Models for Genetic Diversity Generated by Negative Binomial Point Processes

Yuguang F. Ipsen\textsuperscript{1}, Soudabeh Shemehsavar\textsuperscript{2}, and Ross A. Maller\textsuperscript{1}

\textsuperscript{1}Research School of Finance, Actuarial Studies & Statistics, Australian National University

\textsuperscript{2}School of Mathematics, Statistics & Computer Sciences, University of Tehran

May 1, 2019

Abstract

We develop a model based on a generalised Poisson-Dirichlet distribution for the analysis of genetic diversity, and illustrate its use on microsatellite data for the genus \textit{Dasyurus} (the quoll, a marsupial carnivore listed as near-threatened in Australia). Our class of distributions, termed PD\textsuperscript{(r)}\textsubscript{\alpha}, is constructed from a negative binomial point process, generalizing the usual one-parameter PD\textsubscript{\alpha} model, which is constructed from a Poisson point process. Both models have at their heart a Stable(\alpha) process, but in PD\textsuperscript{(r)}\textsubscript{\alpha}, an extra parameter \(r > 0\) adds flexibility, analogous to the way the negative binomial distribution allows for “overdispersion” in the analysis of count data. A key result obtained is a generalised version of Ewens’ sampling formula for PD\textsuperscript{(r)}\textsubscript{\alpha}.

We outline the theoretical basis for the model, and, for the quolls data, estimate the parameters \(\alpha\) and \(r\) by least squares, showing how the extra parameter \(r\) aids in the interpretability of the data by comparison with the standard PD\textsubscript{\alpha} model. The methods potentially have implications for the management and conservation of threatened populations.

Keywords: Poisson-Dirichlet distribution, negative binomial point process, Ewens sampling formula, genetic diversity of quolls

\textsuperscript{*}Email: Yuguang.Ipsen@anu.edu.au

\textsuperscript{†}Email: Shemehsavar@khayam.ut.ac.ir. Dr Shemehsavar thanks the Research School of Finance, Actuarial Studies & Statistics for facilities and support during a sabbatical visit there.

\textsuperscript{‡}Research partially supported by ARC Grant DP1092502; Corresponding author: Email: Ross.Maller@anu.edu.au
1 Introduction

In this era of big data analyses there still remain situations where sample sizes are necessarily small and numbers of variables are limited. This is especially the case in conservation genetics where study populations are typically restricted and data is difficult and expensive to obtain. The level of genetic variation among members of a population and how it differs between populations are fundamental objects of interest and of special significance for guiding conservation strategies, and it is important to make optimal use of what observations are available. Consequently there is a need for specialized techniques for the analysis and interpretation of gene sampling data in conservation genetics studies.

In the past a number of models for genetic diversity have been based on the Poisson-Dirichlet distribution and the associated Ewens sampling formula (Ewens (1972)). As its name suggests, the Poisson-Dirichlet has as an underlying building block a Poisson point process. A two-parameter class of distributions, recently proposed by Ipsen & Maller (2016, 2018), is based on a negative binomial rather than a Poisson point process, an extension which adds flexibility for modelling purposes, analogous to the way the negative binomial allows for over-dispersion in the analysis of count data.

In this paper we use genetic data on species of Australian quolls to illustrate how the modified model can be used to quantify diversity within a population and make soundly based statistical comparisons between populations. Our analysis of the small quolls data set detects differences not found with standard types of analysis. Apart from the data analyses, we present theoretical rationales for the proposed methodology and study the properties of the parameter estimates by simulations.

1.1 Quolls Data

The quoll is a marsupial carnivore which survives in a limited number of locations across the Australian continent, New Guinea and Tasmania. In Australia all species are listed as near-threatened. A recent paper by Firestone, Elphinstone, Sherwin, & Houlden (2000) contains genetic data on 4 species of quoll, a subset of which we analyse for genetic diversity. Due to its near-threatened status the numbers of animals sampled are necessarily small and the genetics data is accordingly restricted. We concentrate on samples of a single species (the western quoll) from 2 locations in Western Australia and analyse microsatellite sample data on 2 loci. There were a maximum of 15 and 34 animals available in the 2 geographical locations.
## 2 Methods

Assuming “selective neutrality” in a gene fitness model, Ewens (1972) derived a distribution for vectors of the type \( M = (M_1, M_2, \ldots, M_n) \), where \( M_j \) counts the number of times an allele of type \( j \) occurs in a sample of size \( n \). This kind of model is appropriate for experimental data in which the number of types, or what types exist, in a population, are unknown, but it is possible in a sample of size \( n \), say, to recognize that there are \( k \) types with a number \( M_j \) of type \( j \), \( j = 1, 2, \ldots, n \). (See Watterson (1974) for further discussion.) In Fisher, Corbet & Williams (1943), R.A. Fisher, also strongly motivated by genetics, was concerned with measuring species diversity using a model based on the negative binomial distribution. Fundamentally, the Dirichlet distribution underlies both approaches.

These two important streams of statistical modelling methodology came together in the paper of Kingman (1975). By taking a limit of finite Dirichlet distributions, analogous to the way in which the Poisson distribution is obtained as a limit of binomials, Kingman derived a class of models referred to as the Poisson-Dirichlet distributions. One member of these is the \( \text{PD}_\alpha \) distribution, which is obtained as the limit of the ranked vector of normalised jumps of a driftless stable subordinator with a single parameter \( \alpha \) in \((0,1)\). The subordinator jumps can be regarded as a Poisson point process in time and space, and taking this point of view opens up a wide variety of applicable theory which has subsequently been expanded in many directions. In particular, this approach was further developed for the \( \text{PD}_\alpha \) model by Perman (1990, 1993).

Recently Ipsen & Maller (2016, 2018) extended the Kingman-Perman \( \text{PD}_\alpha \) model by replacing the Poisson point process with a negative binomial point process, thereby bringing in an extra parameter, \( r > 0 \). The new class, termed \( \text{PD}^{(r)}_\alpha \), is amenable to analysis and among other results a generalised Ewens sampling formulae can be obtained for it. In what follows we outline the basic structure underlying \( \text{PD}^{(r)}_\alpha \), especially relating to the negative binomial point process, suggest a weighted least squares method of fitting it to data, and apply the method to analyse the quolls data. The resulting analyses provide extra insight over and above that obtainable from fitting a \( \text{PD}_\alpha \) model. Some simulations verify that the estimators have reasonable properties and this is further demonstrated in a theoretical appendix.

### 2.1 Data Structure and \( \text{PD}^{(r)}_\alpha \) Model

Consider a sample of size \( n \) from a population with, nominally, infinitely many blocks, whose proportions are governed by a distribution on the simplex \( \nabla_\infty := \{x_i \geq 0, i = 1, 2, \ldots, \sum_{i \geq 1} x_i = 1\} \). In the quolls data, “blocks” represent allele
types.

Corresponding to a given partition of \( \mathbb{N}_n := \{1, 2, \ldots, n\} \) into \( k \) blocks, let \( M_j \) denote the number of blocks in the sample which have exactly \( j \) representatives. The vector \( M = (M_1, \ldots, M_n) \) consists of non-negative, integer-valued random variables satisfying \( \sum_{j=1}^n jM_j = n \) (sample size) and \( \sum_{j=1}^n M_j = k \) (the number of distinct alleles in the sample). Let \( n_i \) be the number of representatives (alleles) of block type \( i \) (allele type \( i \)), \( 1 \leq i \leq k \), in the sample, with \( \sum_{i=1}^k n_i = n \).

Ewens’ sampling formula (Ewens (1972)) gives the probability of a particular realization of the blocks count vector \( M \) under the assumption of selective neutrality. Our aim is to present a generalised version of it for \( PD^{(r)}_\alpha \) and illustrate its usefulness for data analysis.

2.2 Negative Binomial Point Processes

The \( PD^{(r)}_\alpha \) class is based on a negative binomial point process (NBPP), denoted by \( B^{(r)} \), as defined by Gregoire (1984). This point process has Laplace functional

\[
\Phi(f) = \mathbb{E}\left(e^{-B^{(r)}(f)}\right) = \left(1 + \int_{\mathbb{R}_+} (1 - e^{-f(x)}) \Lambda(dx)\right)^{-r},
\]

for nonnegative measurable functions \( f \) on \( \mathbb{R}_+ \). In (2.1), \( r > 0 \) is a parameter and \( \Lambda \) is a measure on the Borel sets of \( \mathbb{R}_+ \). Gregoire devised the NBPP in order to define a process for which all finite-dimensional distributions associated with disjoint bounded Borel sets in \( \mathbb{R}_+ \) are negative binomial.

2.3 The \( PD^{(r)}_\alpha \) Class

To construct \( PD^{(r)}_\alpha \), we choose a measure having density

\[
\rho(x)dx := \alpha x^{-\alpha - 1}dx, \quad \text{for some } 0 < \alpha < 1,
\]

and take a driftless stable subordinator \( (S_t)_{t \geq 0} \) of index \( \alpha \) with \( S_0 = 0 \), whose Lévy measure is given by (2.2). Its jump process is \( (\Delta S_t := S_t - S_{t-})_{t > 0} \), with \( \Delta S_0 = 0 \), and the jumps up till time 1 are ordered as \( \Delta S_1^{(1)} \geq \Delta S_1^{(2)} \geq \cdots \). Let \( S_1 = S_1 - \Delta S_1^{(1)} - \cdots - \Delta S_1^{(r)}, \ r = 0, 1, 2, \ldots \), be the “trimmed” subordinator.

Define a random point measure on the Borel sets of \( (0,1) \) by

\[
\mathbb{B}^{(r)} = \sum_{i \geq 1} \delta_{J_i(r)}, \quad \text{where } J_i(r) = \frac{\Delta S_i^{(r+i)}}{\Delta S_1^{(r)}}, \ i = 1, 2, \ldots, r \in \mathbb{N}.
\]

Thus \( \mathbb{B}^{(r)} \) is defined in terms of ratios of successive jumps of \( S_1 \), rather than in terms of the jumps themselves. In Proposition 2.1 of Ipsen & Maller (2016) it is
proved that $\mathbb{B}^{(r)}$ as defined is distributed as a negative binomial point process with a measure whose density is $\alpha x^{-\alpha-1}1_{(0<x\leq 1)}$. Note that, by comparison with (2.2), this density is truncated to $(0,1]$. (2.3) motivates the further definition:

**Definition 2.1.** For each $r = 1, 2, \ldots$, the vector

$$
(V_n^{(r)})_{n \geq 1} := \left( \frac{\Delta S_1^{(r+1)}}{(r) S_1^{(r+2)}}, \frac{\Delta S_1^{(r+2)}}{(r) S_1^{(r+3)}}, \ldots \right) = \left( \frac{J_r(1)}{\sum_{i \geq 1} J_i(r)}, \frac{J_r(2)}{\sum_{i \geq 1} J_i(r)}, \ldots \right) \quad (2.4)
$$
on $\nabla_\infty$ is said to have a $\text{PD}^{(r)}_{\alpha}$ distribution.

Although the derivation of $\text{PD}^{(r)}_{\alpha}$ via the normalised jumps of a subordinator requires $r$ to be a positive integer, the formulae in what follows are valid for any $r > 0$ with the obvious interpretations, and subsequent results hold in this generality.

We refer to Ipsen & Maller (2016, 2018) for further details on the stick-breaking properties, Palm characterisation and joint densities of the size biased permutations of distributions constructed from negative binomial processes.

## 3 Ewens Sampling Formula for a $\text{PD}^{(r)}_{\alpha}$-Partition

Using the notation in Subsection 2.1, consider a sample of size $n$ from a population with infinitely many blocks, whose proportions are governed by the $\text{PD}^{(r)}_{\alpha}$ distribution. Corresponding to a given partition $\Pi_n = (A_1, \ldots, A_k)$ of $n$ into $k$ blocks, the components $M_j$ of the vector $M$ represent the number of blocks (allele types) of $\Pi_n$ in the sample which have exactly $j$ representatives.

The next theorem presents a version of Ewens’ sampling formula for $\text{PD}^{(r)}_{\alpha}$.

**Theorem 3.1.** The Ewens random partition structure derived from $\text{PD}^{(r)}_{\alpha}$ for a sample of size $n$ partitioned into $k$ blocks has distribution

$$
\mathbb{P}(M = m := (m_1, m_2, \ldots, m_n)) = \alpha^{k} r[k] \prod_{j=1}^{n} \frac{1}{m_j!} \left( \frac{\Gamma(j-\alpha)}{j!} \right)^{m_j} \times \int_{0}^{\infty} \frac{\lambda^{k-1}}{\Psi(\lambda)^r + k} \prod_{j=1}^{n} (G_{j-\alpha}(\lambda))^{m_j} d\lambda \mathbf{1}_{\{\sum_{j=1}^{n} m_j = k\}}, \quad (3.1)
$$

where $\sum_{j=1}^{n} j m_j = n$, $r[k] = r(r+1) \cdots (r+k-1)$ denotes the ascending factorial,

$$
\Psi(\lambda) := 1 + \alpha \int_{0}^{1} (1 - e^{-\lambda x}) x^{-\alpha-1} dx, \quad (3.2)
$$

and $G_{j-\alpha}(\lambda)$ is the incomplete gamma distribution function defined by

$$
G_{j-\alpha}(\lambda) := \frac{1}{\Gamma(j-\alpha)} \int_{0}^{\lambda} x^{j-\alpha-1} e^{-x} dx, \quad \lambda > 0. \quad (3.3)
$$
Remarks. (i) A proof of Theorem 3.1 is in Appendix A.1. Formula (3.1) looks forbidding (and is computationally challenging) but in fact has a readily interpretable and reasonably tractable structure as we show in Appendix A.2.

(ii) It’s important to note that the integral in (3.2) is truncated to (0, 1), whereas integrals in the original PD\(\alpha\) class are over the whole range (0, \(\infty\)). This is a significant distinguishing feature between the two classes.

4 Parameter Estimation Methodology

To estimate the parameters in PD\(\alpha\)(\(r\)) we minimise the weighted sum of squares

\[ S := \sum_{j=1}^{n} w_j (m_j - E_j)^2 \]  

(4.1)

for variations in \(\alpha\) and \(r\). Here \(w_j > 0\) are weights (we chose \(w_j = j\)) and \(E_j = E_j(\alpha, r, k)\) is the expected value of the allele type count \(M_j\), conditional on \(\sum_{j=1}^{n} M_j = k\), based on the current values of \(\alpha\) and \(r\) and the PD\(\alpha\)(\(r\)) model. We have no analytical formula for the \(E_j\), so we estimate them by simulation.

This is done as follows. Given starting values \(\alpha\) in (0, 1), \(r > 0\) and a number of blocks, \(k\), sample from PD\(\alpha\)(\(r\)) by generating realisations of (\(\Gamma_j\))\(j\geq1\) random variables (as successive sums of i.i.d. unit exponential rvs), then setting

\[ \tilde{p}_j = \frac{\Gamma_j^{1/\alpha}}{\sum_{\ell=1}^{J} \Gamma_\ell^{1/\alpha}}, \quad j = 1, 2, \ldots, J, \]  

(4.2)

where \(J\) is a large number; we used \(J = 1000\) and 5000. Then proceed as follows.

1. For the given number of blocks, \(k\), set population probabilities \((p_1, \ldots, p_k)\) as

\[ p_j = \frac{\tilde{p}_{r+j}}{\sum_{\ell=1}^{k} \tilde{p}_{r+\ell}}, \quad j = 1, 2, \ldots, k, \]  

(4.3)

based on the \(\tilde{p}_j\) in (4.2).

2. Follow the procedure outlined in Appendix A.1 to form a partition by drawing \(k\) observations on a random variable \(Y\) having an arbitrary diffuse base distribution \(G_0\) which fixes the block labels (we used \(G_0 = N(0, 1)\)). For a sample of size \(n\), select \(n\) members \((X_i)_{1\leq i\leq n}\) with replacement from those \(k\) numbers according to the probabilities \((p_1, \ldots, p_k)\). Use the \(X_i\) to partition \(n\) into integers \(n_1, \ldots, n_k\), where \(n_i = \#\{\ell : X_\ell = X_i\}, 1 \leq i \leq k\). Then set \(\hat{m}_j = \#\{i : n_i = j, 1 \leq i \leq k\}, 1 \leq j \leq n\). These form the first replacement sample, \((\hat{m}_j(\ell), 1 \leq j \leq n),\) indexed as \(\ell = 1\).
3. Repeat this step $L$ times (we used $L = 1000$), obtaining the estimates $\hat{m}_j(\ell)$, for $1 \leq j \leq n$, $\ell = 1, 2, \ldots, L$. We describe this procedure as “resampling”. After this calculate

$$\hat{E}_j = \frac{1}{L} \sum_{\ell=1}^{L} \hat{m}_j(\ell), \ j = 1, 2, \ldots, n. \quad (4.4)$$

4. During Steps 1 and 2, we can obtain resampling estimates of the variability of $\alpha$ and $r$ by estimating them at each stage to obtain $(\hat{\alpha}(\ell), \hat{r}(\ell))$, $\ell = 1, \ldots, L$. The standard errors in Tables 3 and 4 and the histograms in Figure 4 were calculated from these samples.

The $\hat{E}_j$ in (4.4), calculated with the current values of $\alpha$ and $r$, are substituted for the expected values in (4.4). Minimising the quantity $S$ in (4.41) is done by a grid search over values $\alpha = 0.05, 0.1, \ldots, 0.95$ and $r = 0.1, 0.2, \ldots$. The resulting values $\hat{\alpha}_n$ and $\hat{r}_n$ are our weighted least squares estimates. Note that this analysis is conditional on the observed number $k$ of species.

The method is very highly computer intensive but has the virtue of providing as close a fit as possible to the data, in a weighted least squares sense. In Appendix A.2 we present an argument to suggest that the estimates $(\hat{\alpha}_n, \hat{r}_n)$ will be approximately normally distributed in large samples. This is consistent with what we observe empirically in the resampling histograms.

5 Quolls Data Analysis

The paper by Firestone et al. (2000) contains genetic data on 4 species of quolls (tiger, eastern, northern and western quolls) sampled in 20 locations across Australia. At each location allele counts at 6 genetic loci are available, with around 15-20 allele types per locus. We concentrate on the western quoll data which are from 2 locations in Western Australia (Perth and Batalling State Forest) and consider loci numbers 1.3 and 3.3.1. There were a maximum of 15 and 34 individuals available in the 2 locations. We chose these particular locations for analysis from the available data as they have the most allelic variability, and contain the larger numbers of animals. We refer to Firestone et al. (2000) for further information on the methods of collection of samples and other interesting aspects of the data.

In Table 1 for Locus 1.3, we see in the first column for Perth, that there were 2 allele types having 1 representative, 2 allele types having 2 representatives, $\ldots$, 1 allele type having 9 representatives. Thus the more common alleles occur in the right of the tables, rarer alleles are in the left. (The tables omit values of $j$ where
Total numbers of alleles at Locus 1.3 for Perth were $n = 30$ with $k = 9$ distinct allele types. Similarly, for Batalling, $n = 68$ and $k = 8$, with the numbers in Table 1 for Locus 1.3. Table 2 has the information for Locus 3.3.1, both locations. Since the animals are diploids, the numbers $n$ are double the numbers of individual animals sampled.

Tables 3 and 4 contain the parameter estimates and their standard errors obtained by the method explained in Section 4.

Based on these estimates we see that, at Locus 1.3, $r$ is significantly larger for Batalling than for Perth, while $\alpha$ is significantly larger for Perth than for Batalling. At Locus 3.3.1, the $r$ estimates are not significantly different between Perth and Batalling, but for $\alpha$, the difference approaches significance (larger for Perth). Thus there is some evidence that Locus 1.3 discriminates between Perth and Batalling, whereas Locus 3.3.1 discriminates marginally at best. We discuss the interpretation of the parameters further in Subsection 5.1.

Figure 1 displays the resampling histograms (5000 resamples) for the Locus 3.3.1...
estimates. Even given such a small data set, the distributions are reasonably close to normal and their spreads are well summarised by the standard deviations in Table 4. For Locus 1.3, we omit the histograms which were essentially similar but less well determined as is to be expected from the small sample size at this locus.

5.1 Interpretation of parameters $\alpha$ and $r$

The important distinguishing feature of the PD$_{\alpha}^{(r)}$ distribution over PD$_\alpha$ is its allowance for over-dispersion in the data. This arises from its being based on an underlying negative binomial point process rather than a Poisson point process. To investigate what features of the data the parameter estimates capture, we compare the $\alpha$ values with those obtained from a fit of the original Kingman-Perman PD$_\alpha$ model. The parameter $\alpha$ in PD$_\alpha$ can be regarded as a measure of diversity (“$\alpha$-diversity” Pitman (2006), p.71).

For Locus 1.3, there is significant evidence of differences between locations in the estimates of both $\alpha$ and $r$ from PD$_{\alpha}^{(r)}$ (Table 3). At this locus, for PD$_{\alpha}^{(r)}$ we have $\hat{\alpha} = 0.4158$ (SE=0.0410) for Perth, and $\hat{\alpha} = 0.2190$ (SE=0.0208) for Batalling. For comparison, the $\alpha$ estimates for the PD$_\alpha$ model for this locus are:

for Perth : $\hat{\alpha} = 0.3880$ (SE = 0.1208); for Batalling : $\hat{\alpha} = 0.2676$ (SE = 0.0990).

These are quite similar to those from the PD$_{\alpha}^{(r)}$ model, but the standard errors for $\hat{\alpha}$ from PD$_\alpha$ are considerably larger than from PD$_{\alpha}^{(r)}$. Interpreted as a diversity measure, the $\alpha$ estimates from either model suggest that diversity is greater for Perth than for Batalling for this locus. To demonstrate this alternatively, we can calculate the Shannon entropy index for each location, using the formula

$$1H = -\sum_{i=1}^{k} \pi_i \ln \pi_i$$

(5.1)

where $\pi_i = n_i/n$, $1 \leq i \leq k$, are the allele proportions. For Perth we obtain 1.99 for $1H$, for Batalling we get 1.86. Although not significantly different these are in accord with the $\hat{\alpha}$ values.

Now consider gene Locus 3.3.1. The $\alpha$ estimates for the PD$_\alpha$ model for this are:

for Perth : $\hat{\alpha} = 0.5260$ (SE = 0.1134); for Batalling : $\hat{\alpha} = 0.4026$ (SE = 0.1009).

These compare with estimates 0.4814 (SE=0.0512) and 0.3718 (SE=0.0310), respectively, for PD$_{\alpha}^{(r)}$ (Table 4). We see that the $\hat{\alpha}$ for PD$_\alpha$ do not differ significantly

See W.B. Sherwin, Chao, Jost, & Smouse (2017) for a recent review of the use of entropy and related indices in measuring allelic diversity.
between locations, and are quite similar to those from the PD_{\alpha}^{(r)} model. Again the standard errors for $\hat{\alpha}$ from PD_{\alpha} are considerably larger than from PD_{\alpha}^{(r)}$. Likewise, the estimates of $r$ from PD_{\alpha}^{(r)}$, namely, $\hat{r} = 2.08$ (SE=0.52) for Perth, and $\hat{r} = 2.27$ (SE=0.29) for Batalling, do not differ significantly between locations, for Locus 3.3.1.

For the parameter $r$ we can provide a nice intuitive interpretation. From Gregoire’s formulation of the negative binomial process we see that, for a Borel subset $A$ of $(0, \infty)$,

$$E(B^{(r)}(A)) = r\Lambda(A), \tag{5.2}$$

where $\Lambda(dx) = \alpha x^{-\alpha-1}dx1_{0<x<1}$, and

$$\text{Var}(B^{(r)}(A)) = r(\Lambda(A) + \Lambda^2(A)) \tag{5.3}$$

(see Proposition 3.3 of Gregoire (1984)). So expectations and variances are proportional to $r$, and in (5.3) we also see the extra-Poisson dispersion effect of the negative binomial. This scaling effect of $r$ transfers through to the data analyses.

PD_{\alpha}^{(r)} does not reduce to PD_{\alpha} when $r = 0$; rather, the convergence of PD_{\alpha}^{(r)} to a Poisson-Dirichlet model takes place as $r \to \infty$ (see Ipsen, Maller & Shemehsavar (2018)), consistent with the convergence of a negative binomial variable to a Poisson for large values of the appropriate index. There is no evidence of the $r$ values going out of bounds in Tables 3 and 4.

In summary, there is reasonable evidence that both $\alpha$ and $r$ in the PD_{\alpha}^{(r)} model discriminate between locations for Locus 1.3, but not for Locus 3.3.1. But the PD_{\alpha} model provides no discrimination at either locus. By contrast, Firestone et al. (2000) found a tendency for, but no significant difference in, allelic diversity between the Perth and Batalling quolls.

6 Simulations

In this section we report on some simulations aimed at examining the properties of the parameter estimates. The procedure is the same as described in Section 4 but we stipulated “true” values $\alpha = 0.2$ and $r = 1$ rather than estimated ones. There are two levels of variability to consider, which we term “population” and “resampling” variability.

First we describe the resampling scheme, then how the population simulation is superimposed on it. The “resampling” part is as described in Steps 1–4 of Section 4 having fixed a number of blocks, $k$, set the population probabilities $(p_1, \ldots, p_k)$ as in 1.2, based on the $\tilde{p}_j$ in 1.2. Draw the $L$ samples $(\tilde{m}_j(\ell))$, $j = 1, 2, \ldots, n$, $\ell = 1, 2, \ldots, L$, and from them find least squares estimates, denoting them as $(\hat{\alpha}_1, \hat{r}_1)$. These form the first simulation estimates, indexed as $q = 1$. Repeat this entire
procedure $Q$ times to obtain the simulation sample $(\hat{\alpha}_q, \hat{r}_q)$, $q = 1, 2, \ldots, Q$. We can average these replications to form a simulation estimate of the original $(\alpha, r) = (0.2, 1)$ and display them in a histogram to assess precision.

At the “population” simulation stage, repeat this entire procedure. Still with the same “true” $\alpha$ and $r$, draw a new set of $\tilde{p}_j$ values from (4.2), recalculate the corresponding $p_j$ from (4.3), and then resample $Q$ times as described in the resampling stage. Repeat this whole procedure a total of $N$ times.

The resulting entries in Table 5 are the averages over the $N = 100$ population simulations of the $L = 1000$ resampling estimates in each population simulation.

| Resampling estimates | $k = 2$ $k = 5$ $k = 10$ | $k = 2$ $k = 5$ $k = 10$ | $k = 2$ $k = 5$ $k = 10$ |
|----------------------|-----------------|-----------------|-----------------|
|                      | $n = 20$ $n = 40$ | $n = 100$ $n = 200$ | $n = 200$ $n = 400$ |
| $\hat{\alpha}$      | 0.198 0.185     | 0.221 0.210     | 0.254 0.239     |
| SE$_R(\hat{\alpha})$| 0.023 0.032     | 0.037 0.030     | 0.075 0.055     |
| SE$_P(\hat{\alpha})$| 0.028 0.033     | 0.052 0.043     | 0.086 0.059     |
| $\hat{r}$           | 1.24 1.23       | 1.25 1.31       | 1.19 1.43       |
| SE$_R(\hat{r})$      | 0.45 0.52       | 0.47 0.56       | 0.32 0.66       |
| SE$_P(\hat{r})$      | 0.49 0.53       | 0.51 0.58       | 0.36 0.67       |

Table 5: Simulation results: True values: $\alpha = 0.2$, $r = 1$. 1000 resamples (R) and 100 population redraws (P).

7 Discussion

Properties of the PD$^{(r)}_{\alpha}$ estimators. Given the paucity of the data from which we are trying to make inferences, the estimates are surprisingly good. The resampling procedure produces distributions consistent with the asymptotic analysis in Appendix A.2 which suggests approximate normality should apply in large samples. The simulations reinforce this view. Overall the results suggest that the weighted least squares method provides reliable and robust estimates.

It is notable that the $\alpha$ estimates are very similar for PD$_{\alpha}$ and PD$^{(r)}_{\alpha}$ for the quolls data. This suggests the presence of an underlying Stable($\alpha$) structure for the population in which overdispersion is captured by the introduction of parameter $r$. We observe that introducing $r$ accounts for much of the variability in estimating $\alpha$.

Other Applications. Besides species and gene sampling models, there are many other applications of Poisson-Dirichlet distributions, among which we mention the areas of Bayesian nonparametrics and machine learning, which are currently under vigorous development. We expect the PD$^{(r)}_{\alpha}$ model to be useful as a prior distribution
when over-dispersion is present in the data. Many machine learning models such as the latent Dirichlet allocation models (Blei, Ng & Jordan (2003)) or mixed membership models (Ghahramani & Griffiths (2006)) deal with data, e.g. corpora of documents, which possess large underlying variation. With the flexibility provided by the over-dispersion parameter $r$, one could in principle select a more appropriate prior distribution for analysis.

Concluding Remarks. The methods we illustrate potentially have implications for the management and conservation of threatened populations. Measuring the level of genetic diversity of a population, and comparing levels between populations, are of major importance in determining conservation strategies and providing guidance for reintroduction programs. The study of Firestone et al. (2000) is the first to attempt to determine the level of diversity of the western quolls, with a specific objective of being able to distinguish genetically between the two subgroups and between other Australian quoll species. The data in this case is necessarily small-sample, but the $\text{PD}_\alpha^{(r)}$ analysis contributes useful information regarding diversity even in this case. Similar small-scale data sets are commonly found in the conservation literature: see, e.g., Campos, Posada & Moran (2006), Khosravi, Hemami, Malekian, Silva, Rezaei & Brito (2018). Without sound knowledge of the genetic diversity within and between .. populations ... it is difficult to determine which populations are valuable sources for reintroductions. (Firestone et al. (2000)).

Our $\text{PD}_\alpha^{(r)}$ model is an extension of the $\text{PD}_\alpha$ model, which is a subclass of the general $\text{PD}^{(\alpha, \theta)}$ models due to Pitman & Yor (1997). Statistical inference for the range of $\text{PD}^{(\alpha, \theta)}$ models, even for the one-parameter $\text{PD}_\alpha$ and $\text{PD}^{(\theta)}$ models, is still little understood. Carlton (1999) gives some discussion of estimation problems in these particular models. Our investigations suggest that the two-parameter $\text{PD}_\alpha^{(r)}$ holds promise of being a practical and useful alternative.

References

Blei, D.M., Ng, A. Y. & Jordan, M.I. (2003). Latent Dirichlet allocation. Journal of Machine Learning Research, 3(Jan), 99–1022.

Campos, J.L., Posada, D. and Morán, P. (2006) Genetic variation at MHC, mitochondrial and microsatellite loci in isolated populations of Brown trout (Salmo trutta). Conservation Genetics 7, 515–530.

Carlton, M. (1999). Applications of the Two-Parameter Poisson-Dirichlet Distribution. Unpublished Ph.D. thesis, Department of Statistics, University of California, Los Angeles.
Ewens, W. (1972). The sampling theory of selectively neutral alleles. *Theoret. Pop. Biol.*, 3, 87–112.

Firestone, K., Elphinstone, M.S., Sherwin, W.B., Houlden B.A. (2000) Variability and differentiation of microsatellites in the genus *Dasyurus* and conservation implications for the large Australian carnivorous marsupials. *Conservation Genetics* 1, 115–133.

Fisher, R.A., Corbet, A.S., Williams, C.B. (1943) The relation between the number of species and the number of individuals in a random sample of an animal population, *J. Anim. Ecol.*, 12, 42-58.

Ghahramani, Z. & Griffiths, T. L. (2006). Infinite latent feature models and the Indian buffet process. In: *Advances in Neural Information Processing Systems*, 475–482, MIT Press.

Gregoire, G. (1984). Negative binomial distributions for point processes. *Stoch. Proc. Appl.*, 16, 179–188.

Ipsen, Y.F. & Maller, R.A. (2016). Generalised Poisson-Dirichlet distributions and the negative binomial point process. *arXiv:1611.09980*.

Ipsen, Y.F. & Maller, R.A. (2018). Negative binomial construction of random discrete distributions on the infinite simplex. *Theory of Stochastic Processes*, 22, 34–46.

Ipsen, Y.F., Maller, R.A. & Shemehsavar, S. (2018) Limiting forms of some generalised Poisson-Dirichlet distributions, *submitted*.

Khosravi, R., Hemami, M-R., Malekian, M., Silva, T.L., Rezaei, H.R. and Brito, R.C. (2018) Effect of landscape features on genetic structure of the goitered gazelle (*Gazella subgutturosa*) in Central Iran. *Conservation Genetics* 19, 323–336.

Kingman, J. F. C. (1975). Random discrete distributions. *J. Roy. Statist. Soc. B*, 37, 1–22.

Patil, C.P. & Taillie, C. (1977). Diversity as a concept and its implications for random communities. *Bull. Int. Statist. Inst.* 47, 497–515.

Perman, M. (1990). *Random discrete distributions derived from subordinators*. PhD Thesis. Dept. of Statist., Univ. of California (Berkeley, CA).

Perman, M. (1993). Order statistics for jumps of normalised subordinators. *Stochastic Process. Appl.*, 46(2), 267–281.

Pitman, J. (2003). Poisson-Kingman partitions. *IMS Lecture Notes*, 40, 1–34.

Pitman, J. (2006). *Combinatorial Stochastic Processes*. Springer.

Pitman, J. & Yor, M. (1997). The two-parameter Poisson–Dirichlet distribution derived from a stable subordinator. *Ann. Prob.*, 25, 855–900.
8 Appendix A.1: The PD_{\alpha}^{(r)} Methodology

Here we outline the technology leading to (3.1), referring to the basic theory developed in [Ipsen & Maller (2016, 2018)] where necessary. Let \((P_i)_{i \in \mathbb{N}}\) be a random sequence in \(\nabla_{\infty}\) and \(G\) a random discrete distribution generated from it. With \(\delta_x\) denoting a point mass at \(x \in \mathbb{R}\), \(G\) can be written as

\[
G = \sum_{i \geq 1} P_i \delta_{Y_i},
\]

(8.1)

where \(P_1 \geq P_2 \geq \cdots\), and the \((Y_i)\) are independent and identically distributed (i.i.d.) random variables, independent of the \((P_i)\), having a “base” distribution \(G_0\).

Let \((X_i)\) be independent samples from \(G\). They generate a random partition \(\Pi\) consisting of non-overlapping “blocks”, by assigning \(i\) and \(j\) to the same block when \(X_i = X_j\). Then, conditionally given \(G\), the \(X_i\) are i.i.d. with distribution \(G\). We call \(G\) a species sampling model.

Let \(\Pi_n\) be the restriction of \(\Pi\) to the finite set \(\mathbb{N}_n := \{1, 2, \ldots, n\}\). The distribution of \(\Pi_n\) is such that for each partition \(\{A_1, A_2, \ldots, A_k\}\) of \(\mathbb{N}_n\) with \(#A_i = n_i, 1 \leq i \leq k\), we have

\[
P(\Pi_n = \{A_1, A_2, \ldots, A_k\}) =: p(n_1, \ldots, n_k)
\]

(8.2)

for some symmetric function \(p(\cdot)\) of sequences of positive integers, where \(n_i \geq 1\) and \(\sum_{i=1}^k n_i = n\). The function \(p(n_1, \ldots, n_k)\) is called the exchangeable partition probability function (EPPF) of \(\Pi\).

With \(P_i > 0\) denoting the size of the \(i\)th largest atom of a species sampling model \(G\), as in (8.1), let \(\tilde{P}_i\) denote the size of the \(i\)th atom discovered in the process of random sampling; equivalently, \(\tilde{P}_i\) is the asymptotic frequency of the \(i\)th class of \(\Pi\) in order of its appearance in the sample. The sequence \((\tilde{P}_i)\) is then a size biased permutation of \((P_i)\), and the EPPF in (8.2) satisfies [Pitman (2003), Eq. (3)]

\[
p(n_1, \ldots, n_k) = \mathbb{E}
\left(
\prod_{i=1}^k \tilde{P}_i^{n_i-1} \prod_{i=1}^{k-1} \left(1 - \sum_{j=1}^i \tilde{P}_j\right)
\right).
\]

(8.3)
Now let \( \{A_1, \ldots, A_k\} \) be a partition of \( \mathbb{N}_n \) generated by \( \text{PD}_\alpha^{(r)} \) and let \( (\tilde{J}_i)_{i \in \mathbb{N}} \) be the size biased permutation of the sequence \( J := (J_i)_{i \in \mathbb{N}} \) defined in \((2.3)\); thus, conditionally on \( J \), \( \tilde{J}_i \) takes value \( J_i \) with probability \( J_i/(r)T \); while for \( n \geq 1 \), conditional on \( J \) and \( \{\tilde{J}_1, \ldots, \tilde{J}_n\} \), \( \tilde{J}_{n+1} \) takes value \( J_i \) with probability \( J_i/(r)T - \sum_{j=1}^{i-1} J_j \). Here \((r)T \) is the sum of the points in \( \mathbb{B}(r) \). It has density \( g_r(t) \) (see \cite{Ipsen & Maller, 2016}) and compare with \((2.1)\) satisfying
\[
\int_0^\infty e^{-\lambda x} g_r(x) \, dx = \left( 1 + \int_0^\infty (1 - e^{-\lambda x}) \lambda \, dx \right)^{-1}, \quad \lambda > 0.
\] (8.4)

The next lemma gives a formula for the joint distribution of the \( \text{PD}_\alpha^{(r)} \)-partition with the size biased \( \tilde{J}_i \) and their sum. Recall \( \rho(x) = \alpha x^{\alpha-1} \mathbf{1}_{\{0 < x \leq 1\}} \) in \((2.2)\).

**Lemma 8.1.** For a partition \( \{A_1, A_2, \ldots, A_k\} \) of \( \mathbb{N}_n \) such that \( \#A_i = n_i \), \( 1 \leq i \leq k \), we have
\[
\mathbb{P}(\Pi_n = \{A_1, A_2, \ldots, A_k\}, \tilde{J}_i \in dx_i, 1 \leq i \leq k, (r)T \in dt) = r^k \left[ t^\sum_{i=1}^k x_i \right]^{\#A_k-1} \rho(x) \prod_{i=1}^k x_i^{n_i} \, dt.
\] (8.5)
where \( 0 \leq x_i \leq 1, x_i \neq x_j, 1 \leq i, j \leq k, \) and \( t > \sum_{i=1}^k x_i \).

**Proof of Lemma 8.1.** For \( \text{PD}_\alpha^{(r)} \), the \( \tilde{P} \) in \((2.3)\), conditional on \( \tilde{J}_i \) and \((r)T \), are replaced by \( \tilde{J}_i/(r)T \) so we have for the LHS of \((8.5)\)
\[
\prod_{i=1}^{k-1} \left[ \left( \frac{x_i}{t} \right)^{\#A_i-1} \left( 1 - \sum_{\ell=1}^{i-1} x_\ell \right) \right] \left( \frac{x_k}{t} \right)^{\#A_k-1} \rho \left( \tilde{J}_i \in dx_i, 1 \leq i \leq k, (r)T \in dt \right)
\]

Letting \( n_i := \#A_i, 1 \leq i \leq k, \) with \( \sum_{i=1}^k n_i = n \), the first factor on the RHS equals
\[
\frac{1}{t^{n-1}} \left( \prod_{i=1}^k x_i^{n_i-1} \right) \left( \prod_{i=1}^k \left( t - \sum_{\ell=1}^{i-1} x_\ell \right) \right).
\] (8.6)
Using Proposition 2.3 in \cite{Ipsen & Maller, 2016}, the second factor on the RHS equals
\[
r^k g_r(t) \left( t - \sum_{i=1}^k x_i \right) dt \prod_{i=1}^k \frac{\rho(x_i) x_i \, dx_i}{t - \sum_{\ell=1}^{i-1} x_\ell}.
\] (8.7)
Multiply \((8.6)\) and \((8.7)\) after observing that
\[
\prod_{i=1}^k \frac{1}{t - \sum_{\ell=1}^{i-1} x_\ell} = \prod_{i=1}^k \frac{1}{t - \sum_{\ell=1}^{i-1} x_\ell} = \prod_{i=1}^{k-1} \frac{1}{t - \sum_{\ell=1}^{i-1} x_\ell}
\]
(with the convention \( \sum_{\ell=1}^0 = 0 \)) to get \((8.5)\).

Next we derive expressions for the EPPF generated by \( \text{PD}_\alpha^{(r)} \).
Theorem 8.1. Two formulae for the EPPF of a PD\(^{(r)}\) \(-\)partition are:

\[
p(n_1, \ldots, n_k) = r[k] \int_{x_1=0}^{\infty} \cdots \int_{x_k=0}^{\infty} \frac{g_{r+k}(w) \prod_{i=1}^{k} \rho(x_i)x_i^{n_i}}{(w + \sum_{i=1}^{k} x_i)^n} \, dw \, dx_1 \cdots dx_k \quad (8.8)
\]

and

\[
p(n_1, \ldots, n_k) = \alpha^{k}r[k] \frac{\prod_{i=1}^{k} \Gamma(n_i)}{\Gamma(n)} \prod_{i=1}^{k} \Gamma(n_i - \alpha) \times \int_{\lambda=0}^{\infty} \frac{\lambda^{n_i-1}}{\Psi(\lambda)^{r+k}} \prod_{i=1}^{k} G_{n_i-\alpha}(\lambda) \, d\lambda, \quad (8.9)
\]

where \(\Psi\) is defined in (3.2) and \(G_{n_i-\alpha}(\lambda)\) is the incomplete gamma function.

Proof of Theorem 8.1: Use the change of variable \(t - \sum_{i=1}^{k} x_i = w\) in (8.5), then integrate out with respect to \(x_i\), \(1 \leq i \leq k\), and \(w\), to get (8.8). Formula (8.9) follows by applying the identity

\[
\int_{\lambda=0}^{\infty} \frac{\lambda^{n_i-1} e^{-\lambda w}}{(\Psi(\lambda)^{r+k})} \prod_{i=1}^{k} G_{n_i-\alpha}(\lambda) \, d\lambda
\]

valid for any \(x > 0\), to (8.8) to deduce

\[
\int_{w=0}^{\infty} \frac{g_{r+k}(w) \prod_{i=1}^{k} \rho(x_i)x_i^{n_i}}{(w + \sum_{i=1}^{k} x_i)^n} \, dw
\]

\[
= \frac{1}{\Gamma(n)} \int_{\lambda=0}^{\infty} \lambda^{n_i-1} e^{-\lambda (w + \sum_{i=1}^{k} x_i)} g_{r+k}(w) \, d\lambda
\]

\[
= \frac{1}{\Gamma(n)} \int_{\lambda=0}^{\infty} (\Psi(\lambda)^{r+k})^{-1} \prod_{i=1}^{k} e^{-\lambda x_i} \, d\lambda \quad \text{by (8.4)}.
\]

Multiply this by \(r[k] \prod_{i=1}^{k} \rho(x_i)x_i^{n_i}\) then integrate with respect to the \(x_i\) to obtain (8.9). □

Proof of Theorem 3.1: Choose \(m = (m_1, m_2, \ldots, m_n)\) such that \(\sum_{j=1}^{n} m_j = k\) and \(\sum_{j=1}^{n} jm_j = n\). The probability that \(\Pi_n\) has \(m_j\) blocks of size \(j\), \(1 \leq j \leq n\), is

\[
\mathbb{P}(M = m) = n! \prod_{j=1}^{n} m_j! p(n_1, \ldots, n_k) = \frac{n!}{\prod_{j=1}^{n} j!^{m_j} m_j!} p(n_1, \ldots, n_k),
\]

where \(N_k(m)\) is the number of orderings of the sample prescribed by \(m\). Substitute for \(p(n_1, \ldots, n_k)\) from (8.9), then observe that \(\prod_{i=1}^{k} G_{n_i-\alpha}(\lambda) = \prod_{j=1}^{n} (G_{j-\alpha}(\lambda))^{m_j}\), and adjust the constant factor, to get (3.1). □

9 Appendix A.2: Asymptotics of Estimators

In this section we derive a useful representation for the distribution of the species counts as a mixture of independent Poisson rvs, and use it to suggest the asymptotic
distribution of the least squares estimators. Write (3.1) in the form
\[
P(M = m) = \int_0^\infty \frac{r^k \lambda^{k-1}}{\Psi(\lambda)^{r+k}} \prod_{j=1}^n \frac{1}{m_j!} \left( \frac{1}{j!} \int_0^\lambda \alpha^{j-\alpha-1} e^{-x} \, dx \right)^{m_j} \, d\lambda
\]
\[
= \int_0^\infty \frac{r^k \lambda^{k-1}}{\Psi(\lambda)^{r+k}} \prod_{j=1}^n \left( \frac{e^{-F_j(\lambda)} F_j(\lambda)^{m_j}}{m_j!} \right) e^{T_n(\lambda)} \, d\lambda,
\]
where the \( F_j(\lambda) \) and \( T_n(\lambda) \) are defined by
\[
F_j(\lambda) := \frac{1}{j!} \int_0^\lambda \alpha^{j-\alpha-1} e^{-x} \, dx
\]
and
\[
T_n(\lambda) := \sum_{j=1}^n F_j(\lambda) = \alpha \int_0^\lambda \sum_{j=1}^n \frac{x^{j-\alpha-1}}{j!} e^{-x} \, dx.
\]
Let \( (N_j(\lambda))_{1 \leq j \leq n} \) be \( n \) independent Poisson rvs with \( \mathbb{E} N_j(\lambda) = F_j(\lambda) \). Then
\[
P(M = m) = \int_0^\infty \frac{r^k \lambda^{k-1}}{\Psi(\lambda)^{r+k}} P(N_j(\lambda) = m_j, 1 \leq j \leq n) e^{T_n(\lambda)} \, d\lambda
\]
(9.2) represents the distribution of \( M_n \) as a mixture of independent Poisson rvs. As \( n \to \infty \) we have the finite limit
\[
\lim_{n \to \infty} T_n(\lambda) = \alpha \int_0^\lambda x^{-\alpha-1} (1 - e^{-x}) \, dx := T(\lambda).
\]
(9.3)
To estimate \( \alpha \) and \( r \) we minimise the sum of squares in (4.1) by setting its derivative equal to 0. So to obtain \( (\hat{\alpha}_n, \hat{r}_n) \) we solve the unbiased estimating equation
\[
\sum_{j=1}^n w_j (M_j - E_j(\alpha, r)) \frac{\partial E_j(\alpha, r)}{\partial (\alpha, r)} = 0.
\]
(9.4)
A Taylor expansion and standard arguments show that \( (\hat{\alpha}_n, \hat{r}_n) \) will be asymptotically normally distributed under some regularity conditions if the LHS of (9.4) is asymptotically normal.

From (9.2), we can obtain an expression for linear combinations such as those in (9.3) as corresponding linear combinations of the \( N_j(\lambda) \), and it is then plausible that the estimates will be close to normally distributed after appropriate norming and centering. Working this out in detail is a highly technical exercise; we have to keep in mind the restrictions \( \sum_{j=1}^n m_j = k \) and \( \sum_{j=1}^n jm_j = n \) implicit in (9.2), and the regularity conditions that have to be imposed are not at all transparent, as is often the case with this kind of analysis.

We do not attempt to write these conditions out explicitly since they cannot be checked easily in practice anyway, but the distributions of the estimates shown in the resampling histograms are reasonably close to normal with a spread which is accurately described by the resampling standard errors. In practice, we should check on closeness to normality by using a resampling scheme such as we suggest, or some other bootstrapping or similar method.
Figure 1: Resampling Histograms of Estimates ($\hat{\alpha}, \hat{r}$) for Quolls, Locus 3.3.1