THE MUTATION PROCESS ON THE ANCESTRAL LINE UNDER SELECTION

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1. Abstract

We consider the Moran model of population genetics with two types, mutation, and selection, and investigate the line of descent of a randomly-sampled individual from a contemporary population. We trace this ancestral line back into the distant past, far beyond the most recent common ancestor of the population (thus connecting population genetics to phylogeny) and analyse the mutation process along this line.

To this end, we use the pruned lookdown ancestral selection graph, which consists of the set of potential ancestors of the sampled individual at any given time. A crucial observation is that a mutation on the ancestral line requires that the ancestral line occupy the top position in the graph just ‘before’ the event (in forward time). Relative to the neutral case (that is, without selection), we obtain a general bias towards the beneficial type, an increase in the beneficial mutation rate, and a decrease in the deleterious mutation rate. This sheds new light on previous analytical results.

We discuss our findings in the light of a well-known observation at the interface of phylogeny and population genetics, namely, the difference in the mutation rates (or, more precisely, mutation fluxes) estimated via phylogenetic methods relative to those observed in pedigree studies.

2. Introduction

 Phylogeny is concerned with species trees and basically works according to the following principle (for a review of the field, see Ewens and Grant (2005, Chs. 14 and 15)). One starts with a collection of contemporary species, samples one individual of each species, and sequences a gene (or a collection of genes) in each of these individuals. From this sequence information, one infers the species tree. This tree lives on the long time scale of (usually many) millions of years. If one zooms in on the species tree (see Figure 1), one sees population genetics emerge: species (or populations) consist of individuals, and the individual genealogies, on a shorter time scale of, say, a few hundreds of thousands of years, are embedded within the species tree. The connection is made by the ancestral line that starts from the randomly-chosen individual in each species and goes back into the past, beyond the most recent common ancestor of the population, and meets ancestral lines coming from other species.

 Coalescence theory is concerned with the individual genealogies within species. It is a major part of modern population genetics theory; for a review of the area, see Wakeley (2009). Our goal in this article is to investigate the ancestral line just described, and in this way shed some light on the interface between phylogeny and population genetics. Specifically, we will be concerned with the mutation process on the ancestral line. As a motivation, we address the well-known observation that mutation rates (or, more precisely, mutation fluxes) estimated via pedigree studies (that is, via direct comparison of close relatives) tend to be about a factor of ten higher than those inferred via
phylogenetic methods (Connell et al. 2022) and references therein; see specifically the early work by Parsons et al. (1997); Sigurðardóttir et al. (2000) and Santos et al. (2005)). One of the many possible explanations discussed is purifying selection (Parsons et al. 1997; Sigurðardóttir et al. 2000; Santos et al. 2005; Connell et al. 2022). This means that a deleterious mutation away from the wildtype is selected against; thus an individual carrying it has a lower chance of survival than a wildtype individual. So such mutations, although visible in pedigree studies, have a smaller probability of being observed in phylogenetic analyses.

In this article, we investigate the mutation process on the ancestral line in a prototype model of population genetics, namely the Moran model with two types under mutation and selection. We first describe the model forward in time, starting with the finite-population version and proceeding to its deterministic and diffusion limits (Section 3). In Section 4, we introduce our main tool, namely the pruned lookdown ancestral selection graph (see Baake and Wakolbinger (2018) for a review), which delivers the type distribution of the ancestor, in the distant past, of an individual sampled randomly from the present population. In Section 5, we use this concept to tackle the mutation process on the ancestral line. In the diffusion limit, we recover previous results obtained via analytical methods by Fearnhead (2002) and Taylor (2007) and give them a new meaning in terms of the graphical construction. Section 6 analyses in more detail the probability fluxes of mutation and coalescence events involving the ancestral line. Section 7 is devoted to exploring the dependence on the parameters; we discuss the biological implications in Section 8.

3. The Model

Consider a population consisting of a fixed number \( N \in \mathbb{N} \) of individuals and assume that each of them carries a type, which can be either 0 or 1. At any instant in continuous time, every individual may reproduce or mutate. Individuals of type 0 reproduce at rate \( 1 + s^N \) with \( s^N \geq 0 \) (where the superscript indicates the dependence on population size), while individuals of type 1 reproduce at rate 1. This means that individuals of type 0 have a selective advantage of \( s^N \); for this reason type 0 is referred to as the fit or beneficial type, whereas type 1 is the unfit or deleterious type. When an individual reproduces, it produces a single offspring, which inherits the parent’s type and replaces a uniformly chosen individual, possibly its own parent; this way, population size is kept constant. Every individual mutates at rate \( u^N > 0 \). When it does so, the
new type is \( j \in \{0, 1\} \) with probability \( \nu_j \), where \( 0 < \nu_j < 1 \) and \( \nu_0 + \nu_1 = 1 \). This allows for the possibility of silent mutations, where the type is the same before and after the event.

The Moran model can be represented as an interactive particle system (IPS) as in Figure 2. Every individual is represented by a horizontal line, lines are labelled 1, \ldots, \( N \) from top to bottom, time runs from left to right, and the events are indicated by graphical elements: reproduction events are depicted as arrows connecting two lines with the parent at the tail, the offspring at the tip, and the latter replacing the individual at the target line (arrows pointing to their own tails have no effect and are omitted); beneficial and deleterious mutation events are depicted by circles and crosses, respectively, on the lines. We will first describe the \emph{untyped} version of the IPS, where all graphical elements appear at constant rates, regardless of the types, see Figure 2. We distinguish two kinds of arrows: \emph{neutral} ones have filled arrowheads and appear at rate 1 per individual (or, equivalently, at rate \( 1/N \) per ordered pair of lines), while arrows with hollow heads refer to selective reproduction events and appear at rate \( sN/N \) per ordered pair of lines. Finally, circles (crosses) appear at rate \( uN\nu_0 \) (at rate \( uN\nu_1 \)) on every line. The untyped version may be turned into a typed one (see Figure 3) by assigning an initial type to every line at time 0 and then propagating the types forward according to the following rules. All individuals use the neutral arrows to place offspring, whereas selective arrows are used only by fit individuals and are ignored by unfit ones. A circle (a cross) turns the type into 0 (into 1). Now let \( Y^N_t \) be the number of type-1 individuals at time \( t \). The process \( Y^N = (Y^N_t)_{t \geq 0} \) is a continuous-time birth-death process on \([N]_0\) with transition rates

\[
q_{Y^N}(k, k + 1) = k\frac{N - k}{N} + uN\nu_1(N - k),
\]
\[
q_{Y^N}(k, k - 1) = (1 + sN)(N - k)\frac{k}{N} + uN\nu_0 k,
\]

where \( k \in [N]_0 \) (note that \( q_{Y^N}(N, N + 1) = 0 = q_{Y^N}(0, -1) \) by construction); throughout, for \( i, j \in \mathbb{N} \), we denote by \([i]\) the set \( \{1, \ldots, i\} \), by \([i]_0\) the set \( \{0, \ldots, i\} \), and by \([i : j]\) the set \( \{i, \ldots, j\} \). The (reversible) stationary distribution of \( Y^N \), namely \( \pi^N = (\pi^N_k)_{k \in [N]_0} \), is given by

\[
\pi^N_k := C^N \prod_{j=1}^{k-1} \frac{q_{Y^N}(j, j + 1)}{q_{Y^N}(j + 1, j)},
\]
where the empty product is 1 and $C_N$ is a normalising constant chosen so that $\sum_{k=1}^N \pi_k^N = 1$. We denote by $Y_{\infty}^N$ a random variable with distribution $\pi^N$.

One is usually interested in the limiting behaviour as $N \to \infty$. The most commonly used limits are the deterministic limit (or dynamical law of large numbers) and the diffusion limit, which we will briefly describe below. However, our emphasis will be on finite $N$ for the sake of more generality, so as to also allow for scalings intermediate between deterministic and diffusion. For example, the scaling with $N^\alpha$ for $0 < \alpha < 1$ has recently received considerable attention, see, for example, Boenkost et al. (2021, 2022) or Baake et al. (2023).

### 3.1. Diffusion limit

In the diffusion limit, the selective reproduction rate and the mutation rate depend on $N$ and satisfy

$$
\lim_{N \to \infty} Ns^N = \sigma, \quad \lim_{N \to \infty} Nu^N = \theta
$$

with $0 \leq \sigma, \theta < \infty$. Furthermore, it is assumed that $\lim_{N \to \infty} Y_{\infty}^N / N = \eta_0$. As $N \to \infty$, the process $(Y_{\infty}^N / N)_{N \geq 0}$ is then well known to converge in distribution to $Y := (Y_t)_{t \geq 0}$, the Wright–Fisher diffusion on $[0, 1]$ that solves the stochastic differential equation

$$
\frac{dY_t}{\sqrt{2}} = \left( -\sigma Y_t(1 - Y_t) - \theta \eta_0 Y_t + \theta \eta_1 (1 - Y_t) \right) dt + \sqrt{2} Y_t (1 - Y_t) dW_t
$$

(3.3)

with initial value $\eta_0$, where $(W_t)_{t \geq 0}$ is standard Brownian motion. For $\theta > 0$, the process has a stationary measure $\pi$, known as Wright’s distribution, with density

$$
\pi(\eta) := C \eta^{\theta \eta_1 - 1}(1 - \eta)^{\theta \eta_0 - 1} e^{-\sigma \eta}, \quad 0 < \eta < 1,
$$

(3.4)

where $C := \int_0^1 \eta^{\theta \eta_1 - 1}(1 - \eta)^{\theta \eta_0 - 1} e^{-\sigma \eta} d\eta$ is a normalising constant. Proofs of the above statements can be found in Durrett (2008, Chapter 7). Furthermore, we also have convergence of the stationary distribution,

$$
\pi^N \xrightarrow{N \to \infty} \pi,
$$

(3.5)

where the convergence is in distribution; indeed, the proof of the corresponding result for the Wright–Fisher model by Ethier and Kurtz (1986, Chapter 10, Theorem 2.2) works in exactly the same way for the Moran model. We denote by $Y_{\infty}$ a random variable that has the distribution $\pi$. 

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**Figure 3.** A typed version of the realisation in Figure 2.
3.2. Deterministic limit. In this case one lets $N \to \infty$ without any rescaling of time or of parameters; so $u^N \equiv u$ and $s^N \equiv s$. If $Y_0^N/N \to y_0 \in [0,1]$ when $N \to \infty$, then $(Y_t^N/N)_{t \geq 0}$ converges to $(y(t))_{t \geq 0}$, where $(y(t))_{t \geq 0}$ solves the Riccati differential equation

$$
\dot{y} = -sy(1-y) - u\nu_0 y + u\nu_1 (1-y)
$$

with initial value $y(0) = y_0$; the convergence is uniform on compact time intervals in probability (Cordero 2017b, Prop. 3.1). The solution, which is known explicitly (see for example Cordero 2017b), converges to the unique equilibrium

$$
y_{\infty} := \lim_{t \to \infty} y(t) = \begin{cases} 
\frac{1}{2} \left(1 + \frac{s}{2} - \sqrt{(1 - \frac{s}{2})^2 + 4\frac{s}{2} \nu_0}\right), & s > 0, \\
\nu_1, & s = 0,
\end{cases}
$$

independently of $y_0$. The law of large numbers carries over to the equilibrium in the sense that

$$
\pi_N \to \delta_{y_{\infty}}^{N \to \infty}
$$

in distribution (Cordero 2017b, Corollary 3.6), where $\delta_{y_{\infty}}$ denotes the point measure on $y_{\infty}$.

4. The pruned lookdown ancestral selection graph

In this section, we first recall the pruned lookdown ancestral selection graph in the finite case and then take the deterministic and the diffusion limits.

4.1. The finite case. Based on the the concept of ancestral selection graph (ASG) introduced by Krone and Neuhauser (1997) to study ancestries and genealogies of individuals (see also Etheridge (2011, Sec. 5.4)), the pruned lookdown ancestral selection graph (pLD-ASG) is the tool that allows us to study the ancestral line of an individual chosen randomly from a population at any given time $t \geq 0$, to which we refer as the present. It was introduced by Lenz et al. (2015) for the diffusion limit of the Moran model, extended to the finite population setting and the deterministic limit by Cordero (2017a, Section 4) and Baake et al. (2018), reviewed by Baake and Wakolbinger (2018), and extended to certain forms of frequency-dependent selection by Baake et al. (2023).

The idea behind it is to start with the untyped setting again, follow the history of a single individual (one of the lines at the right of Figure 2) backward in time, and keep track of all potential ancestors this individual may have. These potential ancestors are ordered according to a hierarchy that keeps track of which line will be the true ancestor once the types have been assigned to the lines at any given time point in the past. Therefore, at any time $r$ before the present, the pLD-ASG consists of a random number $L_r \in [N]$ of lines, the potential ancestors, with labels $1, \ldots, L_r$ that reflect the ordering. The ordering contains elements of the lookdown construction of Donnelly and Kurtz (1999) (see also Etheridge (2000, Sec. 5.3)), hence the name; and it is usually visualised by identifying the labels with equidistant levels (starting at level 1), on which the lines are placed. Here we will use a somewhat simpler visualisation in the spirit of the ordered ASG of Lenz et al. (2015), which also arranges lines from bottom to top according to their labels, but does not insist on equidistant spacing or a start at level 1. But this is only a minor difference in wording and visualisation of an underlying identical concept.

We throughout denote forward and backward time by $t$ and $r$, respectively; backward time $r$ corresponds to forward time $t - r$. The concept of the pLD-ASG is captured in the following definition, which is adapted from Cordero (2017a, Sec. 4.6) and Baake and Wakolbinger (2018, Def. 4) and illustrated in Figures 4 and 5.
Figure 4. Transitions in the finite-$N$ pLD-ASG. The vertical positions reflect the labels, with label 1 at the bottom and label $L_r^N$ at the top. The immune line is bold. From left to right: coalescence, branching, collision, deleterious mutation of a line that is not immune, deleterious mutation of the immune line, beneficial mutation.

Figure 5. A realisation of the finite-$N$ pLD-ASG. The immune line is bold, and the ancestral line is marked light blue.

Definition 4.1. The pruned lookdown ASG in a population consisting of $N$ individuals starts with a single line chosen uniformly from the population at backward time $r = 0$ and proceeds in direction of increasing $r$. At each time $r \geq 0$, the graph consists of a finite number $L_r^N$ of lines, one of which is distinguished and called immune. The lines are labelled $1, \ldots, L_r^N$ from bottom to top; the label of the immune line is $M_r^N \in [L_r^N]$ with $M_0^N = 1$. The process $(L_r^N, M_r^N) := (L_r^N, M_r^N)_{r \geq 0}$ lives on the state space $[N]^2$ and evolves via the following transitions when $(L_r^N, M_r^N) = (\ell, m)$ (see again Figure 4).

1. Coalescence. Every line $j \in [2 : \ell]$ coalesces into every line $i \in [1 : j - 1]$ at rate 2, that is, label $j$ is removed and $\ell$ decreases by one. The remaining lines are relabelled to fill the gap while retaining their original order. The event affects the immune line in the same way as all others.

2. Branching. Every line $i \in [1 : \ell]$ branches at rate $s_N r^\ell$, and a new line, namely the incoming branch (the one at the tail of the arrow) is inserted at label $i$ and all previous labels $k \in [i : \ell]$ increase to $k + 1$; in particular, the label of the continuing branch (the one at the tip of the arrow) increases from $i$ to $i + 1$, and $\ell$ increases to $\ell + 1$. If $m \in [i : \ell]$, then $m$ increases to $m + 1$; otherwise, it remains unchanged.

3. Collision. Every line labelled $i \in [1 : \ell]$ collides with the line labelled $j$ and is hence relabelled $j$, at rate $s_N r^\ell$ for every $j \in [1 : i - 1]$. Upon this, all labels previously in $[j : i - 1]$ increase by one. In this case, $\ell$ does not change, and the relabelling applies to the immune line in the same way as to all others.

4. Deleterious mutation. Every line $i \in [1 : \ell]$ experiences deleterious mutations at rate $u_N r_1$. If $i \neq m$, the line labelled $i$ is pruned, and the remaining lines (including the immune
line) are relabelled to fill the gap (again in an order-preserving way), thus rendering the
transition of \( \ell \) to \( \ell - 1 \). If, however, \( i = m \), then the line affected by the mutation is not
pruned, but \( m \) is set to \( \ell \), which remains unchanged. All labels previously in \([i+1: \ell]\)
decrease by one, so that the gaps are filled.

(5) Beneficial mutation. Every line \( i \in [1: \ell] \) experiences beneficial mutations at rate \( u^N \nu_0 \).
In this case all lines labelled \( > i \) are pruned, resulting in a transition from \( \ell \) to \( i \). Also \( m \)
is set to \( i \).

The line-counting process \( L^N := (L_t^N)_{r \geq 0} \), that is, the marginal of \((L_t^N, M_t)_{r \geq 0} \), is itself Markov,
with state space \([N]\). As a consequence of Definition 4.1, \( L^N \) has transition rates
\[
q_{L^N}(n, n+1) = s^N n \frac{N-n}{N}, \\
q_{L^N}(n, n-1) = n \frac{n-1}{N} + u^N \nu_1 (n-1) + u^N \nu_0 \mathbb{1}_{\{u>1\}}, \\
q_{L^N}(n, j) = u^N \nu_0,
\]
for \( n \in [N] \) and \( j \in [n-2] \).

The pLD-ASG encodes potential and true ancestry in the following way. One first samples the
initial types (that is, at forward time \( t = 0 \), so backward time \( r = t \)) for the \( L_t^N \) lines without
replacement according to \((N - Y_0^N, Y_0^N)\), which means a population with \( N - Y_0^N \) type-0 and
\( Y_0^N \) individuals; then one then propagates the types forward along the graph according to the
decay, branching, and mutation events as in the original Moran model. At any instant of
time, the true ancestor is the lowest type-0 line in the graph if there is such a line; when all
individuals are unfit, the ancestor is the immune line \((\text{Cordero 2017a, Prop. 4.6})\).

Let now \( A_r^N \in \{0, 1\} \) be the type of the true ancestor at backward time \( r \leq t \) of the single
individual chosen uniformly at \( r = 0 \). It is then clear that
\[
\mathbb{P}(A_r^N = 1 \mid Y_0 = k) = \sum_{n=1}^{N} \mathbb{P}(L_c^N = n) \frac{k_n}{N^N},
\]
where, for \( k \in \mathbb{Z} \) and \( n \in \mathbb{N} \), \( k_n := k(k-1) \ldots (k-n+1) \) is the falling factorial, and so
\[
\mathbb{P}(A_r^N = 1) = \sum_{n=1}^{N} \mathbb{P}(L_c^N = n) \mathbb{E}\left(\frac{(Y_0^N)^n}{N^N}\right),
\]
We are interested in the ancestral type in the distant past, that is, in the limit \( r \to \infty \) following
\( t \to \infty \). Thanks to being irreducible and recurrent, \( L^N \) converges to a unique stationary distribution
as \( r \to \infty \); we denote by \( L^N_{\infty} \) a random variable with this stationary distribution and denote
the latter by \( w^N := \left( w_n^N \right)_{n>0} \) with \( w_n^N := \mathbb{P}(L^N_{\infty} = n) \). It is well known \(\text{Cordero 2017a}\) that \( w^N \) is characterised via the following recursion for the tail probabilities \( a_n^N := \mathbb{P}(L^N_{\infty} > n) = \sum_{t>n} w_t^N \):
\[
\left( \frac{n+1}{N} + s \frac{N-n}{N} + u \right) a_n^N = \left( \frac{n+1}{N} + u \nu_1 \right) a_{n+1}^N + s \frac{N-n}{N} a_{n-1}^N
\]
for \( 0 < n < N \), together with the boundary conditions \( a_0^N = 1 \) and \( a_N^N = 0 \).

We additionally assume that, thanks to a long evolutionary prehistory, \( Y_0^N \) is stationary (that is,
have the distribution \( \pi^N \) of (3.2)); as a consequence, \( Y_t^N \) is stationary for all \( t \geq 0 \). It is then
clear from (4.3) that \( A_r^N \) will also become stationary as \( r \to \infty \). We write \( A^N_{\infty} \) for a random
variable with this stationary distribution. Due to (4.2) and (4.3), it satisfies
\[
\gamma_k^N := \mathbb{P}(A^N_{\infty} = 1 \mid Y_0 = k) = \sum_{n=1}^{N} w_n^N \frac{k_n}{N^N}
\]
and
\[ P(A_N^\infty = 1) = \sum_{n=1}^{N} w_n^N b_n^N, \tag{4.6} \]
where
\[ b_n^N := E(\frac{Y_N^N}{Nw}). \tag{4.7} \]

It will be shown in the Appendix (Section 9) that the \( b_n^N \) satisfy the sampling recursion
\[ \left(\frac{n-1}{N} + s \frac{N-n}{N} + u\right)b_n^N = \left(\frac{n-1}{N} + u^N \nu_1\right)b_{n-1}^N + s \frac{N-n}{N} b_{n+1}^N, \quad 0 < n \leq N, \tag{4.8} \]
which is complemented by the boundary conditions \( b_0^N = 1 \) and \( b_{N+1}^N = 0 \). Likewise, we can rewrite (4.3) as
\[ 1 - \gamma_k^N = P(A_N^\infty = 0 \mid Y_0 = k) = \sum_{n=1}^{N} a_{n-1}^N \frac{k^{n-1}}{Nw} (N - k), \tag{4.9} \]
which reflects the decomposition according to the lowest type-0 line. In words: the ancestral line is of type 0 if, for some \( n \), there are at least \( n \) lines, the first \( n - 1 \) receive type 1, and line \( n \) receives type 0. This leads to the counterpart of (4.6),
\[ P(A_N^\infty = 0) = \sum_{n=1}^{N} a_{n-1}^N (b_{n-1}^N - b_n^N), \tag{4.10} \]

since
\[ b_{n-1}^N - b_n^N = E\left(\frac{Y_N^N}{Nw}\right)(N - Y_N^\infty). \]

Also note for later use that, by (4.5) and (4.9),
\[ P(\text{any given type-1 individual is the ancestor} \mid Y_N^\infty = k) = \frac{\gamma_k^N}{k} = \sum_{n=1}^{N} w_n^N \frac{(k-1)^{n-1}}{Nw}, \tag{4.11} \]
as well as
\[ P(\text{any given type-0 individual is the ancestor} \mid Y_N^\infty = k) = 1 - \gamma_k^N = \sum_{n=1}^{N} a_{n-1}^N \frac{k^{n-1}}{Nw}; \tag{4.12} \]
see also Khuth et al. (2013), where these relations were derived via a descendant process forward in time.

Let us now compare \( P(A_N^\infty = 1) \), the probability for an unfit ancestor, with \( b_1^N \), which, by (4.7), is the expected probability to sample an unfit individual when drawing a single individual from the present population. Both quantities are presented as functions of \( s^N \) in Figure 6. To produce this plot (and the others contained in this paper), we have computed the \( a_n^N \) and \( b_n^N \) by solving numerically the linear systems given by the recursions (4.4) and (4.8), together with the approximation \( a_n^N = b_n^N = 0 \) for \( n \geq 100 \). This yields accurate results since, for small \( s^N \) and large \( n \), the \( a_n^N \) and \( b_n^N \) decrease to zero rapidly, as confirmed by the fact that the results do not change by moving the truncation point to, for example, 50 or 200.

It is clear that, for any \( s^N > 0 \), we have \( P(A_N^\infty = 1) < b_1^N \), simply because \( b_n^N < b_1^N \) for \( n > 1 \), and so \( \sum_{n=1}^{N} w_n^N b_n^N < b_1^N \) (compare (4.7) and (4.6)). This reflects the fact that an unfit individual has a lower chance that its descendants survive in the long run than does a fit individual; this produces the bias towards type 0 in the past relative to the present.
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Figure 6. The expected proportion of type-1 individuals in the population (that is, \(b_N^Y = \mathbb{E}(Y^N/N)\)) and the probability of a type-1 ancestor, in the finite case as a function of \(s^N\). Further parameters: \(N = 10^4, w^N = 8 \cdot 10^{-4}, \nu_1 = 0.99\).

4.2. Diffusion limit. In the diffusion limit, the pLD-ASG of Definition 4.1 changes in the obvious way: (1) coalescence events occur as before (that is, every line coalesces into every line below it at rate 2), (2) branching events happen at rate \(\sigma\) to every line, (3) collision events are absent, (4) deleterious mutations and (5) beneficial mutations happen at rate \(\theta \nu_1\) and \(\theta \nu_0\) per line, respectively, and so do the resulting pruning and relocation operations. In each of these events, the immune line behaves as in the finite case. See Lenz et al. (2015) and Baake and Wakolbinger (2018) for a comprehensive description. Here we restrict ourselves to the process \((L, M) := (L_r, M_r)_{r \geq 0}\), where \(L_r\) and \(M_r\) are the number of lines and the label of the immune line of the limiting pLD-ASG at backward time \(r\). \((L, M)\) lives on the state space \(\mathbb{N}^2\), and the marginal \(L := (L_r)_{r \geq 0}\) is again Markov with rates that emerge as the limit of the rates (4.1) with the proper rescaling of time and parameters:

\[
q_L(n, n + 1) = n\sigma, \\
q_L(n, n - 1) = n(n - 1) + (n - 1)\theta \nu_1 + \theta \nu_0 \mathbb{1}_{n>1}, \\
q_L(n, j) = \theta \nu_0,
\]

with \(j \in [n - 2], n \in \mathbb{N}\). The process \(L\) is non-explosive (because it is dominated by a Yule process with parameter \(\sigma\), which is non-explosive), and the sequence of processes \((L^N_{Nt})_{r \geq 0}\) converges to it in distribution as \(N \to \infty\); this is a special case of Proposition 2.18 of Baake et al. (2023). The ancestral type \(A_r \in \{0, 1\}\) at backward time \(r \leq t\) is constructed in analogy with the finite case, except that one works with the pLD-ASG in the diffusion limit and samples the types according to \((1 - Y^\infty, Y^\infty)\) in an i.i.d. manner.

The process \(L\) again has a unique stationary distribution; we denote by \(\mathbb{L}_\infty\) a random variable with this stationary distribution and let \(\omega := (\omega_n)_{n \geq 0}\) with \(\omega_n := \mathbb{P}(L_\infty = n) = \lim_{N \to \infty} w^N_n\). It is well known (Lenz et al. 2015) that the tail probabilities \(\alpha_n := \mathbb{P}(L_\infty > n) = \lim_{N \to \infty} a^N_n\) are the unique solution to the recursion

\[
(n + 1 + \sigma + \theta)\alpha_n = (n + 1 + \theta \nu_1)\alpha_{n+1} + \sigma \alpha_{n-1} \quad \text{for } n > 0,
\]

(4.14) together with \(\alpha_0 = 1\) and \(\lim_{n \to \infty} \alpha_n = 0\). Due to (3.5), we also have that \(b^N_n \to \beta_n\) as \(N \to \infty\) for \(n \geq 0\), where \(\beta_n := \mathbb{E}(Y^\infty_n)\). The \(\beta_n\) satisfy the sampling recursion

\[
(n - 1 + \sigma + \vartheta)\beta_n = \sigma \beta_{n+1} + (n - 1 + \vartheta \nu_1)\beta_{n-1}, \quad n \in \mathbb{N},
\]

(4.15)
complemented by the boundary conditions $\beta_0 = 1$ and $\lim_{n \to \infty} \beta_n = 0$, see Krone and Neuhauser (1997) or Baake and Wakolbinger (2018).

For $r \to \infty$, the ancestral type becomes stationary again; we denote by $A_\infty$ a random variable with this stationary distribution. The stationary probability of a type-1 ancestor is given by the analogue of (4.6), that is,

$$P(A_\infty = 1) = \sum_{n=1}^{\infty} \omega_n \beta_n,$$

and the analogue of (4.10) reads

$$P(A_\infty = 0) = \sum_{n=1}^{\infty} \alpha_{n-1}(\beta_{n-1} - \beta_n).$$

4.3. Deterministic limit. In the deterministic limit, the pLD-ASG of Definition 4.1 changes as follows. (1) coalescence events are absent, (2) every line experiences branching events at rate $s$, (3) collisions are absent, (4) deleterious mutations and (5) beneficial mutations happen at rates $w_1$ and $w_0$, respectively, on every line, and so does the resulting pruning. In each of these events, the immune line behaves as in the finite case; see Cordero (2017a) and Baake and Wakolbinger (2018).

It is important to note that, in this limit, the immune line is always the top line in the graph, that is at level $L_r$ for all $r \geq 0$. This is because coalescence events are absent, and thus there is no opportunity to move the immune line away from the top. It therefore suffices to consider the limiting line-counting process $L := (L_r)_{r \geq 0}$, which has state space $\mathbb{N}$ and is characterised by the rates (obtained in the limit from (4.1)):

$$q_L(n, n+1) = ns,$$

$$q_L(n, n-1) = (n-1)w_1 + w_0 \mathbb{1}_{\{n>1\}},$$

$$q_L(n, j) = w_0,$$

with $j \in [n-2], n \in \mathbb{N}$. The sequence of line-counting processes $(L^N)_{N>0}$ converges in distribution to $L$ as $N \to \infty$ (Cordero 2017a, Prop. 5.3). The limiting ancestral type $A_r \in \{0, 1\}$ at backward time $r \leq t$ is constructed in analogy with the finite case, except that one works with the pLD-ASG in the deterministic limit and samples the types according to $(1 - y_\infty, y_\infty)$ in an i.i.d. manner.

The process $L$ is once more non-explosive and has a unique stationary distribution (Cordero 2017a, Lemmas 5.1 and 5.2). We denote by $L_\infty$ a random variable with this distribution, and write $w := (w_n)_{n \geq 0}$ with $w_n := P(L_\infty = n) = \lim_{N \to \infty} w_n^N$. The tail probabilities now are $a_n := P(L_\infty > n) = \lim_{N \to \infty} a_n^N$. Due to the absence of coalescences and collisions in the pLD-ASG, the distribution is available explicitly, see Baake et al. (2018, Prop. 11) and Baake and Wakolbinger (2018, Prop. 7):

$$w_n = p^{n-1}(1 - p), \quad a_n = p^n$$

(so $L_\infty$ has the geometric distribution $Geo(1-p)$ with parameter $p$), where

$$p := \begin{cases} \frac{1}{2} \left( \frac{w_1}{w_1} - \sqrt{\left( \frac{w_1}{w_0} \right)^2 - 4 \frac{w_0}{w_1}} \right), & \nu_1 > 0, \\ \frac{1}{2} \frac{w_0}{w_1}, & \nu_1 = 0. \end{cases}$$

This also includes the limiting case $p = 0$ for $s = 0$, where $L_\infty \equiv 1$; the latter is due to the fact that there are neither branching nor coalescence nor pruning events. Furthermore, due to (3.8), we also have $b_n := \lim_{N \to \infty} b_n^N = y_\infty^p$.

For $r \to \infty$, the ancestral type becomes stationary again; we denote by $A_\infty$ a random variable with this stationary distribution. The stationary probability of a type-1 ancestor is given by the
analogue of (4.6), that is,
\[ P(A_\infty = 1) = \sum_{n=1}^{\infty} w_n b_n, \] (4.20)
and the analogue of (4.10) reads
\[ P(A_\infty = 0) = \sum_{n=1}^{\infty} a_{n-1}(b_{n-1} - b_n). \]

5. The type process on the ancestral line

In this section, we analyse the mutation process of the ancestral line. We again start with the finite case and, from there, proceed to the diffusion and deterministic limits.

5.1. The finite case. Consider the ASG decorated with the mutation events but without pruning and start it with all \( N \) individuals of the population, rather than with a single individual as assumed so far, see Figure 7. In analogy with the start with a single line, this captures all potential ancestors of the \( N \) individuals. Since the corresponding line-counting process is positive recurrent, it will, with probability 1, reach a state with just a single line (a bottleneck) in finite time. At this point (at the latest), all individuals in the population share a common ancestor. The line of true ancestors leading back into the past from the most recent common ancestor onwards is defined as the common ancestor line (Fearnhead 2002; Taylor 2007; Lenz et al. 2015). The common ancestor type process is the mutation process on this very line, in the forward direction of time. The ancestral line together with its type can be constructed via a two-step (backward-forward) scheme in any finite time window \([0, T]\), in a way analogous to that described below (4.1). To this end, denote by \( t, r \in [0, T] \) the forward and backward running time, respectively (i.e. \( r = T - t \)). Run the pLD-ASG from time \( r = 0 \) until time \( r = T \) started with the \( N \) lines, assign a type to each of the \( L_T^N \) lines by sampling without replacement from \((N - Y_0^N, Y_0^N)\), determine the ancestral line at time \( r = T \) (as the lowest type-0 line if there is such a line, or the immune line otherwise), and propagate types and ancestry forward as in the original Moran model until time \( r = 0 \). At any time \( r \), the true ancestral line is, by construction, the lowest type-0 line

\[ T \quad r_b \quad 0 \]

**Figure 7.** Sketch of a population of size 8, its (unpruned) ASG decorated with mutations, the common ancestor line (bold), and the common ancestor type process. The first bottleneck is indicated by \( r_b \). The coloured lines (dark green for type 0, light brown for type 1) are the true ancestors. The thin black lines are ASG lines that turn out as nonancestral. Among them, the dotted black lines are pruned away in the pLD-ASG.
Figure 8. A realisation of the untyped Moran IPS in \([0, T]\) for \(N = 5\) enriched with the (forward) type configuration \((\xi^N_t)_{t \in [0,T]}\) (given by the colours of the lines: dark green for type 0, light brown for type 1) and the (backward) label processes \((\eta^N_r)_{r \in [0,T]}\) (given by numbers between \(\{0, \ldots, 5\}\) located above each line; the immune line has the additional label \(\star\)). The ancestral line (light blue) is extracted from the typed and labelled IPS by the rule that it is, at any time, the line with the lowest label \(i > 0\) with type 0, or the immune line otherwise.

Our aim is to construct the common ancestor type process at stationarity in the forward direction of time. Let us anticipate that this process is not Markov; the additional information contained in the graphical representation is required to turn it into a Markov process. As before, let us first consider a finite time window \([0, T]\). We denote by \(G_T\) the untyped Moran IPS in \([0, T]\) and by \(G_T(I)\) its restriction to \(I \subseteq [0, T]\). Recall that, given an initial type configuration of the lines present at time 0 in \(G_T\), types can be propagated forward in time along the lines in the IPS by respecting the mutation and reproduction events; the so-constructed type-configuration process \((\xi^N_t)_{t \in [0,T]}\) \((\xi_t \in \{0, 1\}^N\) contains the types on all lines at time \(t)\) is coupled to \(G_T\). The pLD-ASG can also be coupled to (embedded in) \(G_T\) by assigning, at any time \(r\), a label in \(\{0, \ldots, N\}\) to each line in the following way. A label 0 means that the line is currently not in the pLD-ASG, whereas a label \(i \geq 1\) indicates the level of that line in the pLD-ASG; an additional label \(\star\) is used to identify the immune line. We call the resulting process \((\eta^N_r)_{r \in [0,T]}\) the label process. This construction allows us to embed the (forward) type-configuration and the (backward) label processes in the same picture (i.e. coupled through \(G_T\)), see Fig. 8. The process \(A^N := (A^N_t)_{t \in [0,T]}\), defined via \(A^N_t := (\xi^N_t, \eta^N_{(T-t)-})\), contains all the information required to determine at any time \(t \in [0, T]\): (1) the number \(Y^N_t\) of individuals of type 1, (2) the number \(\bar{L}^N_t := \bar{L}^N_{T-t}\) of lines in the time-reversed pLD-ASG, (3) the label \(\bar{M}^N_t := \bar{M}^N_{T-t}\) of the immune line, and (4) the type \(A^N_t \in \{0, 1\}\) of the ancestral line.

By construction, the label process is irreducible, and due to the existence of bottlenecks, it is also positive recurrent. Hence it has a unique stationary distribution. Assume now that \(\xi^N_0\) and \(\eta^N_0\) are chosen independently according to their stationary distributions. Since \(\xi^N_t\) and \(\eta^N_{(T-t)-}\) are deterministic functions of their initial configurations and of \(G_T([0, t])\) and \(G_T((t, T])\), respectively,
they are independent (for any fixed time $t$, but not as processes). Thus, the law of $\Lambda_N^t$ is invariant in time and has product form (the product of the corresponding stationary distributions). It is not difficult to prove that, started with this invariant distribution, the process $\Lambda_N^t$ has Markovian transitions (and, in particular, we can define it in $[0, \infty)$). Since we are only interested in the transitions of $\Lambda_N^t$ involving a type change in the ancestral line, we refrain from giving a full description of the rates, and rather focus on the marginal rates of the (non-Markovian) process $A^N$ in the sense of the following definition.

**Definition 5.1.** Consider a stationary Markov process $X := (X_t)_{t \geq 0}$, where $X$ has a finite state space $E$. Let $g : E \rightarrow F$ be a surjective bounded function. Suppose the rate of $X$ from $x$ to $x' \neq x$ is given by $q_X(x, x')$. We define the marginal probability fluxes of $Z := g(X)$ (which may be non-Markovian) as

$$f_Z(z, z') := \sum_{x \in g^{-1}(\{z\}), x' \in g^{-1}(\{z'\})} q_X(x, x') \pi_X(x), \quad z \neq z', \quad (5.1)$$

where $\pi_X$ is the stationary distribution of $X$. The corresponding marginal rates are understood to be

$$q_Z(z, z') := \frac{f_Z(z, z')}{\pi_X(g^{-1}(\{z\}))}. \quad (5.2)$$

**Remark 5.2.** It is easily verified that $f_Z(z, z')$ and $q_Z(z, z')$ also satisfy

$$f_Z(z, z') = \lim_{\varepsilon \to 0} \frac{1}{\varepsilon} P(Z_{t+\varepsilon} = z' | Z_t = z) \quad \text{and}$$

$$q_Z(z, z') = \lim_{\varepsilon \to 0} \frac{1}{\varepsilon} P(Z_{t+\varepsilon} = z' | Z_t = z) \quad (5.3)$$

for any $t \geq 0$; they thus agree with the standard notions of flux and rate if $Z$ itself is Markov.

**Remark 5.3.** If the process $Z$ of Definition 5.1 is of the form $g(h(X))$, where now $h : E \rightarrow H$ and $g : H \rightarrow F$ are surjective bounded functions, then one can compute the marginal fluxes and rates of $Z$ via an intermediate marginalisation step (i.e. first computing the marginal rates of $Y = h(X)$ and then the marginal rates of $g(Y)$) and obtain (5.1) and (5.2).

After these preparations, we can provide explicit expressions for the marginal fluxes and rates of the pair $(\check{L}^N, A^N)$ and of $A^N$ at stationarity.

**Theorem 5.4.** In the marginal process $(\check{L}^N, A^N)$ at stationarity, the transition rates involving a beneficial $(1 \rightarrow 0)$ or deleterious $(0 \rightarrow 1)$ mutation on the ancestral line are given by

$$q_{(\check{L}^N, A^N)}((\ell, 1), (\ell + k, 0)) = u^N \nu_0 \frac{w^N_{\ell+k}}{w^N_{\ell}}, \quad \ell \in [N], \quad k \in [N - \ell]_0, \quad (5.5)$$

and

$$q_{(\check{L}^N, A^N)}((\ell, 0), (\ell, 1)) = u^N \nu_1 \frac{b^N_{\ell-1} - b^N_{\ell}}{\sum_{j=1}^N (b^N_{\ell-1} - b^N_j)}, \quad \ell \in [N]. \quad (5.6)$$

The marginal mutation fluxes are

$$f_{A^N}(1, 0) = u^N \nu_0 \sum_{n=1}^N b^N_{n-1} a^N_n b^N_n = u^N \nu_1 \sum_{n=1}^N w^N_n (b^N_{n-1} - b^N_n) = f_{A^N}(0, 1), \quad (5.7)$$

and the marginal mutation rates read

$$q_{A^N}(1, 0) = u^N \nu_0 \frac{\sum_{n=1}^N a^N_{n-1} b^N_n}{\sum_{k=1}^N w^N_k b^N_k} \quad \text{and} \quad q_{A^N}(0, 1) = u^N \nu_1 \frac{\sum_{n=1}^N w^N_n (b^N_{n-1} - b^N_n)}{\sum_{k=1}^N a^N_{k-1} (b^N_{k-1} - b^N_k)}. \quad (5.8)$$
We will give a graphical proof, which is based on the following crucial observation. It is obvious from Figures 5 and 9 that the ancestral line can display a mutation only if, right before the mutation (in the forward direction, that is, reading the figures from left to right), the ancestral line is at the top and coincides with the immune line. Indeed, the pruning of all lines above a beneficial mutation in the backward direction, and the induced movement of the immune line to the top, means that, forward in time, beneficial mutations can only appear on a top line that is immune. And the only deleterious mutations on the ancestral line are those appearing on the immune line. But a deleterious mutation on the immune line induces movement of the latter to the highest level (in the backward direction), which means that deleterious mutations, too, can only be observed on an immune top line when going forward. With this observation in mind we are ready for the proof.

Proof of Theorem 5.4. Let \( \varepsilon > 0 \) and consider the joint event \( \{ \tilde{L}_t^N = \ell, A_t^N = 1, \tilde{L}_{t+\varepsilon}^N = \ell + k, A_{t+\varepsilon}^N = 0 \} \) for some \( \ell \in [N] \) and \( k \in [N - \ell]_0 \), as shown in the left panel of Figure 9. Up to terms of order \( O(\varepsilon) \), this event happens whenever \( \tilde{E}_t^N = \ell + k \) (that is, at the right of the picture; probability \( \nu_{t, k}^N \)), a beneficial mutation occurs on line \( \ell \) between \( t \) and \( t + \varepsilon \) (probability \( \varepsilon u^N \nu_0 \)) and all \( \ell \) lines are unift at the left of the picture (probability \( b_\ell^N \)). Indeed, since the immune line is at the top to the left of the mutation, this ensures that line \( \ell \) is ancestral and has type 1 to the left and type 0 to the right of the event. The corresponding probability thus reads

\[
P(\tilde{L}_t^N = \ell + k, A_{t+\varepsilon}^N = 0, \tilde{L}_{t+\varepsilon}^N = \ell, A_{t+\varepsilon}^N = 1) = \varepsilon u^N \nu_0 \nu_{t+k}^N b_\ell^N + O(\varepsilon). \tag{5.9}
\]

In a similar way, the joint event \( \{ \tilde{L}_{t+\varepsilon}^N = \ell, A_{t+\varepsilon}^N = 1, \tilde{L}_t^N = k, A_t^N = 0 \} \), as shown in the right panel of Figure 9, occurs when, up to terms of order \( O(\varepsilon) \), \( \tilde{E}_{t+\varepsilon}^N = \ell \) (probability \( \nu_{t}^N \)), a deleterious mutation event occurs on the immune line between \( t \) and \( t + \varepsilon \) (probability \( \varepsilon u^N \nu_1 \)), and line \( \ell \) to the left of the event is the lowest type-0 level (probability \( b_{\ell-1}^N - b_{\ell}^N \)). Indeed, since the immune line is relocated to the top due to the mutation, this combination of events ensures that line \( \ell \) at the left is ancestral and has type 0 to the left and type 1 to the right of the event. The corresponding probability thus reads

\[
P(\tilde{L}_{t+\varepsilon}^N = \ell, A_{t+\varepsilon}^N = 1, \tilde{L}_t^N = k, A_t^N = 0) = \varepsilon u^N \nu_1 \nu_{t+k}^N (b_{\ell-1}^N - b_{\ell}^N) + O(\varepsilon). \tag{5.10}
\]

Eqs. (5.5) and (5.6) follow immediately from (5.9) and (5.10) via (5.4) since \( P(\tilde{L}_t^N = \ell, A_t^N = 1) = \nu_0^N b_\ell^N \) and \( P(\tilde{L}_t^N = \ell, A_t^N = 0) = \nu_1^N \sum_j (b_{j-1}^N - b_j^N) \), respectively. We get to the marginal mutation fluxes by marginalising over \( \ell \) (and \( k \) in (5.9) and (5.10) using (5.3)); the marginal fluxes are equal due to stationarity. They lead to the marginal rates via Eqs. (4.6), (4.10), and (5.2).

It is important to emphasise that the rates \( q_{AN}(1, 0) \) and \( q_{AN}(0, 1) \) are averaged over \( \tilde{L}^N \). And let us mention an alternative argument that leads to the mutation fluxes via the prospective view.

Figure 9. Beneficial and deleterious mutations on the ancestral line. Dark green: type 0; light brown: type 1; bold: immune line.
Namely, conditional on $Y_N^\infty = k$, any of the $k$ unfit individuals turns into a fit one at rate $u^N\nu_0$, and then it is ancestor with the probability (4.12); the latter automatically implies that the unfit predecessor is also ancestral. We therefore get

$$f_{A^N}(1, 0) = u^N\nu_0 \sum_{k=1}^{N} \sum_{n=1}^{N} a_{n-1}^N \frac{(k-1)^{n-1}}{N^k} P(Y_N^\infty = k)$$

$$= u^N\nu_0 \sum_{n=1}^{N} a_{n-1}^N b_n^N.$$ 

Likewise, with (4.11) for deleterious mutations,

$$f_{A^N}(0, 1) = u^N\nu_1 \sum_{k=1}^{N} (N-k) \sum_{l=1}^{N} w_l^N \frac{k^{n-1}}{N^k} P(Y_N^\infty = k)$$

$$= u^N\nu_1 \sum_{n=1}^{N} w_n^N (b_n^{N-1} - b_n^N).$$

5.2. Diffusion limit. Since the process $\mathcal{L}$ is once more positive recurrent, the notion of common ancestor is again well posed in the diffusion limit (Lenz et al. 2015), and in the long term, the common ancestor agrees with the true ancestor in our construction. Let us now consider the fluxes and rates along the (common) ancestor line in this limit, which are defined as $f_A(1, 0) := \lim_{N \to \infty} Nf_{A^N}(1, 0)$, $q_A(1, 0) := \lim_{N \to \infty} Nq_{A^N}(1, 0)$ (due to the rescaling of time), and likewise for $f_A(0, 1)$ and $q_A(0, 1)$. As a simple consequence of Theorem 5.4, we obtain

**Corollary 5.5.** The marginal mutation fluxes and rates of the common ancestor type process in the diffusion limit at stationarity read

$$f_A(1, 0) = \theta \nu_0 \sum_{n=0}^{\infty} \alpha_{n-1} \beta_n,$$

$$f_A(0, 1) = \theta \nu_1 \sum_{n=0}^{\infty} \omega_n (\beta_{n-1} - \beta_n),$$

$$q_A(1, 0) = \theta \nu_0 \sum_{n=0}^{\infty} \alpha_{n-1} \beta_n, \text{ and}$$

$$q_A(0, 1) = \theta \nu_1 \sum_{n=0}^{\infty} \omega_n (\alpha_{n+1} - \alpha_n)(\beta_{n+1} - \beta_n)$$

with $\alpha_n$, $\beta_n$, and $\omega_n$ as in Section 4.2.

**Proof.** The explicit expressions follow from (5.7) and (5.8) together with the convergence of $Nu^N$ to $\theta$ and the $a_{n}^N$ and $b_{n}^N$ to $\alpha_n$ and $\beta_n$, respectively. 

Note that the mutation rates in the corollary were already discovered by (Fearnhead 2002, Corollary 2) by mainly analytical methods; rederiving them here on the grounds of the pLD-ASG greatly simplifies the calculation and provides a transparent and intuitive meaning. Note also that the non-Markovian nature of the process prevails in the diffusion limit.

5.3. Deterministic limit. In contrast to the diffusion limit and the finite case, a common ancestor of the entire population does not exist in the deterministic limit; this is due to the absence of coalescence events. Nevertheless, the ancestral line of a randomly-chosen individual continues to be well defined. Let us now consider the rates along this line, which are defined as
\[ q_A(1,0) := \lim_{N \to \infty} q_{A^n}(1,0), \] and likewise for \( q_A(0,1). \) They once more follow from Theorem 5.4.

**Corollary 5.6.** The marginal mutation rates of the type process on the ancestral line in the deterministic limit at stationarity read

\[
q_A(1,0) = u\nu_1 \frac{1}{1-p} \quad \text{and} \quad q_A(0,1) = u\nu_1 (1-p)
\]

with \( p \) of (4.19).

**Proof.** The result is immediate if we take the limits in (5.8) and use (3.8) as well as (4.18):

\[
q_A(1,0) = u\nu_1 \sum_{n=0}^{\infty} w_n y_n^{(1)} = u\nu_1 \frac{1}{1-p} \quad \text{and}
\]

\[
q_A(0,1) = u\nu_1 \sum_{n=0}^{\infty} w_n y_n^{(1)(1)} = u\nu_1 (1-p)
\]

with \( a_n, b_n, \) and \( w_n \) as in Section 4.3. \( \Box \)

**Remark 5.7.** Let us mention that the resulting process is similar in spirit to the retrospective process of Georgii and Baake (2003, Definition 3.1), which describes the mutation process on the ancestral line of a typical individual in a multitype branching process. Despite the different model, the retrospective process (when specialised to 2 types and \( \lambda = 0 \)) is closely related to the one described here, since \( \gamma(y_\infty)/y_\infty \) and \((1 - \gamma(y_\infty))/(1 - y_\infty)\) have the meaning of the long-term offspring expectation of a type-1 and a type-0 individual, respectively, in a stationary population.

### 6. Flux identities

It is clear (and has already been mentioned in Theorem 5.4) that, at stationarity, the mean beneficial and deleterious mutation fluxes on the ancestral line balance each other. However, this is anything but obvious from the explicit expressions. Specifically, it turns out that, in general, the equality in (5.7) does not hold term by term, that is,

\[
f_{A^N}^{(n)}(1,0) := u^N \nu_0 a_n (b_n^{N} - b_n^{N-1}) \neq u^N \nu_1 w_n (b_n^{N} - b_n^{N-1}) =: f_{A^N}^{(n)}(0,1), \quad n > 0,
\]

as illustrated in Figure 10. Hence, the beneficial and deleterious mutation fluxes do not balance each other on every individual line. It is therefore interesting to investigate these individual fluxes.

**Proposition 6.1** (flux identities). In the finite system, the individual fluxes at stationarity satisfy

\[
f_{A^N}^{(n)}(1,0) + \sum_{i=n+1}^{N} w_i^N (b_i^{N} - b_{i-1}^{N}) = f_{A^N}^{(n)}(0,1) + (n-1)w_n^N (b_n^{N} - b_{n-1}^{N}), \quad n > 0.
\]

**Proof.** We start from the recursions (4.4) and (4.8) and rewrite them as

\[
(i+1)_N + u^N \nu_1) (a_i^{N} - a_{i+1}^{N}) = s_N \frac{N-i}{N} (a_{i-1}^{N} - a_i^{N}) - u^N \nu_0 a_i^{N}, \quad 0 < i < N,
\]

\[
(i-1)_N + u^N \nu_1) (b_i^{N} - b_{i-1}^{N}) = s_N \frac{N-i}{N} (b_{i-1}^{N} - b_i^{N}) + u^N \nu_0 b_i^{N}, \quad 0 < i \leq N.
\]

We now introduce

\[
S_n := \sum_{i=n}^{N} (a_i^{N} - a_{i-1}^{N})(b_{i-1}^{N} - b_i^{N}).
\]

We then have

\[
\sum_{n=1}^{N} S_{n+1} = \sum_{i=1}^{N} (i-1)(a_{i-1}^{N} - a_i^{N})(b_{i-1}^{N} - b_i^{N}).
\]
over pushing the first fit line upwards (forward in time) equals the flux of deleterious mutations at level $n$ together with one-half the flux of coalescences related to arrows sent out by an unfit line $\nu$. Summing over $i \in [k : N]$ on both sides leads to

$$u^N \nu_0 a_{n-1}^N b_n^N = u^N \nu_0 a_{N+k}^N b_{N+1}^N + \frac{1}{N} S_{n+1}$$

considering that $a_N^N = a_{N+1}^N = 0 = b_{N+1}^N$, this yields the claim.

Note that the proposition says that the individual fluxes balance each other together with certain coalescence events, see Figure 11. In detail, the flux of beneficial mutations at level $n$ together with one-half the flux of coalescences related to arrows sent out by an unfit line $n$ and pushing the first fit line upwards (forward in time) equals the flux of deleterious mutations at level $n$ together with one-half the flux of coalescences that push line $n$ upwards when it is the first fit line. Summing both sides in the statement of Proposition 6.1 over $n$, we find:

$$u^N \nu_0 \sum_{n=1}^{N} a_{n-1}^N b_n^N$$

$$= \frac{1}{N} \sum_{n=1}^{N} (n-1) w_n^N (b_{n-1}^N - b_n^N) - \frac{1}{N} S_{n+1} + \sum_{n=1}^{N} u^N \nu_1 w_n^N (b_{n-1}^N - b_n^N),$$

Figure 10. The individual mutation fluxes of (6.1). Parameters: $N = 10^4$, $s^N = 1.5 \cdot 10^{-3}$, $u^N = 8 \cdot 10^{-4}$, $\nu_1 = 0.99$. Using (6.2) in the first step and (6.3) in the last one, we get for $i \geq 1$:

$$u^N \nu_0 a_{i}^N + \left(\frac{i+1}{N} + u^N \nu_1\right)(a_{i}^N - a_{i+1}^N) \frac{b_{i}^N - b_{i+1}^N}{N^i} + u^N \nu_0(a_{i-1}^N - a_i^N)b_i^N$$

$$= s^N \left[ \frac{i}{N^i} (a_{i-1}^N - a_i^N)(b_{i}^N - b_{i+1}^N) + u^N \nu_0(a_{i-1}^N - a_i^N)b_i^N \right]$$

$$= \left(\frac{a_{i-1}^N}{a_{i}^N} - \frac{a_i^N}{a_{i+1}^N}\right) \frac{b_{i}^N - b_{i+1}^N}{N} \left(\frac{i-1}{N} + u^N \nu_1\right).$$

This can be rewritten as

$$u^N \nu_0(a_{i-1}^N b_i^N - a_i^N b_{i+1}^N) + \frac{1}{N} (a_{i-1}^N - a_i^N) (b_i^N - b_{i+1}^N)$$

$$= \left(\frac{a_{i-1}^N}{a_{i}^N} - \frac{a_i^N}{a_{i+1}^N}\right) \frac{b_{i}^N - b_{i+1}^N}{N} \left(\frac{i-1}{N} + u^N \nu_1\right) - \left(\frac{i}{N} + u^N \nu_1\right)(a_{i-1}^N - a_i^N)(b_i^N - b_{i+1}^N).$$
which is the identity of the fluxes in Theorem 5.4, as it must be.

The following corollary is an immediate consequence of Proposition 6.1.

**Corollary 6.2.** In the diffusion limit at stationarity, the individual fluxes of Proposition 6.1 turn into

\[
\theta \nu_0 \sigma_{n-1} + \sum_{i>n} \omega_i (\beta_i - \beta_i) = (\theta \nu_1 + (n - 1)) \omega_n (\beta_{n-1} - \beta_n), \quad n > 0,
\]

while in the deterministic limit at stationarity, they read

\[
u_0 \sigma_{n-1} b_n = \nu_1 w_n(b_{n-1} - b_n), \quad n > 0,
\]

that is, the mutation fluxes do balance each other on every line in this case.

7. Parameter dependence

The crucial parameter dependence is the effect of the branching rate on the tail probabilities, the sampling probabilities, and the marginal mutation rates and fluxes.

7.1. The tail probabilities.

**Proposition 7.1.** The tail probabilities \(a_n^N\) (for \(n \in [N]\)), \(a_n\) (for \(n \in \mathbb{N}\)), and \(a_n\) (for \(n \in \mathbb{N}\)) are strictly increasing in \(s, \sigma\), and \(s\), respectively.

**Proof.** We start with the finite case. To this end, we consider two copies of \(L^N\), namely \(L^{N,1}\) and \(L^{N,2}\) with branching rates \(s^{N,1}\) and \(s^{N,2} > s^{N,1}\), respectively, and couple them on the same probability space. More precisely, we consider the process \((L^{N,1}, L^{N,2})\) on \([N] \times [N]\) with transitions

\[
(n, m) \to (n + 1, m + 1) \quad \text{at rate} \quad s^{N,1} \min \left\{ \frac{N-n}{N}, \frac{N-m}{N} \right\},
\]

\[
(n, m) \to (n + 1, m) \quad \text{at rate} \quad s^{N,1} \left( \frac{N-n}{N} - \frac{N-n}{N} \right) \mathbb{1}_{\{m \geq n, s^{N,1}\}},
\]

\[
(n, m) \to (n + 1, m) \quad \text{at rate} \quad s^{N,1} \left( \frac{N-n}{N} - \frac{N-m}{N} \right) \mathbb{1}_{\{n \geq m, s^{N,1}\}},
\]

\[
(n, m) \to (n - 1, m - 1) \quad \text{at rate} \quad \min \left\{ \frac{n-1}{N} + u \nu_1(n-1), \frac{m-1}{N} + u \nu_1(m-1) \right\},
\]

\[
(n, m) \to (n - 1, m) \quad \text{at rate} \quad \left( \frac{n-1}{N} + u \nu_1(n-1) \right) \mathbb{1}_{\{n \geq m\}},
\]

\[
(n, m) \to (n, m - 1) \quad \text{at rate} \quad \left( \frac{m-1}{N} + u \nu_1(m-1) \right) \mathbb{1}_{\{m \geq n\}},
\]

\[
(n, m) \to (j, j) \quad \text{at rate} \quad u \nu_0 \mathbb{1}_{\{0 < j < \min\{n, m\}\}},
\]

\[
(n, m) \to (n, j) \quad \text{at rate} \quad u \nu_0 \mathbb{1}_{\{n \leq j < m\}},
\]

\[
(n, m) \to (j, n) \quad \text{at rate} \quad u \nu_0 \mathbb{1}_{\{m \leq j < n\}}
\]
for all \( n, m \in [N] \); no other transitions are possible (with probability 1). Note that target states outside \([N] \times [N]\) are included in the list, but are attained at rate 0, so may be ignored. Note also that the transitions are not mutually exclusive: for example, \((n, n) \to (n - 1, n - 1)\) may either happen due to coalescence or deleterious mutation, or due to beneficial mutation; so the total rate is \( n \frac{n - 1}{2} + v_n^N v_o (n - 1) + w_n^N v_o \). We have chosen this presentation of the rates to avoid somewhat technical case distinctions.

It is easily verified that the marginalised rates corresponding to \((L_{N,1}^N, L_{N,2}^N)\), respectively, equal those of \(L_{N,1}^N\) and \(L_{N,2}^N\), so \((L_{N,1}^N, L_{N,2}^N)\) is indeed a coupling of \(L_{N,1}^N\) and \(L_{N,2}^N\). There are no transitions \((n, m) \to (k, j)\) for \(m \geq n\) and \(j < k\), so \(S := \{(n, m) : n, m \in [N], m \geq n\}\) is an invariant set for \((L_{N,1}^N, L_{N,2}^N)\); hence the coupling is monotone. Furthermore, the transitions \((n, m) \to (1, 1), (n, m) \to (n + 1, m + 1)\), and \((n, m) \to (n + 1, m + 1)\) occur at positive rates for all \(n, m \in [N]\), so \((L_{N,1}^N, L_{N,2}^N)\) is irreducible and positive recurrent on \(S\). It therefore has a stationary distribution that assigns positive weight to every state in \(S\). Let \(L_{\infty}^N, L_{\infty}^{N,2}\) be random variables on \(S\) that have this stationary distribution. They obviously satisfy \(P(L_{\infty}^{N,2} > L_{\infty}^N) = 1\) and \(P(L_{\infty}^N > L_{\infty}^{N,2}) > 0\). We may thus conclude that, for \(n \in [N]\),

\[
a_n^N := P(L_{\infty}^{N,2} > n) = P(L_{\infty}^N > n) = P(L_{\infty}^{N,2} > L_{\infty}^N > n) = P(L_{\infty}^N > n) = P(L_{\infty}^N > n) = a_n^N,
\]

which proves the claim in the finite setting. In both the diffusion and the deterministic limits, the proofs are analogous with obvious modifications. (In the deterministic limit, one may alternatively invoke the fact that \(a_n = p^n\) and \(p\) is strictly increasing in \(s\), see Proposition 7.7 below.)

7.2. The sampling probabilities. We continue with the sampling probabilities, whose behaviour is unsurprising, as we have already seen in Figure 6. We prove it here for convenience and lack of reference.

Lemma 7.2. The sampling probabilities at stationarity, that is \(b_n^N\) (for \(n \in [N]\)), \(\beta_n\) (for \(n \in \mathbb{N}\)), and \(b_n\) (for \(n \in \mathbb{N}\)), are strictly decreasing in \(s^N, \sigma,\) and \(s\), respectively.

Proof. We start with the finite case, work with the line-counting process of the killed ASG as introduced in the Appendix (Section 9), and use the representation of \(b_n^N\) as the absorption probability of \(R_N\) as presented in Corollary 9.2. In close analogy with the proof of Proposition 7.1, we consider two copies of \(R_N\), namely \(R_{N,1}^N\) and \(R_{N,2}^N\) with \(R_{0,1}^N = R_{0,2}^N = n \in [N]\) and branching rates \(s_{N,1}^N\) and \(s_{N,2}^N > s_{N,1}^N\), respectively, and couple them on the same probability space. Because of the similarity with the previous proof, we do not spell out all the transitions; suffice it to say that one obtains a monotone coupling \((\overline{R}_{N,1}^N, \overline{R}_{N,2}^N)\) on \([N]_{0,\Delta} \times [N]_{0,\Delta}\), where we identify \(\Delta\) with \(N + 1\). The marginals equal \(R_{N,1}^N\) and \(R_{N,2}^N\) in distribution. Since \(q_{R_{N,1}^N}(n, n + 1) > q_{R_{N,2}^N}(n, n + 1)\) for every \(n \in [N - 1]\), it is clear that, while \(P(\overline{R}_{N,1}^N \leq \overline{R}_{N,2}^N) = 1\) for all \(t \geq 0\) by the monotonicity of the coupling, one also has \(P(\overline{R}_{N,1}^N < \overline{R}_{N,2}^N) > 0\) for all \(t > 0\). Let \(\tau\) be the (random) time of absorption of \(\overline{R}_{N,2}^N\). If \(\overline{R}_{N,2}^N = 0\), we also have \(\overline{R}_{N,1}^N = 0\). If \(\overline{R}_{N,2}^N = \Delta\) (which happens with positive probability), we have \(P(\overline{R}_{N,1}^N < \Delta) > 0\); this entails an additional chance for \(\overline{R}_{N,1}^N\) to absorb in 0. More precisely, we altogether have, with \(R_{N,i}^N := \lim_{\tau \to \infty} R_{N,i}^N\) for \(i \in \{1, 2\}\) and likewise for \(\overline{R}_{N,i}^N\):

\[
P(\overline{R}_{N,1}^N = 0) = P(\overline{R}_{N,1}^N = 0) = P(\overline{R}_{N,2}^N = 0) = P(\overline{R}_{N,2}^N = 0) = P(\overline{R}_{N,1}^N = \Delta) P(\overline{R}_{N,2}^N = 0 | \overline{R}_{N,2}^N = \Delta) > P(\overline{R}_{N,2}^N = 0),
\]

which shows the claim in the finite case. In both the diffusion and the deterministic limits, the proofs are analogous with obvious modifications. □
7.3. The ancestral type distribution. We now turn to the ancestral type distribution, whose dependence on the selection parameter is illustrated in Figure 6. In what follows, we use the shorthand $x'$ for $\frac{d}{d\nu} x$, $\frac{d}{ds} x$ or $\frac{d}{d\theta} x$ for a quantity $x$ (there will be no risk of confusion) and suppress the explicit dependence on $s^N$, $\sigma$ and $s$, respectively.

Proposition 7.3. The stationary probabilities for a type-1 ancestor, namely $\mathbb{P}(A_N^N = 1), \mathbb{P}(A_{\infty} = 1)$, and $\mathbb{P}(A_{\infty} = 1)$ from (4.6), (4.16), and (4.20) are strictly decreasing in $s^N, \sigma$, and $s$, respectively.

Proof. For the finite case, differentiating $\mathbb{P}(A_N^N = 1)$ with respect to $s^N$ gives

$$\frac{d}{ds^N} \mathbb{P}(A_N^N = 1) = \frac{d}{ds^N} \sum_{n=1}^{N} w_n^N b_n^N = \sum_{n=1}^{N} (w_n^N)' b_n^N + \sum_{n=1}^{N} w_n^N (b_n^N)'.$$  (7.1)

From Lemma 7.2, we know that $(b_n^N)' < 0$ for all $n \in [N]$, so the second sum is negative. Writing the first sum as

$$\sum_{n=1}^{N} (w_n^N)' b_n^N = \mathbb{E}[h(Y_N^N)]$$

with

$$h(k) := \sum_{n=1}^{N} (w_n^N) k_n N^2 = \frac{d}{ds^N} \left( \sum_{n=1}^{N} w_n^N k_n N^2 \right) = \frac{d}{ds^N} \mathbb{E} \left( \frac{L_N^N}{N^2} \right) < 0,$$

where the inequality is due to the stochastic monotonicity of $L_N^N$ established in Proposition 7.1. It follows that the first sum in (7.1) is negative as well, which establishes the claim in the finite case. The proofs for the diffusion and deterministic limits are perfectly analogous. In the diffusion limit, the result has been shown previously by Taylor (2007, Prop. 2.6) via a different route. □

7.4. The mutation rates. A first inspection of (5.8) shows the following. For $s^N = 0$, we have $a_n^N = \delta_{0,n}$ and $w_n^N = \delta_{1,n}$, where $\delta$ denotes Kronecker’s delta; this entails that

$$\sum_{n=1}^{N} a_{n-1}^N b_n^N = 1 = \sum_{n=1}^{N} w_n^N (b_n^N - b_n^N)$$

and so $q_{A^N}(1,0) = u^N \nu_0^N$ and $q_{A^N}(0,1) = u^N \nu_1^N$ — as is to be expected, since in this case the mutation rates on the ancestral line should equal the neutral rates. In contrast, for $s^N > 0$, we have $a_n^N > 0$ and $w_n^N > 0$ with $a_n^N = w_n^N + a_n^N > w_n^N$ for all $n > 0$, so $q_{A^N}(1,0) > u^N \nu_0^N$ and $q_{A^N}(0,1) < u^N \nu_1^N$. See Fig. 12 for a numerical evaluation of the dependence of $q_{A^N}(1,0)$ and $q_{A^N}(0,1)$ on $s^N$. Indeed, the figure tells us more than our quick inspection: $q_{A^N}(1,0)$ and $q_{A^N}(0,1)$ are strictly increasing and decreasing, respectively. While we only have the numerical evidence for finite $N$, we now give proofs for our two limits.

Theorem 7.4. The marginal mutation rates on the ancestral line in the diffusion limit at stationarity satisfy

$$q_A'(1,0) = \frac{\theta \nu_0 \beta_1}{(\sum_{m>0} (\alpha_m - \alpha_m) \beta_m)^2} \sum_{n>0} \alpha_n^N \beta_n > 0,$$  (7.2)

$$q_A'(0,1) = -\frac{\theta \nu_1 (\beta_0 - \beta_1)}{(\sum_{m>0} (\alpha_m - \alpha_m) (\beta_m - \beta_m))^2} \sum_{n>0} \alpha_n^N (\beta_{n-1} - \beta_n) < 0.$$  (7.3)

The proof requires two lemmas.
The tail probabilities of Lemma 7.5.

Proof. Set \( \bar{\alpha}_0 = 1 \) and

\[
\bar{\alpha}_n := \frac{\sigma^n (\beta_{n+1} - \beta_{n+2})}{F_n (\beta_1 - \beta_2)}, \quad n \in \mathbb{N},
\]

where \( F_n := \prod_{j=1}^n (j + \theta \nu_1) \). Since \( \alpha := (\alpha_n)_{n \geq 0} \) is the unique solution to the system of equations (4.14) with boundary conditions \( \alpha_0 = 1 \) and \( \lim_{n \to \infty} \alpha_n = 0 \), it suffices to show that \( \bar{\alpha} := (\bar{\alpha}_n)_{n \geq 0} \) solves the same system of equations and boundary conditions. By definition \( \bar{\alpha}_0 = 1 \). Moreover, since \( \lim_{n \to \infty} \beta_n = 0 \) and \( \lim_{n \to \infty} \sigma^n / F_n = 0 \), we deduce that \( \lim_{n \to \infty} \bar{\alpha}_n = 0 \). It remains to show that \( \bar{\alpha} \) satisfies (4.14).

Let \( n > 0 \). Using the definition of \( \bar{\alpha}_n \) and (4.15) first for \( n + 2 \) and then for \( n + 1 \), we obtain

\[
(n + 1 + \sigma + \vartheta) \bar{\alpha}_n = (n + 1 + \sigma + \vartheta) \frac{\sigma^n (\beta_{n+1} - \beta_{n+2})}{F_n (\beta_1 - \beta_2)}
\]

\[
= \frac{\sigma^n}{F_n (\beta_1 - \beta_2)} [(n + 1 + \sigma + \vartheta) \beta_{n+1} - (\sigma \beta_{n+3} + (n + 1 + \vartheta \nu_1) \beta_{n+1})]
\]

\[
= \frac{\sigma^n}{F_n (\beta_1 - \beta_2)} [(n + \sigma + \vartheta) \beta_{n+1} - (\sigma \beta_{n+3} + (n + \vartheta \nu_1) \beta_{n+1})]
\]

\[
= \frac{\sigma^n}{F_n (\beta_1 - \beta_2)} [\beta_{n+2} + (n + \vartheta \nu_1) \beta_n - (\sigma \beta_{n+3} + (n + \vartheta \nu_1) \beta_{n+1})]
\]

\[
= \frac{\sigma^n}{F_n (\beta_1 - \beta_2)} [(n + \vartheta \nu_1) (\beta_n - \beta_{n+1}) + \sigma (\beta_{n+2} - \beta_{n+3})]
\]

\[
= \frac{\sigma^{n+1} (\beta_n - \beta_{n+1})}{F_{n-1} (\beta_1 - \beta_2)} + (n + 1 + \vartheta \nu_1) \frac{\sigma^{n+1} (\beta_{n+2} - \beta_{n+3})}{F_{n+1} (\beta_1 - \beta_2)}
\]

\[
= \sigma \bar{\alpha}_{n-1} + (n + 1 + \vartheta \nu_1) \bar{\alpha}_{n+1}.
\]
This ends the proof. \hfill \Box

**Lemma 7.6.** The derivatives of the tail and sampling probabilities with respect to \( \sigma \) read

\[
\alpha'_n = \alpha_{n-1} - K \alpha_n \quad \text{and} \quad \beta'_n = \beta_1 \beta_n - \beta_{n+1}
\]

for \( n > 0 \), where \( K := \frac{1}{\sigma}[(1 + \theta \nu_1)(\alpha_0 - \alpha_1) + \sigma + \theta \nu_0] \).

**Proof.** Recall from (3.4) that

\[
\beta_n = \mathbb{E}[\mathcal{Y}_\infty] = \int_0^1 \eta^n \pi(\eta) \, d\eta = \frac{d_n}{d_0},
\]

where

\[
d_n := \int_0^1 \eta^{n+\theta \nu_1 - 1}(1 - \eta)^{\theta \nu_0 - 1} e^{-\sigma \eta} \, d\eta, \quad n \geq 0.
\]

Clearly, \( d'_n = -d_{n+1} \), and so

\[
\beta'_n = \frac{-d_{n+1}d_0 + d_n d_1}{d_0^2} = \beta_1 \beta_n - \beta_{n+1}
\]

in line with (7.5). We now turn to the \( \alpha'_n \). A straightforward application of differentiation rules together with (7.5) to the expression for \( \alpha_n \) in Lemma 7.5 brings us to

\[
\alpha'_n = \frac{n \sigma^{n-1} \beta_{n+1} - \beta_{n+2}}{F_n} = \frac{\sigma^n \beta_{n+1} - \beta_{n+2} - \beta_2 - \beta_3}{F_n} = \frac{-\sigma^n \beta_{n+2} - \beta_{n+3}}{F_n} - \frac{\sigma^n \beta_{n+2} - \beta_{n+3}}{F_n} + \frac{n + 1 + \theta \nu_1}{\sigma} \alpha_n = \frac{n + 1 + \theta \nu_1}{\sigma} \alpha_n - \frac{n + 1 + \theta \nu_1}{\sigma} \alpha_{n+1}.
\]

Applying now (4.14), we obtain

\[
\alpha'_n = \frac{1}{\sigma} [\sigma \alpha_{n-1} - (1 + \sigma + \theta) \alpha_n + (1 + \theta \nu_1) \alpha_1 \alpha_n] = \alpha_{n-1} - K \alpha_n,
\]

which shows (7.4). \hfill \Box

**Proof.** of Theorem 7.4. Let us first consider \( q_A(1, 0) \) and rewrite it as

\[
q_A(1, 0) = \theta \nu_0 \frac{A}{A + B} \quad \text{with} \quad A := \sum_{n>0} \alpha_{n-1} \beta_n \quad \text{and} \quad B := \sum_{n>0} \alpha_n \beta_n.
\]

Differentiating yields

\[
q'_A(1, 0) = \theta \nu_0 \frac{B' A - A' B}{(A - B)^2}.
\]

Evaluating \( A' \) with the help of Lemma 7.6 gives

\[
A' = \sum_{n>0} (\alpha'_n - \beta_n + \alpha_{n-1} \beta'_n) = \sum_{n>0} \alpha'_n \beta_{n+1} + \sum_{n>0} \alpha_{n-1} \beta'_n
\]

\[
= \sum_{n>0} (\alpha_{n-1} - K \alpha_n) \beta_{n+1} + \sum_{n>0} \alpha_{n-1} (\beta_1 \beta_n - \beta_{n+1})
\]

\[
= -K \sum_{n>0} \alpha_n \beta_{n+1} + \beta_1 \sum_{n>0} \alpha_{n-1} \beta_n = (\beta_1 - K) A + K \beta_1,
\]

where we have used in the second step that \( \alpha_0 = 1 \), so \( \alpha'_0 = 0 \). In a similar way, we obtain

\[
B' = \sum_{n>0} (\alpha_{n-1} - K \alpha_n) \beta_n + \sum_{n>0} \alpha_n (\beta_1 \beta_n - \beta_{n+1})
\]

\[
= \beta_1 + (\beta_1 - K) \sum_{n>0} \alpha_n \beta_n = (\beta_1 - K) B + \beta_1.
\]
Figure 13. Marginal fluxes at stationarity in the finite system as a function of $s^N$ for two values of $\nu_1$. Further parameters: $N = 10^4$, $u^N = 8 \cdot 10^{-4}$.

So (7.6) evaluates to

$$q'_A(1, 0) = \frac{\theta \nu_0 \beta_1}{(A - B)^2} (A - KB) = \frac{\theta \nu_0 \beta_1}{(A - B)^2} \sum_{n>0} \alpha'_n \beta_n,$$

where we have used in the second step that, by the definitions of $A$ and $B$ as well as Lemma 7.6, we have

$$A - KB = \sum_{n>0} (\alpha_{n-1} - K \alpha_n) \beta_n = \sum_{n>0} \alpha'_n \beta_n.$$

This proves (7.2).

To verify (7.3), we write

$$q_A(0, 1) = \theta \nu_1 \frac{D - E}{D}$$

with $D := \sum_{n>0} \alpha_{n-1} \delta_n$ and $E := \sum_{n>0} \alpha_n \delta_n$,

where, for $n > 0$, $\delta_n := \beta_{n-1} - \beta_n$. Noting that $\delta'_n = \beta_1 \delta_n - \delta_{n+1}$, one then proceeds in a way perfectly analogous to the proof of (7.2).

The signs of $q'_A(1, 0)$ and $q'_A(0, 1)$ now follow immediately since, for all $n > 0$, $\alpha'_n > 0$ by Proposition 7.1 and $\delta_n > 0$.

□

Proposition 7.7. The marginal mutation rates on the ancestral line in the deterministic limit at stationarity satisfy

$$q'_A(1, 0) = uw_0 \frac{p'}{(1 - p)^2} > 0 \quad \text{and} \quad q'_A(0, 1) = -uw_1 p' < 0,$$

where

$$p' = \frac{1}{2u \nu_1} \left(1 - \frac{u + s - 2u \nu_1}{\sqrt{u^2 + s^2 - 4s u \nu_1}}\right) > 0.$$

Proof. The expressions for $p'$, $q'_A(1, 0)$, and $q'_A(0, 1)$ follow from (4.19) and Corollary 5.6 via elementary differentiation. Their signs then result from the fact that

$$u + s - 2u \nu_1 = \sqrt{(u + s - 2u \nu_1)^2} = \sqrt{(u + s)^2 - 4u \nu_1 (u + s) + 4(u \nu_1)^2} < \sqrt{(u + s)^2 - 4s u \nu_1}.$$
7.5. The mutation fluxes. The mutation fluxes are important for comparison with experiment. We concentrate on the finite-$N$ case here; the behaviour in the limits is very similar. Recall that $f_{A^N}(0,1) = f_{A^N}(1,0)$ due to stationarity. Figure 13 shows how the fluxes vary with $s^N$. In the realistic case of large $\nu_1$ (and hence small $\nu_0$), the mutation flux initially increases and then decreases with $s^N$. This may seem astonishing at first sight but has a simple explanation, which reveals itself when comparing with Figures 6 and 12. At $s^N = 0$, the common ancestor type process is simply the neutral mutation process at rates $q_{A^N}(0,1) = u^N \nu_1$ and $q_{A^N}(1,0) = u^N \nu_0$, respectively, so the proportion of type-1 individuals on the ancestral line at stationarity is $P(A_\infty^N = i) = \nu_i$ for $i \in \{0,1\}$, and the stationary flux is $u^N \nu_1 \nu_0 = u^N (1 - \nu_0) \nu_0$ in either direction. Consider now the case $\nu_1 = 0.99$ (see Section 8 for why we chose the parameter values). With increasing $s^N$, $P(A_\infty^N = 0)$ increases; but type-0 individuals have the larger of the two mutation rates, namely $u^N \nu_1$, so the flux initially increases. This behaviour is only reversed at higher values of $s^N$, where type 0 has become dominant and the flux mainly experiences the decrease with $s^N$ of the deleterious mutation rate. In contrast (and unsurprisingly), the flux decreases monotonically with $s^N$ in the unrealistic situation where $\nu_0$ is close to 1.

8. Theory and experiment

It is not our intention to provide a close comparison with the experimental data — after all, this is impossible with the simple two-type model. Nevertheless, we can indeed gain some principal insight in the spirit of a proof of concept, in particular by linking a well-known observation with our ancestral structures.

The two-type model may be considered a toy model for a situation with a single wildtype allele or a set of such alleles, which are selectively favoured and lumped into type 0; and a large number of mutant alleles, which are disfavoured and lumped into type 1. The mutation rates between wildtype and mutant are then approximated by $u^N \nu_0$ and $u^N \nu_1$; mutations within the wildtype and mutant classes are ‘invisible’ in the two-type picture. Let us now discuss our results in the light of the mutation rates (or, more precisely, the mutation fluxes) observed in phylogeny and pedigree studies.

The choice of mutation rates underlying most of our figures is motivated by the mitochondrial control region, to which most of the comparisons of pedigree and phylogenetic mutation rates refer (for example, Parsons et al. (1997); Sigurðardóttir et al. (2000); Santos et al. (2005); the recent study of mutation rates in pedigrees and phylogeny in complete mitochondrial genomes by Connell et al. (2022) reports very similar results). We set $u^N = 8 \cdot 10^{-4}$; this is motivated by the overall mutation probability of $\approx 1.6 \cdot 10^{-6}$ per site and generation in the mitochondrial genome (Connell et al. 2022), the size of the control region of $\approx 1000$ base pairs, and the guess that roughly half the mutations of the wildtype are neutral, that is, remain within the wildtype class (and are hence invisible in our model). The choice of $\nu_1 = 0.99$ and $\nu_0 = 0.01$ in our figures then amounts to assuming that every wildtype allele turns into a deleterious mutant at probability $7.9 \cdot 10^{-4}$ per generation, while every allele in the unfit class is restored to wildtype with probability $8 \cdot 10^{-6}$ per generation. These figures can, of course, be only wild guesses. The inverted choice $\nu_1 = 0.01$ and $\nu_0 = 0.99$ is present in our figures for comparison only, not meant to be biologically reasonable in any way.

We are now ready to discuss the mutation fluxes. Indeed, the experiments do measure what we call mutation fluxes (as opposed to rates), namely the mean number of mutation events per individual and generation, regardless of the parent and offspring’s types. The fluxes in the two-type model only account for the deleterious and beneficial mutations; they are blind to the neutral
mutations within the wildtype and the mutant classes. This is why Figure 13 does not tell the whole story and we have to start afresh.

In the pedigree study, one observes all mutations, from any members of the population (or the sample) to any offspring, regardless of whether they will survive or not. In phylogeny, one only sees the mutations on the ancestral line, that is, from parents that are ancestors to offspring that have also survived in the long run. But which value of $s^N$ should be considered?

We are, of course, in no position to estimate the selection coefficient or to at least come up with an educated guess; firstly, because this is one of the most difficult tasks in population genetics, and second, because this requires working with a half-way reasonable fitness landscape on a large allelic space, rather with a two-type model. What we can say, however, is that, for type 0 and 1 to be interpretable as wildtype and mutant, type 0 must be the dominant type by far and make up, say, at least 90% of the population. Figure 6 tells us that, with our mutation parameters, this requires $s^N \gtrapprox 0.008$. For definiteness, we set $b^N_1 = 0.9$ and correspondingly $s_* \approx 0.008$, with which we work from now on. By Figure 6, we also have $\mathbb{P}(A^N_\infty = 1) \approx 1.3 \cdot 10^{-4}$. Figure 12 further tells us that $q^N_{A^N}(0, 1) \approx 9 \cdot 10^{-7}$ and $q^N_{A^N}(1, 0) \approx 0.007$.

In the pedigree setting, it is reasonable to assume that the total mutation rate (that is, neutral plus selective) is the same in both classes. Let $v_0$ and $v_1$ be the neutral mutation rate of type-0 and type-1 individuals, respectively. In agreement with the observed total mutation rate, we therefore arrange $v_0$ and $v_1$ so that $v_0 + u^N v_1 = 1.6 \cdot 10^{-3} = v_1 + u^N v_0$, so $v_0 \approx 8.08 \cdot 10^{-4}$ and $v_1 \approx 1.592 \cdot 10^{-3}$. The total expected mutation flux in the pedigree setting therefore is

$$(1 - b^N_1) [v_0 + N v_1] + b^N_1 [v_1 + N v_0] = 1.6 \cdot 10^{-3}. \quad (8.1)$$

In contrast, the total mutation flux in the phylogenetic setting is

$$\mathbb{P}(A^N_\infty = 0)[v_0 + q^N_{A^N}(0, 1)] + \mathbb{P}(A^N_\infty = 1)[v_1 + q^N_{A^N}(1, 0)] \approx 8.1 \cdot 10^{-4}. \quad (8.2)$$

This reduction by a factor of close to 2 simply comes from the fact that most of the (descendants of the) deleterious mutations are lost in the long run in the phylogenetic setting, due to purifying selection. More generally, neglecting the small terms in (8.1) and (8.2) (see the numerical values above), we see that the phylogenetic reduction factor is close to $(v_0 + u^N)/v_0$ and can be arranged to take any value $\geq 1$ by a suitable choice of $v_0$. To explain the observed factor of about 10 solely by purifying selection, one needs $v_0 \approx u^N/9$, which seems unrealistically small. Nevertheless, the results of the toy model make the effect of selection on the mutation fluxes very transparent and confirm that purifying selection may substantially contribute to the observed phenomenon.

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9. Appendix: recursions for the sampling probabilities $b^N_0$ As previously done for the diffusion and deterministic limits (see Baake and Wakolbinger (2018) and Baake et al. (2018)), Baake et al. (2023, Section 2.2) have introduced the killed ASG in the finite case, a modification of the ASG that allows to determine whether all individuals in a sample at present are unfit. The crucial observation is that the type of an individual at present is uniquely determined by the most recent mutation that appears on its ancestral line. Hence, when we encounter, going backwards in time, a mutation on a line in the ASG, we do not need to follow it further into the past and we can prune it. Furthermore, if the mutation is beneficial, we know that at least one of the potential ancestors is fit and, because fit lines have the priority at every branching event, the individual at present will also be fit. We therefore kill the process, that is,
Figure 14. The killed ASG either absorbs in the state 0 (left) or in the cemetery state ∆ due to a beneficial mutation (right).

send it to a cemetery state ∆. (Note that we cannot say whether this particular line is the actual ancestor.) This leads us to the following definition.

Definition 9.1 (killed ASG, finite case). The killed ASG in the finite case starts with one line emerging from each of the \( n \in [N] \) individuals present in the sample. Each line branches at rate \( s^N (N - n)/N \), every ordered pair of lines coalesces at rate \( 1/N \), every line is pruned at rate \( u^N \nu_1 \), and the process moves to ∆ at rate \( u^N \nu_0 \) per line.

Let now \( R^N := (R^N_t)_{t \geq 0} := \{R^N_{\tau_n} = \{n \} \in [N]_{\geq 0} \} \) be the line-counting process of the killed ASG. It is a continuous-time Markov chain on \( \mathbb{N}_0 \cup \Delta := [N]_{\geq 0} \cup \Delta \) and with transition rates

\[
q_{R^N}(n, n + 1) = s^N n \frac{N - n}{N}, \quad q_{R^N}(n, n - 1) = n \frac{n - 1}{N} + u^N \nu_1 n, \quad q_{R^N}(n, \Delta) = u^N \nu_0 n
\]

for \( n \in [N] \); all other rates are 0. Clearly, the states 0 and ∆ are absorbing while all other states are transient. Figure 14 shows the two different absorption scenarios. The connection with the sampling probabilities is now made by a special case of Corollary 2.4 of Baake et al. (2023).

Corollary 9.2 (sampling probabilities are absorption probabilities). We have

\[
b^N_n = \mathbb{E}\left[ \frac{Y^N_{\tau_n}}{N + 1} \right] = \mathbb{P}(R^N \text{ absorbs in } 0 \mid R^N_0 = n), \quad n \in [N]_{\geq 0}.
\]

Via this connection, we can now obtain the sampling recursion.

Corollary 9.3 (sampling recursion). The sampling probabilities satisfy the recursion

\[
\left( \frac{n - 1}{N} + s^N \frac{N - n}{N} + u^N \right) b^N_n = \left( \frac{n - 1}{N} + u^N \nu_1 \right) b^N_{n-1} + s^N \frac{N - n}{N} b^N_{n+1}, \quad n \in [N], \quad (9.1)
\]

together with the boundary conditions \( b^N_0 = 1 \) and \( b^N_{N+1} = 0 \).

Proof. The proof is a simple first-step decomposition of the absorption probabilities for \( n \in [N] \), which respects the fact that ∆ is absorbing. \( \square \)

References


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