A Bayesian nonparametric method for the LR assessment in case of rare type match

Giulia Cereda

October 8, 2015

Giulia Cereda ()

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• Crime case

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$$\frac{\Pr(H_p \mid D)}{\Pr(H_d \mid D)} = \underbrace{\frac{\Pr(D \mid H_p)}{\Pr(D \mid H_d)}}_{LR} \frac{\Pr(H_p)}{\Pr(H_d)}$$

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 - H_p : The suspect left the stain
 - H_d : Someone else left the stain
- Background (B): database of DNA profiles from the population of possible perpetrators

DNA profiles

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A DNA profile is a list of integers h = (4 - 5 - 2 - 10) that code some characteristics in some portions of the DNA sequence of an individual: different persons can share the same profile.

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For H_p the match is a sure event,

For H_d the match is a random event with probability p_h = frequency of the profile h of the suspect in the population of possible perpetrators.

DNA database

Database: a list of DNA profiles from a sample from the population of possible perpetrators

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DNA database

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```
DATABASE of size 10
Person 1 (4 - 10 - 6 - 7)
Person 2 (3 - 5 - 6 - 8)
Person 3 (3 - 7 - 8 - 10)
Person 4 (10 - 1 - 4 - 5)
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The database is used to find out the rarity of the matching profile.

• A match between the suspect's DNA profile and the crime stain's DNA profile.

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- This profile is not contained in the database B.

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Especially if the database is big, the profile seems to be rare. How rare?

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Frequentist model: (Cereda 2015) Frequentist approach to LR assessment in case of rare haplotype match arXiv:1502.04083

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 Bayesian model: (Cereda 2015) Full Bayesian approach to LR assessment in case of rare haplotype match *arXiv:1502.02406*

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- Bayesian model: (Cereda 2015) Full Bayesian approach to LR assessment in case of rare haplotype match arXiv:1502.02406
- (Cereda 2015) Nonparametric Bayesian approach to LR assessment in case of rare haplotype match *arXiv:1506.08444*

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Parameter: $\mathbf{p} = (p_t | t \in T)$, T an infinite countable set, $p_t > 0$, $\sum p_t = 1$, to represent the (unknown) frequencies of all DNA types in Nature.

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Assumption 2

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"DNA types" or "colors" is now the same.

Let [n] denote the set $[n] = \{1, 2, ..., n\}$.

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Assumption 2 \rightarrow data can be replaces by the equivalence classes on the indices of the relation "to have the same DNA type". This is a partition of the set [n] : {{1}, {2,9}, {3,5,6,8,10}, {4}, {7}}

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 $\mathcal{D} = \pi_{[n+2]}$ partition of the set $\{1, 2, ..., n+2\}$

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Example: Database $\rightarrow \pi_{[10]} = \{\{1\}, \{2, 9\}, \{3, 5, 6, 8, 10\}, \{4\}, \{7\}\}$

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Example: Database $\rightarrow \pi_{[10]} = \{\{1\}, \{2,9\}, \{3,5,6,8,10\}, \{4\}, \{7\}\}$ $\mathcal{D} \rightarrow \pi_{[12]} = \{\{1\}, \{2,9\}, \{3,5,6,8,10\}, \{4\}, \{7\}, \{11,12\}\}$

$$\mathcal{D} = \pi_{[n+2]}$$
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 $\mathcal{D} \rightarrow \pi_{[12]} = \{\{1\}, \{2, 9\}, \{3, 5, 6, 8, 10\}, \{4\}, \{7\}, \{11, 12\}\}$

We can see the data as a random variable. In that case,

$$\mathcal{D}=\Pi_{[n+2]}.$$

The distribution of $\mathcal{D} = \prod_{[n+2]}$ depends on **p**. However, it does not depend on the order of the p_i .

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We can consider directly the ordered vector $\mathbf{p} \in \nabla_{\infty} = \{(p_1, p_2, ...), p_1 \ge p_2 \ge ... > 0, \sum p_i = 1\}.$

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For instance, p_3 = the frequency of the third most frequent DNA type in Nature.

Bayesian nonparametrics: we need a prior for the parameter **p**.

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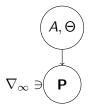
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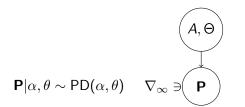
Parameters: $0 < \alpha < 1, \ \theta > -\alpha$

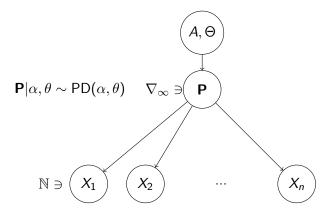
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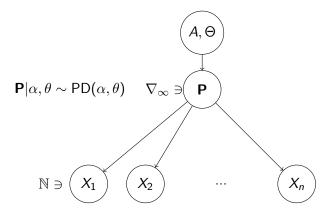






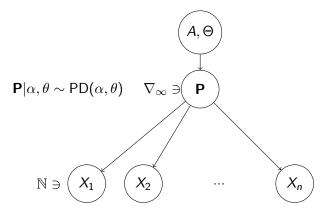
 $X_i = j \rightarrow$ the i-th observation has the *j*th most common type in Nature.

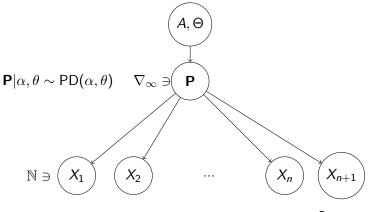
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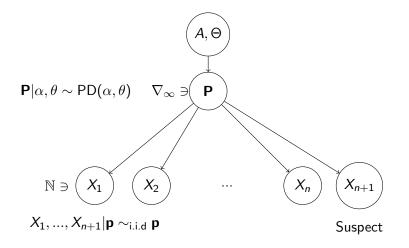
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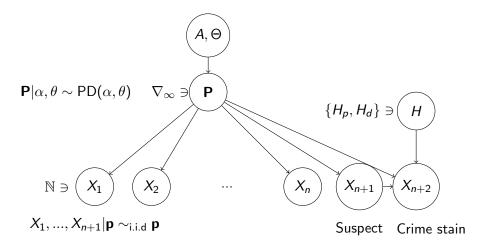
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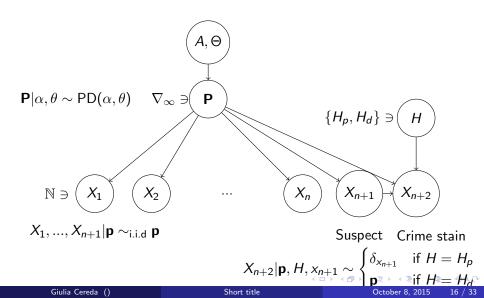


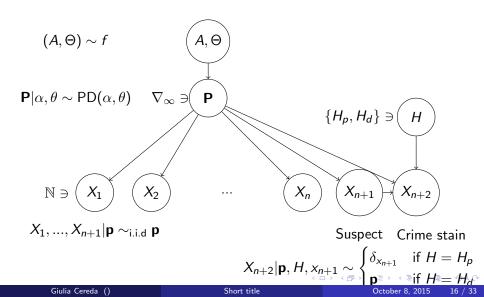


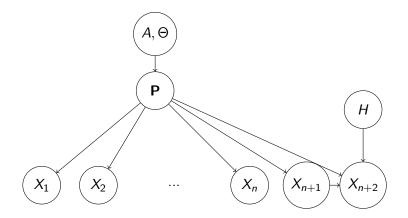
Suspect











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Some notation:

Given $X_1, ..., X_n \in \mathbb{N}$, random variables, $\prod_{[n]}(X_1, X_2, ..., X_n)$ is the random partition defined by the equivalence classes of $i \sim j$ iff $X_i = X_j$.

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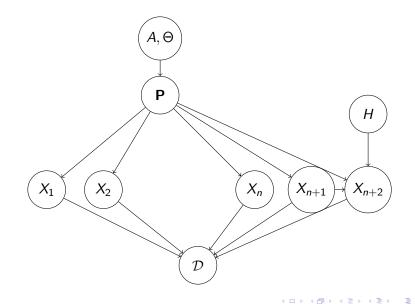
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$$\begin{array}{cccc} X_1, \dots, X_n & \longrightarrow & \Pi_{[n]} = \pi_{[n]}^{\mathrm{Db}} \\ X_1, \dots, X_n, X_{n+1} & \longrightarrow & \Pi_{[n+1]} = \pi_{[n+1]}^{\mathrm{Db}+} \\ X_1, \dots, X_n, X_{n+1}, X_{n+2} & \longrightarrow & \Pi_{[n+2]} = \pi_{[n+2]}^{\mathrm{Db}++} \end{array}$$

 $X_1, ..., X_n$ are not observed, but generates the same partition as the original database.

Data can be defined as $\mathcal{D} = \prod_{[n+2]}$.

The complete model



Pitman sampling formula

Image: A mathematical states and a mathem

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Pitman sampling formula

$$\mathbf{P} \sim \mathsf{PD}(lpha, heta)$$

 $X_1, X_2, ..., X_n | \mathbf{P} = \mathbf{p} \sim_{ ext{i.i.d}} \mathbf{p}$

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$$\Pr(\Pi_{[n]} = \pi_{[n]} | \alpha, \theta) = \mathbb{P}_{\alpha, \theta}^{n}(\pi_{[n]}) = \frac{[\theta + \alpha]_{k-1;\alpha}}{[\theta + 1]_{n-1;1}} \prod_{i=1}^{k} [1 - \alpha]_{n_i - 1;1},$$

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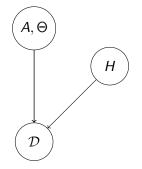
In our model

$$\Pr(D|\alpha,\theta,h) = \Pr(\Pi_{[n+2]} = \pi_{[n+2]}^{Db++} | \alpha,\theta,h) = \begin{cases} \mathbb{P}_{\alpha,\theta}^{n+2}(\pi_{[n+2]}^{Db++}) & \text{if } h = H_d \\ \mathbb{P}_{\alpha,\theta}^{n+1}(\pi_{[n+1]}^{Db+}) & \text{if } h = H_p \end{cases}$$

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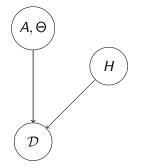
The model, simplified



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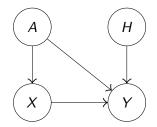
The model, simplified



 $\mathcal{D}=\Pi_{[n+2]}.$

Lemma

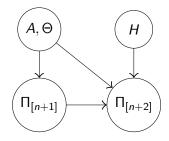
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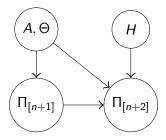
Lemma

Given four random variables A, H, X and Y, as above, the likelihood function for h, given X = x and Y = y, satisfies

$$lik(h \mid x, y) \propto \mathbb{E}(p(y \mid x, A, h) \mid X = x).$$



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 $lik(h \mid \pi_{[n+1]}, \pi_{[n+2]}) \propto \mathbb{E}(p(\pi_{[n+2]} \mid \pi_{[n+1]}, A, \Theta, h) \mid \Pi_{[n+1]} = \pi_{[n+1]}).$

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$$\mathsf{LR} = \frac{p(\pi_{[n+2]}|H_p)}{p(\pi_{[n+2]}|H_d)} = \frac{p(\pi_{[n+1]}, \pi_{[n+2]}|H_p)}{p(\pi_{[n+1]}, \pi_{[n+2]}|H_d)} = \frac{\mathsf{lik}(H_p|\pi_{[n+1]}, \pi_{[n+2]})}{\mathsf{lik}(H_d|\pi_{[n+1]}, \pi_{[n+2]})}$$

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Lemma allows to write

$$\mathsf{LR} = \frac{\mathbb{E}(\overbrace{p(\pi_{[n+2]} \mid \pi_{[n+1]}, A, \Theta, H_p)}^{1} \mid \Pi_{[n+1]} = \pi_{[n+1]})}{\mathbb{E}(\underbrace{p(\pi_{[n+2]} \mid \pi_{[n+1]}, A, \Theta, H_d)}_{\frac{1-A}{n+1+\Theta}} \mid \Pi_{[n+1]} = \pi_{[n+1]})}$$

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Image: A math and A

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$$\mathsf{LR} = \frac{1}{\mathbb{E}\left(\frac{1-A}{n+1+\Theta} \mid \mathsf{\Pi}_{[n+1]} = \pi_{[n+1]}\right)}$$

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We are interested in the distribution of $\Phi, \Theta | \Pi_{[n+1]}$

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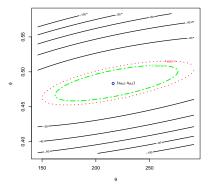
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Log likelihood with ϕ and heta

 $\log_{10} p(\pi_{[n+1]} \mid \phi, \theta)$

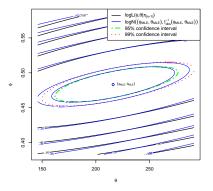


Dutch Y-STR database, 7 loci, N=18,925

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Log likelihood with ϕ and θ

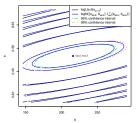
 $\log_{10} p(\pi_{[n+1]} \mid \phi, \theta)$



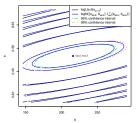
Dutch Y-STR database, 7 loci, N=18,925

Giulia Cereda ()

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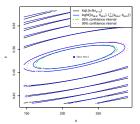
$p(\pi_{[n+1]} \mid \phi, \theta) \approx N((\phi_{MLE}, \theta_{MLE}), I_{MLE}^{-1})$



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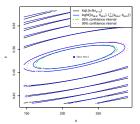
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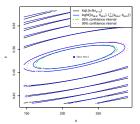


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$$\mathsf{LR} = \frac{n}{\mathbb{E}(\Phi \mid \Pi_{[n+1]} = \pi_{[n+1]})} \approx \frac{n+1+\theta_{\mathsf{MLE}}}{1-\alpha_{\mathsf{MLE}}}$$

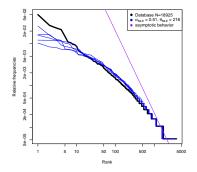
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Sorted relative frequencies: how good is our prior?

Comparison between the spectrum from a big database, and simulations from $PD(\alpha, \theta)$ using MLE estimators of the parameters.

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Comparison between the spectrum from a big database, and simulations from $PD(\alpha, \theta)$ using MLE estimators of the parameters.



Thick black line: ranked relative frequencies in the database. Thin black lines: simulations from the PD($\alpha_{MLE}, \theta_{MLE}$). Dotted line: asymptotics.

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Image: Image:

$$\mathsf{LR}_{|\mathbf{p}} = rac{p(\pi^{\mathsf{Db}++}_{[n+2]}|H_p,\mathbf{p})}{p(\pi^{\mathsf{Db}++}_{[n+2]}|H_d,\mathbf{p})} =$$

Image: Image:

$$\mathsf{LR}_{|\mathbf{p}} = \frac{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_{p},\mathbf{p})}{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_{d},\mathbf{p})} = \mathsf{Applying \ \mathsf{Lemma}} =$$

Image: A matrix and a matrix

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$$\mathsf{LR}_{|\mathbf{p}} = \frac{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_p, \mathbf{p})}{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_d, \mathbf{p})} = \mathsf{Applying \ \mathsf{Lemma}} = \frac{1}{\mathbb{E}(p_{x_{n+1}}|\pi_{[n+1]}^{\mathsf{Db}+}, \mathbf{p})}.$$

Image: Image:

$$\mathsf{LR}_{|\mathbf{p}} = \frac{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_p, \mathbf{p})}{p(\pi_{[n+2]}^{\mathsf{Db}++}|H_d, \mathbf{p})} = \mathsf{Applying \ \mathsf{Lemma}} = \frac{1}{\mathbb{E}(p_{x_{n+1}}|\pi_{[n+1]}^{\mathsf{Db}+}, \mathbf{p})}.$$

How is this compared to the one we get with our method when ${\bf p}$ is unknown?

Database of 2085 Y-STR profiles form Dutch men.

Database of 2085 Y-STR profiles form Dutch men.

Test: Compare the distribution of $\log_{10}(LR_{|p})$ and $\log_{10}LR$ obtained by 100 samples of size 100 from this population.

Results

Compare the distribution of $\log_{10}(\rm LR_{|p})$ and $\log_{10}\rm LR$ obtained by 100 samples of size 100 from this population

