Λ-coalescents arising in a population with dormancy

by Jason Schweinsberg
University of California San Diego

With Fernando Cordero, Adrián González Casanova, and Maite Wilke-Berenguer

Outline of Talk

1. The Wright-Fisher model and Kingman’s coalescent
2. Coalescents with multiple mergers
3. A model involving dormancy
4. Results for the genealogy of the population
The Wright-Fisher Model

One of the earliest models in population genetics, goes back to Fisher (1921) and Wright (1930).

- The population has fixed size $N$.
- Generations do not overlap.
- Each individual has one parent, chosen at random from the individuals in the previous generation.

Sample two individuals in generation 0. The probability that they have the same parent is $1/N$.

Let $T$ be the number of generations we have to go back before they have the same ancestor. Then

$$P(T > Nx) = \left(1 - \frac{1}{N}\right)^{[Nx]} \approx e^{-x}.$$  

$T/N$ has approximately an exponential distribution with rate 1.
**Kingman’s Coalescent (Kingman, 1982)**

Continuous-time Markov chain \((\Pi(t), t \geq 0)\) taking values in the set \(\mathcal{P}_n\) of partitions of \(\{1, \ldots, n\}\).

Here \(\Pi(0)\) consists of \(n\) singletons. Any transition that involves merging two blocks into one happens at rate 1. No other transitions are possible.

When there are \(k\) blocks, the distribution of the time until the next merger is exponential with rate \(k(k - 1)/2\). Then two randomly chosen blocks merge.

One time unit in Kingman’s coalescent corresponds to \(N\) generations in the Wright-Fisher model.
A Limit Theorem

**Theorem** (Kingman, 1982): Suppose a population evolves according to the Wright-Fisher model with population size $N$. Sample $n$ individuals at random from the population in generation zero. Let $\Psi_N(k)$ be the partition of $\{1, \ldots, n\}$ such that $i$ and $j$ are in the same block if and only if the $i$th and $j$th sampled individuals have the same ancestor in generation $-k$. Let $(\Pi(t), t \geq 0)$ be Kingman’s coalescent. Then, as $N \to \infty$,

$$(\Psi_N(\lfloor Nt \rfloor), t \geq 0) \Rightarrow (\Pi(t), t \geq 0).$$

Here $\Rightarrow$ denotes weak convergence of stochastic processes with respect to the Skorohod topology.
Coalescents with multiple mergers (\(\Lambda\)-coalescents)

Introduced by Pitman (1999), Sagitov (1999), and Donnelly and Kurtz (1999). More than two ancestral lines can merge at a time.

Applications of coalescents with multiple mergers:
- Large family sizes (many lineages trace back to an individual with large number of offspring).
- Natural selection (many lineages trace back to an individual who got a beneficial mutation).
Definition and construction of the \( \Lambda \)-coalescent

Let \( \Lambda \) be a finite measure on \([0, 1]\). The \( \Lambda \)-coalescent is a continuous-time \( \mathcal{P}_n \)-valued Markov chain such that when there are \( b \) blocks, each \( k \)-tuple \((2 \leq k \leq b)\) of blocks merges at rate

\[
\lambda_{b,k} = \int_0^1 p^{k-2}(1 - p)^{b-k} \Lambda(dp).
\]

Put \( \Lambda = a\delta_0 + \Lambda_0 \), with \( \Lambda_0(\{0\}) = 0 \).

- Each pair of lineages merges at rate \( a \).
- Construct a Poisson point process on \([0, \infty) \times (0, 1]\) with intensity

\[
dt \times p^{-2}\Lambda_0(dp).
\]

If \((t, p)\) is a point of this Poisson process, then a \( p \)-merger occurs at time \( t \). Each lineage independently participates with probability \( p \).
Cannings models (Cannings, 1974)

- The population has fixed size $N$.
- Generations do not overlap.
- Let $\nu_{1,N}, \ldots, \nu_{N,N}$ denote the numbers of offspring of the $N$ individuals. The distribution of $(\nu_{1,N}, \ldots, \nu_{N,N})$ is exchangeable.
- Family sizes in different generations are independent and identically distributed.

**Ancestral process:** Sample $n$ individuals from generation 0. Let $\Psi_N(k)$ be the partition of $\{1, \ldots, n\}$ such that $i$ and $j$ are in the same block if and only if the $i$th and $j$th sampled individuals have the same ancestor in generation $-k$.

The probability that two individuals have the same parent is

$$c_N = \frac{E[(\nu_{1,N})^2]}{N - 1}, \quad (m)_k = m(m-1) \ldots (m-k+1).$$
Convergence of the genealogy in Cannings models

**Theorem** (Möhle, 2000): Consider a Cannings model in which

$$\lim_{N \to \infty} \frac{E[(\nu_{1,N})_3]}{N^2 c_N} = 0.$$  

Then the ancestral processes $\Psi_N([t/c_N], t \geq 0)$ converge as $N \to \infty$ to Kingman's coalescent.

**Theorem** (Sagitov, 1999; Möhle and Sagitov, 2001): Suppose

1. $\lim_{N \to \infty} c_N = 0$.
2. $\lim_{N \to \infty} \frac{E[(\nu_{1,N})_2(\nu_{2,N})_2]}{N^2 c_N} = 0$.
3. For all $x \in (0, 1)$ such that $\Lambda(\{x\}) = 0$, we have

$$\lim_{N \to \infty} \frac{N}{c_N} P(\nu_{1,N} > Nx) = \int_x^1 y^{-2} \Lambda(dy).$$  

Then the processes $(\Psi_N([t/c_N]), t \geq 0)$ converge to the $\Lambda$-coalescent.
Heavy-tailed offspring distributions

Consider the following population model:

- The population has fixed size $N$.
- Generations do not overlap.
- Numbers of offspring $\xi_1, \ldots, \xi_N$ of the $N$ individuals are i.i.d. positive integer-valued random variables.
- We have $P(\xi_i \geq k) \sim Ck^{-\alpha}$, where $\alpha > 0$.
- Obtain the next generation by sampling $N$ offspring without replacement.

Sample $n$ individuals in generation 0.

Let $\Psi_N(k)$ be the partition of $\{1, \ldots, n\}$ such that $i$ and $j$ are in the same block if and only if the $i$th and $j$th sampled individuals have the same ancestor in generation $-k$. 
Genealogy of the population

**Theorem** (Schweinsberg, 2003): As $N \to \infty$,

- If $\alpha \geq 2$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge to Kingman’s coalescent. When $\alpha > 2$, we have $c_N \sim \sigma^2 / N$, where $\sigma^2$ is the variance of the number of surviving offspring.

- If $1 < \alpha < 2$, the processes $(\Psi_N(\lfloor AN^{\alpha-1}t \rfloor), t \geq 0)$ converge, for some constant $A$, to the $\Lambda$-coalescent, where $\Lambda$ is the Beta($2 - \alpha, \alpha$) distribution (beta coalescent).

- If $\alpha = 1$, the processes $(\Psi_N(\lfloor (\log N)t \rfloor), t \geq 0)$ converge to the $\Lambda$-coalescent, where $\Lambda$ is uniform distribution on $(0, 1)$, called the Bolthausen-Sznitman (1998) coalescent.

- If $0 < \alpha < 1$, the processes $(\Psi_N(k))_{k=0}^{\infty}$ converge as $N \to \infty$ to a discrete-time coalescent process.
Idea of the proof \((1 < \alpha < 2)\)

Let \(\mu = E[\xi_i]\) be the mean of the offspring distribution.

We get a \(p\)-merger with \(p \geq x\) if

\[
\frac{\xi}{\xi + N\mu} \geq x \quad \iff \quad \xi \geq \frac{x}{1-x} \cdot N\mu
\]

The probability of such a family in a given generation is

\[
NP\left(\xi \geq \frac{x}{1-x} \cdot N\mu\right) \sim NC\left(\frac{x}{1-x} \cdot N\mu\right)^{-\alpha}.
\]

The rate of such mergers in the \(\text{Beta}(2 - \alpha, \alpha)\)-coalescent is

\[
\frac{1}{\Gamma(\alpha)\Gamma(2-\alpha)} \int_{x}^{1} p^{-1-\alpha}(1-p)^{\alpha-1} \, dp = \frac{1}{\alpha \Gamma(\alpha)\Gamma(2-\alpha)} \left(\frac{x}{1-x}\right)^{-\alpha}.
\]
In many populations, individuals periodically enter a dormant state, in response to unfavorable environmental conditions.

Wright and Vetsigian postulated that the randomness in the times when individuals emerge from a dormant state could lead to a heavy-tailed offspring distribution.

They found in bacterial experiments that “the heavy-tailed nature of the distribution of descendants can, in our case, be largely explained by phenotypic variability in lag time before exponential growth.”

Our goal: to formulate a population model that incorporates this behavior, and to investigate whether dormancy can give rise to $\Lambda$-coalescent genealogies.
A model involving dormancy

Each year begins at time 0 with \( N \) dormant individuals and lasts for time \( T_N \).

1. **Activation phase (Spring):** Each individual wakes up at some time in \((0, t_N)\), then reproduces at rate \( \lambda_N \), as in a Yule process. The times at which individuals wake up are i.i.d.

2. **Active phase (Summer):** Between times \( t_N \) and \( T_N \), all individuals are awake and reproduce at rate \( \lambda_N \).

3. **Sampling phase (Winter):** At time \( T_N \), all individuals enter a dormant state. We choose \( N \) individuals at random to survive until the start of the next year. All other individuals die.

Sample \( n \) individuals at the beginning of year 0. Let \( \Psi_N(k) \) be the partition of \( \{1, \ldots, n\} \) such that \( i \) and \( j \) are in the same block if and only if the \( i \)th and \( j \)th individuals in the sample have the same ancestor at the beginning of year \( -k \).
A two-point distribution

Let $\tau_{i,N}$ be the time when the $i$th individual emerges from dormancy.

Suppose there is no summer, so $T_N = t_N$. Suppose

$$P(\tau_{i,N} = 0) = \omega_N, \quad P(\tau_{i,N} = T_N) = 1 - \omega_N$$

with

$$\lim_{N \to \infty} N\omega_N = 0.$$

If an individual wakes up at time 0 instead of at time $T_N$, then it will have approximately $We^{\lambda_N T_N}$ descendants at time $T_N$, where $W \sim \text{Exponential}(1)$.

If $\lambda_N T_N = \log(\kappa N)$, then the fraction of the population at the end of the year descended from the individual who wakes up early is approximately

$$Y_\kappa = \frac{\kappa W}{\kappa W + 1}.$$
Genealogy of the population

**Theorem** (Cordero, González Casanova, Wilke-Berenguer, Schweinsberg, 2022):
Assume that $\lambda_N T_N = \beta \log(\kappa N)$. Then:

1. If $\beta > 1$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge as $N \to \infty$ to the star-shaped coalescent, which is the $\Lambda$-coalescent with $\Lambda = \delta_1$.

2. If $\beta = 1$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge as $N \to \infty$ to the $\Lambda_{\kappa}$-coalescent, with $\Lambda_{\kappa}$ being the probability measure characterized by
   \[
y^{-2} \Lambda_{\kappa}(dy) := \frac{1}{E[Y_{\kappa}^2]} P(Y_{\kappa} \in dy).
   \]

3. If $\beta < 1$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge as $N \to \infty$ to Kingman’s coalescent.

Also,
\[
c_N \sim \begin{cases} 
N \omega_N & \text{if } \beta > 1 \\
N \omega_N E[Y_{\kappa}^2] & \text{if } \beta = 1 \\
2\kappa^{-2\beta} \omega_N N^{2\beta-1} & \text{if } \beta < 1
\end{cases}
\]
Exponentially increasing rate of exit from dormancy

Suppose there is no summer, so \( t_N = T_N \). Suppose
\[
\lim_{N \to \infty} \frac{\log N}{T_N} = 0.
\]

Suppose \( \lambda_N = \lambda > 0 \) for all \( N \).

Suppose \( T_N - \tau_{i,N} = \zeta_i \wedge T_N \), where \((\zeta_i)_i=1\) are i.i.d. We assume there exist constants \( \gamma > 0 \) and \( c > 0 \) such that
\[
P(\zeta_i > y) \sim ce^{-\gamma y}, \quad \text{as } y \to \infty.
\]

Let \( X_{i,N} \) be the number of descendants of the \( i \)th individual at the end of spring. The conditional distribution of \( X_{i,N} \) given \( \zeta_i = u < T_N \) is Geometric\( (e^{-\lambda u}) \).

When \( \zeta_i \sim \text{Exponential}(\gamma) \), writing \( \alpha = \gamma/\lambda \),
\[
P(X_{i,N} > k) = \int_0^\infty (1 - e^{-\lambda u})^k \gamma e^{-\gamma u} \, du = \frac{\alpha \Gamma(k + 1) \Gamma(\alpha)}{\Gamma(k + 1 + \alpha)} \sim \Gamma(\alpha + 1)k^{-\alpha}.
\]
Genealogy of the population

**Theorem** (Cordero, González Casanova, Wilke-Berenguer, Schweinsberg, 2022): Under the conditions described on the previous slide with $\alpha = \gamma / \lambda$:

1. If $\alpha \geq 2$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge as $N \to \infty$ to Kingman’s coalescent.

2. If $1 \leq \alpha < 2$, the processes $(\Psi_N(\lfloor t/c_N \rfloor), t \geq 0)$ converge as $N \to \infty$ to the $\Lambda$-coalescent in which $\Lambda$ is the Beta$(2-\alpha, \alpha)$ distribution.

3. If $0 < \alpha < 1$, the processes $(\Psi_N(m))_{m=0}^{\infty}$ converge as $N \to \infty$ to a discrete-time coalescent process.

**Note:** We have $c_N \sim AN^{1-\alpha}$ when $1 < \alpha < 2$ and $c_N \sim 1/(\log N)$ when $\alpha = 1$. 
Classifying the possible limits

**Theorem** (Cordero, González Casanova, Wilke-Berenguer, Schweinsberg, 2022): It is possible for the \( \Lambda \)-coalescent to arise as the limit of the rescaled ancestral processes \( (\Psi_N(\lfloor \rho_N t \rfloor), t \geq 0) \) in this population model if and only if

\[
\Lambda = b_0 \delta_0 + b_1 \delta_1 + \Lambda',
\]

where \( b_0, b_1 \geq 0 \) and \( \Lambda'(dy) = h(y) \, dy \) with

\[
h(y) = \int_{0}^{\infty} \frac{1}{\kappa} \left( \frac{y}{1-y} \right)^2 e^{-\frac{y}{\kappa(1-y)}} \eta(d\kappa)
\]

for all \( y \in (0, 1) \) and \( \int_{0}^{\infty} (1 \wedge \kappa^2) \eta(d\kappa) < \infty \).

**Note:** Recall \( Y_\kappa = \kappa W / (\kappa W + 1) \) and \( y^{-2} \Lambda_\kappa(dy) = E[Y_\kappa^2]^{-1} P(Y_\kappa \in dy) \). Now

\[
\Lambda'(dy) = \int_{0}^{\infty} y^2 P(Y_\kappa \in dy) \eta(d\kappa) = \int_{0}^{\infty} \Lambda_\kappa(dy) E[Y_\kappa^2] \eta(d\kappa).
\]
The effect of summer

Theorem (Cordero, González Casanova, Wilke-Berenguer, Schweinsberg, 2022): Suppose, for a model with no summer, the processes \( (\Psi_N(\lfloor t/c_N \rfloor), t \geq 0) \) converge to a \( \Lambda \)-coalescent. Let \( X_{i,N} \) denote the number of descendants of the \( i \)th individual at the beginning of a given year who are alive at time \( t_N \), and suppose

\[
\lim_{N \to \infty} \frac{1}{c_N} E \left[ \frac{1}{X_{1,N} + \cdots + X_{N,N}} \right] = 0.
\]

Then we get the same limiting genealogy if a summer of any length is added.

Note: Here \( c_N \) is the probability that two lineages coalesce during the spring. If there are \( M \) individuals alive at the end of the spring, the probability that two lineages coalesce during the summer is \( O(1/M) \).
Summary and conclusions

- In many populations, individuals periodically enter a dormant state.
- Individuals that emerge unusually early from dormancy may be able to produce more descendants than other individuals.
- In extreme cases, this could lead to Λ-coalescent genealogies, in which many ancestral lines get traced back to one individual that emerged unusually early from dormancy.
- Wright and Vestigian (2019): “it is unlikely that the variance diverges with population size for the particular species and conditions we examined”. Therefore, at the moment, we do not have evidence that Λ-coalescent genealogies occur in actual biological populations undergoing dormancy.