Latent variable models in biology and ecology

Chapter 2: Mixtures models

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- Mixture model: one of the most simple latent variable models
- Assumptions
  - Observations supposed to be independent,
  - Each observation arises from a given class that is unobserved
- Main goal : retrieve the class from which each observation arises
- Also referred as unsupervised classification as we do not dispose of any observation with known label.
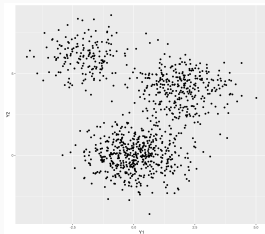
First toy illustration

Observations described by 2 variables
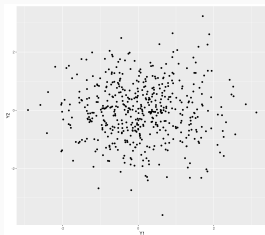
Observation distribution seems easy to model with one Gaussian

First toy illustration

Observations described by 2 variables

Data are scattered and subpopulations are observed

According to the experimental design, there exists no external information about them

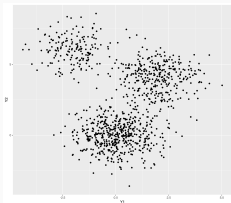**This is an underlying structure observed through the data**

Definition (Mixture model)

It is a probabilistic model for representing the presence of subpopulations within an overall population.
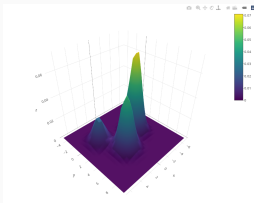
$$Y_i|Z_i = k \sim \mathcal{N}(\mu_k, \Sigma_k), \quad P(Z_i = k) = \pi_k$$

what we observe | the model | the expected results

$Z = ?$ | | $Z : 1 = \bullet, 2 = \bullet, 3 = \bullet$

$\rightarrow$ It is an unsupervised classification method

Applications in biology

Technics of clustering widely used in biology. See the Wikipedia page

Gene expression

- To build groups of genes with related expression patterns (also known as coexpressed genes).
- Often such groups contain functionally related proteins, such as enzymes for a specific pathway, or genes that are co-regulated.
- Y_{tm} gene expression of gene at locus t in condition $m = 1, \ldots, P$ conditions.

(Figure from [Parraga-Alava et al., 2018])

Human genetic

- Better understand the genetic structure of populations
- Relies on the genotyping of large sets of individuals sampled in different places, environments or with different origins
- Genotype Y_{it} of a series of individuals $i \in [1, I]$ at a series of locus $t \in [1, T]$ is measured
- Aim: distinguish sub-populations.

Model without 'admixture'

Each individual i is supposed to belong to one population, labeled Z_i

$$
\begin{aligned}
(Z_i)_i \text{ iid} \quad &\sim \quad \mathcal{M}(1; \pi), \\
(Y_{it})_{i,t} \text{ indep.} \,|\, (Z_i) \quad &\sim \quad \mathcal{M}(1; \gamma_{Z_i t}),
\end{aligned}
$$

γ_{kt} is the vector of the allelic frequencies at locus t in population k

which makes explicit the fact that, if individual i belongs to population k, its genotype is generated with the allelic frequencies of its population.

Model with 'admixture'

$$(Y_{it})_{i,t} \text{ indep.} \,|\, (S_{it}) \;\sim\; \mathcal{M}(1; \gamma_{S_it})$$
$$(S_{it})_{i,t} \text{ indep.} \,|\, (Q_i) \;\sim\; \mathcal{M}(1; Q_i),$$
$$(Q_i)_i \text{ iid} \;\sim\; \mathcal{D}(1; \alpha)$$

About Q_i: individual preferential trends characterized

- Dirichlet distribution whose support is the the simplex of $\mathbb{R}^K$.
- D_i is the position of individual i in the simplex, the vertices of which correspond to fictitious individuals purely issued from each population.

Hidden variable is hence $Z = (Q, S)$.

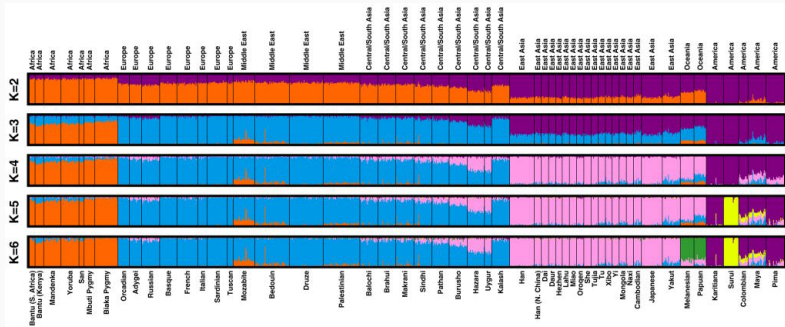## Model with 'admixture': reformulation

The model can be rewritten also after marginalization over S_{it}:

$$
\begin{aligned}
(Q_i)_i \text{ iid} &\sim \mathcal{D}(1; \alpha), \\
(Y_{it})_{i,t} \text{ indep.} \,|\, (Q_i) &\sim \mathcal{M}\left(1; \sum_k Q_{ik}\gamma_{kt}\right).
\end{aligned}
$$

The latent variable reduces then to $Z = (Q)$.

See [Pritchard et al., 2000] for more details.

Expected results

Population origine of series of human genomes with varying number of groups K. Each column corresponds to an individual. Each individual is represented by a thin vertical line partitioned into K colored segments that represent the fractions of the individual's genome estimated to belong to the K clusters. From [Rosenberg, 2011].

Plant and animal ecology i

To describe and to make spatial and temporal comparisons of communities (assemblages) of organisms in heterogeneous environments.

- Y_{is}: abundancy of species i at location s.
- Not the same repartitions with respect to species.

Plant and animal ecology ii
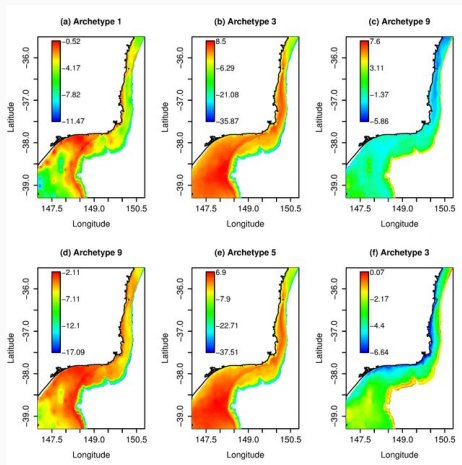
[Dunstan et al., 2013]

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Definition

- Let $(Y_i)_{i=1,\ldots,n}$ be independent variables
- For each individual i assumes the existence an unknown (or latent) label Z_i that can take a finite number of values among $[1, K]$.
- The distribution of Y_i depends on the value Z_i.

Definition

An independent K mixture model is defined as follows: $\forall i = 1, \ldots, n$

$$
\begin{aligned}
P(Z_i = k) &= \pi_k, \quad (i.i.d) \\
Y_i|(Z_i = k) &\sim_{i.i.d} \mathcal{F}_k = \mathcal{F}(\gamma_k),
\end{aligned}
\tag{1}
$$

where $\sum_{k=1}^{K} \pi = 1$.

Let $f_k(\cdot) = f(\cdot; \gamma_k)$ be the pdf of distribution of $\mathcal{F}(\gamma_k)$.

- $Y_i|(Z_i = k) \sim \mathcal{F}(\gamma_k)$ is equivalent to $Y_i|Z_i \sim \mathcal{F}(\gamma_{Z_i})$
- Let $Z_{ik} = \mathbf{1}_{\{Z_i = k\}}$

$$(Z_{ik})_{k=1,\ldots,K} \sim \mathcal{M}(1, \boldsymbol{\pi})$$

where $\mathcal{M}$ is the multinomial distribution $\boldsymbol{\pi} = (\pi_1, \ldots, \pi_K)$

About the mixture proportions

- $\pi_k =$ proportion of the population k
- Sometimes called prior probabilities although this denomination may be misleading in a non-Bayesian context.
- Also often refereed to as the proportions of the mixture.

About the emission distribution

- Conditionally on $\{Z_i = k\}$, Y_i has a parametric distribution $\mathcal{F}_k = \mathcal{F}(\gamma_k)$ with probability distribution function (pdf) $f_k(\cdot) = f(\cdot; \gamma_k)$.
- $\mathcal{F}_k$ is called the emission distribution in class k
- It describes how observed data arising from class k are emitted.
- f_k is called the emission pdf.

Useful notations

- $\mathbf{Z} = (Z_1, \ldots, Z_n)$
- $\mathbf{Y} = (Y_1, \ldots, Y_n)$
- $\boldsymbol{\pi} = (\pi_k)_{k=1,\ldots,K}$
- $\boldsymbol{\gamma} = (\gamma_k)_{k=1,\ldots,K}$
- $\theta = (\boldsymbol{\pi}, \boldsymbol{\gamma})$

Conditional distributions

$$
\begin{array}{rcccl}
p_\theta(\mathbf{Z}) &=& \prod_{i=1}^n \pi_{Z_i} &=& \prod_{i=1}^n \prod_{k=1}^K (\pi_k)^{Z_{ik}}, \\
p_\theta(\mathbf{Y}|\mathbf{Z}) &=& \prod_{i=1}^n f(Y_i, \gamma_{Z_i}) &=& \prod_{i=1}^n \prod_{k=1}^K f(Y_i, \gamma_k)^{Z_{ik}},
\end{array}
$$

Marginal distribution

Marginal pdf. of Y_i is the mixture distribution

$$g(y) = \sum_{k=1}^{K} \pi_k f(y; \gamma_k).$$

Example of a mixture of $K = 3$
Gaussian distributions
$\frac{1}{3}\mathcal{N}(1,1) + \frac{1}{6}\mathcal{N}(3,1) + \frac{1}{2}\mathcal{N}(5,3^2)$

Label switching
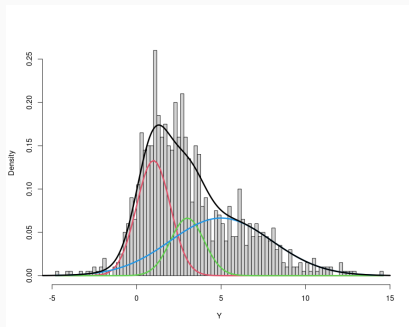
Since the (Z_i) are not observed, the model is invariant for any permutation of the labels $[1, K]$.

Therefore, the mixture model with K classes has $K!$ equivalent definitions.

Number of parameters

- Depends on both the dimension of the data and the number of groups
- $\sum_{k=1}^{K} \pi_k = 1$, $\boldsymbol{\pi}$ involves only $K - 1$
- About $\boldsymbol{\gamma} = (\gamma_1, \ldots, \gamma_K)$, its dimension is typically proportional to the number of groups K
- For $\mathcal{F}_k$: univariate Poisson distributions with respective mean γ_k, $\boldsymbol{\gamma}$ of dimension $K \Rightarrow 2K - 1$ parameters
- For $\mathcal{F}_k$: d-variate normal distributions (with respective mean vector μ_k and variance Σ_k):

$$(K - 1) + Kd + Kd(d + 1)/2 \simeq Kd^2/2$$

parameters

Dependency structures

- The (Z_i) are independent;
- the (Y_i) are independent conditionally to $\mathbf{Z} = (Z_i)_{i=1,\ldots,n}$;
- the couples $\{(Y_i, Z_i)\}_i$ are iid.
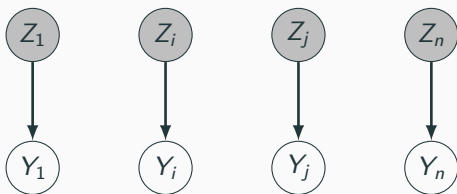
Graphical representation of a mixture model

Remarks

1. Because the $\{(Y_i, Z_i)\}_i$ are independent, we have that

$$p_\theta(Z_i|\mathbf{Y}) = p_\theta(Z_i|Y_i)$$

   which means that the information about the classification of individual i is contained in the observation Y_i.

2. Note that the variables (Y_i, Y_j) are *not* independent conditionally on the event $Z_i = Z_j$.

Two tasks

- For a fixed number of class K, estimating the parameters

$$\boldsymbol{\pi} = (\pi_1, \ldots, \pi_K), \quad \boldsymbol{\gamma} = (\gamma_1, \ldots, \gamma_K)$$

$$\theta = (\boldsymbol{\pi}, \boldsymbol{\gamma})$$

$\Rightarrow$ (Maximum likelihood) estimation

- Would be great to obtain a classification of the observations
- Choosing the number of classes $K \Rightarrow$ Model selection

Parameter estimation

- General introduction to finite mixture models and their inference can be found in [McLachlan and Peel, 2000]
- Most popular inference method: maximum likelihood approach
- Specificity of latent variable models : the observed data $\mathbf{Y} = (Y_i)_{i=1,\dots,n}$ seen as incomplete, as the latent variables $\mathbf{Z} = (Z_i)_{i=1,\dots,n}$ are not observed
- Often referred to as incomplete data models.

Likelihoods

Definition

The observed data log-likelihood is the marginal log-likelihood of the observed variables $\mathbf{Y}$:

$$\log p_\theta(\mathbf{Y}).$$

The complete data log-likelihood is the joint log-likelihood of the observed $\mathbf{Y}$ and latent $\mathbf{Z}$ variables:

$$\log p_\theta(\mathbf{Y}, \mathbf{Z}).$$

Expression of the likelihoods

Proposition (Likelihoods)

For the mixture model (1), the log-likelihood is

$$\log p_\theta(\mathbf{Y}) = \sum_{i=1}^{n} \log \left[\sum_{k=1}^{K} \pi_k f(Y_i; \gamma_k) \right],$$

and, denoting $Z_{ik} = \mathbf{1}_{\{Z_i = k\}}$, the complete log-likelihood is

$$\log p_\theta(\mathbf{Y}, \mathbf{Z}) = \sum_{i=1}^{n} \sum_{k=1}^{K} Z_{ik} \left[\log \pi_k + \log f(Y_i; \gamma_k) \right].$$

Proof

The dependency structure described in previously ensures that

$$
\begin{aligned}
\log p_\theta(\mathbf{Y}) &= \sum_{i=1}^{n} \log p_\theta(Y_i) = \sum_{i=1}^{n} \log g(Y_i) \\
\text{and} \qquad \log p_\theta(\mathbf{Y}, \mathbf{Z}) &= \sum_{i=1}^{n} \log p_\theta(Y_i, Z_i) \\
&= \sum_{i=1}^{n} \left[\log p_\theta(Z_i) + \log p_\theta(Y_i | Z_i) \right].
\end{aligned}
$$

Remark: $\log p_\theta(Y_i)$ not easy to optimize

About the EM algorithm

- First proposed by [Dempster et al., 1977] for a large class of incomplete data models, including mixture models.
- Based on a decomposition of the incomplete data likelihood.

Proposition (Decomposition of the log-likelihood)

For any θ and θ'

$$\log p_\theta(\mathbf{Y}) = \mathbb{E}_{\theta'}\left[\log p_\theta(\mathbf{Y}, \mathbf{Z})|Y\right] - \mathbb{E}_{\theta'}\left[\log p_\theta(\mathbf{Z}|\mathbf{Y})|\mathbf{Y}\right].$$

Proof

It suffices to develop

$$\mathbb{E}_{\theta'}\left[\log p_\theta(\mathbf{Z}|\mathbf{Y})|\mathbf{Y}\right] \quad = \quad \mathbb{E}_{\theta'}\left[\log p_\theta(\mathbf{Y},\mathbf{Z}) - \log p_\theta(\mathbf{Y})|\mathbf{Y}\right]$$

reminding that $\mathbb{E}_{\theta'}\left[\log p_\theta(\mathbf{Y})|\mathbf{Y}\right] = \log p_\theta(Y)$.

Remarks

1. Decomposition of Slide 35 is convenient bacause makes a connexion between $\log p_\theta(\mathbf{Y})$ (often intractable) and $\log p_\theta(\mathbf{Y}, \mathbf{Z})$ (generally more manageable).

2. if $\theta' = \theta$, the second term is the entropy of the latent variables $\mathbf{Z}$ given the observed $\mathbf{Y}$:

$$\mathcal{H}[p_\theta(\mathbf{Z}|\mathbf{Y})] := -\mathbb{E}_\theta[\log p_\theta(\mathbf{Z}|\mathbf{Y})|\mathbf{Y}]$$

EM Algorithm

$$\widehat{\theta} = \arg \max_{\theta} \log p_\theta(\mathbf{Y}).$$

Algorithm (EM)

Repeat until convergence:

__Expectation step__ (E-step) given the current estimate θ^h of θ, compute $p_{\theta^h}(\mathbf{Z}|\mathbf{Y})$, or at least all the quantities needed to compute $\mathbb{E}_{\theta^h}[\log p_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}]$;

__Maximization step__ (M-step) update the estimate of θ as

$$\theta^{h+1} = \arg \max_{\theta} \mathbb{E}_{\theta^h}[\log p_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}].$$

Proposition ([Dempster et al., 1977])

The log-likelihood of the observed data $\log p_\theta(\mathbf{Y})$ *increases at each step:*

$$\log p_{\theta^{h+1}}(\mathbf{Y}) \geq \log p_{\theta^h}(\mathbf{Y}).$$

Proof i

Because $\theta^{h+1} = \arg\max_\theta \mathbb{E}_{\theta^h}[\log p_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}]$, we have

$$
\begin{align}
0 &\leq \mathbb{E}_{\theta^h}[\log p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}] - \mathbb{E}_{\theta^h}[\log p_{\theta^h}(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}] \tag{2} \\
&= \mathbb{E}_{\theta^h}\left[\log \frac{p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z})}{p_{\theta^h}(\mathbf{Y}, \mathbf{Z})}|\mathbf{Y}\right] \tag{3} \\
&\leq \log \mathbb{E}_{\theta^h}\left[\frac{p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z})}{p_{\theta^h}(\mathbf{Y}, \mathbf{Z})}|\mathbf{Y}\right] \tag{4}
\end{align}
$$

by Jensen's inequality.

Proof ii

We further develop $\log \mathbb{E}_{\theta^h} \left[p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z}) / p_{\theta^h}(\mathbf{Y}, \mathbf{Z}) \,|\mathbf{Y}\right]$ as

$$
\begin{aligned}
\log \int \frac{p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z})}{p_{\theta^h}(\mathbf{Y}, \mathbf{Z})} p_{\theta^h}(\mathbf{Z}|\mathbf{Y}) \, d\mathbf{Z} &= \log \int \frac{p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z})}{p_{\theta^h}(\mathbf{Y}, \mathbf{Z})} \frac{p_{\theta^h}(\mathbf{Y}, \mathbf{Z})}{p_{\theta^h}(\mathbf{Y})} \, d\mathbf{Z} & (5) \\
&= \log \left[\frac{1}{p_{\theta^h}(\mathbf{Y})} \int p_{\theta^{h+1}}(\mathbf{Y}, \mathbf{Z}) \, d\mathbf{Z} \right] & (6) \\
&= \log \left[\frac{p_{\theta^{h+1}}(\mathbf{Y})}{p_{\theta^h}(\mathbf{Y})} \right] & (7)
\end{aligned}
$$

Finally :

$$
\log \left[\frac{p_{\theta^{h+1}}(\mathbf{Y})}{p_{\theta^h}(\mathbf{Y})} \right] \geq 0
$$

Convergence

There is no general guaranty about the convergence of the EM algorithm towards the MLE $\widehat{\theta}$. The main property is that the observed likelihood increases at each iteration step.

Although, in practice : very sensible to the initialisation point.
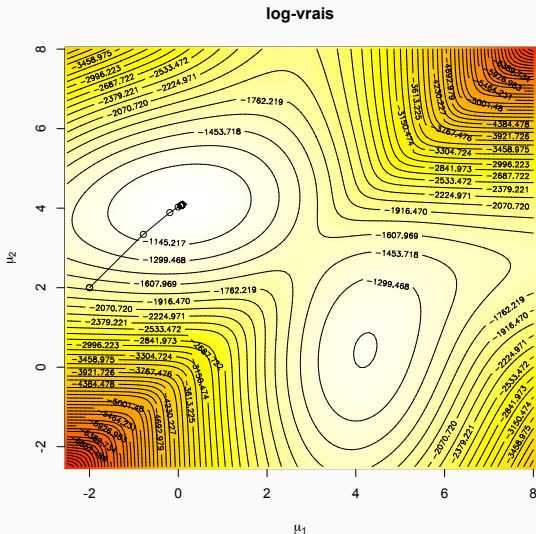
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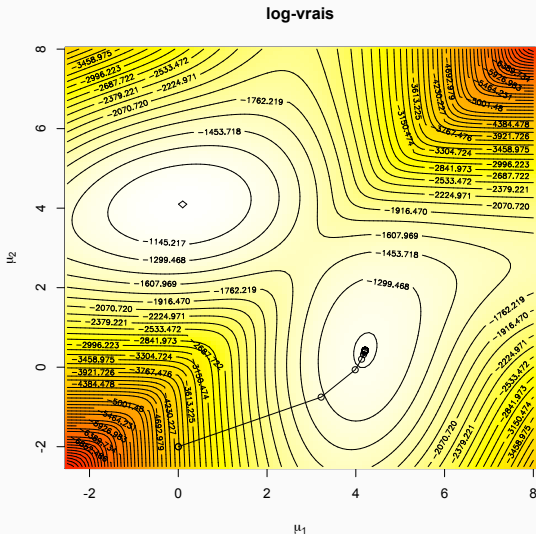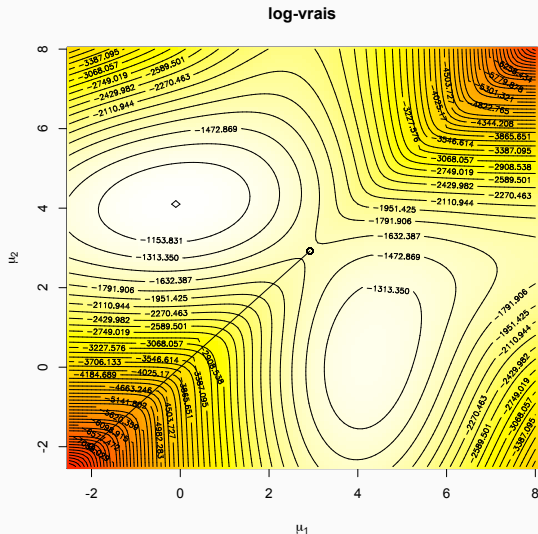
Application for the mixture model : E step

E-step is straightforward for independent mixture models.

Proposition

In a mixture model (1)*, the hidden states Z_i are independent conditional on the observations:*

$$p_\theta(\mathbf{Z}|\mathbf{Y}) = \prod_{i=1}^{n} p_\theta(Z_i|Y_i)$$

and, denoting $Z_{ik} = \mathbf{1}_{\{Z_i=k\}}$, the conditional distribution of each Z_i is given by

$$\tau_{ik} := P_\theta(Z_i = k|Y_i) = \mathbb{E}_\theta(Z_{ik}|Y_i) = \frac{\pi_k f_k(Y_i)}{\sum_{\ell=1}^{K} \pi_\ell f_\ell(Y_i)}.$$

Proof i

- First result is a direct consequence of Slide 28
- Second result follows from the Bayes formula

$$
\begin{aligned}
\tau_{ik} &= P_\theta(Z_i = k | Y_i) = \frac{P_\theta(Z_i = k) p_\theta(Y_i | Z_i = k)}{p_\theta(Y_i)} \\
&= \frac{P_\theta(Z_i = k) p_\theta(Y_i | Z_i = k)}{\sum_\ell P_\theta(Z_i = \ell) p_\theta(Y_i | Z_i = \ell)}.
\end{aligned}
$$

- $P_\theta(Z_i = k | Y_i) = \mathbb{E}_\theta(Z_{ik} | Y_i)$ because Z_{ik} is binary.

Proof ii

The update formula's of the τ_{ik} at the $(h+1)$-th E-step is then

$$\tau_{ik}^{h+1} = \frac{\pi_k^h f(Y_i; \gamma_k^h)}{\sum_\ell \pi_\ell^h f(Y_i; \gamma_\ell^h)}$$

where θ^h stands for the current estimate of θ resulting from the h-th M step.

Remark

Conditional probability τ_{ik} is sometimes referred to as the **posterior probability** for observation i to belong to class k (as opposed to the **prior probability** π_k).

Again this phrase is misleading in a non-Bayesian context and 'conditional probability' should be preferred.

M-step for the mixture model

$$\theta^{h+1} = \arg\max_\theta \mathbb{E}_{\theta^h}[\log p_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}]$$

We use Proposition 33 to get an explicit formula for this quantity

$$
\begin{aligned}
\mathbb{E}_{\theta^h}[\log p_\theta(Y, Z)|Y] &= \mathbb{E}_{\theta^h}\left[\sum_{i=1}^{n}\sum_{k=1}^{K} Z_{ik}[\log \pi_k + \log f(Y_k; \gamma_k)]|\mathbf{Y}\right] \\
&= \sum_{i=1}^{n}\sum_{k=1}^{K} \mathbb{E}_{\theta^h}(Z_{ik}|Y_i)[\log \pi_k + \log f(Y_k; \gamma_k)] \\
&= \sum_{i=1}^{n}\sum_{k=1}^{K} \tau_{ik}^{h}[\log \pi_k + \log f(Y_k; \gamma_k)].
\end{aligned}
$$

Application for the mixture model : M step

- Has to be maximized with respect to $\theta = (\boldsymbol{\pi}, \boldsymbol{\gamma})$, the τ_{ik} being fi

- For the proportions, one straightforwardly get

$$\pi_k^{h+1} = \frac{1}{n} \sum_{i=1}^{n} \tau_{ik}^h.$$

- For $\boldsymbol{\gamma}$: solution of this optimization problem has no general form as it strongly depends on the model at hand

- Some general formula can be derived in the case of the exponential family, as we will see in Section 3

About the entropy

$$\mathcal{H}[p_\theta(\mathbf{Z}|\mathbf{Y})] = -\mathbb{E}_\theta[\log p_\theta(\mathbf{Z}|\mathbf{Y})|\mathbf{Y}]$$

Can be calculated using the conditional independence of the Z_i given the data $\mathbf{Y}$:

$$
\begin{aligned}
\mathcal{H}[p_\theta(\mathbf{Z}|\mathbf{Y})] &= \sum_{i=1}^{n} H[p_\theta(Z_i|Y_i)] \\
&= -\sum_{i=1}^{n} \mathbb{E}_\theta[\log P(Z_i = k|Y_i)|Y_i] \qquad (8) \\
&= -\sum_{i=1}^{n} \sum_{k=1}^{K} \tau_{ik} \log \tau_{ik}.
\end{aligned}
$$

Definition ((Exponential family of distribution))

The distribution p belongs to exponential family with *canonical parameter* θ if

$$p_\theta(y) = \exp[\theta^\mathsf{T} t(y) - a(y) - b(\theta)]$$

where $t(y)$ is the vector of the *sufficient statistics*.

Maximum likelihood for the exponential family

Two general properties that show connections between maximum likelihood estimates and moment estimates for this class of distribution.

Proposition

$b'(\theta) = \mathbb{E}_\theta[t(Y)]$.

Proposition

For an iid sample $(Y_1, \ldots Y_n)$, the MLE $\widehat{\theta}$ of θ satisfies

$$b'(\widehat{\theta}) = \frac{1}{n} \sum_{i=1}^{n} t(Y_i) =: \overline{t}(Y).$$

This shows that the MLE $\widehat{\theta}$ is also the moment estimate of θ based on the mean of the sufficient statistics.

Proof in appendix slides 78 and 80.

EM for the exponential family

Proposition

If all emission distributions $\mathcal{F}_k$ belong to the exponential family with respective sufficient statistics t_k and normalizing functions a_k and b_k, the maximization in the M step results in the weighted moment estimates based on the expectation of the sufficient statistics, i.e. γ_k^{h+1} satisfies:

$$\mathbb{E}_{\gamma_k^{h+1}}[t_k(U)] = \frac{T_k^{h+1}}{N_k^{h+1}}$$

where

- $U \sim f(\cdot, \gamma_k^{h+1})$,
- $\tau_{ik}^{h+1} = \mathbb{E}_{\theta^{h+1}}(Z_{ik}|Y_i)$,
- $N_k^{h+1} = \sum_{i=1}^n \tau_{ik}^{h+1}$
- *and* $T_k^{h+1} = \sum_{i=1}^n \tau_{ik}^{h+1} t_k(Y_i)$.

Proof i

Complete-likelihood for exponential family

$$
\begin{aligned}
\log p_\theta(Y, Z) &= \sum_{i=1}^{n} \sum_{k=1}^{K} Z_{ik}[\log \pi_k + \log f_k(Y_i)] \\
&= \sum_{i=1}^{n} \sum_{k=1}^{K} Z_{ik}[\log \pi_k + \gamma_k^\mathsf{T} t_k(Y_i) - a_k(Y_i) - b_k(\gamma_k)]
\end{aligned}
$$

So conditional expectation is

$$
\begin{aligned}
&\mathbb{E}[\log p_\theta(Y, Z)|Y] = \\
&= \mathbb{E}\left[\sum_{i=1}^{n} \sum_{k=1}^{K} Z_{ik}\left[\log \pi_k - b_k(\gamma_k)\right]|Y\right] + \mathbb{E}\left[\sum_{i=1}^{n} \sum_{k=1}^{K} Z_{ik}\left[\gamma_k^\mathsf{T} t_k(Y_i) - a_k(Y_i)\right]|Y\right] \\
&= \sum_{k=1}^{K} N_k[\log \pi_k - b_k(\gamma_k)] + \sum_{k=1}^{K} \gamma_k^\mathsf{T} T_k - \sum_{i=1}^{n} \tau_{ik} a_k(Y_i).
\end{aligned}
$$

The derivative with respect to γ_k is null iff $b'_k(\gamma_k) = T_k/N_k$ and the result follows from the general properties of the exponential family given in Propositions slide 54.

- $\frac{T_k^{h+1}}{N_k^{h+1}}$ is an empirical weighted moment of the Y_i
- So the estimate of γ_k resulting from Proposition slide 54 is a moment-type estimate
- Depending on the form of $\mathbb{E}_{\gamma_k}[t_k(U)]$ as a function of γ_k, this estimate can have a close form or not

Expression for some popular models

- Poisson mixture: $\mathcal{F}_k = \mathcal{P}(\gamma_k)$:

$$\widehat{\gamma}_k = \frac{1}{N_k} \sum_{i=1}^{n} \tau_{ik} Y_i.$$

- Gaussian mixture: $\mathcal{F}_k = \mathcal{N}(\mu_k, \sigma_k^2)$:

$$\widehat{\mu}_k = \frac{1}{N_k} \sum_{i=1}^{n} \tau_{ik} Y_i, \qquad \widehat{\sigma}_k^2 = \frac{1}{N_k} \sum_{i=1}^{n} \tau_{ik} (Y_i - \widehat{\mu}_k)^2.$$

- Multinomial mixture: $\mathcal{F}_k = \mathcal{M}(1; \gamma_k)$, denoting $Y_{ia} = \mathbf{1}_{\{Y_i = a\}}$:

$$\widehat{\gamma}_{ka} = \frac{1}{N_k} \sum_{i=1}^{n} \tau_{ik} Y_{ia}.$$

Fisher information and asymptotic variance of the ML

Asymptotic variance of the maximum likelihood estimate

$$\widehat{\theta} = (\widehat{\pi}, \widehat{\gamma})$$

is provided by the Fisher information matrix I by

$$\mathbb{V}_{\infty}(\widehat{\theta}) = I_{\theta}^{-1}$$

where

$$
\begin{aligned}
S_{\theta}(\mathbf{Y}) &= \partial_{\theta} \log p_{\theta}(\mathbf{Y}) \\
I_{\theta} &= \mathbb{E}[S_{\theta}(\mathbf{Y}) S_{\theta}(\mathbf{Y})^{\mathsf{T}}] = -\mathbb{E}_{\mathbf{Y}}\left[\partial_{\theta^2}^2 \log p_{\theta}(\mathbf{Y})\right].
\end{aligned}
$$

Problem: Evaluation of $S'_{\theta}(\mathbf{Y}) = \partial_{\theta^2}^2 \log p_{\theta}(\mathbf{Y})$ because $p_{\theta}(\mathbf{Y})$ is a sum.

Louis's formula

[Louis, 1982] provides a convenient way to compute the Hessian matrix

$$S'_\theta(\mathbf{Y}) = \partial^2_{\theta^2} \log p_\theta(\mathbf{Y}),$$

which only uses by-products of the EM algorithm.

Proposition ([Louis, 1982])

$$
\begin{aligned}
S'_\theta(\mathbf{Y}) &= \mathbb{E}[S'_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}] + \mathbb{E}\left[S_\theta(\mathbf{Y}, \mathbf{Z})S_\theta(\mathbf{Y}, \mathbf{Z})^\intercal|\mathbf{Y}\right] \\
&\quad - \mathbb{E}[S_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}]\mathbb{E}[S_\theta(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}]^\intercal.
\end{aligned}
$$

Proof is given in Appendix on Slide 81.

Two main interests:

- Involve the complete likelihood and can, most of the times, be easily computed (see example in Apppendix Slide 85)
- Last term null when $\theta = \widehat{\theta} = \arg\max \log p_\theta(\mathbf{Y})$ since $p'_\theta(\mathbf{Y})|_{\widehat{\theta}} = 0$.

How many states?

- K is not known general
- A model with $K-1$ classes is nested in a model with K classes : the likelihood increases as well
- Likelihood not a relevant criterion to estimate K
- Dimension of the parameter θ increases with K.

Penalized likelihood criteria

Penalized likelihood criterion

- Let $\widehat{\theta}_K$ be the maximum likelihood estimate of θ for a model with K components:

$$\widehat{\theta}_K = \arg \max_{\theta \in \Theta_K} \log p_\theta(\mathbf{Y})$$

where Θ_K : parameter space for a K-mixture model

- Penalized likelihood estimate of K:

$$\widehat{K} = \arg \max_K \left(\log p_{\widehat{\theta}_K}(Y) - \text{pen}(K) \right).$$

Bayesian information criterion

- Most commonly used criterion [Schwarz, 1978]
- Originally defined in a Bayesian framework

Three levels of hierarchy:

1. a prior distribution $p(K)$ for the number of components;
2. a conditional distribution $p(\theta|K)$ for the parameter θ given the number of components;
3. a likelihood $p_\theta(\mathbf{Y})$ which corresponds to the conditional distribution of the observations $\mathbf{Y}$ given the parameters: $p_\theta(\mathbf{Y}) = p(\mathbf{Y}|\theta, K)$.

Posterior probability of K

- Model selection problem relies on conditional distribution of K given the observations:

$$p(K|\mathbf{Y}) = \frac{p(\mathbf{Y}, K)}{p(\mathbf{Y})} = \frac{p(K)p(\mathbf{Y}|K)}{p(\mathbf{Y})}.$$

- Ideally, one would choose

$$\widehat{K} = \arg\max_K p(K|\mathbf{Y}) = \arg\max_K (\log p(K) + \log p(\mathbf{Y}|K))$$

- But $\log p(Y|K) = \log \int p(\mathbf{Y}|\theta, K)p(\theta|K) \, \mathrm{d}\theta$
  - Difficult to evaluate
  - Laplace approximation

Laplace approximation

Proposition (Laplace approximation)

Under regularity conditions,

$$\log p(\mathbf{Y}|K) = \log p_{\widehat{\theta}_K}(\mathbf{Y}) - \frac{d_K}{2} \log n + \mathcal{O}(1).$$

where d_K denotes the number of independent parameters in a model with K components.

- Detailed proof: [Lebarbier and Mary-Huard, 2004], together with precise comparative study between BIC and another popular model selection criterion: AIC.
- The term $\log p(K)$ remains fix when n grows large: neglected

BIC

Definition

$$\widehat{K}_{BIC} = \arg \max_K \left(\log p_{\widehat{\theta}_K}(\mathbf{Y}) - \frac{d_K}{2} \log n \right).$$

From BIC to Integrated Complete Likelihood (ICL)

Using Proposition 35

$$\log p_{\widehat{\theta}_K}(\mathbf{Y}) = \mathbb{E}_{\widehat{\theta}_K}\left[\log p_{\widehat{\theta}_K}(\mathbf{Y}, \mathbf{Z})|\mathbf{Y}\right] - \underbrace{\mathbb{E}_{\widehat{\theta}_K}\left[\log p_{\widehat{\theta}_K}(\mathbf{Z}|\mathbf{Y})|\mathbf{Y}\right]}_{(1)}$$

- (1): entropy of the classification distribution
- Entropy is small when observation are classified with reasonable confidence.
- [Biernacki et al., 2000]: account for the classification uncertainty in the selection of K
- Penalize value of K with large entropy

ICL

Definition (ICL)

$$
\begin{aligned}
\widehat{K}_{ICL} &= \arg\max_K \left(\log p_{\widehat{\theta}_K}(Y) - \mathcal{H}[p_{\widehat{\theta}_K}(\mathbf{Z}|\mathbf{Y})] - \frac{d_K}{2} \log n \right) \\
&= \arg\max_K \left(\mathbb{E}_{\widehat{\theta}_K} \left[\log p_{\widehat{\theta}_K}(\mathbf{Y}, \mathbf{Z})|\mathbf{Y} \right] - \frac{d_K}{2} \log n \right)
\end{aligned}
$$

Illustration

Unsupervised classification

- Often the main aim when using a mixture model.
- Maximum likelihood inference provides estimates of θ
- By-product of EM : conditional distribution of the hidden classes Z conditional to the observed data Y

Soft classification

$$\tau_{ik} = P_{\widehat{\theta}}(Z_i = k | \mathbf{Y})$$

- Gives a measure of the confidence with which an observation could be classified into a given group
- Uncertainty of the classification summarized by:

$$\mathcal{H}[p_{\widehat{\theta}}(Z_i | Y)] = \mathcal{H}[p_{\widehat{\theta}}(Z_i | Y_i)] = -\sum_{k=1}^{K} \tau_{ik} \log \tau_{ik}.$$

  Sometimes referred to as the *classification uncertainty*

- Entropy of the whole conditional distribution of Z given Y: sum of all the individual's uncertainties

Hard classification

When observations need to be classified into groups, the most common rule is the 'maximum a posteriori' (MAP) rule.

Definition

The MAP classification rule is given by:

$$\widehat{\mathbf{Z}} = \arg\max_z p_\theta(\mathbf{Z} = z | \mathbf{Y}).$$

- The MAP rule can be applied to each observation label Z_i as

$$\widehat{Z}_i = \arg\max_k \tau_{ik}$$

- In the case of mixture, equivalent:

$$\widehat{Z} = \arg\max_z p_\theta(Z = z | Y) = (\widehat{Z}_i)_i$$

since the Z_i are independent conditionally on Y.

Conclusion

- Idea really simple.
- Example of R package : mixtools
- Used in many context, even for complexe data. The emission distribution has to be adapted.
- Next chapter : Hidden Markov Models

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Proposition
$b'(\theta) = \mathbb{E}_\theta[t(Y)]$.

Remind that the moment generating function of V

$$m(z) = \mathbb{E}[e^{z^\intercal V}]$$

with $m'(0) = \mathbb{E}(V)$

For the exponential family, consider the moment generating function of the sufficient statistics

$$
\begin{aligned}
m(z) &:= \mathbb{E}[e^{z^\intercal t(Y)}] = \int e^{z^\intercal t(y)} p_\theta(y) \, \mathrm{d}y \\
&= \int \exp[(z + \theta)^\intercal t(y) - a(y) - b(\theta)] \, \mathrm{d}y.
\end{aligned}
$$

Appendix. Properties of the exponential family ii

Because p_θ is a pdf, $e^{b(\theta)}$ is a normalizing constant:

$$\int \exp[\theta^\mathsf{T} t(y) - a(y)] \, \mathrm{d}y = e^{b(\theta)}$$

$$\Rightarrow \qquad \int \exp[(z+\theta)^\mathsf{T} t(y) - a(y)] \, \mathrm{d}y = e^{b(z+\theta)}$$

so

$$m(z) = e^{-b(\theta)} \int \exp[(z+\theta)^\mathsf{T} t(y) - a(y)] \, \mathrm{d}y = e^{b(z+\theta) - b(\theta)}.$$

The result follows from the fact that $m'(z) = b'(\theta + z) \Rightarrow m'(0) = b'(\theta)$.

Proposition

For an iid sample $(Y_1, \ldots Y_n)$, the MLE $\widehat{\theta}$ of θ satisfies

$$b'(\widehat{\theta}) = \frac{1}{n} \sum_{i=1}^{n} t(Y_i) =: \overline{t}(Y).$$

Take the derivative of the log-likelihood

$$\sum_i \log p(Y_i; \theta) = \sum_i [\theta^\mathsf{T} t(Y_i) - a(Y_i)] - nb(\theta)$$

with respect to θ.

Proposition ([Louis, 1982])

$$
\begin{aligned}
S'_\theta(Y) &= \mathbb{E}[S'_\theta(Y,Z)|Y] + \mathbb{E}\left[S_\theta(Y,Z)S_\theta(Y,Z)^\intercal|Y\right] \\
&\quad - \mathbb{E}[S_\theta(Y,Z)|Y]\mathbb{E}[S_\theta(Y,Z)|Y]^\intercal.
\end{aligned}
$$

Demonstration

Recalling that

$$
\log p_\theta(Y) = \log\left[\sum_z p_\theta(Y,z)\right],
$$

Appendix. Asymptotic variance ii

we have

$$
\begin{aligned}
S_\theta(Y) &= p'_\theta(Y)/p_\theta(Y) = \sum_z p'_\theta(Y,z)/p_\theta(Y) \\
&= \sum_z \frac{p'_\theta(Y,z)}{p_\theta(Y,z)} p_\theta(Y,z)/p_\theta(Y) = \sum_z \frac{p'_\theta(Y,z)}{p_\theta(Y,z)} p_\theta(z|Y) \\
&= \mathbb{E}\left[\frac{\partial}{\partial\theta}\log p_\theta(Y,z)\right] = \mathbb{E}[S_\theta(Y,Z)|Y].
\end{aligned}
\tag{9}
$$

Because the second derivative of $\log f$ is

$$
(\log f)'' = \frac{f''}{f} - \left(\frac{f'}{f}\right)\left(\frac{f'}{f}\right)^{\mathsf{T}}
\tag{10}
$$

Appendix. Asymptotic variance iii

the second derivative of $\log p_\theta(Y)$ is

$$
\begin{aligned}
S'_\theta(Y) &= \frac{\partial^2}{\partial \theta^2} \log \sum_z p_\theta(Y, z) \\
&= \frac{\sum_z p''_\theta(Y, z)}{p_\theta(Y)} - \left[\frac{\sum_z p'_\theta(Y, z)}{\sum_z p_\theta(Y, z)} \right] \left[\frac{\sum_z p'_\theta(Y, z)}{\sum_z p_\theta(Y, z)} \right]^\mathsf{T} \\
&= \frac{\sum_z p''_\theta(Y, z)}{p_\theta(Y)} - \mathbb{E}[S_\theta(Y, Z)|Y]\mathbb{E}[S_\theta(Y, Z)|Y]^\mathsf{T}.
\end{aligned}
$$

Appendix. Asymptotic variance iv

The same trick as in (9) can be combines with (10) for the first term to get

$$
\begin{aligned}
\frac{\sum_z p_\theta''(Y,z)}{p_\theta(Y)} &= \sum_z \left[\frac{p_\theta''(Y,z)}{p_\theta(Y,z)} - \left(\frac{p_\theta'(Y,z)}{p_\theta(Y,z)} \right) \left(\frac{p_\theta''(Y,z)}{p_\theta(Y,z)} \right)^\mathsf{T} + \left(\frac{p_\theta'(Y,z)}{p_\theta(Y,z)} \right) \left(\frac{p_\theta'(Y,z)}{p_\theta(Y,z)} \right)^\mathsf{T} \right] \\
&\quad \times \frac{p_\theta(Y,z)}{p_\theta(Y)} \\
&= \sum_z \left[\frac{\partial^2}{\partial \theta^2} \log p_\theta(Y,z) + \left(\frac{p_\theta'(Y,z)}{p_\theta(Y,z)} \right) \left(\frac{p_\theta'(Y,z)}{p_\theta(Y,z)} \right)^\mathsf{T} \right] p_\theta(z|Y) \\
&= \mathbb{E}[S_\theta'(Y,Z)|Y] + \mathbb{E} \left[S_\theta(Y,Z) S_\theta(Y,Z)^\mathsf{T} | Y \right]
\end{aligned}
$$

which completes the proof.

Appendix 2: Assymptotic variance for the Poisson emission distribution i

- Mixture model (1) where $F(\gamma_k) = \mathcal{P}(\gamma_k)$.
- Complete log-likelihood

$$\log p_\theta(Y, Z) = \sum_{i,k} Z_{ik} \left[\log \pi_k - \gamma_k + Y_i \log \gamma_k - \log(Y_i!)\right]$$

  where $\pi_K = 1 - \sum_{k<K} \pi_k$.

- First derivatives
  - $\partial_{\pi_k} \log p_\theta(Y, Z) = \frac{\sum_{i=1}^n Z_{ik}}{\pi_k} - \frac{\sum_{i=1}^n Z_{iK}}{\pi_K}$
  - $\partial_{\gamma_k} \log p_\theta(Y, Z) = -\sum_{i=1}^n Z_{ik} + \frac{\sum_{i=1}^n Z_{ik} Y_i}{\gamma_k}$

Appendix 2: Assymptotic variance for the Poisson emission distribution ii

- Second derivatives:
  - $\partial^2_{\pi_k^2} \log p_\theta(Y, Z) = -\frac{\sum_{i=1}^n Z_{ik}}{\pi_k^2} + \frac{\sum_{i=1}^n Z_{iK}}{\pi_K^2}$,
  - $\partial^2_{\pi_k, \pi_\ell} \log p_\theta(Y, Z) = \frac{\sum_{i=1}^n Z_{iK}}{\pi_K^2}$
  - $\partial^2_{\gamma_k^2} \log p_\theta(Y, Z) = -\frac{\sum_{i=1}^n Z_{ik} Y_i}{\gamma_k^2}$,
  - $\partial^2_{\gamma_k, \gamma_\ell} \log p_\theta(Y, Z) = 0$.

The first term of Prop slide 61 requires the calculation of the following moments, denoting here $\mathbb{E}^Y(\cdot) = \mathbb{E}(\cdot | Y)$:

$$
\begin{array}{rcl}
\mathbb{E}^Y\left(\sum_{i=1}^n Z_{ik}\right) & = & \sum_{i=1}^n \tau_{ik} =: N_k, \\
\mathbb{E}^Y\left(\sum_{i=1}^n Z_{ik} Y_i\right) & = & \sum_{i=1}^n \tau_{ik} Y_i =: S_k.
\end{array}
$$

The second term requires these of

Appendix 2: Assymptotic variance for the Poisson emission distribution iii

$$
\begin{aligned}
\mathbb{E}^Y\left[\left(\sum_{i=1}^n Z_{ik}\right)\left(\sum_{i=1}^n Z_{i\ell}\right)\right] &= \mathbb{E}^Y\left(\sum_{i=1}^n Z_{ik}Z_{i\ell} + \sum_{i\neq j} Z_{ik}Z_{j\ell}\right) \\
&= \sum_{i=1}^n \mathbb{E}^Y(Z_{ik}Z_{i\ell}) \\
&\quad + \sum_{i\neq j} \mathbb{E}^Y(Z_{ik})\mathbb{E}^Y(Z_{j\ell}) \\
&=^* \sum_{i=1}^n \mathbf{1}_{\{k=\ell\}}\tau_{ik} + \sum_{i\neq j}\tau_{ik}\tau_{j\ell} \\
&= \mathbf{1}_{\{k=\ell\}}N_k + N_k N_\ell - \sum_{i=1}^n \tau_{ik}\tau_{i\ell}, \\
\mathbb{E}\left[\left(\sum_{i=1}^n Z_{ik}Y_i\right)\left(\sum_{i=1}^n Z_{i\ell}\right)\right] &= \mathbf{1}_{\{k=\ell\}}S_k + S_k N_\ell - \sum_{i=1}^n Y_i \tau_{ik}\tau_{i\ell}, \\
\mathbb{E}^Y\left[\left(\sum_{i=1}^n Z_{ik}Y_i\right)\left(\sum_{i=1}^n Z_{i\ell}Y_i\right)\right] &= \mathbf{1}_{\{k=\ell\}}Q_k + S_k S_\ell - \sum_{i=1}^n Y_i^2 \tau_{ik}\tau_{i\ell},
\end{aligned}
$$

where $Q_k = \sum_{i=1}^n Y_i^2 \tau_{ik}$ and * because $Z_{ik}Z_{i\ell} = 0$ if $k \neq \ell$.