Detecting Outliers in Hidden Markov modeling through Relative Entropy: Applications to Change-Point Detection

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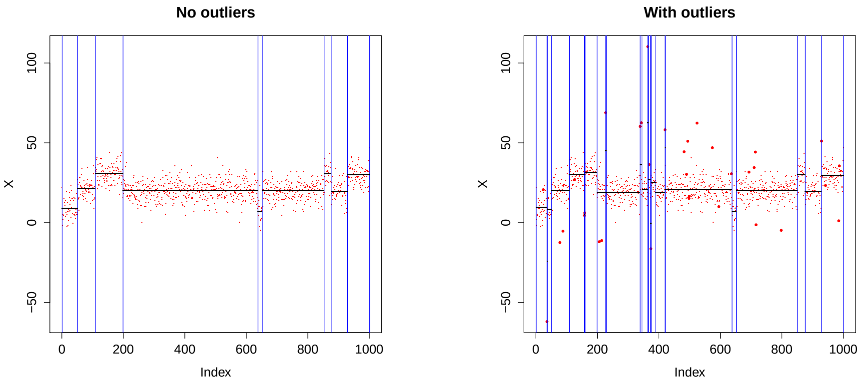
Change-point detection, HMMs and outliers

- Given an heterogeneous sequence: find the segments in which the signal is homogeneous
- Here: Hidden Markov modeling
- Segmentation models are sensitive to the presence of outliers



Hidden Markov Models

- X_i observed variable
- S_i hidden variable, for $i = 1 \dots, n$

$$\begin{tikzcd}
S_1 \arrow[r] \arrow[d] & S_2 \arrow[r] \arrow[d] & S_3 \arrow[r] \arrow[d] & S_4 \arrow[r] \arrow[d] & S_5 \arrow[d] \\
X_1 & X_2 & X_3 & X_4 & X_5
\end{tikzcd}$$

Factorization of the joint probability distribution

$$\mathbb{P}(S_{1:n} = s_{1:n}, X_{1:n} = x_{1:n}) = \mathbb{P}(S_1 = s_1) \prod_{i=2}^{n} \mathbb{P}(S_i = s_i | S_{i-1} = s_{i-1}) \prod_{i=1}^{n} \mathbb{P}(X_i = x_i | S_i = s_i)$$

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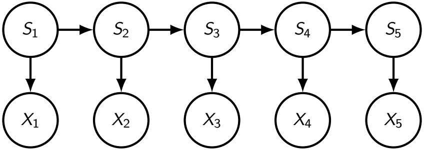

Factorization of the joint probability distribution

$$\mathbb{P}(S_{1:n}, X_{1:n}) = \mathbb{P}(S_1) \prod_{i=2}^{n} \mathbb{P}(S_i | S_{i-1}) \prod_{i=1}^{n} \mathbb{P}(X_i | S_i)$$

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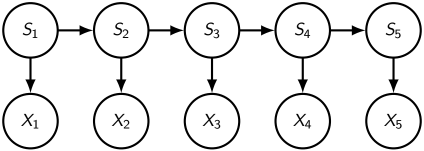

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Example (Application to change point detection)

- Level-based: S_i = underlying level of observation X_i
- Segment-based: S_i = segment of X_i with $S_1 = 1$ and $S_n = \#$ segments

Inference in HMMs

If $\mathcal{E} = \{X_{1:n} = x_{1:n}\}$ observed, compute $\mathbb{P}(\mathcal{E}), \mathbb{P}(S_i|\mathcal{E}), \mathbb{P}(S_i|S_{i-1}, \mathcal{E})$...

Backward and Forward recursions (Baum-Welch algorithm)

Standard inference problems solved by *combining* the Forward and Backward quantities

- $F_i(S_i) := \mathbb{P}(S_i, X_{1:i} = x_{1:i})$
- $B_i(S_i) := \mathbb{P}(X_{i+1:n} = x_{i+1:n}|S_i)$,

which are computed recursively:

- $F_i(S_i) = \sum_{S_{i-1}} F_{i-1}(S_{i-1})\mathbb{P}(S_i|S_{i-1})\mathbb{P}(X_i|S_i)$
- $B_{i-1}(S_{i-1}) = \sum_{S_i} \mathbb{P}(S_i|S_{i-1})\mathbb{P}(X_i|S_i)B_i(S_i)$.

E.g. $\mathbb{P}(S_i, \mathcal{E}) = F_i(S_i)B_i(S_i)$

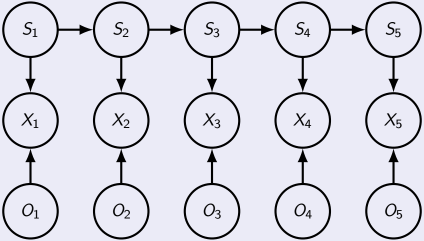
Parameter estimation

EM algorithm: Backward/Forward quantities provide explicit update formulas

Ad hoc model for outlier detection in HMMs

- X_i is an outlier if it is not generated by the underlying HMM
- $\Rightarrow$ extend the HMM with variables for the outliers status [Shah 2006]

Topology and conditional dependencies (homoscedastic Gaussian case)



- $O_i = 1$ iff X_i is outlier; $\mathbb{P}(O_i = 1) = \rho$
- $\mathbb{P}(S_1)$; $\mathbb{P}(S_i | S_{i-1})$: same as for underlying HMM
- $\mathbb{P}(X_i | S_i, O_i = 0) = \mathcal{N}(\mu_{S_i}, \sigma^2)$: same as for underlying HMM
- $\mathbb{P}(X_i | S_i, O_i = 1) = \mathcal{N}(\mu_{S_i}, \sigma^2) + \mathcal{N}(0, \delta^2)$

Inference in the ad hoc model

Inferring outlier posterior probabilities

$$\mathbb{P}(O_i = 1|\mathcal{E}) = \sum_{S_i} \left(\frac{\rho\mathbb{P}(X_i{=}x_i|S_i,O_i{=}1)}{\rho\mathbb{P}(X_i{=}x_i|S_i,O_i{=}1)+(1-\rho)\mathbb{P}(X_i{=}x_i|S_i,O_i{=}0)} \cdot \mathbb{P}(S_i|\mathcal{E}) \right)$$

where

$$\mathbb{P}(S_i|\mathcal{E}) = \frac{F_i(S_i)B_i(S_i)}{\sum_{S_i} F_i(S_i)B_i(S_i)}$$

$$F_i(S_i) = \sum_{S_{i-1}} F_{i-1}(S_i)\mathbb{P}(S_i|S_{i-1})\mathbb{P}(X_i|S_i)$$

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$$F_i(S_i) = \sum_{S_{i-1}} F_{i-1}(S_i)\mathbb{P}(S_i|S_{i-1}) \sum_{o=0,1} \mathbb{P}(X_i = x_i|S_i, O_i = o)\mathbb{P}(O_i = o)$$

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EM algorithm for the ad hoc model

Parameter updates: 2 new parameters (ρ, δ^2)

- Transition parameters have same update formulas as in plain HMM
- $\rho = \frac{\sum_{i=1}^{n} \mathbb{P}(O_i{=}1|\mathcal{E})}{n}$
- $\mu_s, \sigma^2, \delta^2$ found as fixed points:

$$\begin{cases} \mu_s & = \frac{\sum_i x_i[\mathbb{P}(S_i{=}s,O_i{=}1|\mathcal{E})\sigma^2+\mathbb{P}(S_i{=}s,O_i{=}0|\mathcal{E})(\sigma^2+\delta^2)]}{\sum_i[\mathbb{P}(S_i{=}s,O_i{=}1|\mathcal{E})\sigma^2+\mathbb{P}(S_i{=}s,O_i{=}0|\mathcal{E})(\sigma^2+\delta^2)]} \\ \sigma^2 & = \frac{\sum_i\sum_s(x_i-\mu_s)^2\mathbb{P}(S_i{=}s,O_i{=}0|\mathcal{E})}{\sum_i\sum_s\mathbb{P}(S_i{=}s,O_i{=}0|\mathcal{E})} \\ \sigma^2+\delta^2 & = \frac{\sum_i\sum_s(x_i-\mu_s)^2\mathbb{P}(S_i{=}s,O_i{=}1|\mathcal{E}))}{\sum_i\sum_s\mathbb{P}(S_i{=}s,O_i{=}1|\mathcal{E})} \end{cases}$$

where $\mathbb{P}(S_i, O_i|\mathcal{E}) = \frac{\rho\mathbb{P}(X_i{=}x_i|S_i,O_i{=}1)}{\rho\mathbb{P}(X_i{=}x_i|S_i,O_i{=}1)+(1-\rho)\mathbb{P}(X_i{=}x_i|S_i,O_i{=}0)} \cdot \mathbb{P}(S_i|\mathcal{E})$

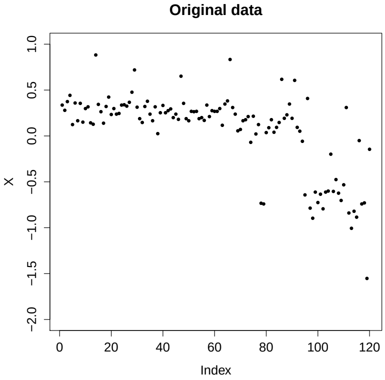
Initialization

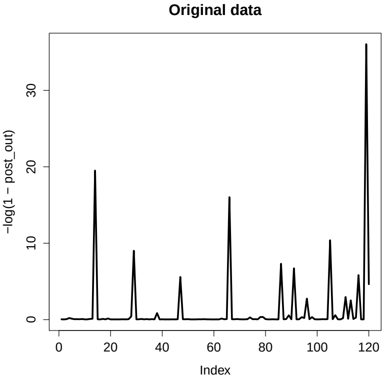
k-means algorithm and z-score

Application of the ad hoc model to real data

Ad hoc model performs well on data simulated by... ad hoc model. What about real data?

- CNV dataset from breast cancer cell line BT474 [Snijders 2001]
- Level-based model: S_i = level of observation X_i
- Parameters in the ad hoc model estimated with the EM algorithm





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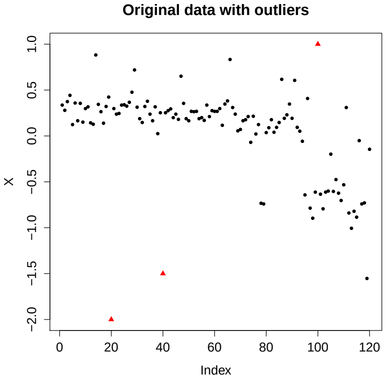

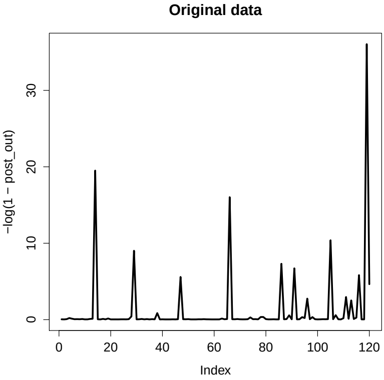

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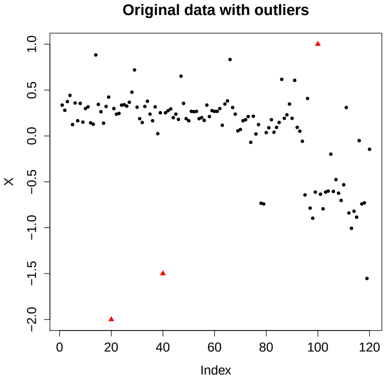
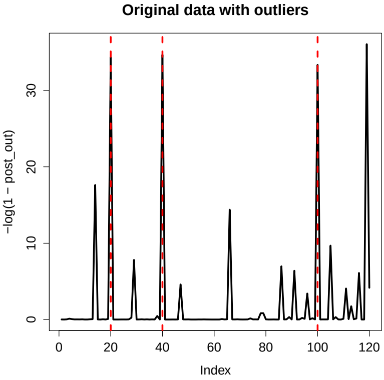




Outlier detection through relative entropy

Intuition

- If $X_i = x_i$ is an outlier than it must have a strong influence on $\mathbb{P}(S_{1:n}|\mathcal{E}) = \mathbb{P}(S_{1:n}|X_{1:n} = x_{1:n})$
- As a consequence, $\mathbb{P}(S_{1:n}|X_{1:n} = x_{1:n})$ must differ significantly from $\mathbb{P}(S_{1:n}|X_{-i} = x_{-i})$
- We can try to use the relative entropy

$$K_i := \sum_{S_{1:n}} \mathbb{P}(S_{1:n}|X_{-i} = x_{-i}) \log \frac{\mathbb{P}(S_{1:n}|X_{-i} = x_{-i})}{\mathbb{P}(S_{1:n}|X_{1:n} = x_{1:n})}$$

for outlier detection: *"the higher K_i the more likely $X_i = x_i$ is an outlier"*

- Technical problem: *how to compute K_i?*

Computing K_i

- By using naively back/forw recursions, the complexity for computing K_i for a given i is $O(n)$ $\Rightarrow$ the overall complexity is $O(n^2)$

Linear time algorithm for computing K_i for all $i = 1, \dots, n$

$$K_i = \sum_{S_i} \mathbb{P}(S_i | X_{-i} = x_{-i}) \log \frac{\mathbb{P}(S_i | X_{-i} = x_{-i})}{\mathbb{P}(S_i | X_{1:n} = x_{1:n})},$$

with

$$\mathbb{P}(S_i | X_{-i} = x_{-i}) = \frac{F_i^*(S_i) B_i(S_i)}{\sum_{S_{i-1}} F_i^*(S_{i-1}) B_i(S_{i-1})}$$

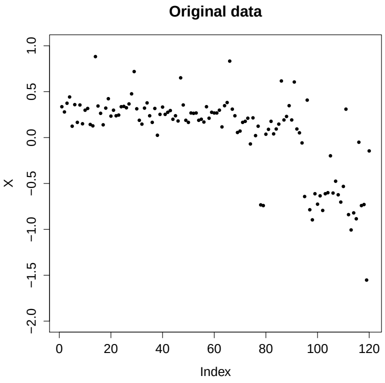
where B and F are the standard back/forw quantities for HMMs and

$$F_i^*(S_i) = \sum_{S_{i-1}} F_{i-1}(S_{i-1}) \mathbb{P}(S_i | S_{i-1}).$$

$\Rightarrow$ the overall complexity is $O(n)$

Application of the relative entropy based method to real data

- Same CNV dataset as before
- Parameters in the underlying HMM estimated with the EM algorithm



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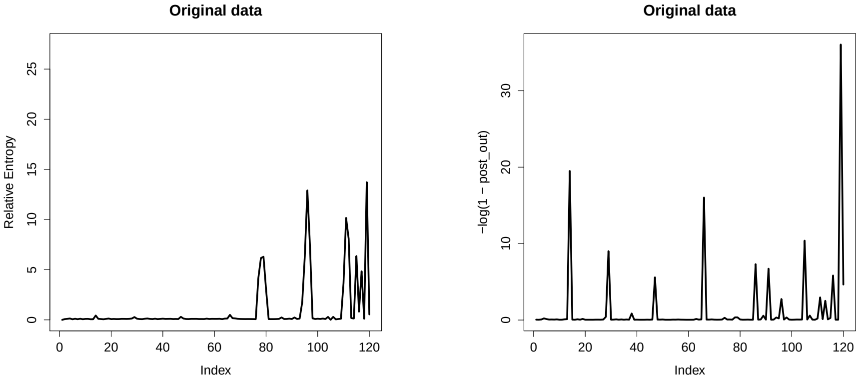

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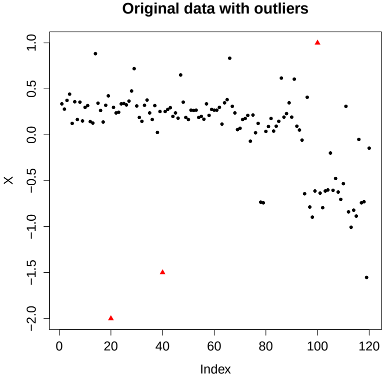

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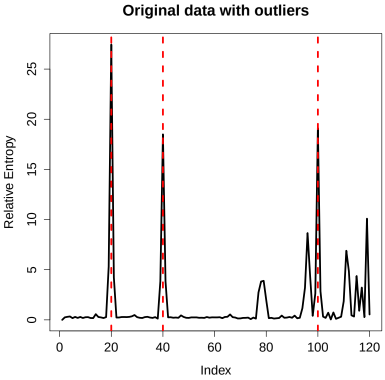
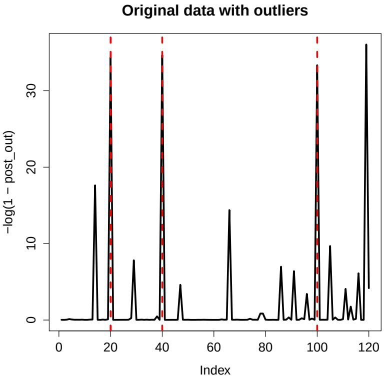
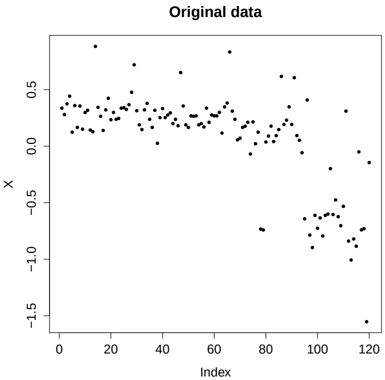




Comparison on real CNV dataset

- Original data: $n = 120$ observations
- H0: random samples of $n/2$ observations from the original dataset
- H1: $\rho \times n/2$ outliers added with $\mathcal{N}(0, \delta^2)$ $(\rho = 0.05, \delta = 6)$



- Parameters estimated
- Global statistics for ad hoc model: $T = \max_{i=1,\dots,n} \mathbb{P}(O_i = 1|\mathcal{E})$
- Global statistics for relative entropy method: $S = \max_{i=1,\dots,n} K_i$

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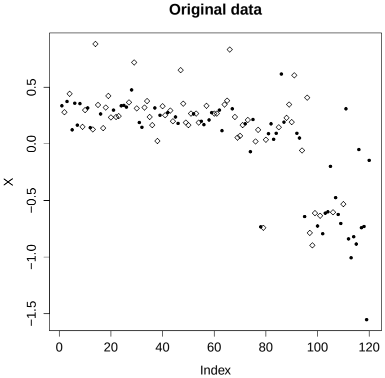

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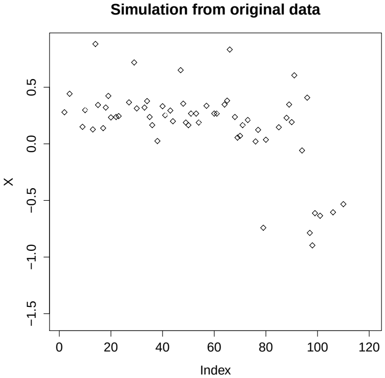

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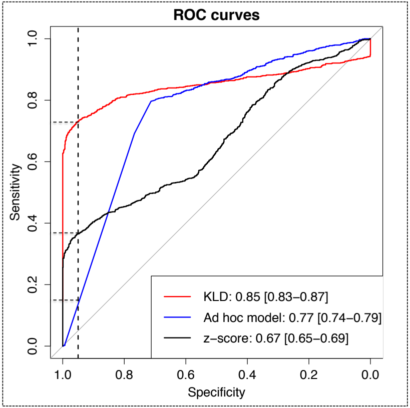
Results of comparison on real data



Discussion

- When data is not generated accordingly to the ad hoc model, the method based on relative entropy is more performant
- The method based on z-score is not satisfactory

Final word

Conclusions

- Ad hoc model:
  - + Outlier explicit modeling, convenient for simulating
  - − Intricate EM algorithm
  - − Very sensitive, false positives
- Method based on relative entropy:
  - + Model free
  - + Parameter estimation simple to implement and fast
  - + Robust

Perspectives

- Comparison with standard outliers detection methods (e.g. LOF)
- Local statistics for outlier detection based on relative entropy
- Application to biological data

References

J. Fridlyand et al. *Hidden Markov models approach to the analysis of array CGH data*; Journal of multivariate analysis, 2004.

A. Olshen et al. *Circular binary segmentation for the analysis of array-based DNA copy number data*; Biostatistics, 2004.

J. Bilmes. *A gentle tutorial of the EM algorithm and its application to parameter estimation for Gaussian mixture and hidden Markov models*; International Computer Science Institute, 1998.

S.S. Shah et al. *Integrating copy number polymorphisms into array CGH analysis using a robust HMM*; Bionformatics, 2006.

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Appendix

Initialization of the EM algorithm for the ad hoc model

Using z-score

- Cluster the observations with the k-means algorithm
- μ_{S_i} = mean of all the observations within the same cluster
- $\sigma^2 = \frac{\sum_{i=1}^{n}(X_i - \mu_{S_i})^2}{n}$
- Compute for each observation its z-score:

$$z_i = \frac{X_i - \mu_{S_i}}{\sigma} \sim \mathcal{N}(0,1)$$

- If $|z_i| > 1.96$, then $O_i = 1$
- Compute ρ and δ^2

Relative entropy

Definition

Given two probability distributions p and q their relative entropy (Kullback-Leibler divergence) is

$$D_{KL}(p||q) := \int_z p(z) \log \frac{p(z)}{q(z)} dz = \mathbb{E}_p[\log p] - \mathbb{E}_p[\log q]$$

Properties

- $D_{KL}(p||q)$ is a non-symmetric measure of the distance between p and q: it measures the extra number of bits required for encoding events sampled from p using a code based on q
- Monte Carlo estimation: if $x_1, \ldots, x_N$ is a sample of p, then

$$D_{KL}(p||q) \approx \frac{\sum_{j=1}^{N} \log p(x_j) - \log q(x_j)}{N}$$

- For common (e.g. normal) distributions: exact closed forms of D_{KL}

Parameters for simulations

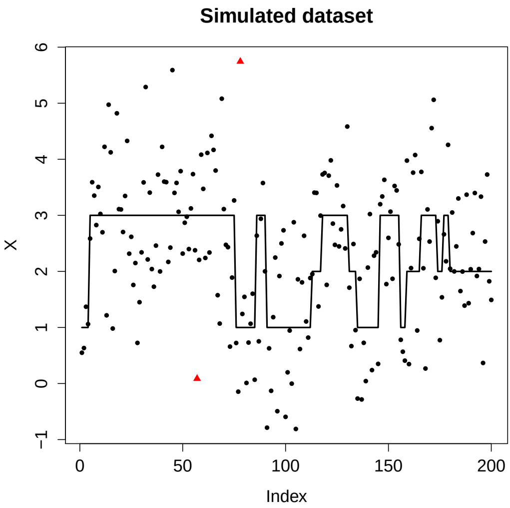
Homoscedastic Gaussian ad hoc model

- $\mathbb{P}(O_i = 1) = \rho$
- $\mathbb{P}(X_i | S_i, O_i = 0) = \mathcal{N}(\mu_{S_i}, \sigma^2)$
- $\mathbb{P}(X_i | S_i, O_i = 1) = \mathcal{N}(\mu_{S_i}, \sigma^2) + \mathcal{N}(0, \delta^2)$

Parameters

- $S_i \in \{1, 2, 3\}$
- $\mathbb{P}(S_1 = 1) = 1$
- $\mathbb{P}(S_i | S_{i-1}) = \begin{pmatrix} 1-\eta & \eta/2 & \eta/2 \\ \eta/2 & 1-\eta & \eta/2 \\ \eta/2 & \eta/2 & 1-\eta \end{pmatrix}$ with $\eta = 0.05$; i=2,...,n
- $\mu_1 = 1$, $\mu_2 = 2$, $\mu_3 = 3$
- $\sigma = 1$
- $\rho = 0.05$
- δ `in seq(from=0.00,to=4.50,by=0.50)`

Example of simulation under H1



Validation of the ad hoc model on a toy example

- Simulations done with the homoscedastic Gaussian ad hoc model:
  - H0: no outlier ($\delta = 0$)
  - H1: presence of outliers ($\delta \neq 0$)
- Global statistics: $T = \max_{i=1,\dots,n} \mathbb{P}(O_i = 1|\mathcal{E})$
- $\mathbb{P}(O_i|\mathcal{E})$ computed using the true parameters

δ	$AUC(T)$
0.00	0.52 [0.48,0.55]
0.50	0.49 [0.46,0.53]
1.00	0.56 [0.52,0.59]
1.50	0.74 [0.71,0.78]
2.00	0.87 [0.85,0.89]
2.50	0.93 [0.91,0.95]
3.00	0.97 [0.95,0.98]
3.50	0.99 [0.98,0.99]
4.00	0.99 [0.99,1.00]
4.50	0.99 [0.99,1.00]

Validation of the relative entropy method on the toy example and comparison

- Global statistics: $S = \max_{i=1,\ldots,n} K_i$

δ	$AUC(S)$	$AUC(T)$
0.00	0.50 [0.47,0.54]	0.52 [0.48,0.55]
0.50	0.50 [0.46,0.53]	0.49 [0.46,0.53]
1.00	0.52 [0.49,0.56]	0.56 [0.52,0.59]
1.50	0.55 [0.52,0.59]	0.74 [0.71,0.78]
2.00	0.69 [0.66,0.73]	0.87 [0.85,0.89]
2.50	0.76 [0.73,0.79]	0.93 [0.91,0.95]
3.00	0.84 [0.81,0.87]	0.97 [0.95,0.98]
3.50	0.87 [0.85,0.90]	0.99 [0.98,0.99]
4.00	0.92 [0.91,0.94]	0.99 [0.99,1.00]
4.50	0.96 [0.95,0.97]	0.99 [0.99,1.00]