



LABORATOIRE DE PROBABILITÉ, STATISTIQUES ET MODÉLISATION

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Internship report notes

Master 2 - Data Science

Alexandre CHAUSSARD

1 Results to check and/or review

List of results that would require a **review**:

- Proposition 6.5: check the validity of the fixed point algorithm part using the provided reference.

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2 Introduction

Given observations of random variables (X, Y) , we suppose that there exist another set of random variables Z that we do not observe, yet that characterize (X, Y) conditionally to Z . Z is then called a latent variable, or a hidden variable.

For instance, if we observe the weights of a given population through X , and that we aim at inferring their height Y , knowing the sex of each individual through Z could improve our predictions on Y . Hence, assuming that there exist a latent variable to a given model adds structure to the model while improving the explainability, as Z characterizes the behavior of our dataset. Typically, clustering methods like KMeans or Gaussian Mixtures Models (GMM) provide a discrete Z given the observations, which are interesting in the sense that they provide a categorical representation of our data.

However, finding such Z given the observations is not straight forward, and not all dataset respond to a latent process, and may be not even a discrete one.

During this research internship, we aim at exploring latent models with discrete latent space in order to analyze the microbiota structure. Our first focus will be on various methods to conceive latent models like Expectation-Maximization and variational models.

3 Expectation-Maximization

3.1 Overview

The Expectation-Maximization (EM) algorithm, first introduced in [3], is vast class of latent models. It is based on the following smart decomposition of the data:

$$\log p_\theta(X) = \underbrace{\mathbb{E}_{p_{\hat{\theta}}(Z|X)}[\log p_\theta(X, Z)|X]}_{Q(\hat{\theta}, \theta)} - \mathbb{E}_{p_{\hat{\theta}}(Z|X)}[\log p_\theta(Z|X)|X]$$

The idea behind this decomposition is that $\log p_\theta(X)$ is generally not tractable since it's an integral, while the complete likelihood $p_\theta(X, Z)$ is generally manageable. Note that since $\log p_\theta(X)$ can not be computed, $\log p_\theta(Z|X)$ can't either by extension. Hence, we introduce $\log p_{\hat{\theta}}(Z|X)$ where $\hat{\theta}$ is the maximum likelihood estimator of θ :

$$\hat{\theta} = \arg \max_{\theta} \log p_\theta(X)$$

The main trick of the EM algorithm relies in the idea that $Q(\hat{\theta}, \theta)$ is sufficient to compute a maximum likelihood estimator of θ . Indeed, consider the following algorithm:

Algorithm 1 Expectation-Maximization

Require: $\hat{\theta}$

Repeat until convergence

Expectation: compute $p_{\hat{\theta}}(Z|X)$ to compute $Q(\hat{\theta}, \cdot)$

Maximization: $\hat{\theta} = \arg \max_{\theta} Q(\hat{\theta}, \theta)$

return $\hat{\theta} = 0$

If we denote by $\hat{\theta}^h$ the iterates of this algorithm, one can show using Jensen's inequality that:

$$\log p_{\hat{\theta}^{h+1}}(X) \geq \log p_{\hat{\theta}^h}(X)$$

As a result, the EM algorithm maximizes the likelihood, producing an estimator $\hat{\theta}$ that is an MLE of θ . Note that we don't have a convergence certainty towards the best maximizer of the likelihood, only to a local maxima. Hence, the EM algorithm is heavily sensitive to the initialization we pick.

All is required now is to choose a latent model so we can perform the EM algorithm, meaning that we have to define the distributions of the followings:

- $Z \sim \mathcal{B}(K, \pi)$, conveniently set to a binomial of parameter π so that it's discrete and simple to manage.
- $X|Z = k \sim p_{\gamma(k)}(X|Z = k)$, which is where we have the most choice to make.

In such models, $\theta = (\pi, \gamma(0), \dots, \gamma(K))$. In the next section, we will study a specific fork architecture of the gaussian mixture case.

3.2 Gaussian Mixture Linear Classifier

3.2.1 Framework and computations

Consider the case for which we observe $(X_i, Y_i)_{1 \leq i \leq n}$ i.i.d samples, where X_i denotes a feature vector and Y_i a label in a classification framework. We aim at introducing a linear classifier that exploits a latent structure over (X, Y) , so that we have the following latent model:

- $Z_i \sim \mathcal{B}(K, \pi)$, we note $\pi_k = \mathbb{P}(Z_i = k)$
- $X_i | Z_i = k \sim \mathcal{N}(\mu_k, \sigma_k I)$, we denote by $f_k(X_i)$ its density.
- $\mathbb{P}(Y_i = 1 | X_i, Z_i = k) = \sigma(W_{e,k}^T e_k + W_{x,k}^T X_i) = p_k(X_i)$, where e_k denotes a vector from the canonical basis of \mathbb{R}^K .

As we want to perform the EM algorithm find an MLE of

$$\theta = (\pi, \mu_1, \dots, \mu_K, \sigma_1, \dots, \sigma_K, W_{e,1}, \dots, W_{e,K}, W_{x,1}, \dots, W_{x,K})$$

we start by computing the **expectation** step by assessing $p_{\hat{\theta}}(Z_i = k | X_i, Y_i)$, using Bayes rules ($\hat{\pi}, \hat{p}, \hat{f}$ signify that we evaluate these quantities using the current estimate $\hat{\theta}$):

$$\begin{aligned} p_{\hat{\theta}}(Z_i = k | X_i, Y_i) &= \frac{\hat{\pi}_k \hat{f}_k(X_i) (Y_i \hat{p}_k(X_i) + (1 - Y_i)(1 - \hat{p}_k(X_i)))}{\sum_{j=1}^K \hat{\pi}_j \hat{f}_j(X_i) (Y_i \hat{p}_j(X_i) + (1 - Y_i)(1 - \hat{p}_j(X_i)))} \\ &= \tau_{ik} \end{aligned}$$

Now, we can safely evaluate $Q(\hat{\theta}, \theta)$ for any θ :

$$\begin{aligned} Q(\hat{\theta}, \theta) &= \mathbb{E}_{p_{\hat{\theta}}(Z|X)}[\log p_{\theta}(X, Y, Z) | X] \\ &= \sum_{i=1}^n \sum_{k=0}^K \log p_{\theta}(X_i, Y_i, Z_i = k) \tau_{ik} \\ &= \sum_{i=1}^n \sum_{k=0}^K (\log \pi_k + \log f_k(X_i) + Y_i \log p_k(X_i) + (1 - Y_i) \log(1 - p_k(X_i))) \tau_{ik} \end{aligned}$$

The **maximization** step now consists in deriving $Q(\hat{\theta}, \theta)$ regarding each parameters in θ so that we obtain an either explicit value or iterative procedure to compute the next iterate of $\hat{\theta}$.

- Maximization regarding π_k under constraint that $\sum_{k=0}^K \pi_k = 1$ can be solved explicitly using Lagrange duality:

$$\pi_k^* = \frac{1}{n} \sum_{i=1}^n \tau_{ik}$$

- Maximization regarding (μ_k, σ_k) is given by the maximum of likelihood estimator on $\sum_{i=1}^n \tau_{ik} \log f_k(X_i)$:

$$\begin{aligned} \mu_k^* &= \frac{1}{\sum_{i=1}^n \tau_{ik}} \sum_{i=1}^n \tau_{ik} X_i \\ \sigma_k^* &= \frac{1}{\sum_{i=1}^n \tau_{ik}} \sum_{i=1}^n \tau_{ik} (X_i - \mu_k^*)(X_i - \mu_k^*)^\top \end{aligned}$$

-
- Maximization regarding $(W_{e,k}, W_{x,k})$ is not explicit, and requires a fixed point algorithm like gradient descent to determine an estimate of the optimal parameters. The iterations using full batch gradient descent are given below, with learning rate α :

$$W_{e,k}^{l+1} \leftarrow W_{e,k}^l - \alpha \sum_{i=1}^n (p_k(X_i) - Y_i) \tau_{ik} e_k$$

$$W_{x,k}^{l+1} \leftarrow W_{x,k}^l - \alpha \sum_{i=1}^n (p_k(X_i) - Y_i) \tau_{ik} X_i$$

Each iteration should be confronted to the maximization criterion, so that each iterate improves $Q(\hat{\theta}, \theta)$:

$$Q(\hat{\theta}^{l+1}, \theta) \geq Q(\hat{\theta}^l, \theta)$$

In the end, only the best improvement iterate is kept for $W_{e,k}^*$ and $W_{x,k}^*$. Note that other methods could be used like SGD or CMAES as implemented during the internship.

After the maximization, we can update $\hat{\theta}$ with the previously computed parameters, and redo the (E) and (M) steps up to convergence. The convergence can be measured relatively to $Q(\hat{\theta}, \theta)$, so that for a threshold ϵ , we can use the following stopping criterion:

$$\frac{|Q(\hat{\theta}^{h+1}, \hat{\theta}^h) - Q(\hat{\theta}^h, \hat{\theta}^h)|}{|Q(\hat{\theta}^h, \hat{\theta}^h)|} \leq \epsilon$$

We now have a ready-to-go Gaussian Mixture Linear Classifier that we can benchmark on an suited dataset against other common methods.

3.2.2 Dataset generation

Before we benchmark the Gaussian Mixture Linear Classifier, we need to generate a dataset that is well suited to its usage. Hence, we set random parameters for θ and generate a new dataset following the latent framework we have set previously:

- For all $k \leq K$, generate n/K points following a gaussian parameterized by $(\mu_k, \sigma_k I)$. X is given by each point coordinate, Z by the gaussian from which the point was generated.
- For each sample, characterized by (X_i, Z_i) , draw the label of the sample as $Y_i \sim \mathcal{B}(\sigma(W_{e,Z_i}^T e_{Z_i} + W_{x,Z_i}^T X_i))$.

The following figure illustrates a generation with $K = 2$:

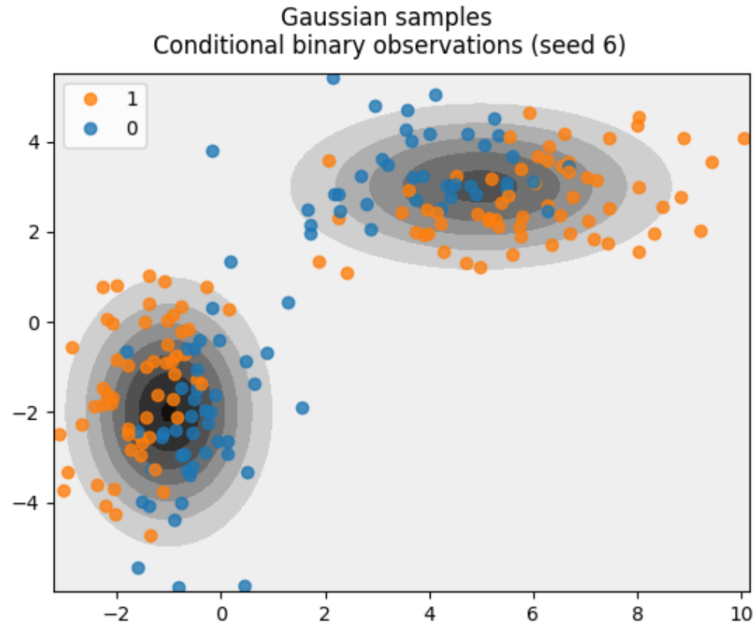


Figure 1: Generated samples out of $K = 2$ gaussians, labeled using a logistic model (label 1 or 0). The density of the hidden gaussians is represented in the background using shades of grey (black intense, white almost 0)

Note on the previous figure 1 that the limit between the labels in each subset gaussian is not sharp, as it is sorted out of a probabilistic modelisation (logistic). Consequently, an interesting observation can be made as we force the gaussians to have small values of mean and variance. Indeed, as shown on the next figure, if we take the same parameters as previously and divide them by a factor 10, the logistic model is ill conditioned.

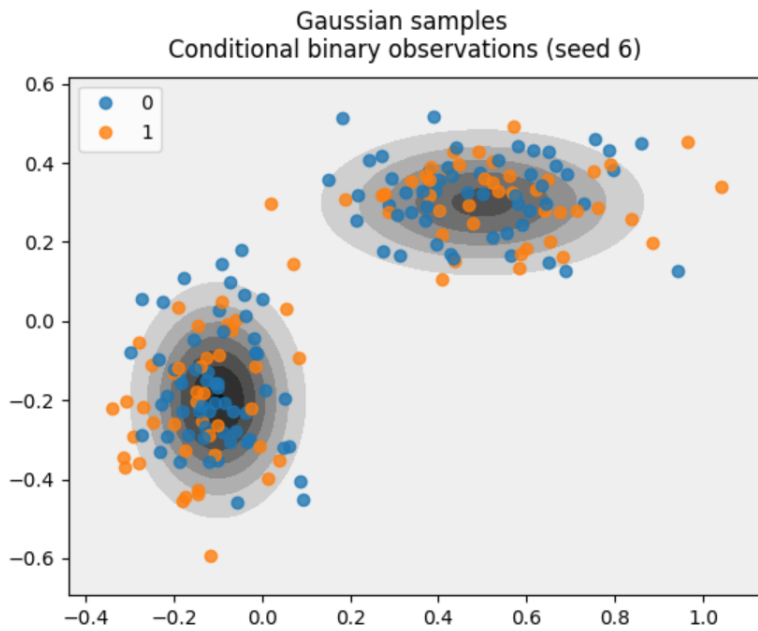


Figure 2: Generated samples out of $K = 2$ gaussians, using the same parameters as for figure 1 divided by a factor 10.

All points are close to the frontier in terms of norm on figure 2. Since we do not exploit any normalization term in the sigmoid modelization, this leads to a blurry area in which the linear separation model is not useful at all. This situation is heavily problematic as it prevents us from performing a general benchmark on that dataset. Indeed, the further larger the variance is, with sufficient samples, the better the linear approximation will be and therefore the better the modelization gets. On the contrary, with smaller variance the linear model isn't descriptive of the generated samples as they are all close to the frontier, therefore with heavily noisy labelization.

3.3 Dirichlet Mixture Linear Classifier

3.3.1 Framework and objectives

In the previous section, we have derived a very classical model (Gaussian Mixture Model) into a linear classifier using the EM algorithm. However, gaussian latent modelization is far from general, and may not be suited to our specific usage on the microbiota. Indeed, after performing the previous method onto our microbiota dataset, it turned out to be performing just as bad as the classical logistic regression, no matter the chosen latent space dimension. Therefore, we deduce that the gaussian latent modelization is not adapted to our practical settings.

As we analyze the data, we observe that each X_i belongs to the simplex. A natural distribution supported on the simplex is the Dirichlet distribution, parameterized by $\alpha = (\alpha_1, \dots, \alpha_p)$ where p is the dimension of X_i . We denote the dirichlet distribution by $\mathcal{D}(\alpha)$, for which the density is given by the following:

$$f(x|\alpha) = \frac{\Gamma\left(\sum_{j=1}^p \alpha_j\right)}{\prod_{j=1}^p \Gamma(\alpha_j)} \prod_{j=1}^p x_j^{\alpha_j-1}$$

where Γ denotes the gamma function. For notation simplicity, we also introduce the digamma function that will play a key role in our model:

$$\psi(\alpha) = \frac{d}{d\alpha} \log \Gamma(\alpha) = \frac{\Gamma'(\alpha)}{\Gamma(\alpha)}$$

Since we are doing a Dirichlet mixture model, we have K Dirichlet distributions to handle. We introduce the notation $\alpha^{(k)}$ to parameterize the k -th Dirichlet distribution.

As previously, we first perform the **expectation** step obtain the same result with a different conditional a priori distribution on $X|Z$:

$$\begin{aligned} p_{\hat{\theta}}(Z_i = k | X_i, Y_i) &= \frac{\hat{\pi}_k f(X_i | \hat{\alpha}^{(k)}) (Y_i \hat{p}_k(X_i) + (1 - Y_i)(1 - \hat{p}_k(X_i)))}{\sum_{j=1}^K \hat{\pi}_j f(X_i | \hat{\alpha}^{(j)}) (Y_i \hat{p}_j(X_i) + (1 - Y_i)(1 - \hat{p}_j(X_i)))} \\ &= \tau_{ik} \end{aligned}$$

We can now evaluate $Q(\hat{\theta}, \theta)$ for any θ , which enables us to perform the **maximization** step:

-
- The maximization over π_k under the simplex constraint $\sum_{k=1}^K \pi_k = 1$ is again given by Lagrange duality as:

$$\pi_k^* = \frac{1}{n} \sum_{i=1}^n \tau_{ik}$$

- The maximization over $\alpha^{(k)j}$ is not straightforward on the other hand, and requires a fixed point algorithm. Indeed, deriving over $\alpha^{(k)j}$ we obtain:

$$\begin{aligned} \partial_{\alpha_j^{(k)}} Q(\hat{\theta}, \theta) &= \sum_{i=1}^n \left(\psi \left(\sum_{l=0}^K \alpha_l^{(k)} \right) - \psi(\alpha_j^{(k)}) + \log x_{ij} \right) \tau_{ik} \\ &= \left(\psi \left(\sum_{l=0}^K \alpha_l^{(k)} \right) - \psi(\alpha_j^{(k)}) \right) \sum_{i=1}^n \tau_{ik} + \sum_{i=1}^n \tau_{ik} \log x_{ij} \end{aligned}$$

Hence, as we look for $\partial_{\alpha_j^{(k)}} Q(\hat{\theta}, \theta) = 0$, we obtain:

$$\psi(\alpha_j^{(k)}) - \psi \left(\sum_{l=0}^K \alpha_l^{(k)} \right) = \frac{\sum_{i=1}^n \tau_{ik} \log x_{ij}}{\sum_{i=1}^n \tau_{ik}}$$

Thankfully, [7] provides a few tricks to solve iteratively such equation, so that we can iterate as follows (5 steps are sufficient to obtain high-accuracy solution according to [7]):

$$\begin{aligned} \alpha_j^{(k)} &\leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \tau_{ik} \log x_{ij}}{\sum_{i=1}^n \tau_{ik}} + \psi \left(\sum_{l=0}^K \hat{\alpha}_l^{(k)} \right) \right) \\ \hat{\alpha}_j^{(k)} &\leftarrow \alpha_j^{(k)} \end{aligned}$$

However, this solution is a lower bound to the true objective, which makes our EM a generalized version of it.

- The maximization over $(W_{e,k}, W_{x,k})$ is also given by a fixed point algorithm, which ends up being the same computation as previously for the Gaussian case:

$$\begin{aligned} W_{e,k}^{l+1} &\leftarrow W_{e,k}^l - \alpha \sum_{i=1}^n (p_k(X_i) - Y_i) \tau_{ik} e_k \\ W_{x,k}^{l+1} &\leftarrow W_{x,k}^l - \alpha \sum_{i=1}^n (p_k(X_i) - Y_i) \tau_{ik} X_i \end{aligned}$$

3.3.2 Dataset generation

Now that we have defined the EM algorithm in the previous section, we aim at generating a dataset to benchmark the dirichlet mixture classifier. Following a similar procedure as for the gaussian case, we are able to generate a dataset that matches a dirichlet mixture and is labeled following the sigmoid modelisation. The next figure illustrates a given generation:

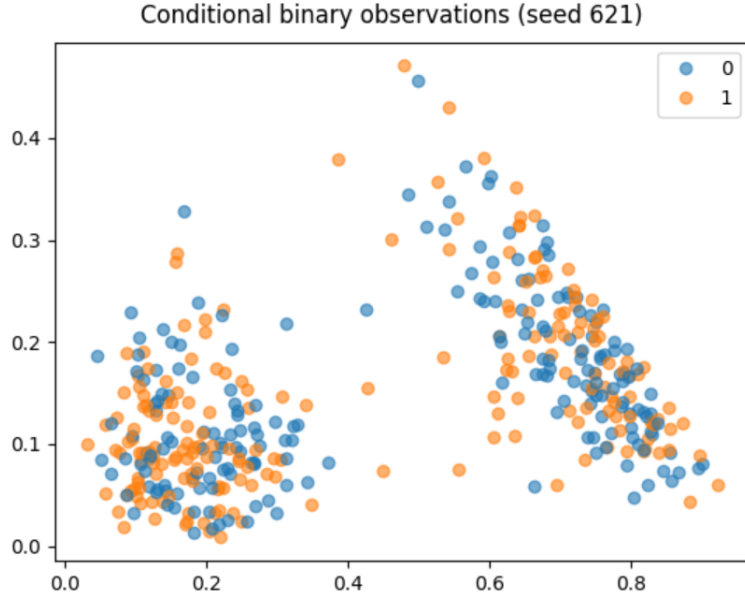


Figure 3: Generated samples out of $K = 2$ dirichlet distributions, labeled using the sigmoid modelisation on $\mathbb{P}(Y_i = 1)$.

As previously, since the data lives in the simplex, they are too close to the border of the next label set, which ends up creating a blurry dataset for which the linear model is not relevant anymore.

4 Variational methods

In this section, we aim at resourcing some variational methods that we are going to use in this study.

4.1 Variational Auto-Encoders

4.1.1 Framework and optimization objective

We are interested in another kind of latent models, this time based on variational inference results to achieve a new kind of deep latent structure: the Variational Auto-Encoder (VAE). These latent models were introduced in 2013 by Kingma, better described in a more in depth paper in 2019: see [6]

Once again, we assume the observations X to be modelizable by a given distribution parameterized by θ :

$$X \sim p_{\theta}(x)$$

Determining θ holds to find one θ^* that would optimize a given objective, generally chosen as the maximum of likelihood. Indeed, if $\theta^* \in \arg \max_{\theta} p_{\theta}(x)$, then such θ^* maximizes the density around the dense areas of the observations, which makes them highly likely to happen under such distribution p_{θ^*} . Hence, the maximum likelihood is a natural criterion:

$$\theta^* \in \arg \max_{\theta} p_{\theta}(x)$$

However, such modelization does not include a latent structure. As a result, we try to enforce it by rewriting the objective as follows:

$$p_{\theta}(x) = \int_{\mathcal{Z}} p(x, z) dz$$

Using Bayes decomposition, we obtain the following objective:

$$p_{\theta}(x, z) = p_{\theta}(z) p_{\theta}(x|z)$$

Recall that the prior $p_{\theta}(z)$ and the a priori $p_{\theta}(x|z)$ are defined by the framework (ex: Bernoulli prior and Gaussian posterior gives the Gaussian mixture framework). However, the computation of the evidence $p_{\theta}(x)$ is generally intractable in practice, which also leads to a non-tractable posterior distribution: $p_{\theta}(z|x)$. As a result, not being able to compute the evidence leads to not being able to provide a gradient regarding θ , so we can not perform the backpropagation in a deep learning approach.

Note that there exist approximate inference techniques to compute the evidence and the posterior, but these are quite expensive and often yield poor convergence results.

To overcome this issue, we introduce a smart rewriting of the objective using variational

inference. Indeed, let $q_\Phi(z|x) \approx p_\theta(z|x)$ to be learnt over Φ , one can write:

$$\begin{aligned} \log p_\theta(x) &= \mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x)] \\ &= \mathbb{E}_{q_\Phi(z|x)} \left[\log \frac{p_\theta(x)}{q_\Phi(z|x)} \frac{q_\Phi(z|x)}{p_\theta(z|x)} \right] \\ &= \underbrace{\mathbb{E}_{q_\Phi(z|x)} \left[\log \frac{p_\theta(x)}{q_\Phi(z|x)} \right]}_{ELBO(q_\Phi(z|x), p_\theta(x, z))} + D_{KL}[q_\Phi(z|x) || p_\theta(z|x)] \end{aligned}$$

The first term of that decomposition is generally called the Evidence Lower BOund (ELBO), as it marks a lower bound to the evidence $\log p_\theta(x)$ since the KL divergence is a positive quantity:

$$\log p_\theta(x) \geq ELBO(q_\Phi(z|x), p_\theta(x, z))$$

$q_\Phi(z|x)$ is an approximation of the true posterior $p_\theta(z|x)$ that we aim at learning in a family of distributions. For instance,

$$q_\Phi(\cdot|x) \sim \mathcal{N}(\mu(x), \Sigma(x))$$

would be an approximation of the true posterior by a Gaussian distribution. Notice that the true posterior may very not likely be Gaussian, which creates a first complexity error in our model.

Despite being a lower bound on the true maximum likelihood objective, the ELBO is actually tractable. Indeed, as we continue the computation:

$$\begin{aligned} ELBO(q_\Phi(z|x), p_\theta(x, z)) &= \mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x, z)] - \mathbb{E}_{q_\Phi(z|x)}[\log q_\Phi(z|x)] \\ &= \mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x|z)] - D_{KL}[\log q_\Phi(z|x) || p_\theta(z)] \end{aligned}$$

Another remarkable fact, is that when maximizing the ELBO, we are actually minimizing the KL divergence between the estimated and the true posterior. Hence, one can define the ELBO as a suboptimal objective to our problem that we get to maximize to obtain (Φ^*, θ^*) , the parameters of our model.

4.1.2 Reparameterization trick

Even though the gradient of the ELBO is well defined for θ , it is not possible to compute the differential relatively to Φ yet, as it requires samples from the approximation to the posterior $q_\Phi(z|x)$ to compute $\mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x|z)]$.

Since, sampling is not a differentiable operation, we make use of the change of variable formula, so that for a bijective transformation $z = \phi_x(\epsilon)$, we get:

$$p(z) = p(\epsilon) \det \left| \frac{\partial \epsilon}{\partial z} \right|$$

Hence, if we take ϵ a random variable of density $p(\epsilon)$ that does not depend on θ , Φ nor x , so that $z = \phi_x(\epsilon)$, for any L_1 function f ,

$$\mathbb{E}_{q_\Phi(z|x)}[f(z)] = \mathbb{E}_{p(\epsilon)}[f(z)]$$

As a result, the samples are not obtained through q_Φ anymore but through $p(\epsilon)$, so that can safely perform derivation of the ELBO relatively to Φ and backpropagate our gradient through the network.

4.1.3 Architecture

The vanilla architecture of the VAE is described by the following illustration:

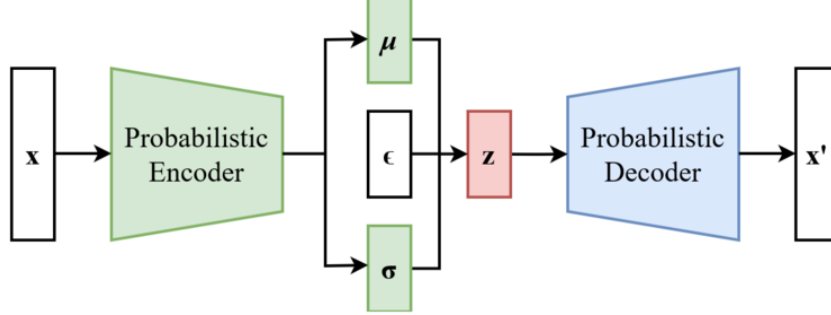


Figure 4: Illustration of a VAE with Gaussian prior (wikipedia)

The first part is generally called the encoder, as it turns a sample x into its latent representation z by modeling the posterior $q_{\Phi}(z|x)$. The second part is then called the decoder, as it throws a latent representation in the sample space. The latest can even serve as a generative architecture, as one can sample from the latent space through $q_{\Phi}(z|x)$, and decode it to obtain a new sample.

As we can see more clearly in that illustration, we can see that Φ and θ are trained jointly through the ELBO, both serving for one part of the VAE at a time.

The training procedure is straightforward: the entry is a sample x and the output objective is the same sample x . We aim at train the VAE for learning the data space and its latent representation by learning how to reconstruct the samples through it.

4.1.4 Limits for our problem

As we have seen through the reparameterization trick, training a VAE architecture requires to be able to backpropagate the gradient of the ELBO at each step. We namely had to perform the reparameterization trick to circumvent the randomness operation which is not differentiable. As a result, learning a discrete posterior is not possible with such architecture, since we would have to perform a projection of the output of the encoder on a discrete space, which is not a differentiable operation.

Yet, learning discrete representation of our data seems much more natural than continuous latent ones. As we tend to categorize things as much as we can, describing behaviors with words for instance. Furthermore, a discrete representation facilitates the interpretation of the latent space by ordering data distribution in simple bins.

The next architecture, called the VQ-VAE, stands as a first fork option to the VAE with discrete posterior.

4.2 VQ-VAE

4.2.1 Quick overview

Introduced in [10], VQ-VAE architecture provides a framework to compute discrete posterior distributions $q_\Phi(z|x)$. To compare that model with the VAE, we start by introducing the architecture of the model for which an illustration is given below:

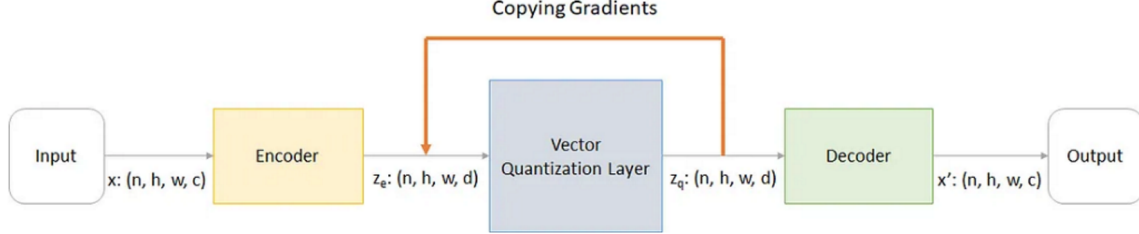


Figure 5: VQ-VAE architecture (source: Medium)

As we can see on figure 5, the major difference with the vanilla VAE architecture lies in the vector quantization step which enables to project the output of the encoder denoted by $z_e(x)$ onto a discrete embedding dictionary (e_1, \dots, e_K) by a simple distance argument:

$$k = \arg \min_j \|z_q(x) - e_j\|_2, \quad z_q(x) = e_k$$

The projection of $z_e(x)$ on that discrete dictionary is denoted by $z_q(x)$, and serves as the input of the decoder. For further visual representation of the vector quantization layer, an illustration is given below.

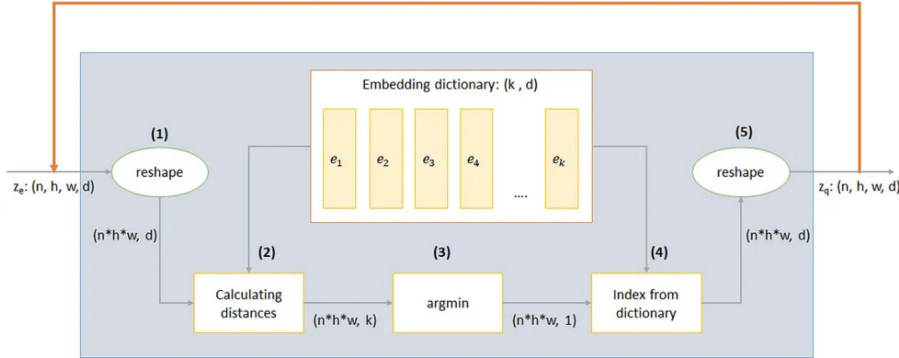


Figure 6: Architecture of the VQ-VAE: quantization layer (source: Medium)

Looking at the previous figures, we can grasp the challenge of backpropagation in such model with discrete prior and posterior. In the next section, we enter in the mathematical definition of the objective and how to train this architecture.

4.2.2 Framework and optimization objective

Contrary to the vanilla VAE, we have the following categorical distributions assumption:

-
- The prior $p_\theta(z)$ is categorical. In the original paper, it is taken as uniform supported in $\{1, \dots, K\}$ during the training. When the training is over, it is fit to an autoregressive distribution through a PixelCNN (see [9]). It is left as an exploration research field to be able to learn the prior while training the model.
 - The posterior $q_\Phi(z|x)$ is categorical and set to the following:

$$q_\Phi(k|x) = \mathbb{1}_{\{k=\arg \min_j \|z_q(x)-e_j\|_2\}}$$

This modelization of the posterior enables to obtain a discrete latent space, but it does not allow to differentiate regarding Φ . As a result, the authors suggest two possible strategies:

- *Straight-through*: propagate the gradient through the discrete part (vector quantization layer) without changing it. The intuition is that the gradient propagated from the encoder contains sufficient information to update the encoder accordingly, but this is just intuition.
- *Subgradient*: compute the subgradient of the quantization layer (unexplored yet)

The optimization objective of the VQ-VAE is based on the ELBO, that one can compute as follow:

$$\begin{aligned} ELBO(q_\Phi(z|x), p_\theta(z|x)) &= \mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x, z)] - \mathbb{E}_{q_\Phi(z|x)}[\log q_\Phi(z|x)] \\ &= \mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x|z)] - D_{KL}[q_\Phi(z|X)||p_\theta(z)] \end{aligned}$$

Notice then that:

- Since $q_\Phi(k|x) = \mathbb{1}_{\{k=\arg \min_j \|z_q(x)-e_j\|_2\}}$, we have:

$$\mathbb{E}_{q_\Phi(z|x)}[\log p_\theta(x|z)] = \log p_\theta(x|z_q(x))$$

- Since $Z \sim \mathbb{U}(\{1, \dots, K\})$, $\mathbb{P}(Z = k) = \frac{1}{K}$. Also, notice that $q_\Phi(z_q(x)|x) = 1$ by definition of $q_\Phi(z|x)$ and $z_q(x)$. Combining those results, we obtain:

$$\begin{aligned} D_{KL}[q_\Phi(z|x)||p_\theta(z)] &= \mathbb{E}_{q_\Phi(z|x)} \left[\log \frac{q_\Phi(z|x)}{p_\theta(z)} \right] \\ &= \log \frac{q_\Phi(z_q(x)|x)}{p_\theta(z_q(x))} \\ &= \log K \end{aligned}$$

Therefore, this KL divergence does not impact the optimization objective as it does not depend on (θ, Φ) .

Computing the previous quantities, we would obtain the following suboptimal objective for the VQ-VAE:

$$ELBO(\Phi, \theta) = \log p_\theta(x|z_q(x))$$

However, such objective does not enable to learn the dictionary since we use a straight-through approach over the quantization layer. To update the dictionary, the authors suggest to add the following term to the loss:

$$\|sg[z_e(x)] - e\|_2^2$$

Where sg denotes the stop-gradient operator, meaning we do not consider any gradient after the given operation.

Finally, to prevent embedding space over expansion, the authors suggest the addition of a commitment loss parameterized by $\beta > 0$:

$$\beta \|z_e(x) - sg[e]\|_2^2$$

Indeed, the embedding space was unconstrained so far, so its dimension could grow arbitrarily. Intuitively, adding that term forces the model to commit to a given embedding.

Overall, we obtain this final objective to maximize:

$$\mathcal{L}(\theta, \Phi) = \log p_\theta(x|z_q(x)) + \|sg[z_e(x)] - e\|_2^2 + \beta \|z_e(x) - sg[e]\|_2^2$$

4.2.3 Discussion over some limiting aspects

Even though the previous objective seems natural, we only rigorously justified the first term thanks to the ELBO. Indeed, the dictionary update loss as well as the commitment loss keep dropping out from nowhere, while they seem necessary to ensure the training of our model.

Furthermore, we did not really deal with the non differentiability of the vector quantization loss, and the straight-through estimator can definitely be criticized about what information it actually provides to the encoder to update appropriately.

Finally, the usage of the PixelCNN after the training seems very unnatural, and could significantly boost the model as the PixelCNN did show amazing performances so far.

4.3 PixelCNN

4.3.1 Overview

Introducing the VQ-VAE, we have seen that an under-table tool that was being used is the Pixel CNN, first introduced in [9]. In the context of the VQ-VAE, it plays a major role after training by learning the prior $p(z)$, that was set to a discrete uniform previously. This makes a drastic difference with the VAE, as we are not setting the prior ourselves as it's being learnt and modeled by the PixelCNN in this process.

Indeed, the PixelRNN/PixelCNN architectures are sequential deep neural networks that aims at modeling the distribution of a data space in an autoregressive fashion. Their main characteristic is that they take advantage of the structure of an image to learn the data space distribution.

- *PixelRNN*: Bi-directional recurrent networks with variable smart directions are used to model the spatial dependencies between pixels. (3 architectures are presented in the original paper, but our focus will be on the PixelCNN here).
- *PixelCNN*: the dependencies between the pixels is modeled through stacking of masked convolution layers (it's faster since the receptive field is bounded by the size of the convolution), no pooling layer is used. The masking ensures that we keep an autoregressive estimation of a new pixel, without seeing the future pixels.

The following figure illustrates the masked convolution technique.

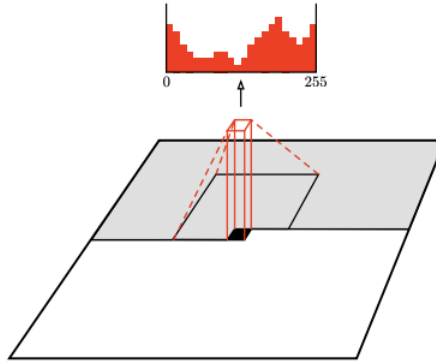


Figure 7: Masked convolution on an image (largest square), the big square represents the receptive field of the black pixel, the white area is masked

Hence, given x an image represented by its pixels $x = (x_1, \dots, x_n)$, as usual for these distribution modeling problems, we aim at finding $\theta^* = \arg \max_{\theta} p_{\theta}(x)$ in a family of distributions parameterized by θ : the maximum of likelihood. Contrary to the usual independent framework, we consider a local dependency between pixels given by the receptive field of our convolution. This local dependency is limited to the already seen pixels only as well, thanks to the masked convolution.

Furthermore, rather than using continuous outputs, these architecture use a softmax layer to determine the pixel of a given generation, leading to a discrete prior rather than a continuous

one, which is required for the VQ-VAE for instance. As a result, the distribution of a pixel conditionally to the ones in its receptive field is given by a multinomial in $\{0, \dots, 256\}$.

4.3.2 Usage in the VQ-VAE

Once we have trained the VQ-VAE, the original paper states that we can replace the uniform prior on $p(z)$ by a PixelCNN to model the prior. To perform the training of the PixelCNN, we turn the samples x in their latent representations z , and train the PixelCNN over the latent representations z . This way, we have created dependency between the z in the latent feature mapping, and we obtain a prior over their distribution modeled by the PixelCNN.

5 Microbiota analysis

This section aims at using the previous elements to design microbiota adapted latent methods.

5.1 Microbiota dataset

As we look into the microbiota data, we notice a major phylogenetic architecture to describe various levels of precision in the microbiota composition. Indeed, such phylogenetic structure can be represented as a tree as on the following figure:

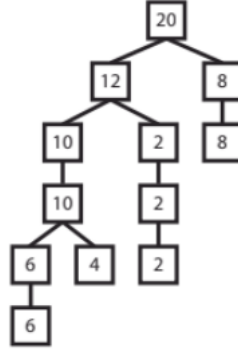


Figure 8: Phylogenetic tree example with abundance data (in the nodes) at each layer of the tree. Each node represents a bacterium species at a given precision layer in the tree. From [5].

Such structure can not be used directly in a machine learning system since it's not a vectorizable representation. Hence, we first suggest to transform the tree in a matrix to image structure as in the following example:

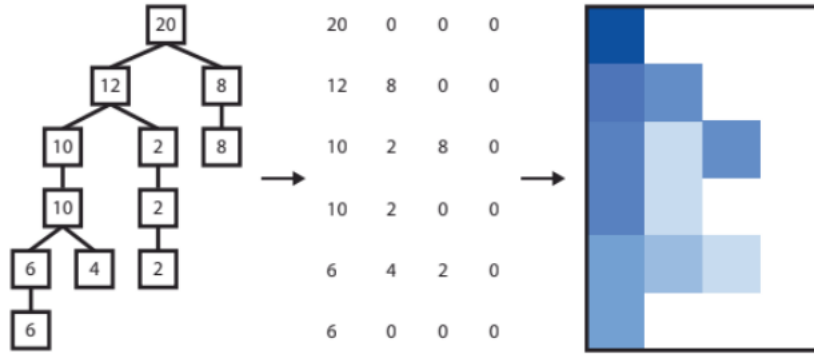


Figure 9: Phylogenetic tree to image representation: opacity of the pixel relates to the abundance of the species at the given level of precision (normalized between 0 and 1). From [5].

To give some notations, we introduce the following framework:

- The maximum depth of a phylogenetic tree is D , the maximum number of nodes N_T , the maximum amount of unique species at any level is U .
- X_i is the abundance matrix of the individual i (image of size $D \times U$), which is observed.

- T_i is the adjacent matrix of the phylogenetic tree of individual i , of size $N_T \times N_T$, which is observed. We could use any other encoding of a tree (contour function, depth function, ...).
- For a given tree, we will denote by U_ℓ the number of species at a given precision layer ℓ of the tree.
- We assume that we have n individuals observed through $(X_i, T_i)_{1 \leq i \leq n}$ i.i.d samples.

Note that all tree follow the same global architecture, with branches and nodes not always being activated. The following figure illustrates the global architecture at precision layer 4:

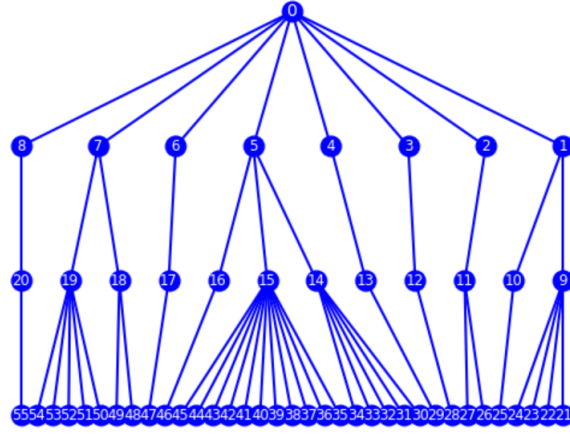


Figure 10: Taxonomy of the microbiota dataset (precision 4)

Notice that at this precision layer, we don't have any missing entries, which facilitates the current modelisation. The next figure illustrates a sample from the dataset as well, as taxa-abundance data represented through opacity.

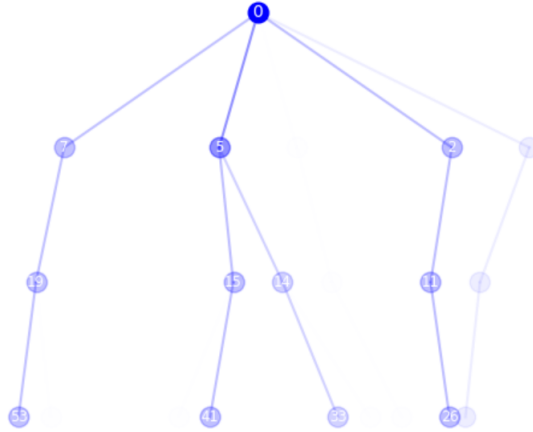


Figure 11: Taxa-abundance sample from the dataset (opacity relates the abundance value)

5.2 Mathematical context and objective without latent variables

In first approximation, we would like to define a generative model that does not exploit any latent structure. Such model, parameterized by θ , aims at finding an optimal distribution in

the sense of the maximum of likelihood, within a family of distributions yet to be defined. The maximum likelihood objective is given below as:

$$\theta^* = \arg \max_{\theta} p_{\theta}(X, T)$$

One can rewrite the joint distribution as follow:

$$\begin{aligned} p_{\theta}(X, T) &= \prod_{i=1}^n p_{\theta}(X_i, T_i) \\ &= \prod_{i=1}^n p_{\theta}(T_i) p_{\theta}(X_i | T_i) \end{aligned}$$

As a result, to compute this objective, we need to define a prior $p_{\theta}(T_i)$ that generates trees, and a posterior distribution $p_{\theta}(X_i | T_i)$ that generates abundance data from a sampled tree.

5.3 Markovian model without latent variables

5.3.1 Design of the prior

We aim at defining a parameterized distribution $p_{\theta}(T)$ from which one can sample trees. We would also like this distribution to model the trees of the microbiota dataset, meaning that the generated trees should look like the ones from the dataset as well, and respect the phylogenetic constraints.

Consequently, we introduce a first simple generative process to characterize $p_{\theta}(T)$ that we would call the markovian parenting tree generator. Before describing the generation method, let us introduce the framework of it:

- Describe T as a succession of L layers: $T = (T^{(1)}, \dots, T^{(L)})$. We assume that we have no missing data, so to say, all the leaves the tree are reaching the precision layer L .
- Describe a given layer ℓ as a discrete vector in $\{0, 1\}^U$. To each possible node at layer ℓ we can associate an index k so that we denote the nodes by $u_k^{(\ell)} \in \{0, 1\}$.

$$T^{(\ell)} = (u_1^{(\ell)}, \dots, u_{K_{\ell}}^{(\ell)})$$

A node $u_k^{(\ell)}$ is activated if it is valued as 1 in $T^{(\ell)}$, otherwise it is not.

- We introduce the function \mathcal{P} that takes a node $u_k^{(\ell)}$ as input, and output the parent of the node if it is well defined in T , otherwise it outputs 0 as if the parent was not activated:

$$\mathcal{P}(u_k^{(\ell)}) = \begin{cases} 1 & \text{if parent of } u_k^{(\ell)} \text{ exists and is activated} \\ 0 & \text{otherwise} \end{cases}$$

Now that the framework is clear and defined, we describe the generative process:

-
- The root node of the tree is deterministic, since all trees begin to the same root ancestor. Hence, we have:

$$p(T^{(1)}) = \delta_{e_1}(T^{(1)})$$

- For all $l \geq 2, k \in \{1, \dots, U\}$, we assume that the activation of the node $u_k^{(\ell)}$ is distributed as a Bernoulli conditionally to its parent activation, parameterized by $\pi_k^{(\ell)}$:

$$u_k^{(\ell)} | \left\{ \mathcal{P}(u_k^{(\ell)}) = 1 \right\} \sim \mathcal{B}(\pi_k^{(\ell)})$$

To respect the tree architecture, we assume that if $\mathcal{P}(u_k^{(\ell)}) = 0$ then the probability for the children to be activated is deterministic and set to 0, as a child can not exist without his parent.

- For now, we make the major assumption that all nodes are independent within a given layer conditionally to their parents, and they only depend on their respective parent, so that:

$$p\left(u_1^{(\ell)}, \dots, u_{K_\ell}^{(\ell)} | \mathcal{P}(u_1^{(\ell)}), \dots, \mathcal{P}(u_{K_\ell}^{(\ell)})\right) = \prod_{k=1}^{K_\ell} p\left(u_k^{(\ell)} | \mathcal{P}(u_k^{(\ell)})\right)$$

- The dependency between the layers of the tree is markovian:

$$p_\theta(T^{(\ell+1)} | T^{(1:\ell)}) = p_\theta(T^{(\ell+1)} | T^{(\ell)})$$

Noting these framework properties, we can describe the log-likelihood of such prior model on the trees using proposition 6.1:

$$p_\theta(T_1, \dots, T_n) = \sum_{i=1}^n \sum_{l=1}^{L-1} \sum_{k=1}^{K_\ell} \mathcal{P}(u_k^{(\ell+1)}) \left[u_k^{(\ell+1)} \log \pi_k^{(\ell+1)} + (1 - u_k^{(\ell+1)}) \log(1 - \pi_k^{(\ell+1)}) \right]$$

Looking at the previous formula, we obtain that such prior is parameterized by the activation probabilities $\pi_k^{(\ell)}$ of each node $u_k^{(\ell)}$. Furthermore, due to the indicator function expressed through $\mathcal{P}(u_k^{(\ell+1)})$, the update of a given activation probability will only be impacted by the trees which have the node $u_k^{(\ell)}$ in any branch.

Now that the prior of the trees is well defined, we would like to compute an optimal value of $\pi^{(\ell)} = (\pi_1^{(\ell)}, \dots, \pi_U^{(\ell)})$ in the sense of the maximum of likelihood. Using proposition 6.2, we obtain

$$\left(\pi_k^{(\ell)}\right)^* = \frac{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) u_{k,i}^{(\ell)}}{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)})}$$

This estimator is actually the common MLE for a Bernoulli parameter estimation, except that it limits the computation of the estimation to all trees that respect the root constraint, and that could possess the node $u_k^{(\ell)}$ since they must have the parent node $\mathcal{P}(u_k^{(\ell)})$.

5.3.2 Design of the posterior and maximum likelihood estimator

Now that we have a way to generate trees, we need to define an explicit stochastic relationship between the abundance data and the structure of the tree. Such inevitably exists, as when observing T , the abundance of an entity that isn't present in T is necessarily 0. Similarly, it is highly likely that when observing the presence of certain entities in T that induces a high abundance of another neighbour entity (interaction between bacteria).

For context, we recall that X is a matrix of shape (L, U) , where L is the maximum precision level of the trees (assumed to be the same for all trees for now) and U the maximum number of entities at each depth of the trees. We denote by $X^{(\ell)}$ the ℓ -th line of the abundance matrix, for which up to U_ℓ elements should be non-zero.

We assume the following framework:

- Let $X = (X^1, \dots, X^L)$ a given abundance matrix associated to a tree T .
- We denote $X^{(\ell)} = (x_1^{(\ell)}, \dots, x_{K_\ell}^{(\ell)})$ the abundance vector at layer ℓ .
- For an abundance node $x_k^{(\ell)}$, we denote by $\mathcal{C}(x_k^{(\ell)})$ the set of abundance children associated to that node. Also, we introduce the notation $T_k^{(\ell+1)}$, the vector of activation states $(u_j^{(\ell+1)})_j$ of the children of the node $u_k^{(\ell)}$ in T (restriction of $T^{(\ell)}$ to the children of the node $u_k^{(\ell)}$).
- $X^{(1)} = [1, 0, \dots, 0]$, since it's the root of the tree, only one entity gets the whole weight.
- Since we would like a simple explicit model at first, we design a posterior $p_\theta(X|T)$ which is markovian relatively to the layers of the tree, so that the abundance at the next layer are only impacted by the previous layers abundance for now:

$$p_\theta(X^{(\ell)}|X^{(1:\ell-1)}, T) = p_\theta(X^{(\ell)}|X^{(\ell-1)}, T^{(\ell)})$$

- In addition to the markovianity, we assume that at a given layer, the abundance only depends of their respective parent and their siblings:

$$p(X^{(\ell+1)}|X^{(\ell)}, T^{(\ell+1)}) = \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T^{(\ell+1)})$$

- Each value in $X^{(\ell)}$ is restricted by the following set of constraints due to the nature of the abundance in a tree structure:
 - If node k at layer $\ell - 1$ has one child, then its abundance value is the same for the child node.
 - If node k at layer $\ell - 1$ has at least two children, the children abundance sums to the parent's abundance value.

- Since we deal with proportions in abundance vectors, it seems natural to use the Dirichlet distribution in first assumption. Hence, we assume that for all $l \geq 2$, if $|\mathcal{C}(x_k^{(\ell)})| > 1$,

$$\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T \sim x_k^{(\ell)} \mathcal{D}(\alpha_k^{(\ell)} \odot T_k^{(\ell+1)})$$

We denote by $f_{\alpha_k^{(\ell)} \odot T^{(\ell+1)}}$ the density of this distribution, parameterized by $\alpha_k^{(\ell)} \odot T^{(\ell+1)}$ which denotes a masked version of $\alpha_k^{(\ell)}$ relatively to the activated nodes of the tree at $T^{(\ell)}$. Notice that setting this distribution framework enables us to verify the constraints given above by renormalizing the layer with the obtained weights. Naturally, if $|\mathcal{C}(x_k^{(\ell)})| = 1$, we have $\mathcal{C}(x_k^{(\ell)}) \sim \delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)}))$ to respect the constraints.

Noting the previous framework, the whole abundance distribution conditionally to a tree T can be written as (see proposition 6.4):

$$p(X|T) = \delta_{e_1}(X^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)})) \mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|=1} \frac{1}{x_k^{(\ell)}} f_{\alpha_k^{(\ell)} \odot T^{(\ell+1)}} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right)^{\mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|>1}} \right)$$

One can then compute a maximum likelihood estimator of each $\alpha_k^{(\ell)}$ using a fixed point iterative algorithm that is described in the appendix as proposition ??:

$$\alpha_{k,v}^{(\ell)} \leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,u}^{(\ell)} \right) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right] \right]}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)}} \right)$$

5.3.3 Optimization of the objective

Recall the optimization objective, written under the maximum of the log-likelihood:

$$\begin{aligned} \theta^* &= \arg \max_{\theta} \log p_{\theta}(X, T) \\ &= \arg \max_{\theta} \sum_{i=1}^n \log p_{\theta}(X_i, T_i) \\ &= \arg \max_{\theta} \sum_{i=1}^n \log p_{\theta}(T_i) + \log p_{\theta}(X_i|T_i) \end{aligned}$$

Notice that, in our context, this objective is separable since the prior and posterior do not share any common parameter in θ . Hence, the optimal θ^* is given by the concatenation of the MLE from the prior on the trees and the posterior of the abundance knowing the trees, which we both have computed in the previous sections.

5.3.4 Experiments

We implement the previous model to form a baseline to which we can compare the upcoming latent models to. Before we work with real data, we generate an artificial dataset based on the following structure.

We define the true parameters of the model:

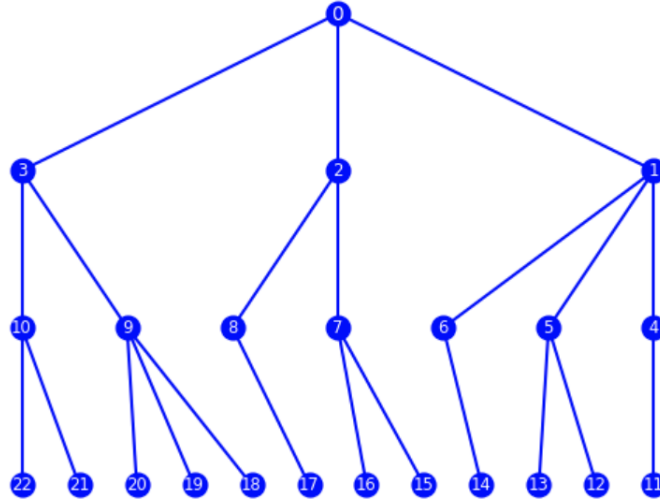


Figure 12: Global artificial tree structure

- The activation probabilities for each node: $\pi_k^{(\ell)}$
- The abundance parameters for each parent node: $\alpha_k^{(\ell)}$

Using the true parameters, we define the prior and posterior of the previous model to generate a dataset. The following figure illustrates a sample of that dataset.

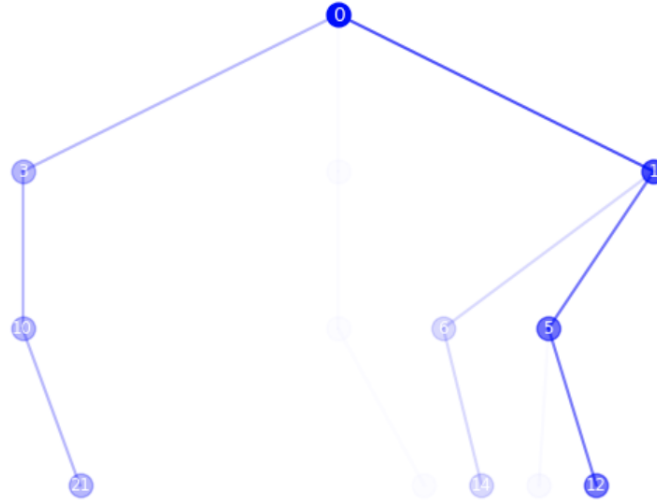


Figure 13: Artificial taxa-abundance data following the global structure of figure 12. Opacity accounts for the value of the abundance.

Now that we have a dataset, we train another set of 10 priors and posteriors onto the artificial data study the convergence of the model and the absolute error to the parameters. The following figure illustrates the convergence of the 10 models for the maximum likelihood objective, relatively to the the number of iterations done for the fixed point algorithm to compute the abundance parameters.

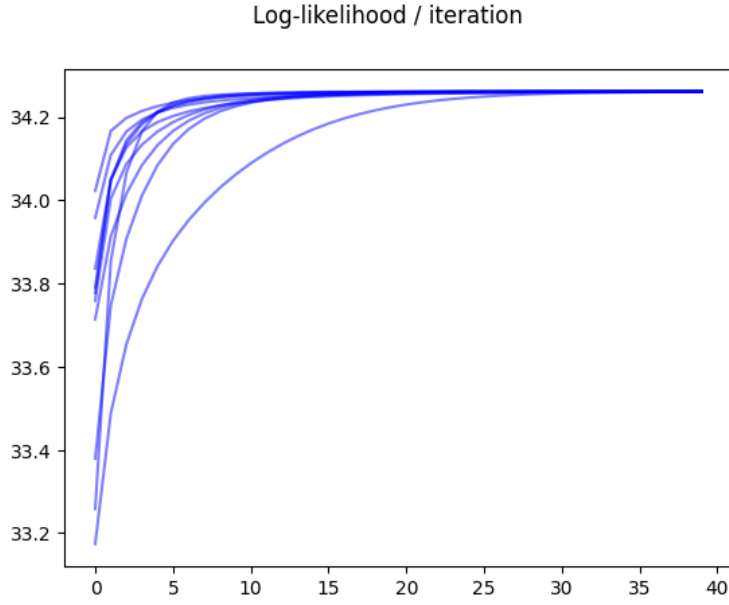


Figure 14: Log-likelihood per iteration of the optimization algorithm for 10 models being trained with random initializations.

Looking at figure 14, it seems that the optimization algorithm is converging to a given $\hat{\theta}^*$. We would then like to compare such $\hat{\theta}^*$ to the true set of parameters θ^* . The following figure illustrates the mean distance of the parameters to the original one: $\|\theta^* - \hat{\theta}^*\|_2$.

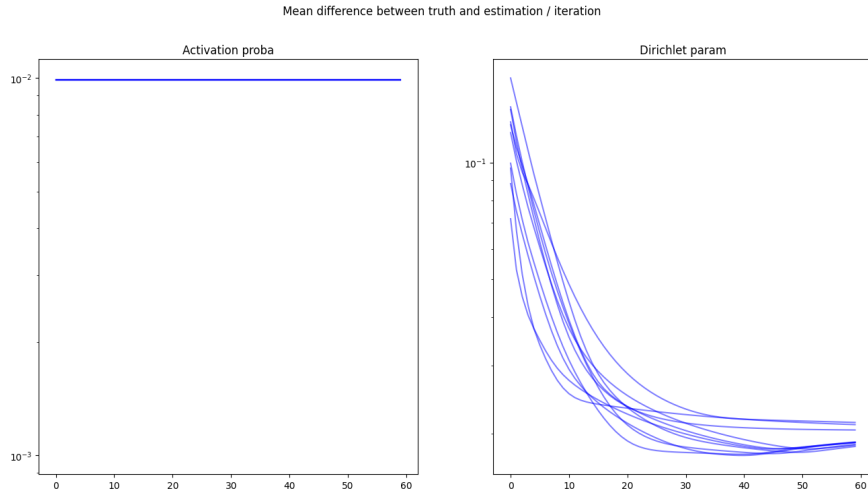


Figure 15: Average log optimization error to the real parameters for 10 models initialized with random parameters, per iteration.

Since the optimal value of $\pi_k^{(\ell)}$ is explicit, the error does not change over the iterations of the fixed point algorithm. On the other hand, the dirichlet parameters $\alpha_k^{(\ell)}$ are optimized coordinate per coordinate in the fixed point algorithm, leading to the convergence profile of figure 15, which shows that there is a convergence to a given $\hat{\theta}^*$ that is close to θ^* with mean error rate of approximately 10^{-2} .

The next graph illustrates the error per coordinate of the estimated parameter and the true one for the 10 trained models.

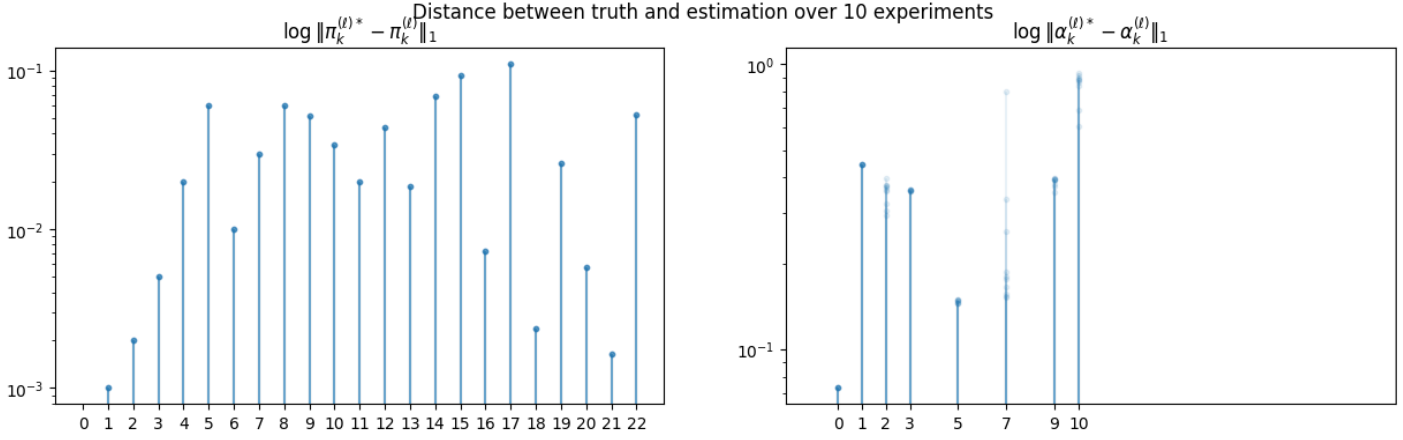


Figure 16: Coordinate-wise log optimization error to the real parameters for 10 models initialized with random parameters, 100 samples, for 40 steps of the fixed point algorithm.

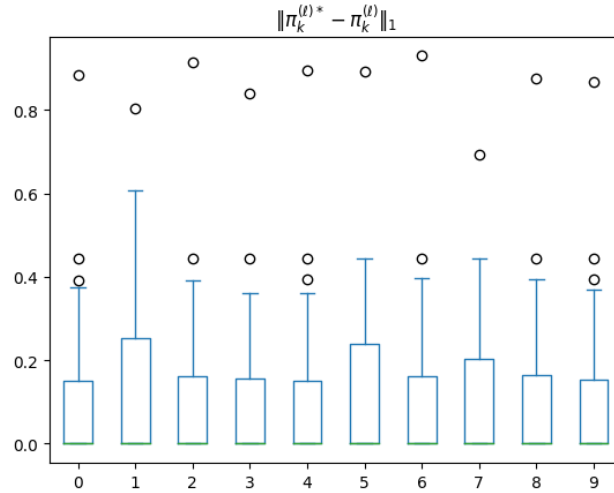


Figure 17: Boxplots of the coordinate-wise optimization error to the real parameters of the dirichlet distribution for 10 runs initialized with random parameters, for 40 steps of the fixed point algorithm.

As it seems, some coordinates are much more accurately determined than others, which highly relates to the structure of the tree (Markovian) and the general presence of each node in the dataset. Indeed, the less likely a node is activated, the less samples we have to compute its activation probability. Likewise, the deeper a node is in the tree, the less likely it is to be present in a tree due to the conditional probability to the parent. That can then explain why deeper nodes are harder to grasp, and for which the parameter estimation is less good than the one of higher entities in the hierarchy of the tree.

Furthermore, note that we used an initialization of the $\pi_k^{(\ell)}$ to 0, which artificially boosts the estimation quality of some node like the 16-th one, which has a probability of activation of

0.1 in our artificial model. Hence, while the estimation seems to be great for that node, this is just biased by the fact that this node rarely is present and that the initialization is quite close to the truth already.

Finally, we look into the average optimization error as we vary the amount of samples in figure 18.

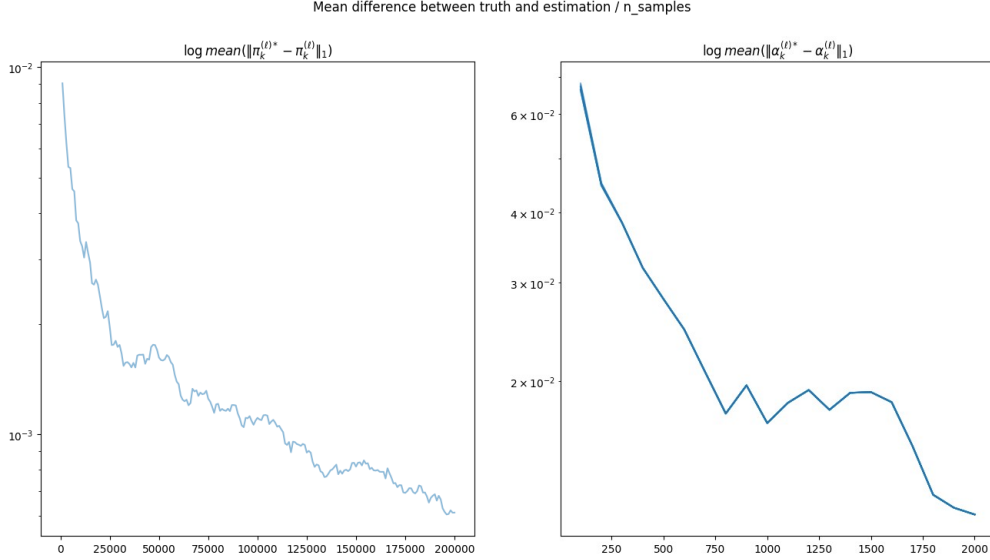


Figure 18: Average log optimization error to the real parameters for 10 models initialized with random parameters, per samples in the dataset.

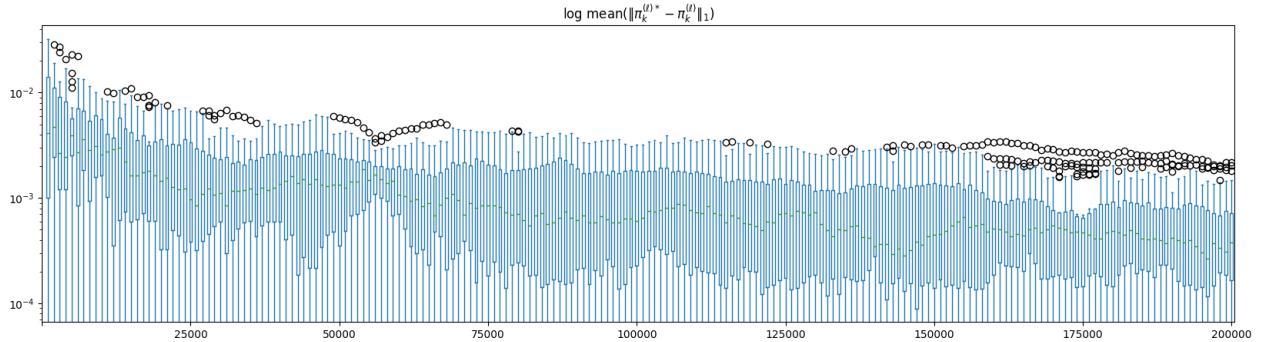


Figure 19: Average log optimization error as boxplots, for 10 models initialized randomly, per samples in the dataset.

As it seems, the more samples we have the better the estimation gets, which seems natural as we look at the estimators. Furthermore, we observe that the variance decreases with the amount of samples, which is reassuring.

5.3.5 Conclusions

This first model is interesting as it provides a benchmark baseline for our upcoming tree-structured models. However, it clearly lacks of complexity:

-
- The nodes are modeled as independent, which prevents any modelisation of correlation between entities. As for many ecosystems, we would expect some bacteria to have symbiotic relationship, or domination roles, especially when it comes to critical systems like disease detection in which some bacteria may proliferate over others. One idea could be to model an interaction graph (see [8]) and use it as a correlation restriction between our bacteria.
 - The abundance data generation takes the tree constraints into account, but the Dirichlet distribution is not quite expressive. Mixture models could enable us to provide more expressive priors and model multiple modes rather than one at the moment, to the cost of more parameters though.
 - So far we have only considered trees without any missing entries at precision level L , which is not what we observe for microbiota datasets. Inference for missing data implementation could be interesting in the future.

5.4 Taxonomy clustering: mixture model

5.4.1 Context

A first natural extension to improve the model introduced in 5.3 would be to take into account a latent clustering of the microbiota. Indeed, before looking at functional areas, it is highly likely that some people may share a similar taxonomic architecture.

Introducing that latent clustering can be done by modelizing a hidden variable that we will generally denote as Z . We then define the following framework, which stands as a mixture of the previously introduced model 5.3 over the latent clusters:

- Denote $Z \in \{1, \dots, C\}$ the latent taxonomic clustering variable.
- Denote $p(Z = c) = \gamma_c$ the probability of cluster c .
- Assume that T and X distributions parameters are now conditioned by the value of Z :

$$\begin{aligned}
 p(Z = c) &= \gamma_c \\
 p\left(u_k^{(\ell)} = 1 | \mathcal{P}(u_k^{(\ell)}) = 1, Z = c\right) &= \pi_{c,k}^{(l)} \\
 p\left(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, T^{(\ell+1)}, Z = c\right) &\sim x_k^{(\ell)} \mathcal{D}(\alpha_{c,k}^{(l)} \odot T_k^{(\ell+1)}) \quad \text{if } |\mathcal{C}(x_k^{(\ell)})| > 1
 \end{aligned}$$

5.4.2 Optimization

The usual optimization objective for this model would be the maximum likelihood regarding the observations, such that:

$$\theta^* = \arg \max_{\theta} p_{\theta}(T, X)$$

However, this does not implement the latent structure that we are aiming for, as we do not observe Z . Hence, using the Expectation-Maximization algorithm presented in 3, we introduce a suboptimal objective on the complete joint distribution:

$$\theta^* = \arg \max_{\theta} \underbrace{\mathbb{E}_{p_{\hat{\theta}}(Z|T,X)} [\log p_{\theta}(T, X, Z) | T, X]}_{Q(\hat{\theta}, \theta)}$$

Such optimal θ^* can be computed through a mix of explicit estimators and a fixed point algorithm detailed in proposition 6.6. Notice that the computed estimator of θ^* is quite close to the one presented in section 5.3, as we simply introduced a global architecture clustering based on a mixture of the simple generative model.

5.4.3 Experiments

5.5 Hidden Markov Models: functional modelization

5.5.1 Context of Hidden Markov Models and Hidden Tree Markov Models

One key goal of our analysis of the microbiota is to highlight functional groups of bacteria in a studied context (reaction to treatment, overall view, disease tracking, post-surgery reaction, ...). Unveiling such groups can be done through various processes, namely by using latent variables.

Previously, we have worked with i.i.d latent variables, meaning that the hidden representation does not implement any typical graph structure that would model latent dependencies between the bacteria (i.e., functional areas). A typical latent model that would admit the simplest graph structure of a chain is given by the Hidden Markov Models.

In a nutshell, Hidden Markov Models (HMM) are a class of generative models such that the latent variable follows a Markov Chain. Formally, for an observed variable $Y = (Y_1, \dots, Y_n)$, $Z = (Z_1, \dots, Z_n)$ is a discrete HMM relatively to Y if we have the following:

- Z is a Markov Chain (MC) with discrete states:

$$\mathbb{P}(Z_{i+1}|Z_i, Z_{i-1}, \dots, Z_1) = \mathbb{P}(Z_{i+1}|Z_i)$$

- We define a prior on the seed of the MC:

$$\forall c \in \{1, \dots, C\}, \mathbb{P}(Z_1 = c) = \pi_c$$

- We define a transition probability on the states of the MC:

$$\forall c, k \in \{1, \dots, C\}^2, \mathbb{P}(Z_{i+1} = c|Z_i = k) = t_{k,c}^{(i+1)}$$

- We define an emission probability for Y to be in a given state when observing Z :

$$P(Y_i \in A|Z_i = c) = a_c(A)$$

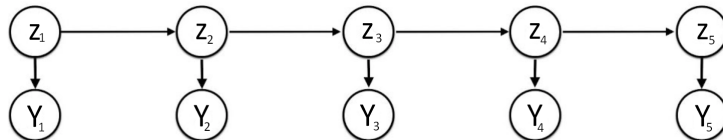


Figure 20: Dependency graph of a HMM (Z is the latent MC, Y the observation)

As a result, we can use HMM to model hidden functional groups of bacteria from one layer to the other.

This idea can even be extended to the specific tree-like structure of taxa-abundance data, introducing the wider class of models given by the Hidden Tree Markov Model (HTMM) [4]. The HTMM provides a dependency graph that accounts for the tree structure, so that instead of a chain, we get a Markov tree. The following figure illustrates a given HTMM, precisely a top-down architecture:

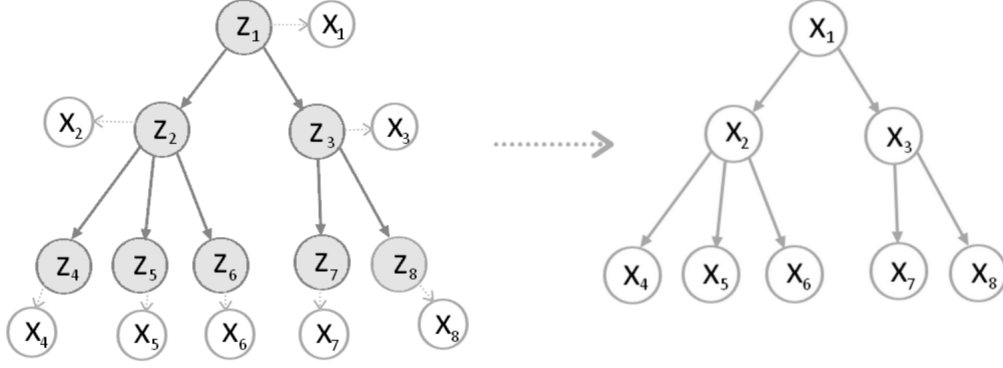


Figure 21: Top-down HTMM example: Z represents the latent Markov tree, X the observed abundance of a given bacteria in the taxonomic tree

The mathematical framework is then slightly different for top-down HTMM:

- Z is a Markov Tree (MT):

$$\mathbb{P}\left(Z_k^{(l)} = c | Z_0^{(\ell)}, \dots, Z_{K_\ell}^{(\ell)}, \mathcal{P}(Z_0^{(\ell)}), \dots, \mathcal{P}(Z_{K_\ell}^{(\ell)}), \dots, Z_1^{(1)}\right) = \mathbb{P}\left(Z_k^{(\ell)} | \mathcal{P}(Z_k^{(\ell)})\right)$$

- We define a prior on the first layer of the MT, relatively to a given tree T :

$$\mathbb{P}\left(Z_1^{(1)} = j | T^{(1)}\right) = a_j$$

- We define the transition probabilities of the MT, relatively to a given tree T :

$$\mathbb{P}\left(\mathcal{C}(Z_k^{(\ell)}) = \nu | Z_k^{(\ell)} = j, T^{(\ell+1)}\right) = t_{\nu,j}^{(\ell)}$$

Notice that this model would be tree dependent, as we want to attach existing nodes to a given hidden state, to generate the abundance accordingly. One could think of a tree generator model by applying the HTMM onto the global taxonomy directly, and defining an "empty" state that would turn off the specific nodes in the final representation.

Some recent work on HTMM have been showing that top-down modelling captures less discriminant detail and conditional dependencies between the nodes than the bottom-up architecture [1]. In this first study though, we will focus on top-down models as a first try, as it makes the generation more natural (since we can not generate from bottom to up for the abundance). Note that bottom-up modeling is still possible, but we can not generate sequentially the abundance with Z . We would need to generate Z entirely, and then generate X in a top-down fashion.

5.5.2 Tree generation: layer-wise Hidden Markov Model

In this section, we aim at modeling the distribution of the trees $(T_i)_{1 \leq i \leq n}$ through a layer-wise Hidden Markov Model. The following figure illustrates the dependency between the latent markov chain and the tree:

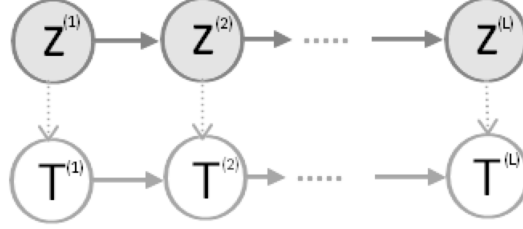


Figure 22: Layer-wise hidden markov model dependency graph: Z is the HMM, $T^{(\ell)}$ is the layer (ℓ) of the tree

As illustrated on figure 22 and by nature of our taxonomic data, the layer of the tree follow at least a Markov process (it could be deeper than markovian). Conversely to the previous situations, the latent variable now follows a markov chain as well, which enables to group the layers of the tree with a dependence on the parent's layer group.

Furthermore, in that approach, we decide to model the layers rather than the nodes to introduce dependency between the nodes of a given layer, so we do not look into the $u_k^{(\ell)}$ anymore but directly into the $T^{(\ell)}$.

Concisely, the mathematical framework can be written as follow:

- Z follows a discrete MC:

$$\begin{aligned}\mathbb{P}(Z^{(1)} = c) &= a_c \\ \mathbb{P}(Z^{(\ell+1)} = k | Z^{(\ell)} = c) &= t_{c,k}^{(\ell+1)}\end{aligned}$$

- T is markovian and parameterized conditionally to Z . We denote by $\mathcal{C}(T^{(\ell)})$ the set of plausible activation vectors at layer $(\ell + 1)$ in the taxonomy, relatively to the parents activation given by $T^{(\ell)}$. We then model the emission probability by:

$$\mathbb{P}(T^{(\ell+1)} = \omega | Z^{(\ell+1)} = c, T^{(\ell)}) = \frac{\pi_c(\omega) \mathbb{1}_{\omega \in \mathcal{C}(T^{(\ell)})}}{\sum_{\nu \in \mathcal{C}(T^{(\ell)})} \pi_c(\nu)}$$

One may notice that such modelisation on T will require a large number of parameters. Indeed, the set of plausible activation vectors at layer $(\ell + 1)$ relatively to $T^{(\ell)}$ can be quite big, as for each activate node $u_k^{(\ell)}$ in $T^{(\ell)}$ we get $2^{|\mathcal{C}(u_k^{(\ell)})|} - 1$ plausible activated vectors of its children (-1 comes from the fact that we would like to avoid missing values). We denote by $\mathcal{S}(Z)$ the discrete state space of the hidden markov chain. Hence, for a given tree T , we obtain a total number of parameters of:

$$n_{\text{params}} = \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} |\mathcal{S}(Z)| \times \left(2^{|\mathcal{C}(u_k^{(\ell)})|} - 1 \right)$$

As usual for latent data models, variational methods come in handy for learning the model. Hence, we provide an Expectation-Maximization algorithm to compute the optimal parameters of the prior over the tree in proposition 6.8.

5.5.3 Abundance generation: Hidden Tree Markov Model

In this section, we aim at modeling the distribution of the abundance data $(X_i)_{1 \leq i \leq n}$ conditionally to its known corresponding trees $(T_i)_{1 \leq i \leq n}$, using a Hidden Tree Markov Model (HTMM). The dependency graph is illustrated by the figure 21, giving us the follow mathematical framework:

- Z is a discrete Markov Tree (MT):
 - We define a prior over the root:

$$\mathbb{P}(Z_1^{(1)} = c | T^{(1)}) = a_j$$

- We define the transition probabilities:

$$\mathbb{P}(\mathcal{C}(Z_k^{(\ell)}) = \nu | Z_k^{(\ell)} = j, T^{(\ell+1)}) = t_{j,\nu}^{(\ell)}$$

- X follows a Markov Tree structure. We define the emission probability of the corresponding abundance relatively to the hidden states and tree:

$$p(\mathcal{C}(x_k^{(\ell)}) | \mathcal{C}(Z_k^{(\ell)}) = \nu, x_k^{(\ell)}, T^{(\ell+1)}) \sim x_k^{(\ell)} \mathcal{D}(\alpha_{\nu,k}^{(\ell)} \odot T_k^{(\ell+1)})$$

The number of parameters of that model is quite large, as it depends on the size of the latent space state and the number of child node of each parent node in the tree structure. Indeed, for each node, the corresponding dirichlet parameter is of the size of the number of children (if it has more than 1 child). Then, we have as many dirichlet parameters as the possible amount of vector ν , for which each coordinate has $|\mathcal{S}(Z)|$ plausible value. That leads us to the following number of parameters:

$$n_{\text{params}} = \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} |\mathcal{S}(Z)|^{|\mathcal{C}(u_k^{(\ell)})|} \times |\mathcal{C}(u_k^{(\ell)})| \times \mathbb{1}_{|\mathcal{C}(u_k^{(\ell)})| > 1}$$

As previously for the tree generation, we propose an Expectation-Maximization algorithm to compute the optimal parameters of that HTMM, detailed in proposition **[TODO]**.

5.5.4 Bottom-Up Hidden Tree Markov Model: general model

The previous approach used a Top-Down HTMM (see figure 21), which is a valid approach to model a parent to child dependency only. Medically speaking, the top-down HTMM can be seen as parent dependent functions: a parent's function would determine the function of its child, which would be independently sorted conditionally to the parent. In addition to that questionable modelisation, related work from [1] as shown that top-down HTMM are capturing less conditional dependencies between nodes, modeling less distributions than a bottom-up approach.

Hence, we are now interested in a bottom-up HTMM, which is illustrated in figure 23.

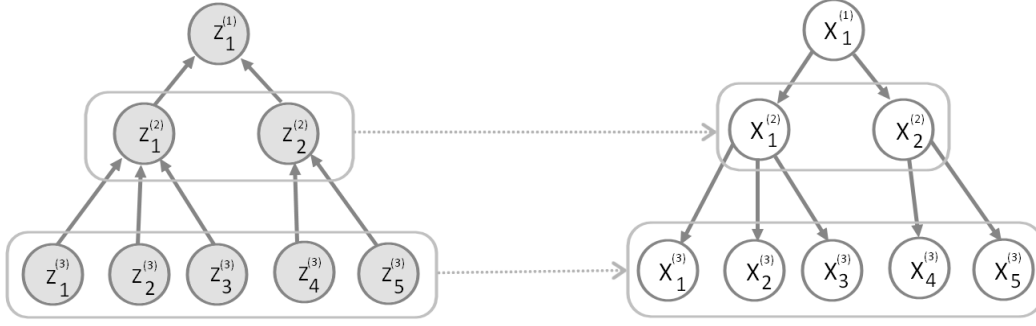


Figure 23: Bottom-down Hidden Tree Markov Model architecture: Z represents the latent space, X a taxa-abundance sample. The pointed out sections and arrows represent the dependencies. Each node is marked by its layer (ℓ) and position within that layer k .

The hidden states Z are taking the graph dependency form of a tree that covers the entire taxonomy, and take value in a discrete set $\mathcal{S}(Z)$. To take in consideration the sparsity of the taxa-abundance data, we introduce a killing state that we denote as s_\emptyset , which filter out a node and its children when appearing.

The general framework is then the following:

- Z is bottom-up HTMM with killing state s_\emptyset so that:

$$p(Z^{(L)} = \nu) = \pi_\nu$$

$$p(Z_k^{(\ell)} = s | \mathcal{C}(Z_k^{(\ell)}) = \nu) = t_{\nu, s}^{(\ell)}$$

- X is a top-down markov tree conditionally to Z :

$$p(X^{(1)}) = \delta_{e_1}(X^{(1)})$$

$$p(X) = p(X^{(1)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)} | X^{(\ell)})$$

$$p(X^{(\ell+1)} | X^{(\ell)}) = \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)})$$

- The conditional distribution of X knowing Z is characterised as follows:

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(z_k^{(\ell)}) = \nu) = x_k^{(\ell)} \mathcal{D}(\alpha_{k, \nu}^{(\ell)})$$

$\alpha_{k, \nu}^{(\ell)}$ are the dirichlet parameters of the distribution, which are vectors of dimension

$$\#\{j \in \nu | j \neq s_\emptyset\}$$

as they only impact the nodes that are still alive according to Z .

To compute the optimal parameters of that model given a dataset of taxa-abundance data $(X_i)_{1 \leq i \leq n}$, we propose an Expectation-Maximization algorithm in proposition 6.10. However, even if this model is covers all types of problems with its general form, it may not be suited to our special use case with very few samples. Hence, the next section provides an under-parameterized alternative model to fit the framework of low samples datasets.

5.5.5 Bottom-Up Hidden Tree Markov Model: low sample regime

The previous bottom-up HTMM model is extremely general, as it provides a parameter for each transition configuration through $t_{\mathcal{C}(Z_k^{(\ell)}), Z_k^{(\ell)}}^{(\ell)}$. However, training such model onto a small dataset may be difficult and could result in overfitting, as we may highly likely not explore all possible configurations for a node and its children, and that all the configuration do not impact each other's transition probabilities in the HTMM.

Hence, building on the previous model, we introduce a low parameterized model, which may not capture all the expressiveness of the general HTMM, but that could be trainable on a low sample regime.

- Let $S \in \mathbb{N}$ the number of possible hidden states, at each layer (ℓ) we introduce the parameter $\pi^{(\ell)} \in \mathbb{R}^S$, which defines the aggregation parameter $\omega_k^{(\ell)}$:

$$\omega_k^{(\ell)} = \sum_{z \in \mathcal{C}(Z_k^{(\ell)})} \sum_{s=1}^S \mathbb{1}_{z=e_s} \pi_s^{(\ell)} e_s$$

where e_s denotes the embedding of the state s into a vector representation (one-hot encoding for instance).

- Z is bottom-up HTMM with killing state s_\emptyset , for which the transition probabilities are defined through $\omega_k^{(\ell)}$:

$$\begin{aligned} Z_k^{(L)} &\sim \text{Cat}(\pi^{(L)}) \\ Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)}) &\sim \text{Cat}(\text{softmax}(\omega_k^{(\ell)})) \end{aligned}$$

- X is a top-down markov tree conditionally to Z :

$$\begin{aligned} p(X^{(1)}) &= \delta_{e_1}(X^{(1)}) \\ p(X) &= p(X^{(1)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)} | X^{(\ell)}) \\ p(X^{(\ell+1)} | X^{(\ell)}) &= \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}) \end{aligned}$$

- The conditional distribution of X knowing Z is characterised as follows, for the nodes that have at least 2 children:

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(z_k^{(\ell)}) = \nu) = x_k^{(\ell)} \mathcal{D}(\alpha_k^{(\ell)} \odot \nu)$$

$\alpha_k^{(\ell)}$ are the dirichlet parameters of the distribution, which are vectors of dimension $\mathcal{C}(Z_k^{(\ell)})$. The hadamard product with ν denotes that we mask the coordinates of $\alpha_k^{(\ell)}$ according to the states that are still alive i.e. different from e_{s_\emptyset} . If the node has 1 child or none, the abundance value has to be the same as the parent.

Note that the hidden states are embedding vectors e_s with $s \in \{1, \dots, S\}$. So, for notation simplicity, we will introduce the following subscript notation. Given θ a vector of dimension S , Z a random variable taking value in the discrete state set $\mathcal{S}(Z)$ of dimension S , the embedding vectors of $\mathcal{S}(Z)$ are denoted by e_s :

$$\theta_Z = \sum_{s=1}^S \mathbb{1}_{Z=e_s} \theta_s$$

Finding the optimal parameters of such model fitting a dataset can be done in the sense of the maximum likelihood, using the proposed Expectation-Maximization algorithm of proposition ???.

5.6 Poisson log-normal models

5.6.1 Introduction to PLN

Abundance data are quite complex to model by nature, as they account both for the presence of a given entity in a given ecosystem and the given amount of the said entity in the studied ecosystem. Extensive studies have been conducted on abundance data, among which we decide to focus on a state-of-art approach, the Poisson Log-Normal (PLN) model introduced in [2].

The PLN model belongs to the family of latent models, as it models the abundance of the entities using a Poisson distribution that is parameterized by a latent gaussian. Formally speaking, if $X \in \mathbb{R}^{n \times d}$ denotes an abundance matrix of d entities among n individuals, we get the following model:

- $Z_i \sim \mathcal{N}(0, \Sigma)$ where $\Sigma \in \mathbb{R}^{d \times d}$ is to be learnt.
- $X_{ij} \sim \mathcal{P}(e^{Z_{ij}})$
- The Z_i are independent, and X_{ij} is independent from all other abundance conditionally to Z_{ij} .

This modelization turns out to have some great mathematical properties, among which one can highlight the faithful correlation, which means that the correlation between X_{ij} and X_{ik} has the same sign as the correlation between Z_{ij} and Z_{ik} . Hence, if the correlation is 0 in the latent space between two entities, we can deduce that the abundance of the two species are always independent. This faithful correlation property then enables us to determine a latent interaction graph between entities, and therefore unveil hidden functional groups in our study.

Furthermore, assuming we have p covariates at our disposal in addition to the abundance for each individual, denoting such covariates by $D \in \mathbb{R}^{n \times p}$, we can account for fixed effects such as clinical variables, or environmental variables. Implementing it in the model is then done through the mean of the latent layer, assuming that the mean for the sample i is then given by $\mu_i = D_i^\top \theta$, where θ is to be learnt.

Finally, one can add an offset to the model to model sampling efforts between sites of interest, or individuals (omitted in our specific case by assumption).

While the modelization appears interpretable and straight-forward, the training of PLN models requires variational inference due to the latent layer in the problem. Indeed, partially

observed models generally make a great use of the E-M algorithm to estimate the parameters of the model, yet the E-step requires to compute $p(Z|X)$ which is not explicit in the general PLN framework.

Instead, the authors of [2] suggest a variational approximation of the E-step. Basically, we introduce a gaussian parameterized by Φ , $q_{\Phi}(Z|X) \sim \prod_{k=1}^d \mathcal{N}(m_k, s_k)$, where the m_k and s_k are to be learnt (they compose Φ). Before performing the estimation step $h + 1$, we then find the optimal variational approximation of the posterior given by

$$\Phi^{h+1} = \arg \max_{\Phi} ELBO(\theta^h, \Phi)$$

As the ELBO turns out to be explicit in the independent gaussian variational approximation (see appendix of [2]). Then, we perform the E-step by replacing the true posterior $p(Z|X)$ by $q_{\Phi^{h+1}}(Z|X)$, then we perform the M-step to compute θ^{h+1} , and so on up until convergence.

This model has proven to be very efficient to model independent samples, in very different context like PCA, Mixture Models, Network inference, for which the respective ELBO varies slightly. The application of interest for our problem is the Network inference, which would enable us to unveil functional interactions between the bacteria of the gut microbiome. The Network inference problem also comes with a sparsity-informed architecture suggested by the authors, by introducing a LASSO penalty on the covariance matrix Σ of the latent layer. Each connected component then forms a functional group in our interpretation.

5.6.2 Layer independent PLN

Recall our specific microbiota framework, we introduce a taxonomy \mathcal{T} , with N nodes, L layers, and K_{ℓ} entities at layer ℓ . Let $(X_i)_{1 \leq i \leq n}$ the taxa-abundance samples of n individuals, supported on the taxonomy \mathcal{T} . We denote by $x_k^{(\ell)} \in \mathbb{N}$ the abundance of the k -th entity in a taxa-abundance sample at layer ℓ .

The layer independent PLN model would then consist of building one PLN model for each layer of the taxonomy. Note beforehand that this model can create non-physical samples, as it is not informed by the previous layer to perform a normalization. The main interest of this model is then to obtain a first functional graph of the entities within the taxonomy without developing external tools, but only using the PLN package. Next sections will focus on new models derivated from PLN.

For each layer ℓ , introducing a latent variable $Z^{(\ell)}$, the formal model is such that $(Z^{(\ell)}, X^{(\ell)}) \sim PLN(0, \Sigma^{(\ell)})$ where $\Sigma^{(\ell)} \in \mathbb{R}^{K_{\ell} \times K_{\ell}}$. We then have to learn L covariance matrices, which can be seen as solving L PLN problems independently due to the layer independence between the $(Z^{(\ell)}, X^{(\ell)})$.

5.6.3 Multi-layer covariance PLN

Assume the same framework as previously. This time we introduce dependency between the layers by making some assumptions on the joint distribution of the $Z^{(\ell)}$, so that:

- $Z = (Z^{(1)}, \dots, Z^{(L)}) \sim \mathcal{N}(0, \Sigma)$, where $\Sigma \in \mathbb{R}^{(K_1 + \dots + K_L)^2}$
- $Z^{(\ell)} \sim \mathcal{N}(0, \Sigma^{(\ell)})$ where $\Sigma^{(\ell)} \in \mathbb{R}^{K_{\ell}^2}$

- $x_k^{(\ell)} \sim \mathcal{P}(e^{Z_k^{(\ell)}})$

This model differs from the following by the join distribution assumption structure that replaces the independence per layer. The consequence is that we are now able to model functional groups between all entities in the taxonomy, leading to inter layer functional group rather than just intra layer.

Using a variational approximation, this model turns out to be tractable (see proposition 6.13).

6 Appendix

6.1 Microbiota analysis

Proposition 6.1. (*Bernoulli tree prior*)

Let $T = (T^{(1)}, \dots, T^{(L)})$ a random tree of depth L .

For all $\ell \in \{1, \dots, L\}$, denote $T^{(\ell)} = (u_1^{(\ell)}, \dots, u_{K_\ell}^{(\ell)})$ the nodes at layer ℓ , such that $u_k^{(\ell)} \in \{0, 1\}$ denotes whether or not the node is activated (1 if activated).

Denote $\mathcal{P}(u_k^{(\ell)})$ the parent of the node $u_k^{(\ell)}$ that outputs 1 if the parent exists and is activated, 0 otherwise.

Assume that:

- $p(T^{(1)}) = \delta_{e_1}(T^{(1)})$
- $\forall l \geq 2, p(T^{(l+1)} | T^{(1:l)}) = p(T^{(l+1)} | T^{(l)})$
- $\forall l \geq 2, k \in \{1, \dots, K_\ell\}, u_k^{(\ell)} | \left\{ \mathcal{P}(u_k^{(\ell)}) = 1 \right\} \sim \mathcal{B}(\pi_k^{(\ell)})$
- $p(u_1^{(\ell)}, \dots, u_{K_\ell}^{(\ell)} | \mathcal{P}(u_1^{(\ell)}), \mathcal{P}(u_{K_\ell}^{(\ell)})) = \prod_{k=1}^{K_\ell} p(u_k^{(\ell)} | \mathcal{P}(u_k^{(\ell)}))$

Then, the distribution of T can be written as:

$$p(T) = \delta_{e_1}(T^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\left(\pi_k^{(\ell+1)} \right)^{u_k^{(\ell+1)}} \left(1 - \pi_k^{(\ell+1)} \right)^{1-u_k^{(\ell+1)}} \right)^{\mathcal{P}(u_k^{(\ell+1)})}$$

Proof.

$$\begin{aligned} p(T) &= p(T^{(1)}, \dots, T^{(L)}) \\ &= p(T^{(1)}) \prod_{l=1}^{L-1} p(T^{(l+1)} | T^{(l)}) \\ &= \delta_{e_1}(T^{(1)}) \prod_{l=1}^{L-1} p(u_1^{(\ell+1)}, \dots, u_{K_\ell}^{(\ell+1)} | \mathcal{P}(u_1^{(\ell+1)}), \dots, \mathcal{P}(u_{K_\ell}^{(\ell+1)})) \\ &= \delta_{e_1}(T^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} p(u_k^{(\ell+1)} | \mathcal{P}(u_k^{(\ell+1)})) \\ &= \delta_{e_1}(T^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\pi_k^{(\ell+1)} u_k^{(\ell+1)} + (1 - \pi_k^{(\ell+1)}) (1 - u_k^{(\ell+1)}) \right)^{\mathcal{P}(u_k^{(\ell+1)})} \\ &= \delta_{e_1}(T^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\left(\pi_k^{(\ell+1)} \right)^{u_k^{(\ell+1)}} \left(1 - \pi_k^{(\ell+1)} \right)^{1-u_k^{(\ell+1)}} \right)^{\mathcal{P}(u_k^{(\ell+1)})} \end{aligned}$$

□

Proposition 6.2 (MLE of the Bernoulli tree prior). *Recall the context of 6.1.*

Let (T_1, \dots, T_n) n trees i.i.d following the bernoulli tree prior. Denote the maximum likelihood objective by

$$\begin{aligned} \arg \max_{\pi_j^{(m)}} & \sum_{i=1}^n \sum_{l=1}^{L-1} \sum_{k=1}^{K_\ell} \mathcal{P}(u_{k,i}^{(\ell+1)}) \left[u_{k,i}^{(\ell+1)} \log \pi_k^{(\ell+1)} + (1 - u_{k,i}^{(\ell+1)}) \log(1 - \pi_k^{(\ell+1)}) \right] \\ \text{s.t. } & \forall k, l, \pi_k^{(\ell)} \in [0, 1] \end{aligned} \quad (1)$$

Then the maximum likelihood estimator is given by

$$\left(\pi_k^{(\ell)} \right)^* = \frac{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) u_{k,i}^{(\ell)}}{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)})}$$

Proof. Simply by deriving the objective we obtain:

$$\begin{aligned} \partial_{\pi_j^{(m)}} \log p(T_1, \dots, T_n) &= \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) \left[u_{j,i}^{(m)} \frac{1}{\pi_j^{(m)}} + (1 - u_{j,i}^{(m)}) \frac{-1}{1 - \pi_j^{(m)}} \right] \\ &= \frac{1}{\pi_j^{(m)}} \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) u_{j,i}^{(m)} - \frac{1}{1 - \pi_j^{(m)}} \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) (1 - u_{j,i}^{(m)}) \end{aligned}$$

Looking for 0 valued gradient, we end up with:

$$\begin{aligned} (1 - \pi_j^{(m)}) \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) u_{j,i}^{(m)} - \pi_j^{(m)} \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) (1 - u_{j,i}^{(m)}) &= 0 \\ \pi_j^{(m)} \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) &= \sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) u_{j,i}^{(m)} \\ \pi_j^{(m)} &= \frac{\sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)}) u_{j,i}^{(m)}}{\sum_{i=1}^n \mathcal{P}(u_{j,i}^{(m)})} \end{aligned}$$

Since the obtained value respects the constraint, that concludes the proof. \square

Proposition 6.3 (Law of aX). *Let $X \sim f(x)$ a random variable with density f .*

Let $a \in (0, 1)$.

The distribution of aX is given by:

$$p_{aX}(u) = \frac{1}{a} f\left(\frac{u}{a}\right)$$

Proof. We use the transfer theorem. Let g a continuous and measurable function.

$$\begin{aligned} \mathbb{E}[g(aX)] &= \int g(ax) f(x) dx \\ &= \int g(u) f\left(\frac{u}{a}\right) \frac{1}{a} du \end{aligned}$$

Hence, one can identify the distribution of aX as $p_{aX}(u) = \frac{1}{a} f\left(\frac{u}{a}\right)$ \square

Proposition 6.4 (Abundance distribution conditionally to the trees). *Let $X = (X^{(1)}, \dots, X^{(L)})$ the abundance matrix of a tree T with depth L .*

For all $\ell \in \{1, \dots, L\}$, denote $X^{(\ell)} = (x_1^{(\ell)}, \dots, x_{K_\ell}^{(\ell)})$ the abundance value of each node of the tree.

Denote $\mathcal{C}(x_k^{(\ell)})$ the vector of children abundances related to $x_k^{(\ell)}$, which is empty if it has no children.

Denote $T_k^{(\ell+1)}$ the vector of activation states $(u_j^{(\ell+1)})_j$ of the children of the node $u_k^{(\ell)}$ in T .

Assume that:

- $p(X^{(1)}) = \delta_{e_1}(X^{(1)})$
- $p(X^{(\ell+1)}|X^{(1:\ell)}, T) = p(X^{(\ell+1)}|X^{(\ell)}, T^{(\ell+1)})$
- $p(X^{(\ell+1)}|X^{(\ell)}, T^{(\ell+1)}) = \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T^{(\ell+1)})$
- For all $l \in \{2, \dots, L\}, k \in \{1, \dots, K_\ell\}$,
 - If $|\mathcal{C}(x_k^{(\ell)})| > 1$: $\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T^{(\ell+1)} \sim x_k^{(\ell)} D(\alpha_k^{(\ell)} \odot T_k^{(\ell+1)})$
 - If $|\mathcal{C}(x_k^{(\ell)})| = 1$, $\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T^{(\ell+1)} \sim \delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)}))$

Then, the distribution of X conditionally to T is given by:

$$p(X|T) = \delta_{e_1}(X^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)})) \mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|=1} \frac{1}{x_k^{(\ell)}} f_{\alpha_k^{(\ell)} \odot T_k^{(\ell+1)}} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right)^{\mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|>1}} \right)$$

Proof.

$$\begin{aligned} p(X|T) &= p(X^{(1)}, \dots, X^{(L)}|T) \\ &= p(X^{(1)}|T^{(1)}) \prod_{l=1}^{L-1} p(X^{(\ell+1)}|X^{(\ell)}, T^{(\ell+1)}) \\ &= \delta_{e_1}(X^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)})|x_k^{(\ell)}, T^{(\ell+1)}) \\ &= \delta_{e_1}(X^{(1)}) \prod_{l=1}^{L-1} \prod_{k=1}^{K_\ell} \left(\delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)})) \mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|=1} \underbrace{\left[\frac{1}{x_k^{(\ell)}} f_{\alpha_k^{(\ell)} \odot T_k^{(\ell+1)}} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right) \right]}_{\text{proposition 6.3}} \right)^{\mathbb{1}_{|\mathcal{C}(x_k^{(\ell)})|>1}} \end{aligned}$$

□

Proposition 6.5 (MLE of the abundance distribution a posteriori). *Consider the context of proposition 6.4.*

We consider a data set $(X_i, T_i)_{1 \leq i \leq n}$ of abundance matrix and trees.

Denote by $\mathcal{V}(\alpha_k^{(\ell)}, T) = \left\{ v \in \mathbb{N} \left[\alpha_k^{(\ell)} \odot T_k^{(\ell+1)} \right]_v \neq 0 \right\}$ the set of indexes so that the coordinate

of $\alpha_k^{(\ell)}$ is not masked by $T^{(\ell+1)}$.

The maximum likelihood estimator of the distribution characterising the abundance conditionally to the tree is then given by the following fixed point algorithm:

$$\forall l \in \{1, \dots, L\}, k \in \{1, \dots, K_\ell\}, v \in \{1, \dots, |\alpha_k^{(\ell)}|\},$$

$$\alpha_{k,v}^{(\ell)} \leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,u}^{(\ell)} \right) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right]}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)}} \right)$$

Proof. The maximum likelihood objective can be written as:

$$\arg \max_{\alpha_{j,v}^{(m)}} \sum_{i=1}^n \sum_{l=1}^{L-1} \sum_{k=1}^{K_\ell} \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \log f_{\alpha_k^{(\ell)} \odot T_{k,i}^{(\ell+1)}} \left(\frac{C(x_{k,i}^{(\ell)})}{x_{k,i}^{(\ell)}} \right)$$

Using the Dirichlet distribution expression, we can write the following:

$$\log f_{\alpha_k^{(\ell)} \odot T_{k,i}^{(\ell+1)}} \left(\frac{C(x_{k,i}^{(\ell)})}{x_{k,i}^{(\ell)}} \right) = \log \Gamma \left(\sum_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,v}^{(\ell)} \right) - \sum_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \Gamma(\alpha_{k,v}^{(\ell)}) + \sum_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} (\alpha_{k,v}^{(\ell)} - 1) \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}}$$

Writing ψ the digamma function, the derivative of the objective relatively to a fixed $\alpha_{k,v}^{(\ell)}$ is given by:

$$\partial_{\alpha_{k,v}^{(\ell)}} \log p(X|T) = \sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,u}^{(\ell)} \right) - \psi(\alpha_{k,v}^{(\ell)}) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right]$$

Looking for 0 valued gradient, we obtain the following equation:

$$\psi(\alpha_{k,v}^{(\ell)}) = \frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,u}^{(\ell)} \right) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right]}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)}}$$

Using the reference trick from [7], one can compute an iterative fixed point algorithm to solve the previous equation, leading to:

$$\alpha_{k,v}^{(\ell)} \leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)} \alpha_{k,u}^{(\ell)} \right) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right]}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_k^{(\ell)}, T_i)}} \right)$$

□

Proposition 6.6 (Expectation-Maximization for Taxonomy Clustering model). *We use the same context as for proposition 6.4 of this section. Assume the following framework changes:*

- $(X_i, T_i)_{1 \leq i \leq n}$ samples of taxa-abundance data (X_i abundance, T_i associated taxonomy)
- Let Z_i a latent variable taking values in $\{1, \dots, C\}$.
- We model the distributions as follow:

$$\begin{aligned}
p(Z_i = c) &= \gamma_c \\
p\left(u_{k,i}^{(\ell)} = 1 | \mathcal{P}(u_{k,i}^{(\ell)}) = 1, Z_i = c\right) &= \pi_{c,k}^{(\ell)} \\
p\left(\mathcal{C}(x_{k,i}^{(\ell)}) | x_{k,i}^{(\ell)}, T^{(\ell+1)}, Z_i = c\right) &\sim x_k^{(\ell)} \mathcal{D}(\alpha_{c,k}^{(\ell)} \odot T_k^{(\ell+1)}) \quad \text{if } |\mathcal{C}(x_k^{(\ell)})| > 1
\end{aligned}$$

Consider the optimization problem:

$$\theta^* = \arg \max_{\theta} \mathbb{E}_{p_{\theta}}[\log p_{\theta}(X, T, Z) | X, T]$$

Then, the Expectation-Maximization algorithm solving that problem is given by (for each sample i and cluster index c):

Expectation step:

$$p_{\hat{\theta}}(Z_i = c | X_i, T_i) = \tau_{ic} = \frac{\hat{\gamma}_c p_{\hat{\theta}}(T_i | Z_i = c) p_{\hat{\theta}}(X_i | T_i, Z_i = c)}{\sum_{c'=1}^C \hat{\gamma}_{c'} p_{\hat{\theta}}(T_i | Z_i = c') p_{\hat{\theta}}(X_i | T_i, Z_i = c')}$$

where:

$$\begin{aligned}
p_{\hat{\theta}}(T_i | Z_i = c) &= \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} \left[\left(\hat{\pi}_{c,k}^{(\ell+1)} \right)^{u_{k,i}^{(\ell+1)}} \left(1 - \hat{\pi}_{c,k}^{(\ell+1)} \right)^{1-u_{k,i}^{(\ell+1)}} \right]^{\mathcal{P}(u_{k,i}^{(\ell+1)})} \\
p_{\hat{\theta}}(X_i | T_i, Z_i = c) &= \delta_{e_1}(X_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} \delta_{x_{k,i}^{(\ell)}} \left(\mathcal{C}(x_{k,i}^{(\ell)}) \right)^{\mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})| > 1}} \left(\frac{1}{x_{k,i}^{(\ell)}} f_{\hat{\alpha}_{c,k}^{(\ell)} \odot T_{i,k}^{(\ell+1)}} \left(\frac{\mathcal{C}(x_{k,i}^{(\ell)})}{x_{k,i}^{(\ell)}} \right) \right)^{\mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})| > 1}}
\end{aligned}$$

Maximization step:

$$\begin{aligned}
\gamma_c^* &= \frac{1}{n} \sum_{i=1}^n \tau_{ic} \\
\left(\pi_{c,k}^{(\ell)} \right)^* &= \frac{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) u_{k,i}^{(\ell)} \tau_{ic}}{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) \tau_{ic}} \\
\left[\alpha_{c,k}^{(\ell)} \right]_v &\leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})| > 1} \mathbb{1}_{v \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \left[\alpha_{c,k}^{(\ell)} \right]_u \right) + \log \frac{\left[\mathcal{C}(x_{k,i}^{(\ell)}) \right]_v}{x_{k,i}^{(\ell)}} \right] \tau_{ic}}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})| > 1} \mathbb{1}_{v \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \tau_{ic}} \right)
\end{aligned}$$

Proof. We study the two parts of the EM algorithm, starting with the expectation step.

Expectation step:

Applying Bayes formula, we obtain:

$$\begin{aligned}
p_{\hat{\theta}}(Z_i = c | X_i, T_i) &= \tau_{ic} \\
&= \frac{p_{\hat{\theta}}(Z_i = c, X_i, T_i)}{\sum_{c'=1}^C p_{\hat{\theta}}(Z_i = c', X_i, T_i)} \\
&= \frac{\hat{\gamma}_c p_{\hat{\theta}}(T_i | Z_i = c) p_{\hat{\theta}}(X_i | T_i, Z_i = c)}{\sum_{c'=1}^C \hat{\gamma}_{c'} p_{\hat{\theta}}(T_i = Z_i = c') p_{\hat{\theta}}(X_i | T_i, Z_i = c')}
\end{aligned}$$

Exploiting the model framework, we obtain the following expression for the conditional distribution of the taxonomy:

$$\begin{aligned}
p_{\theta}(T_i | Z_i = c) &= p_{\theta}(T_i^{(1)}, \dots, T_i^{(L)} | Z_i = c) \\
&= \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\theta}(T_i^{(\ell+1)} | T_i^{(\ell)}, Z_i = c) \\
&= \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} p_{\theta}(u_{k,i}^{(\ell+1)} | \mathcal{P}(u_{k,i}^{(\ell+1)}), Z_i = c) \\
&= \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} \left[\left(\pi_{c,k}^{(\ell+1)} \right)^{u_{k,i}^{(\ell+1)}} \left(1 - \pi_{c,k}^{(\ell+1)} \right)^{1-u_{k,i}^{(\ell+1)}} \right]^{\mathcal{P}(u_{k,i}^{(\ell+1)})}
\end{aligned}$$

Similarly, the conditional abundance distribution is given by:

$$\begin{aligned}
p_{\theta}(X_i | T_i, Z_i = c) &= p_{\theta}(X_i^{(1)}, \dots, X_i^{(L)} | T_i, Z_i = c) \\
&= \delta_{e_1}(X_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\theta}(X_i^{(\ell+1)} | X_i^{(\ell)}, T_i^{(\ell+1)}, Z_i = c) \\
&= \delta_{e_1}(X_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} p_{\theta}(\mathcal{C}(x_{k,i}^{(\ell)}) | x_{k,i}^{(\ell)}, T_i^{(\ell+1)}, Z_i = c) \\
&= \delta_{e_1}(X_i^{(1)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_{\ell}} \delta_{x_{k,i}^{(\ell)}} \left(\mathcal{C}(x_{k,i}^{(\ell)}) \right)^{\mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|=1}} \left(\frac{1}{x_{k,i}^{(\ell)}} f_{\alpha_{c,k}^{(\ell)} \odot T_{i,k}^{(\ell+1)}} \left(\frac{\mathcal{C}(x_{k,i}^{(\ell)})}{x_{k,i}^{(\ell)}} \right) \right)^{\mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1}}
\end{aligned}$$

Maximization step

Recall the general objective:

$$\begin{aligned}
\theta^* &= \arg \max_{\theta} Q(\hat{\theta}, \theta) \\
&= \arg \max_{\theta} \frac{1}{n} \sum_{i=1}^n \sum_{c=1}^C (\log \gamma_c + \log p(T_i | Z_i = c) + \log p(X_i | T_i, Z_i = c)) \tau_{ic}
\end{aligned}$$

We start by optimizing γ . The objective can be written as:

$$\begin{aligned}
& \arg \max_{\gamma_j} \quad \sum_{i=1}^n \sum_{c=1}^C \tau_{ic} \log \gamma_c \\
& \text{s.t.} \quad \sum_{c=1}^C \gamma_c = 1 \\
& \quad \forall c, \gamma_c \geq 0
\end{aligned} \tag{2}$$

This optimization problem can be solved using Lagrange duality. KKT conditions are then written as (optimizing relatively to any γ_c):

$$\begin{aligned}
& \frac{1}{\gamma_c} \sum_{i=1}^n \tau_{ic} + \phi - \psi = 0 \\
& \sum_{c=1}^C \gamma_c = 1 \\
& \psi \geq 0 \\
& \forall c, \gamma_c \geq 0 \\
& \psi \gamma_c = 0
\end{aligned}$$

Using the first identity, we get:

$$\gamma_c = \frac{1}{\psi - \phi} \sum_{i=1}^n \tau_{ic}$$

Let's set $\phi = 0$, which enables use to ensure the last identity. Using the second identity, we then have:

$$\begin{aligned}
\sum_{c=1}^C \gamma_c = 1 &= \sum_{c=1}^C \frac{1}{\psi} \sum_{i=1}^n \tau_{ic} \\
&= \frac{1}{\psi} \sum_{i=1}^n \sum_{c=1}^C \tau_{ic} \\
&= \frac{1}{\psi} \sum_{i=1}^n 1 \\
&= \frac{n}{\psi}
\end{aligned}$$

Hence, we get $\psi = n$, which leads us to the optimal γ_c that verifies the KKT constraints:

$$\gamma_c^* = \frac{1}{n} \sum_{i=1}^n \tau_{ic}$$

Looking at π_c now, the optimization objective can be written as:

$$\begin{aligned}
& \arg \max_{\pi_{c,k}^{(\ell)}} \quad \sum_{i=1}^n \sum_{c=1}^C \tau_{ic} \log p_{\pi_c}(T_i | Z_i = c) \\
& \text{s.t.} \quad \pi_{c,k}^{(\ell)} \in [0, 1]
\end{aligned} \tag{3}$$

This objective is very similar to the MLE computed for proposition 6.2, up to a multiplication by τ_{ic} . Hence, we refer to the proposition 6.2 for the computation, which leads to the following optimal π_c :

$$\left(\pi_{c,k}^{(\ell)}\right)^* = \frac{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) u_{k,i}^{(\ell)} \tau_{ic}}{\sum_{i=1}^n \mathcal{P}(u_{k,i}^{(\ell)}) \tau_{ic}}$$

Similarly, the optimization objective of α_c is close to the one of proposition ??, up to a multiplication by τ_{ic} . Therefore, we refer to ?? for the original computation, that just requires a multiplication by τ_{ic} to obtain the following result:

$$\left[\alpha_{c,k}^{(\ell)}\right]_v \leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \left[\psi \left(\sum_{u \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \left[\alpha_{c,k}^{(\ell)}\right]_u \right) + \log \frac{[\mathcal{C}(x_{k,i}^{(\ell)})]_v}{x_{k,i}^{(\ell)}} \right] \tau_{ic}}{\sum_{i=1}^n \mathbb{1}_{|\mathcal{C}(x_{k,i}^{(\ell)})|>1} \mathbb{1}_{v \in \mathcal{V}(\alpha_{c,k}^{(\ell)}, T_i)} \tau_{ic}} \right)$$

□

Proposition 6.7 (Likelihood of layer-wise hidden markov model tree generator). *Let us assume the following framework:*

- Z follows a discrete Markov Chain, such that for all $c \in \{1, \dots, C\}$:

$$\begin{aligned} \mathbb{P}(Z^{(1)} = c) &= a_c \\ \mathbb{P}(Z^{(\ell+1)} = k | Z^{(\ell)} = c) &= t_{c,k}^{(\ell+1)} \end{aligned}$$

- T denotes a tree, for which the layers are denoted by $(T^{(\ell)})_{1 \leq \ell \leq L}$ and the internal nodes by $u_k^{(\ell)}$ (Bernoulli variables).
- We introduce $\mathcal{C}(T^{(\ell)})$ the set of plausible children in the taxonomy at layer $(\ell + 1)$, conditionally to $T^{(\ell)}$.
- The emission probability is then modeled by:

$$\mathbb{P}(T^{(\ell+1)} = \omega | Z^{(\ell+1)} = c, T^{(\ell)}) = \frac{\pi_c(\omega) \mathbb{1}_{\omega \in \mathcal{C}(T^{(\ell)})}}{\sum_{\nu \in \mathcal{C}(T^{(\ell)})} \pi_c(\nu)}$$

Then, the joint likelihood of such model is given by:

$$p(T, Z) = p(Z^{(1)}) \delta_{e_1}(T^{(1)}) \prod_{\ell=1}^{L-1} p(T^{(\ell+1)} | Z^{(\ell+1)}, T^{(\ell)}) p(Z^{(\ell+1)} | Z^{(\ell)})$$

Proof.

$$\begin{aligned}
p(T, Z) &= p(T^{(1)}, Z^{(1)}, \dots, T^{(L)}, Z^{(L)}) \\
&= p(T^{(1)}, Z^{(1)}) \prod_{\ell=1}^{L-1} p(T^{(\ell+1)}, Z^{(\ell+1)} | T^{(\ell)}, Z^{(\ell)}) \\
&= p(Z^{(1)}) p(T^{(1)} | Z^{(1)}) \prod_{\ell=1}^{L-1} p(T^{(\ell+1)} | Z^{(\ell+1)}, Z^{(\ell)}, T^{(\ell)}) p(Z^{(\ell+1)} | Z^{(\ell)}, T^{(\ell)}) \\
&= p(Z^{(1)}) \delta_{e_1}(T^{(1)}) \prod_{\ell=1}^{L-1} p(T^{(\ell+1)} | Z^{(\ell+1)}, T^{(\ell)}) p(Z^{(\ell+1)} | Z^{(\ell)})
\end{aligned}$$

□

Proposition 6.8 (Expectation-Maximization for layer-wise hidden markov model for tree generation). *Consider the framework of proposition 6.7. Recall the Expectation-Maximization objective:*

$$\theta^* = \arg \max_{\theta} Q(\hat{\theta}, \theta)$$

where $Q(\hat{\theta}, \theta) = \mathbb{E}_{p_{\hat{\theta}}(Z|T)}[\log p(T, Z) | T]$. The E-M algorithm is then given by:

- **Expectation step:**

Let $\nu = (c_1, \dots, c_L)$. We define $\tau_{i\nu} = p(Z_i = \nu | T_i)$. Then we have:

$$\tau_{i\nu} = \frac{p_{\hat{\theta}}(Z_i^{(1)} = c_1) \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c_{\ell+1}, T_i^{(\ell)}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c_{\ell+1} | Z_i^{(\ell)} = c_{\ell})}{\sum_{c'_1=1}^C \dots \sum_{c'_L=1}^C p_{\hat{\theta}}(Z_i^{(1)} = c'_1) \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c'_{\ell+1}, T_i^{(\ell)}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c'_{\ell+1} | Z_i^{(\ell)} = c'_{\ell})}$$

- **Maximization step:**

The prior on the markov chain root can be maximized explicitly:

$$a_c^* = \frac{1}{n} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \mathbb{1}_{c_1=c} \tau_{i\nu}$$

The emission probabilities can be computed through an iterative proximal gradient ascent algorithm.

The transition probabilities can be maximized explicitly:

$$t_{c,k}^{(\ell)*} = \frac{1}{\sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \mathbb{1}_{c_{\ell-1}=c} \mathbb{1}_{c_{\ell}=k}} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \mathbb{1}_{c_{\ell-1}=c} \mathbb{1}_{c_{\ell}=k}$$

Proof. We start by showing the **Expectation step**:

$$\begin{aligned}
\tau_{i\nu} &= p_{\hat{\theta}}(Z_i^{(1)} = c_1, \dots, Z_i^{(L)} = c_L | T_i) \\
&= \frac{p_{\hat{\theta}}(Z_i^{(1)} = c_1, \dots, Z_i^{(L)} = c_L, T_i)}{\sum_{c'_1=1}^C \dots \sum_{c'_L=1}^C p_{\hat{\theta}}(Z_i^{(1)} = c'_1, \dots, Z_i^{(L)} = c'_L, T_i)} \\
&= \frac{p_{\hat{\theta}}(Z_i^{(1)} = c_1) p_{\hat{\theta}}(T_i^{(1)} | Z_i^{(1)} = c_1) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c_{\ell+1}, T_i^{(\ell)}, Z_i^{(\ell)} = c_{\ell}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c_{\ell+1} | Z_i^{(\ell)} = c_{\ell}, T_i^{(\ell)})}{\sum_{c'_1=1}^C \dots \sum_{c'_L=1}^C p_{\hat{\theta}}(Z_i^{(1)} = c'_1) p_{\hat{\theta}}(T_i^{(1)} | Z_i^{(1)} = c'_1) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c'_{\ell+1}, T_i^{(\ell)}, Z_i^{(\ell)} = c'_{\ell}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c'_{\ell+1} | Z_i^{(\ell)} = c'_{\ell}, T_i^{(\ell)})} \\
&= \frac{p_{\hat{\theta}}(Z_i^{(1)} = c_1) \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c_{\ell+1}, T_i^{(\ell)}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c_{\ell+1} | Z_i^{(\ell)} = c_{\ell})}{\sum_{c'_1=1}^C \dots \sum_{c'_L=1}^C p_{\hat{\theta}}(Z_i^{(1)} = c'_1) \delta_{e_1}(T_i^{(1)}) \prod_{\ell=1}^{L-1} p_{\hat{\theta}}(T_i^{(\ell+1)} | Z_i^{(\ell+1)} = c'_{\ell+1}, T_i^{(\ell)}) p_{\hat{\theta}}(Z_i^{(\ell+1)} = c'_{\ell+1} | Z_i^{(\ell)} = c'_{\ell})}
\end{aligned}$$

We now prove the **Maximization step**. Recall the maximization objective:

$$\theta = \arg \max_{\theta} Q(\hat{\theta}, \theta)$$

We use the notation $\nu = (c_1, \dots, c_L)$. Using the likelihood computed at proposition 6.7, the maximization term becomes:

$$\begin{aligned} Q(\hat{\theta}, \theta) &= \mathbb{E}_{p_{\hat{\theta}}(Z|T)} [\log p_{\theta}(T, Z)|T] \\ &= \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \left[\log a_{c_1} + \sum_{\ell=1}^{L-1} \mathbb{1}_{T_i^{(\ell+1)} \in \mathcal{C}(T_i^{(\ell)})} \left(\log \pi_{c_{\ell+1}}^{(\ell+1)}(T_i^{(\ell+1)}) - \log \sum_{\mu \in \mathcal{C}(T_i^{(\ell)})} \pi_{c_{\ell+1}}^{(\ell+1)}(\mu) + \log t_{c_{\ell}, c_{\ell+1}}^{(\ell+1)} \right) \right] \tau_{i\nu} \end{aligned}$$

We obtain an explicit formula to compute the optimal parameters of the markov chain and emission probability:

- The optimization regarding $a = (a_1, \dots, a_C)$ is given by the following objective:

$$\begin{aligned} a_c^* &= \arg \max_{a_c} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \log a_{c_1} \\ \text{s.t.} \quad &\sum_{c=1}^C a_c = 1 \\ &\forall c, a_c > 0 \end{aligned}$$

This optimization problem is solvable through Lagrange duality, leading to the following optimal result:

$$a_c^* = \frac{1}{n} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \mathbb{1}_{c_1=c} \tau_{i\nu}$$

- For $\omega \in \mathcal{T}(\ell)$, where $\mathcal{T}(\ell)$ denotes the possible activation vectors in the global taxonomy \mathcal{T} , the optimization regarding the emission states parameters $\pi_c^{(\ell)}(\omega)$ is given by:

$$\begin{aligned} \pi_c^{(\ell)}(\omega)^* &= \arg \max_{\pi_c^{(\ell)}(\omega)} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \sum_{\ell=1}^{L-1} \mathbb{1}_{T_i^{(\ell+1)} \in \mathcal{C}(T_i^{(\ell)})} \left(\log \pi_{c_{\ell+1}}^{(\ell+1)}(T_i^{(\ell+1)}) - \log \sum_{\mu \in \mathcal{C}(T_i^{(\ell)})} \pi_{c_{\ell+1}}^{(\ell+1)}(\mu) \right) \\ \text{s.t.} \quad &\sum_{\nu \in \mathcal{T}(\ell)} \pi_c^{(\ell)}(\nu) = 1 \\ &\forall \nu \in \mathcal{T}(\ell), \pi_c^{(\ell)}(\nu) > 0 \end{aligned}$$

The derivative relatively to $\pi_c^{(\ell)}(\omega)$ is then given by:

$$\frac{\partial Q(\hat{\theta}, \theta)}{\partial \pi_c^{(\ell)}(\omega)} = \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \mathbb{1}_{\omega \in \mathcal{C}(T_i^{(\ell-1)})} \mathbb{1}_{c_{\ell}=c} \left(\frac{1}{\pi_c^{(\ell)}(\omega)} - \frac{1}{\sum_{\mu \in \mathcal{C}(T_i^{(\ell-1)})} \pi_c^{(\ell)}(\mu)} \right)$$

Since the objective is convex, this leads to a fixed point algorithm to compute the emission states parameters like proximal gradient descent to keep the constraint.

- The optimization objective regarding the transition probabilities of the markov chain $t_{c,k}^{(\ell)}$ is given by:

$$\begin{aligned}
t_{c,k}^{(\ell)*} = & \arg \max_{t_{c,k}^{(\ell)}} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \sum_{\ell=1}^{L-1} \mathbb{1}_{T_i^{(\ell+1)} \in \mathcal{C}(T_i^{(\ell)})} \log t_{c_\ell, c_{\ell+1}}^{(\ell+1)} \\
\text{s.t. } & \sum_{j=1}^C t_{c,j}^{(\ell)} = 1 \\
& \forall j, t_{c,j}^{(\ell)} > 0
\end{aligned}$$

Using Lagrange duality, we can solve the optimization problem to obtain the following optimal parameter:

$$t_{c,k}^{(\ell)*} = \frac{1}{\sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \mathbb{1}_{c_{\ell-1}=c}} \sum_{i=1}^n \sum_{\nu \in \mathcal{S}(Z)} \tau_{i\nu} \mathbb{1}_{c_{\ell-1}=c} \mathbb{1}_{c_\ell=k}$$

□

Proposition 6.9 (Joint distribution of Bottom-Up Hidden Tree Markov Model applied to top-down markov tree data). *Let X a top-down tree markov model. Let Z a Hidden Tree Markov Model defined over X .*

Then the joint likelihood is given by:

$$p(X, Z) = p(X^{(1)})p(Z^{(\ell)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) p(Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)}))$$

Proof.

$$\begin{aligned}
p(X, Z) &= p(X^{(1)}, Z^{(1)}, \dots, X^{(L)}, Z^{(L)}) \\
&= p(X^{(1)})p(Z^{(L)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)}, Z^{(\ell)} | X^{(\ell)}, Z^{(\ell+1)}) \\
&= p(X^{(1)})p(Z^{(L)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}), Z_k^{(\ell)} | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) \\
&= p(X^{(1)})p(Z^{(L)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}), |x_k^{(\ell)}, Z_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) p(Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)})) \\
&= p(X^{(1)})p(Z^{(L)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}), |x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) p(Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)}))
\end{aligned}$$

□

Proposition 6.10 (Expectation-Maximization for Bottom-Up Hidden Tree Markov Model with dirichlet modelization). *Let \mathcal{T} a global taxonomy. Define $(X_i, Z_i)_{1 \leq i \leq n}$ i.i.d latent taxa abundance samples over \mathcal{T} . We assume the following framework:*

- Z is a bottom-up HTMM with killing state s_0 so that:

$$p(Z^{(\ell)} = \nu) = \pi_\nu$$

$$p(Z_k^{(\ell)} = s | \mathcal{C}(Z_k^{(\ell)}) = \nu) = t_{\nu,s}^{(\ell)}$$

- X is a top-down markov tree conditionally to Z :

$$p(X^{(1)}) = \delta_{e_1}(X^{(1)})$$

$$p(X) = p(X^{(1)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)} | X^{(\ell)})$$

$$p(X^{(\ell+1)} | X^{(\ell)}) = \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)})$$

- The conditional distribution of X knowing Z is characterised as follows:

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)}) = \nu) = x_k^{(\ell)} \mathcal{D}(\alpha_{k,\nu}^{(\ell)})$$

$\alpha_{k,\nu}^{(\ell)}$ are the dirichlet parameters of the distribution, which are vectors of dimension

$$\#\{j \in \nu | j \neq s_0\}$$

as they only impact the nodes that are still alive according to Z .

Then we can run an Expectation-Maximization algorithm to compute the optimal parameters of that model onto the dataset, for which the steps are described as follows:

Expectation step:

For all possible Z over the taxonomy \mathcal{T} , we can explicitly compute $p(Z_i = Z | X_i) = \tau_{iZ}$:

$$\tau_{iZ} = \frac{\delta_{e_1}(X_i^{(1)}) p(Z^{(L)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_{i,k}^{(\ell)}) | x_{i,k}^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) p(Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)}))}{\sum_{\tilde{Z} \sim \mathcal{T}} \delta_{e_1}(X_i^{(1)}) p(\tilde{Z}^{(L)}) \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_{i,k}^{(\ell)}) | x_{i,k}^{(\ell)}, \mathcal{C}(\tilde{Z}_k^{(\ell)})) p(\tilde{Z}_k^{(\ell)} | \mathcal{C}(\tilde{Z}_k^{(\ell)}))}$$

Maximization step: For all possible ν over the last layer of the taxonomy $\mathcal{T}^{(L)}$,

$$\pi_\nu^* = \frac{1}{n} \sum_{i=1}^n \sum_{Z \sim \mathcal{T}} \tau_{iZ} \mathbb{1}_{Z^{(L)}=\nu}$$

For all given layer (ℓ) , consider ν a the possible hidden states of a group of children nodes of the taxonomy \mathcal{T} at layer (ℓ) , and s a possible state for the parent of the said group of children,

$$t_{\nu,s}^{(\ell)*} = \frac{1}{\sum_{i=1}^n \sum_{Z \sim \mathcal{T}} \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu} \tau_{iZ}} \sum_{i=1}^n \sum_{Z \sim \mathcal{T}} \mathbb{1}_{Z_k^{(\ell)}=s} \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu} \tau_{iZ}$$

For all parent node indexed by (ℓ, k) , ν a possible hidden state vector over $\mathcal{C}(Z_k^{(\ell)})$, the optimal dirichlet parameter is given by the following fixed point algorithm:

$$\left[\alpha_{k,\nu}^{(\ell)} \right]_j \leftarrow \psi^{-1} \left(\frac{\sum_{i=1}^n \sum_{Z \sim \mathcal{T}} \tau_{iZ} \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu} \log \frac{\mathcal{C}(x_{i,k}^{(\ell)})_j}{x_{i,k}^{(\ell)}}}{\sum_{i=1}^n \sum_{Z \sim \mathcal{T}} \tau_{iZ} \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu}} + \psi \left(\sum_j \left[\alpha_{k,\nu}^{(\ell)} \right]_j \right) \right)$$

Proof. We start by showing the **Expectation step**:

Using Bayes formula, we get:

$$\begin{aligned}\tau_{iZ} &= p(Z|X_i) \\ &= \frac{p(Z, X_i)}{p(X_i)} \\ &= \frac{p(Z, X_i)}{\sum_{\tilde{Z} \sim \mathcal{T}} p(\tilde{Z}, X_i)}\end{aligned}$$

Using proposition 6.9 to compute the joint likelihood, we get the wanted result.

We now look into the **Maximization step**. Recall the EM optimization objective:

$$\theta^* = \arg \max_{\theta} Q(\hat{\theta}, \theta) \quad (4)$$

where $Q(\hat{\theta}, \theta)$ is given by:

$$\begin{aligned}Q(\hat{\theta}, \theta) &= \mathbb{E}_{p_{\hat{\theta}}(Z|X)}[\log p_{\theta}(X, Z)|X] \\ &= \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \log p_{\theta}(X_i, Z_i = Z) \tau_{iZ} \\ &= \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \left(\log \pi_{Z^{(L)}} + \sum_{\ell=1}^{L-1} \sum_{k=1}^{K_{\ell}} \log p_{\alpha_{k, \mathcal{C}(Z_k^{(\ell)})}^{(\ell)}}(\mathcal{C}(x_k^{(\ell)}) | \mathcal{C}(Z_k^{(\ell)}, x_k^{(\ell)})) + \log t_{\mathcal{C}(Z_k^{(\ell)}, Z_k^{(\ell)})}^{(\ell)} \right) \tau_{iZ}\end{aligned}$$

- The optimal prior parameters π_{ν}^* are solutions of the following optimization problem:

$$\begin{aligned}\pi_{\nu}^* &= \arg \max_{\pi_{\nu}} \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \log \pi_{Z^{(L)}} \\ \text{s.t. } &\forall \nu \in \mathcal{T}^{(L)}, \pi_{\nu} > 0 \\ &\sum_{Z \sim \mathcal{T}} \pi_{Z^{(L)}} = 1\end{aligned} \quad (5)$$

This convex problem is solvable through Lagrange duality, for which the key KKT conditions are the following:

$$\sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \mathbb{1}_{Z^{(L)}=\nu} \frac{1}{\pi_{\nu}} - \psi + \phi = 0$$

Setting $\phi = 0$, we get:

$$\pi_{\nu} = \frac{1}{\psi} \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \mathbb{1}_{Z^{(L)}=\nu}$$

Recall the summation constraint over the π_{ν} ,

$$\sum_{Z \sim \mathcal{T}} \pi_{Z^{(L)}} = 1$$

Which implies:

$$\begin{aligned}
\psi &= \sum_{Z \sim \mathcal{T}} \sum_{Z' \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \mathbb{1}_{Z^{(L)}=\nu} \\
&= \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \\
&= \sum_{i=1}^n 1 \\
&= n
\end{aligned}$$

Which gives us the wanted formula.

The optimal transition parameters $t_{\nu,s}^{(\ell)*}$ are solutions of the following optimization problem:

$$\begin{aligned}
t_{\nu,s}^{(\ell)*} &= \arg \max_{t_{\nu,s}^{(\ell)}} \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \sum_{\ell=1}^{L-1} \sum_{k=1}^{K_\ell} \log t_{\mathcal{C}(Z_k^{(\ell)}), Z_k^{(\ell)}}^{(\ell)} \\
\text{s.t. } &\forall s \in \mathcal{S}(Z_k^{(\ell)}), t_{\nu,s}^{(\ell)} > 0 \\
&\sum_{s \in \mathcal{S}(Z_k^{(\ell)})} t_{\nu,s}^{(\ell)} = 1
\end{aligned} \tag{6}$$

As previously, this problem can be solved through Lagrange duality. Using a similar reasoning, we get the normalization value for ψ given by:

$$\psi = \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu} \tau_{iZ}$$

Which gives us the objective result.

The optimal abundance parameters $\alpha_{k,\nu}^{(\ell)*}$, using proposition 6.3 on the conditional distribution, the parameters are solutions of the following optimization problem:

$$\begin{aligned}
\left[\alpha_{k,\nu}^{(\ell)}\right]_j^* &= \arg \max_{\left[\alpha_{k,\nu}^{(\ell)}\right]} \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \sum_{\ell=1}^{L-1} \sum_{k=1}^{K_\ell} \log \frac{1}{x_k^{(\ell)}} f_{\alpha_{k,\mathcal{C}(Z_k^{(\ell)})}^{(\ell)}} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right) \\
\text{s.t. } &\forall j, \left[\alpha_{k,\nu}^{(\ell)}\right]_j > 0
\end{aligned} \tag{7}$$

Deriving the objective we get:

$$\frac{\partial Q(\hat{\theta}, \theta)}{\partial \left[\alpha_{k,\nu}^{(\ell)}\right]_j} = \sum_{Z \sim \mathcal{T}} \sum_{i=1}^n \tau_{iZ} \mathbb{1}_{\mathcal{C}(Z_k^{(\ell)})=\nu} \left(\psi \left(\sum_j \left[\alpha_{k,\nu}^{(\ell)}\right]_j \right) - \psi \left(\left[\alpha_{k,\nu}^{(\ell)}\right]_j \right) + \log \frac{\mathcal{C}(x_k^{(\ell)})_j}{x_k^{(\ell)}} \right)$$

Looking for 0 valued gradient and using the fixed point algorithm provided by [7], we obtain the fix point algorithm we want. □

Proposition 6.11 (Likelihood under-parameterized Bottom-Up HTMM). *Assume Z is a HTMM over the taxonomy \mathcal{T} , X a taxa-abundance sample over \mathcal{T} . Assume that the following holds:*

- *Let $S \in \mathbb{N}$ the number of possible hidden states, at each layer (ℓ) we introduce the parameter $\pi^{(\ell)} \in \mathbb{R}^S$, which defines the aggregation parameter $\omega_k^{(\ell)}$:*

$$\omega_k^{(\ell)} = \sum_{z \in \mathcal{C}(Z_k^{(\ell)})} \sum_{s=1}^S \mathbb{1}_{z=e_s} \pi_s^{(\ell)} e_s$$

where e_s denotes the embedding of the state s into a vector representation (one-hot encoding for instance).

- *Z is bottom-up HTMM with killing state s_0 , for which the transition probabilities are defined through $\omega_k^{(\ell)}$:*

$$\begin{aligned} Z_k^{(L)} &\sim \text{Cat}(\pi^{(L)}) \\ Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)}) &\sim \text{Cat}(\text{softmax}(\omega_k^{(\ell)})) \end{aligned}$$

- *X is a top-down markov tree conditionally to Z :*

$$\begin{aligned} p(X^{(1)}) &= \delta_{e_1}(X^{(1)}) \\ p(X) &= p(X^{(1)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)} | X^{(\ell)}) \\ p(X^{(\ell+1)} | X^{(\ell)}) &= \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}) \end{aligned}$$

- *The conditional distribution of X knowing Z is characterised as follows, for the nodes that have at least 2 children and that are not at hidden state s_0 :*

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(z_k^{(\ell)}) = \nu) = x_k^{(\ell)} \mathcal{D}(\alpha_k^{(\ell)} \odot \nu)$$

Otherwise if $x_k^{(\ell)}$ has 1 child, its abundance value is transmitted deterministically to his child if it is not at state s_0 , otherwise the value is 0.

Then the likelihood of such model is given by:

$$\begin{aligned} p(X, Z) &= \delta_{e_1}(X^{(1)}) \prod_{k=1}^{K_L} \pi_{Z_k^{(L)}}^{(L)} \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} \delta_0(\mathcal{C}(x_k^{(\ell)})) \mathbb{1}_{Z_k^{(\ell)}=s_0} \\ &\quad \delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)})) \mathbb{1}_{\# \mathcal{C}(Z_k^{(\ell)})=1} \mathbb{1}_{Z_k^{(\ell)} \neq s_0} \\ &\quad \left(\frac{1}{x_k^{(\ell)}} f_{\alpha_k^{(\ell)} \odot \mathcal{C}(Z_k^{(\ell)})} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right) \right)^{\mathbb{1}_{\# \mathcal{C}(Z_k^{(\ell)}) > 1} \mathbb{1}_{Z_k^{(\ell)} \neq s_0}} \text{softmax}(\omega_k^{(\ell)})_{Z_k^{(\ell)}} \end{aligned}$$

Proof.

$$\begin{aligned}
p(X, Z) &= p(X^{(1)}, Z^{(1)}, \dots, X^{(L)}, Z^{(L)}) \\
&= \delta_1(X_1^{(1)}) p(Z^{(L)}) \prod_{\ell=1}^{L-1} p(X^{(\ell+1)}, Z^{(\ell)} | X^{(\ell)}, Z^{(\ell+1)}) \\
&= \delta_1(X_1^{(1)}) \prod_{k=1}^{K_L} \pi_{Z_k^L}^{(L)} \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) p(Z_k^{(\ell)} | \mathcal{C}(Z_k^{(\ell)})) \\
&= \delta_1(X_1^{(1)}) \prod_{k=1}^{K_L} \pi_{Z_k^L}^{(L)} \prod_{\ell=1}^{L-1} \prod_{k=1}^{K_\ell} p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) \text{softmax}(w_k^{(\ell)})_{Z_k^{(\ell)}}
\end{aligned}$$

Notice three possible different scenarios:

- If the node is being killed by the HTMM through s_\emptyset :

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) = \delta_0(\mathcal{C}(x_k^{(\ell)}))$$

- If the node has only one child and that it is not killed by the HTMM:

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}, \mathcal{C}(Z_k^{(\ell)})) = \delta_{x_k^{(\ell)}}(\mathcal{C}(x_k^{(\ell)}))$$

- Finally, if the children has two children or more and that it is not killed by the HTMM, using the proposition 6.3:

$$p(\mathcal{C}(x_k^{(\ell)}) | x_k^{(\ell)}) = \frac{1}{x_k^{(\ell)}} f_{\alpha_k^{(\ell)} \odot \mathcal{C}(Z_k^{(\ell)})} \left(\frac{\mathcal{C}(x_k^{(\ell)})}{x_k^{(\ell)}} \right)$$

Marking these three cases through the corresponding indicator functions we obtain the wanted result. \square

Proposition 6.12. (*Expectation-Maximization for under parameterized Bottom-Up HTMM model*) We set ourselves in the framework of 6.11.

Let $(X_i, Z_i)_{1 \leq i \leq n}$ a dataset of respectively taxa-abundance data and hidden functional states defined on the taxonomy \mathcal{T} . We denote by $\mathcal{S}(\mathcal{T})$ the set of possible HTMM realization over \mathcal{T} .

Assume that we want to find the θ^* MLE of the under parameterized bottom-up HTMM model over that dataset, then we provide an Expectation-Maximization approach to compute it:

Expectation-step:

Denote $\tau_{iZ} = p(Z_i | X_i)$, then this quantity is given by:

$$\tau_{iZ} = \frac{p(Z_i, X_i)}{\sum_{Z \in \mathcal{S}(\mathcal{T})} p(Z, X_i)}$$

where $p(Z, X)$ is explicitly provided in proposition 6.11

Maximization-step:

The optimal prior parameter of the HTMM is given by:

$$\pi_s^{(L)*} = \dots$$

The optimal transition parameters ($1 < \ell < L$) of the HTMM are given by:

$$\pi_s^{(\ell)*} = \dots$$

The optimal abundance parameters are obtained through the following fixed point algorithm:

$$\alpha_k^{(\ell)} \leftarrow \psi^{(-1)}(\dots)$$

Proposition 6.13. (Variational Expectation-Maximization objective for multi-layer PLN) Recall our specific microbiota framework, we introduce a taxonomy \mathcal{T} , with N nodes, L layers, and K_ℓ entities at layer ℓ . Let $(X_i)_{1 \leq i \leq n}$ the taxa-abundance samples of n individuals, supported on the taxonomy \mathcal{T} . We denote by $x_k^{(\ell)} \in \mathbb{N}$ the abundance of the k -th entity in a taxa-abundance sample at layer ℓ .

The multi-layer PLN model is defined through the following assumptions:

- $Z = (Z^{(1)}, \dots, Z^{(L)}) \sim \mathcal{N}(0, \Sigma)$, where $\Sigma \in \mathbb{R}^{(K_1 + \dots + K_L)^2}$
- $Z^{(\ell)} \sim \mathcal{N}(0, \Sigma^{(\ell)})$ where $\Sigma^{(\ell)} \in \mathbb{R}^{K_\ell^2}$
- $x_k^{(\ell)} \sim \mathcal{P}(e^{Z_k^{(\ell)}})$

We choose the variational family of independent gaussian kernels:

$$q_\Phi(Z|X) = \prod_{\ell=1}^L q_{\Phi_\ell}(Z^{(\ell)}|X^{(\ell)})$$

where $q_{\Phi_\ell}(Z^{(\ell)}|X^{(\ell)}) \sim \mathcal{N}(m^{(\ell)}, S^{(\ell)})$

Recall the optimization objective, given by the suboptimal maximum of likelihood of the ELBO:

$$\arg \max_{\theta, \Phi} ELBO(\theta, \Phi) = \mathbb{E}_{q_\Phi} [\log p_\theta(X, Z)] - \mathbb{E}_{q_\Phi} [\log q_\Phi(Z|X)]$$

Under these assumptions, the ELBO can then be written as:

$$\begin{aligned} ELBO(\theta, \Phi) = & -\frac{1}{2} \log \frac{|\Sigma|}{|S|} + \frac{1}{2} \sum_{\ell=1}^L K_\ell - \frac{1}{2} \text{tr}(\Sigma^{-1} S) - \frac{1}{2} m^\top \Sigma m \\ & + \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} X_k^{(\ell)} m_k^{(\ell)} - \log X_k^{(\ell)}! - e^{\frac{S_k^{(\ell)2}}{2} + m_k^{(\ell)}} \end{aligned}$$

Proof. Recall the ELBO formula:

$$ELBO(\theta, \Phi) = \underbrace{\mathbb{E}_{q_\Phi} [\log p_\theta(X, Z)]}_{\text{joint distribution}} - \underbrace{\mathbb{E}_{q_\Phi} [\log q_\Phi(Z|X)]}_{\text{entropy}}$$

We start by developing the entropy term:

$$\begin{aligned} H &= -\mathbb{E}_{q_\Phi} [\log q_\Phi(Z|X)] \\ &= -\int \log q_\Phi(Z) q_\Phi(Z) dZ \end{aligned}$$

Then, we develop the joint distribution term, exploiting the conditional independence of the taxa conditionnally to the respective latent variable:

$$\begin{aligned} \mathbb{E}_{q_\Phi} [\log p_\theta(X, Z)] &= \int \log p_\theta(X, Z) q_\Phi(Z) dZ \\ &= \int \log p_\theta(Z) q_\Phi(Z) dZ + \int \log p_\theta(X|Z) q_\Phi(Z) dZ \\ &= \underbrace{\int \log p_\theta(Z) q_\Phi(Z) dZ}_{(A)} + \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} \int \log p_\theta(X_k^{(\ell)} | Z_k^{(\ell)}) q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)} \end{aligned}$$

Notice then:

$$H + (A) = -D_{KL}(q_\Phi \| p_\theta)$$

Leading to a new formulation of the ELBO:

$$\begin{aligned} ELBO(\theta, \Phi) &= E_{q_\Phi} [\log p_\theta(X, Z)] + H \\ &= \underbrace{\sum_{\ell=1}^L \sum_{k=1}^{K_\ell} \int \log p_\theta(X_k^{(\ell)} | Z_k^{(\ell)}) q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)}}_{(B)} - D_{KL}(q_\Phi(Z) \| p_\theta(Z)) \end{aligned}$$

We start by expressing the first term:

$$\begin{aligned} (B) &= \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} \int \log p_\theta(X_k^{(\ell)} | Z_k^{(\ell)}) q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)} \\ &= \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} \int \log \frac{e^{Z_k^{(\ell)} X_k^{(\ell)}}}{X_k^{(\ell)}!} e^{-e^{Z_k^{(\ell)}}} q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)} \\ &= \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} \int (X_k^{(\ell)} Z_k^{(\ell)} - \log X_k^{(\ell)}! - e^{Z_k^{(\ell)}}) q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)} \\ &= \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} X_k^{(\ell)} m_k^{(\ell)} - \log X_k^{(\ell)}! - \underbrace{\int e^{Z_k^{(\ell)}} q_{\Phi_\ell}(Z^{(\ell)}) dZ^{(\ell)}}_{(*)} \end{aligned}$$

Developing (*) we get:

$$\begin{aligned}
(*) &= \int e^{Z_k^{(\ell)}} (2\pi)^{-\frac{K_\ell}{2}} |S^{(\ell)}|^{-\frac{1}{2}} \exp(-(Z^{(\ell)} - m^{(\ell)})^\top \Sigma^{-1} (Z^{(\ell)} - m^{(\ell)})) dZ^{(\ell)} \\
&= \int e^{Z_k^{(\ell)}} (2\pi S_k^{(\ell)2})^{-\frac{1}{2}} \exp\left(-\frac{(Z_k^{(\ell)} - m_k^{(\ell)})^2}{2S_k^{(\ell)2}}\right) dZ_k^{(\ell)} \\
&= (2\pi S_k^{(\ell)2})^{-\frac{1}{2}} \int \exp\left(-\frac{(Z_k^{(\ell)2} - 2Z_k^{(\ell)} m_k^{(\ell)} + m_k^{(\ell)2} - 2Z_k^{(\ell)} S_k^{(\ell)2})}{2S_k^{(\ell)2}}\right) dZ_k^{(\ell)} \\
&= (2\pi S_k^{(\ell)2})^{-\frac{1}{2}} \int \exp\left(-\frac{(Z_k^{(\ell)2} - 2Z_k^{(\ell)}(m_k^{(\ell)} + S_k^{(\ell)2}) + m_k^{(\ell)2})}{2S_k^{(\ell)2}}\right) dZ_k^{(\ell)} \\
&= (2\pi S_k^{(\ell)2})^{-\frac{1}{2}} \int \exp\left(-\frac{(Z_k^{(\ell)} + m_k^{(\ell)} + S_k^{(\ell)2})^2}{2S_k^{(\ell)2}}\right) dZ_k^{(\ell)} \exp\left(\frac{S_k^{(\ell)2}}{2} + m_k^{(\ell)}\right) \\
&= \exp\left(\frac{S_k^{(\ell)2}}{2} + m_k^{(\ell)}\right)
\end{aligned}$$

Hence we get an explicit formula for (B):

$$(B) = \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} X_k^{(\ell)} m_k^{(\ell)} - \log X_k^{(\ell)}! - e^{\frac{S_k^{(\ell)2}}{2} + m_k^{(\ell)}}$$

Finally, using chapter 8.2 of [11], we can compute the KL divergence between the two gaussians q_Φ and p_θ :

$$\begin{aligned}
D_{KL}(q_\Phi(Z) \| p_\theta(Z)) &= \int \left[\frac{1}{2} \log \frac{|\Sigma|}{|S|} - \frac{1}{2} (z - m)^\top S^{-1} (z - m) + \frac{1}{2} z^\top \Sigma^{-1} x \right] q_\Phi(z) dz \\
&= \frac{1}{2} \log \frac{|\Sigma|}{|S|} - \frac{1}{2} \text{tr}(\mathbb{E}_{q_\Phi}[(Z - m)(Z - m)^\top] S^{-1}) + \frac{1}{2} \mathbb{E}_{q_\Phi}[Z^\top \Sigma^{-1} Z] \\
&= \frac{1}{2} \log \frac{|\Sigma|}{|S|} - \frac{1}{2} \text{tr}(I_{K_1+\dots+K_L}) + \frac{1}{2} m^\top \Sigma^{-1} m + \frac{1}{2} \text{tr}(\Sigma^{-1} S) \\
&= \frac{1}{2} \left(\log \frac{|\Sigma|}{|S|} - \sum_{\ell=1}^L K_\ell + \text{tr}(\Sigma^{-1} S) + m^\top \Sigma m \right)
\end{aligned}$$

We then obtain the complete ELBO:

$$\begin{aligned}
ELBO(\theta, \Phi) &= -\frac{1}{2} \log \frac{|\Sigma|}{|S|} + \frac{1}{2} \sum_{\ell=1}^L K_\ell - \frac{1}{2} \text{tr}(\Sigma^{-1} S) - \frac{1}{2} m^\top \Sigma m \\
&\quad + \sum_{\ell=1}^L \sum_{k=1}^{K_\ell} X_k^{(\ell)} m_k^{(\ell)} - \log(X_k^{(\ell)}!) - e^{\frac{S_k^{(\ell)2}}{2} + m_k^{(\ell)}}
\end{aligned}$$

□

7 Bibliography

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