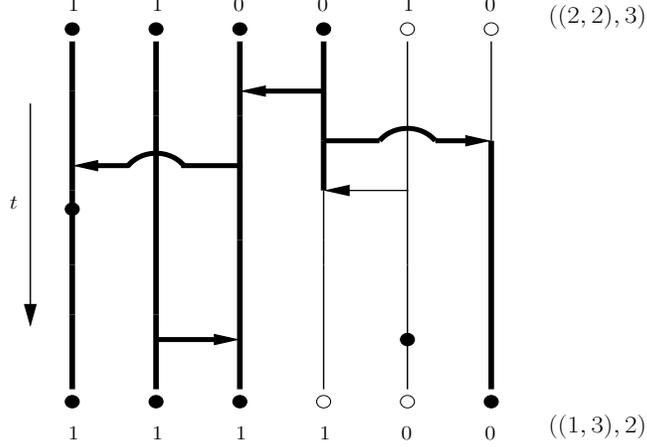
Figure 3: The process $\left(\boldsymbol{M}_t, Z_t^N\right)_{t \geqslant 0}$. The initial sample $\boldsymbol{M}_0 = (2,2)$ in a population of size $N = 6$ (whose number of type-0 individuals is $Z_0^N = 3$) is marked black at the top. Fat lines represent their descendants. At the later time (bottom), 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.

where $|\boldsymbol{m}| = m_0 + m_1$ for a sample $\boldsymbol{m} = (m_0, m_1)$, is a closed (or invariant) set of the Markov chain. (Given a Markov chain $(Y(t))_{t \geqslant 0}$ 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. [17, Ch. 3.2]).)

From now on we restrict ourselves to the initial value $(\boldsymbol{M}_0, Z_0^N) = ((k,0), k)$, 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 of the descendants of the type-0 individuals. In other words, we are interested in the probability that the common ancestor at time 0 belongs to our fit sample $\boldsymbol{M}_0$. Let us 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 $Z_0^N = k$. Equivalently, h_k^N is the absorption probability of $(\boldsymbol{M}_t, Z_t^N)$ in $\mathcal{A}_N$, conditional on $(\boldsymbol{M}_0, Z_0^N) = ((k,0), k)$. Obviously $h_0^N = 0$, $h_N^N = 1$. It is important to note that, given absorption in $\mathcal{A}^N$, the common ancestor is a random draw from the initial sample. Therefore,

$$\mathbb{P}\big(\text{a specific type-0 individual will fix} \mid Z_0^N = k\big) = \frac{h_k^N}{k}. \tag{19}$$

Likewise,

$$\mathbb{P}\big(\text{a specific type-1 individual will fix} \mid Z_0^N = k\big) = \frac{1 - h_k^N}{N - k}. \tag{20}$$

We will now calculate the absorption probabilities with the help of 'first-step analysis' (cf. [17, Thm. 3.3.1], see also [5, Thm. 7.5]). Let us recall the method for convenience.

9

Lemma 1 ('first-step analysis'). *Assume that $(Y(t))_{t \geqslant 0}$ 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 $y \in E$,*

$$\mathbb{P}\left(Y \text{ absorbs in } \mathcal{A} \mid Y(0) = y\right) = \sum_{z \in E: z \neq y} \mathbb{P}\left(Y(T_y) = z \mid Y(0) = y\right)$$

$$\times \mathbb{P}\left(Y \text{ absorbs in } \mathcal{A} \mid Y(0) = z\right).$$

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. 4), state the transition rates and calculate absorption probabilities, based upon the new state. We assume throughout that $0 < k < N$.
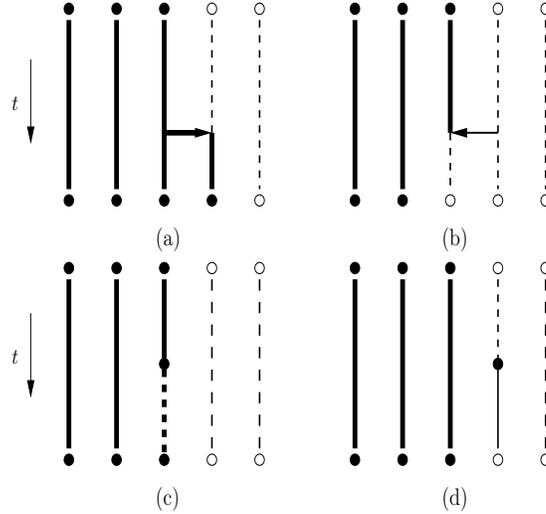

Figure 4: Transitions out of $((k, 0), k)$. Solid lines represent type-0 individuals, dashed ones type-1 individuals. Descendants of type-0 individuals (marked black at the top) are represented by bold lines.

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

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 (M_t, Z_t^N) starts afresh in the new state $((k + 1, 0), k + 1)$, with absorption probability h_{k+1}^N.

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

A type-1 individual reproduces and replaces a (sample) individual of type 0. This occurs at rate $\frac{k(N-k)}{N}$ 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) $((k,0),k) \to ((k-1,1),k-1)$:

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 (20), 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

$$\mathbb{P}\left(\text{absorption in } \mathcal{A}_N \mid \left(\boldsymbol{M}_0, Z_0^N\right) = ((k-1,1),k-1)\right)$$
$$= h_{k-1}^N + \frac{1-h_{k-1}^N}{N-(k-1)}.$$

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

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 (19), we get

$$\mathbb{P}\left(\text{absorption in } \mathcal{A}_N \mid \left(\boldsymbol{M}_0, Z_0^N\right) = ((k,0),k+1)\right)$$
$$= h_{k+1}^N - \frac{h_{k+1}^N}{k+1}.$$

Note that, in steps (c) and (d) (and already in (19) and (20)), 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 $(\boldsymbol{M}_t, Z_t^N)$. 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 λ_k^N and μ_k^N as in (1)):

$$\left(\lambda_k^N + \mu_k^N\right) h_k^N = \lambda_k^N h_{k+1}^N + \mu_k^N h_{k-1}^N + ku_N\nu_1 \frac{1-h_{k-1}^N}{N-(k-1)} - (N-k)u_N\nu_0 \frac{h_{k+1}^N}{k+1}, \quad (21)$$

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

$$\frac{1}{2}\frac{1}{N}\left(\lambda_k^N + \mu_k^N\right) N^2 \left(h_{k+1}^N - 2h_k^N + h_{k-1}^N\right)$$
$$+ \frac{1}{2}\left(\lambda_k^N - \mu_k^N\right)\left(N\left(h_{k+1}^N - h_k^N\right) - N\left(h_{k-1}^N - h_k^N\right)\right) \quad (22)$$
$$+ \frac{k}{N}\frac{N}{N-(k-1)}Nu_N\nu_1\left(1-h_{k-1}^N\right) - \frac{N-k}{N}\frac{N}{k+1}Nu_N\nu_0 h_{k+1}^N = 0.$$

11

Let us now consider a sequence $(k_N)_{N \in \mathbb{N}}$ with $0 < k_N < N$ and $\lim_{N \to \infty} \frac{k_N}{N} = x$. The probabilities $h_{k_N}^N$ converge to $h(x)$ as $N \to \infty$ (for the stationary case a proof is given in the Appendix). Equation (22), with k replaced by k_N, together with (3) and (4) leads to Taylor's boundary value problem (6).

Equations for ψ. As in the previous Section, we consider $\left(\boldsymbol{M}_t, Z_t^N\right)_{t \geqslant 0}$ with start in $((k, 0), k)$, and now introduce the new function $\psi_k^N := h_k^N - \frac{k}{N}$. ψ^N is the part of absorption probability in $\mathcal{A}_N$ that goes back to selective reproductions (in comparison to the neutral case). We therefore speak of ψ^N (as well as of ψ) as the 'extra' absorption probability.

Substituting $h_k^N = \psi_k^N + \frac{k}{N}$ in (21) yields the following difference equation for ψ^N:

$$
\begin{aligned}
\left(\lambda_k^N + \mu_k^N\right) \psi_k^N = {} & \lambda_k^N \psi_{k+1}^N + \mu_k^N \psi_{k-1}^N + \frac{k(N-k)}{N^2} s_N \\
& - k u_N \nu_1 \frac{\psi_{k-1}^N}{N-(k-1)} - (N-k) u_N \nu_0 \frac{\psi_{k+1}^N}{k+1}
\end{aligned}
\tag{23}
$$

$(0 < k < N)$, 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 $\psi_{k+1}^N + \frac{1}{N}$ (rather than h_{k+1}^N). Here, $\frac{1}{N}$ is the neutral fixation probability of the individual just created via the selective event; ψ_{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 (23). Performing $N \to \infty$ in the same way as for h, we obtain Taylor's boundary value problem (11) and now have an interpretation in terms of the graphical representation to go with it.

4.2 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 (21), or equivalently, (23). 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 ψ_k in terms of a recursion.

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

$$
\psi_{N-k} = \frac{k(N-k)}{\mu_{N-k}} \left(\frac{\mu_{N-1}}{N-1} \psi_{N-1} + \frac{\lambda_{N-k+1}}{(k-1)(N-k+1)} \psi_{N-k+1} - \frac{s(k-1)}{N^2} \right).
\tag{24}
$$

Remark 1. *The quantity $\lambda_k/(k(N-k)) = (1+s)/N + u\nu_0/k$ is well-defined for all $1 \leqslant k \leqslant N$, 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 (23) and divide by $i(N - i)$ to obtain

$$\left(\frac{\lambda_i}{i(N - i)} + \frac{\mu_i}{i(N - i)} \right) \psi_i = \left(\frac{1 + s}{N} + \frac{u\nu_0}{i + 1} \right) \psi_{i+1} + \left(\frac{1}{N} + \frac{u\nu_1}{N - (i - 1)} \right) \psi_{i-1} + \frac{s}{N^2}$$

$$= \frac{\lambda_{i+1}}{(i + 1)(N - i - 1)} \psi_{i+1} + \frac{\mu_{i-1}}{(i - 1)(N - i + 1)} \psi_{i-1} + \frac{s}{N^2}.$$

$$(25)$$

Together with

$$\left(\frac{\lambda_1}{N - 1} + \frac{\mu_1}{N - 1} \right) \psi_1 = \frac{\lambda_2}{2(N - 2)} \psi_2 + \frac{s}{N^2}, \tag{26}$$

$$\left(\frac{\lambda_{N-1}}{N - 1} + \frac{\mu_{N-1}}{N - 1} \right) \psi_{N-1} = \frac{\mu_{N-2}}{2(N - 2)} \psi_{N-2} + \frac{s}{N^2}, \tag{27}$$

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

$$\sum_{i=N-k+1}^{N-1} \left(\frac{\lambda_i}{i(N - i)} + \frac{\mu_i}{i(N - i)} \right) \psi_i = \sum_{i=N-k+1}^{N-2} \frac{\lambda_{i+1}}{(i + 1)(N - i - 1)} \psi_{i+1}$$

$$+ \sum_{i=N-k+1}^{N-1} \frac{\mu_{i-1}}{(i - 1)(N - i + 1)} \psi_{i-1} + \frac{s(k - 1)}{N^2},$$

which proves the assertion.

$\square$

Lemma 2 allows for an explicit solution for ψ.

Theorem 1. *For $1 \leqslant l, n \leqslant N - 1$, let*

$$\chi_\ell^n := \prod_{i=\ell}^{n} \frac{\lambda_i}{\mu_i} \quad and \quad K := \sum_{n=0}^{N-1} \chi_1^n. \tag{28}$$

The solution of recursion (24) is then given by

$$\psi_{N-k} = \frac{k(N - k)}{\mu_{N-k}} \sum_{n=N-k}^{N-1} \chi_{N-k+1}^n \left(\frac{\mu_{N-1}}{N - 1} \psi_{N-1} - \frac{s(N - 1 - n)}{N^2} \right) \tag{29}$$

with

$$\psi_{N-1} = \frac{1}{K} \frac{N - 1}{\mu_{N-1}} \frac{s}{N^2} \sum_{n=0}^{N-2} (N - 1 - n) \chi_1^n. \tag{30}$$

An alternative representation is given by

$$\psi_{N-k} = \frac{1}{K} \frac{k(N - k)}{\mu_{N-k}} \frac{s}{N^2} \sum_{\ell=0}^{N-k-1} \sum_{n=N-k}^{N-1} (n - \ell) \chi_1^\ell \chi_{N-k+1}^n. \tag{31}$$

13

Proof. We first prove (29) by induction over k. For $k = 1$, (29) is easily checked to be true. Inserting the induction hypothesis for some $k - 1 \geqslant 0$ into recursion (24) yields

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

which immediately leads to (29). For $k = N$, (29) gives (30), since $\psi_0 = 0$ and $k(N - k)/\mu_{N-k}$ is well-defined by Remark 1. We now check (31) by inserting (30) into (29) and then use the expression for K as in (28):

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

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

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

The first sum is the right-hand side of (31) 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., [5, Thm. 6.1]), but, to the best of our knowledge, have not yet appeared in the literature for the case with mutation.

4.3 The solution of the differential equation

As a little detour, let us revisit the boundary value problem (6). To solve it, Taylor assumes that h can be expanded in a power series in σ. This yields a recursive series of boundary value problems (for the various powers of σ), which are solved by elementary methods and combined into a solution of h (cf. [20]).

However, the calculations are slightly long-winded. In what follows we show that the boundary value problem (6) (or equivalently (11)) 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. (3)) and the diffusion coefficient $b(x)$ (cf. (4)), differential equation (11) reads

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

or, equivalently,

$$\psi''(x) + 2\frac{a(x)}{b(x)}\psi'(x) + 2\frac{c(x)}{b(x)}\psi(x) = -\sigma. \tag{32}$$

Since

$$\frac{c(x)}{b(x)} = \frac{d}{dx}\frac{a(x)}{b(x)} \tag{33}$$

(32) is an exact differential equation (for the concept of exactness, see [10, Ch. 3.11] or [3, Ch. 2.6]). Solving it corresponds to solving its primitive

$$\psi'(x) + 2\frac{a(x)}{b(x)}\psi(x) = -\sigma(x - \tilde{x}). \tag{34}$$

The constant $\tilde{x}$ plays the role of an integration constant and will be determined by the initial conditions later. (Obviously (32) is recovered by differentiating (34) and observing (33).) As usual, we consider the homogeneous equation

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

first. According to [5, Ch. 7.4] and [8, Ch. 4.3], its solution φ_1 is given by

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

(Note the link to the stationary distribution provided by the last expression (cf. [5, Thm. 7.8] and [8, Ch. 4.5]).) Of course the same expression is obtained via separation of variables. Again we will deal with the constant γ later.

Variation of parameters yields the solution φ_2 of the inhomogeneous equation (34):

$$\varphi_2(x) = \varphi_1(x)\int_\beta^x \frac{-\sigma(p-\tilde{x})}{\varphi_1(p)}dp = \sigma\varphi_1(x)\int_\beta^x \frac{\tilde{x}-p}{\varphi_1(p)}dp. \tag{35}$$

Finally, it remains to specify the constants of integration $\tilde{x}$, γ and the constant β to comply with $\varphi_2(0) = \varphi_2(1) = 0$. We observe that the factor γ cancels in (35), thus its

15

choice is arbitrary. $\varphi_1(x)$ diverges for $x \to 0$ and $x \to 1$, so the choice of β 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

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

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. (7)).

We close this Section with a brief consideration of the initial value a_1 of the recursions (15). Since, by (18), $a_1 = -\psi'(1)$, it may be obtained by analysing the limit $x \to 1$ of (34). In the quotient $a(x)\psi(x)/b(x)$, numerator and denominator disappear as $x \to 1$. According to l'Hôpital's rule we get

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

therefore the limit $x \to 1$ of (34) yields

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

Thus, we obtain a_1 without the need to differentiate expression (7).

5 Derivation of Fearnhead's coefficients in the discrete setting

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 $\psi^N = (\psi_k^N)_{0 \leqslant k \leqslant N}$ in (25)-(27). Let

$$\widetilde{\psi}_k^N := \frac{\psi_k^N}{k(N - k)}, \tag{36}$$

for $1 \leqslant k \leqslant N - 1$. In terms of these new variables, (27) reads

$$-\mu_{N-1}^N \widetilde{\psi}_{N-1}^N + \mu_{N-2}^N \widetilde{\psi}_{N-2}^N - \lambda_{N-1}^N \widetilde{\psi}_{N-1}^N + \frac{s_N}{N^2} = 0. \tag{37}$$

We now perform linear combinations of (25)-(27) (again expressed in terms of the $\widetilde{\psi}_{N-k}^N$) to obtain

$$\sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} (\lambda_{N-k}^N + \mu_{N-k}^N) \widetilde{\psi}_{N-k}^N$$

$$= \sum_{k=2}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} \lambda_{N-k+1}^N \widetilde{\psi}_{N-k+1}^N + \sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1} \mu_{N-k-1}^N \widetilde{\psi}_{N-k-1}^N \tag{38}$$

$$+ \frac{s_N}{N^2} \sum_{k=1}^{n-1} (-1)^{n-k-1} \binom{n-2}{k-1},$$

for $n \geqslant 3$. Noting that the last sum disappears as a consequence of the binomial theorem, rearranging turns (38) into

$$\sum_{k=0}^{n-1}(-1)^{n-k-1}\binom{n-1}{k}\mu_{N-k-1}^{N}\widetilde{\psi}_{N-k-1}^{N} + \sum_{k=1}^{n-1}(-1)^{n-k}\binom{n-1}{k}\lambda_{N-k}^{N}\widetilde{\psi}_{N-k}^{N} = 0. \qquad (39)$$

On the basis of equations (37) and (39) for $(\widetilde{\psi}_k^N)_{1\leqslant k\leqslant N-1}$ we will now establish a discrete version of Fearnhead's coefficients, and a corresponding discrete version of recursion (15) and initial value (18). Motivated by the limiting expression (14), we choose the ansatz

$$\psi_{N-k}^{N} = (N-k)\sum_{i=1}^{k} a_i^N \frac{k_{[i]}}{N_{[i+1]}}, \qquad (40)$$

where we adopt the usual notation $x_{[j]} := x(x-1)\ldots(x-j+1)$ for $x \in \mathbb{R}$, $j \in \mathbb{N}$. 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 2. *The a_n^N, $1 \leqslant n \leqslant N-1$, satisfy the following relations: $a_1^N = N\psi_{N-1}$,*

$$(N-2)\left[\left(\frac{2}{N}+u\nu_1\right)a_2^N - \left(\frac{2}{N}+\frac{N-1}{N}s+u\right)a_1^N + \frac{N-1}{N}s\right] = 0, \qquad (41)$$

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

$$(N-n)\left[\left(\frac{n}{N}+u\nu_1\right)a_n^N - \left(\frac{n}{N}+\frac{N-(n-1)}{N}s+u\right)a_{n-1}^N + \frac{N-(n-1)}{N}sa_{n-2}^N\right] = 0. \qquad (42)$$

Proof. At first we note that the initial value a_1^N follows directly from (40) for $k=1$. Then, we remark that, by (36) and (40),

$$\widetilde{\psi}_{N-k} = \frac{1}{k}\sum_{i=1}^{k} a_i^N \frac{k_{[i]}}{N_{[i+1]}} \qquad (43)$$

for $1 \leqslant k \leqslant N-1$. To prove (41) we insert this into (37) 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 (41).

To prove (42) we express (39) in terms of the a_n^N via (43). The first sum of (39) becomes

$$\sum_{k=0}^{n-1}(-1)^{n-k-1}\binom{n-1}{k}\mu_{N-k-1}\widetilde{\psi}_{N-k-1} = \sum_{k=0}^{n-1}(-1)^{n-k-1}\binom{n-1}{k}\mu_{N-k-1}\sum_{i=1}^{k+1}a_i^N\frac{k_{[i-1]}}{N_{[i+1]}}$$

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

17

Analogously, the second sum of (39) turns into

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

Multiplying with $N!$, (39) is thus reformulated as

$$\sum_{i=1}^{n}a_i^N(N-i-1)_{[n-i]}(A_{\mu,i}^n + A_{\lambda,i}^n) = 0, \tag{44}$$

where

$$A_{\mu,i}^n := \sum_{k=i}^{n}(-1)^{n-k}\binom{n-1}{k-1}(k-1)_{[i-1]},\mu_{N-k} \tag{45}$$

$$A_{\lambda,i}^n := \sum_{k=i}^{n-1}(-1)^{n-k}\binom{n-1}{k}(k-1)_{[i-1]}\lambda_{N-k}. \tag{46}$$

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

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

for $i \leqslant k \leqslant n$ and apply this to the right-hand side of (45). 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

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

taken at $N - n$. Since μ is a quadratic function, we conclude that $A_{\mu,i}^n = 0$ for all $1 \leqslant i \leqslant n-3$. 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

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

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

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

18

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 $i \leqslant k \leqslant n-1$, 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
$$\lambda : \{0,\ldots,N-1\} \to \mathbb{R}_{\geqslant 0}, \quad k \mapsto \frac{\lambda_k}{N-k} = \frac{k}{N}(1+s) + u\nu_0$$

taken at $N-(n-1)$. Consequently, $A_{\lambda,i}^n = 0$ for all $1 \leqslant i \leqslant n-3$, and in $A_{\lambda,n-2}^n$ (more precisely in the first difference quotient of λ at $N-(n-1)$) the constant terms cancel 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 (44) with the results for $A_{\mu,i}^n$ and $A_{\lambda,i}^n$ yields the assertion (42). $\qquad\square$

It will not come as a surprise now that the discrete recursions of the a_n^N obtained in Thm. 2 lead to Fearnhead's coefficients a_n in the limit $N \to \infty$. According to Thm. 3 in the Appendix, $\psi_{k_N}^N$ converges to $\psi(x)$ for any given sequence $(k_N)_{N\in\mathbb{N}}$ with $0 < k_N < N$ and $\lim_{N\to\infty}\frac{k_N}{N} = x$. Comparing (40) with (14), we obtain
$$\lim_{N\to\infty} a_n^N = a_n$$

for all $n \geqslant 1$. The recursions (15) of Fearnhead's coefficients then follow directly from the recursions in Thm. 2 in the limit $N \to \infty$.

6 Discussion

More than fifteen years after the discovery of the ancestral selection graph by Neuhauser and Krone [14, 16], ancestral processes with selection constitute an active area of research, see, e.g., the recent contributions [6, 7, 15, 18, 21]. 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 the conditional ASG [22, 23] for an example). Even the special case of a single ancestral line (emerging from a sample of size one) is not yet fully understood. The work by Fearnhead [9] and Taylor [20] established important results about the CAP with the help of diffusion theory and analytical tools, but the particle representation can only be partially recovered behind the continuous limit. In this article, we have therefore made a first step towards complementing the picture by attacking the problem from the discrete (finite-population) side. Let us briefly summarise our results.

The pivotal quantity considered here is the *fixation probability of the offspring of all fit 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 $N \to \infty$, the equation turns into the (second-order ODE) boundary problem obtained via diffusion theory by Taylor [20], 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 (16) into the boundary value problem (11) and performing a somewhat tedious differentiation exercise. Here we have taken a third route that relies on the difference equation (25) and stays entirely within 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 recent work by Pokalyuk and Pfaffelhuber [18], who re-analysed the process of fixation under strong selection (in the absence of mutation) with the help of an ASG in a discrete setting.

Clearly, the present article is only a first step towards a better understanding of the particle picture related to the common ancestor process. 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 (see Section 3.2); but the genealogical content of the recursions (15) remains to be elucidated. It would also be desirable to generalise the results to finite type spaces, in the spirit of Etheridge and Griffiths [6].

Acknowledgement

Appendix

In Section 4.1 we have presented an alternative derivation of the boundary value problem for the conditional probability h. It remains to prove that $\lim_{N\to\infty} h_{k_N}^N = h(x)$, with $x \in [0,1]$, $0 < k_N < N$, $\lim_{N\to\infty} \frac{k_N}{N} = x$ and h as given as in (7).
Since $h_k^N = \frac{k}{N} + \psi_k^N$ and $h(x) = x + \psi(x)$, respectively, it suffices to show the corresponding convergence of the ψ_k^N. For ease of exposition we assume here that the process is stationary.

Lemma 3. *Let $\tilde{x}$ be as in* (8). *Then*

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

Proof. Since the stationary distribution π_Z^N of $\left(Z_t^N\right)_{t\geqslant 0}$ (cf. (2)) satisfies

$$\prod_{i=1}^{n-1} \frac{\lambda_i^N}{\mu_i^N} = \frac{\pi_Z^N(n)}{C_N} \frac{\mu_n^N}{\lambda_0^N}, \tag{47}$$

for $1 \leqslant n \leqslant N$, equation (30) leads to

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

where we have used (1) in the last step. The stationary distribution of the rescaled process $\left(X_t^N\right)_{t\geqslant 0}$ is given by $\left(\pi_X^N\left(\frac{i}{N}\right)\right)_{0\leqslant i\leqslant N}$, where $\pi_X^N\left(\frac{i}{N}\right) = \pi_Z^N(i)$. Besides, the sequence

of processes $\left(X_t^N\right)_{t \geqslant 0}$ converges to $(X_t)_{t \geqslant 0}$ in distribution, hence

$$\lim_{N \to \infty} N \psi_{N-1}^N = \lim_{N \to \infty} \frac{N s_N}{1 + N u_N \nu_1} \frac{\mathbb{E}_{\pi_X^N}\left(X^N \left(1 - X^N\right)^2 \left(1 + \frac{u_N \nu_1}{1 - X^N}\right)\right)}{\mathbb{E}_{\pi_X^N}\left(X^N \left(1 - X^N\right) \left(1 + \frac{u_N \nu_1}{1 - X^N}\right)\right)}$$

$$= \frac{\sigma}{1 + \theta \nu_1} \frac{\mathbb{E}_{\pi_X}\left(X(1 - X)^2\right)}{\mathbb{E}_{\pi_X}\left(X(1 - X)\right)} = \frac{\sigma}{1 + \theta \nu_1}(1 - \tilde{x}),$$

as claimed. $\qquad\square$

Remark 2. *The proof gives an alternative way to obtain the initial value a_1 (cf. (18)) of recursion (15).*

Theorem 3. *For a given $x \in [0, 1]$, let $(k_N)_{N \in \mathbb{N}}$ be a sequence with $0 < k_N < N$ and $\lim_{N \to \infty} \frac{k_N}{N} = x$. Then*

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

where ψ is the solution of the boundary value problem (11).

Proof. Using first Theorem 1, then (47), and finally (1), we obtain

$$\psi_k^N = \frac{k(N - k)}{\mu_k^N} \sum_{n=1}^{N-k} \left(\prod_{i=k+1}^{N-n} \frac{\lambda_i^N}{\mu_i^N}\right) \left(\frac{\mu_{N-1}^N}{N - 1} \psi_{N-1}^N - \frac{s_N(n - 1)}{N^2}\right)$$

$$= \frac{k(N - k)}{\mu_k^N} \left(\mu_{k+1}^N \pi_Z^N(k + 1)\right)^{-1} \sum_{n=0}^{N-k-1} \mu_{N-n}^N \pi_Z^N(N - n) \left(\frac{\mu_{N-1}^N}{N - 1} \psi_{N-1}^N - \frac{s_N n}{N^2}\right)$$

$$= \left(1 + \mathcal{O}\left(\frac{1}{N}\right)\right) \left(\frac{k + 1}{N} \frac{N - k - 1}{N} \pi_Z^N(k + 1)\right)^{-1}$$

$$\times \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N - n) \frac{N - n}{N} \frac{n}{N} \left(1 + \frac{N u_N \nu_1}{n}\right) \left((1 + N u_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N}\right).$$

In order to analyse the convergence of this expression, define

$$S_1^N(k) := \frac{k + 1}{N} \frac{N - k - 1}{N} \pi_Z^N(k + 1),$$

$$S_2^N(k) := \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N - n) \frac{N - n}{N} \frac{n}{N} \left((1 + N u_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N}\right)$$

$$= \int_0^1 T_k^N(y) dy,$$

$$S_3^N(k) := \frac{1}{N} \sum_{n=0}^{N-k-1} \pi_Z^N(N - n) \frac{N - n}{N} u_N \nu_1 \left((1 + N u_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N}\right)$$

$$= \int_0^1 \tilde{T}_k^N(y) dy,$$

22

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

$$T_k^N(y) := \begin{cases} \mathbb{1}_{\{n \leqslant N-k-1\}} \pi_Z^N(N-n) \frac{N-n}{N} \frac{n}{N} \left((1 + N u_N \nu_1) N \psi_{N-1}^N - N s_N \frac{n}{N} \right), \\ \quad \text{if } \frac{n}{N} \leqslant y < \frac{n+1}{N}, n \in \{0, \dots, N-1\}, \\ 0, \text{ if } y = 1, \end{cases}$$

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

Consider now a sequence $(k_N)_{N \in \mathbb{N}}$ as in the assumptions. Then $\lim_{N \to \infty} \pi_Z^N(k_N) = \pi_X(x)$ (cf. [5, p. 319]), and due to Lemma 3

$$\lim_{N \to \infty} S_1^N(k_N) = x(1-x)\pi_X(x),$$

$$\lim_{N \to \infty} T_{k_N}^N(k_N) = \mathbb{1}_{\{y \leqslant 1-x\}} \pi_X(1-y)(1-y)y(\sigma(1-\tilde{x}) - \sigma y),$$

$$\lim_{N \to \infty} \tilde{T}_{k_N}^N(k_N) = 0.$$

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

$$\lim_{N \to \infty} S_2^N(k_N) = \int_0^{1-x} \pi_X(1-y)(1-y)y(\sigma(1-\tilde{x}) - \sigma y)dy,$$

$$\lim_{N \to \infty} S_3^N(k_N) = 0,$$

thus

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

Substituting on the right-hand side yields

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

$$= (x(1-x)\pi_X(x))^{-1} \sigma \left[\int_0^1 \pi_X(y)y(1-y)(y-\tilde{x})dy + \int_0^x \pi_X(y)y(1-y)(\tilde{x}-y)dy \right]$$

$$= (x(1-x)\pi_X(x))^{-1} \sigma \int_0^x \pi_X(y)y(1-y)(\tilde{x}-y)dy = \psi(x),$$

where the second-last equality goes back to the definition of $\tilde{x}$ in (8), and the last is due to (7), (10) and (2). $\qquad \square$

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