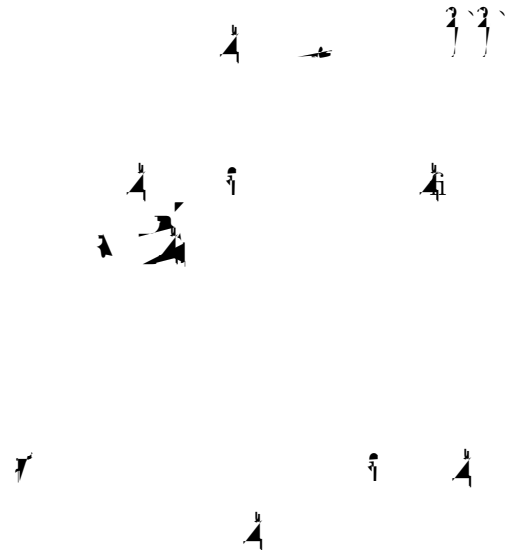


A pedigree-transmission likelihood for multiplex families

Tianyu Yang



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SIMON FRASER UNIVERSITY

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Declaration of Committee

Name: Tianyu Yang

Degree: Master of Science

Thesis title: A pedigree-transmission likelihood for multiplex families











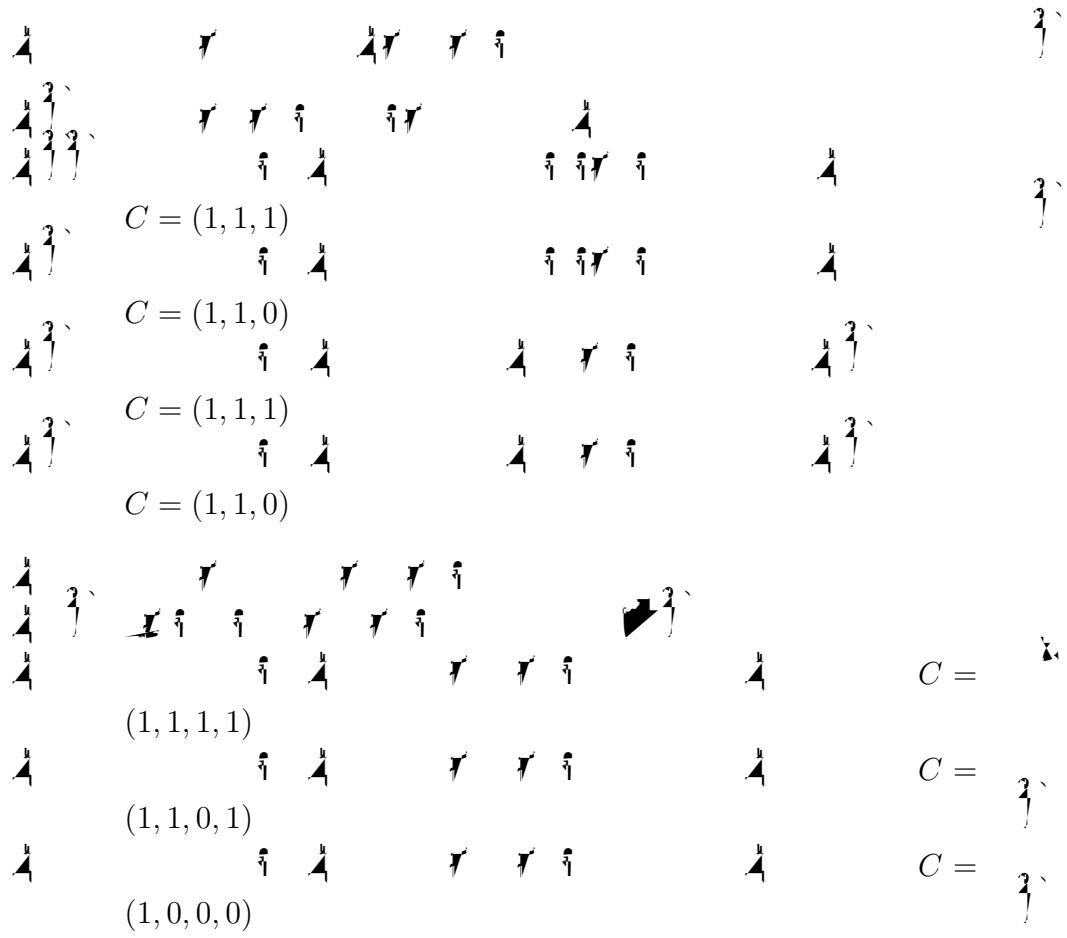
Committee: Chair:  

Jinko Graham   
Rhonda Rosychuk    
Brad McNeney  

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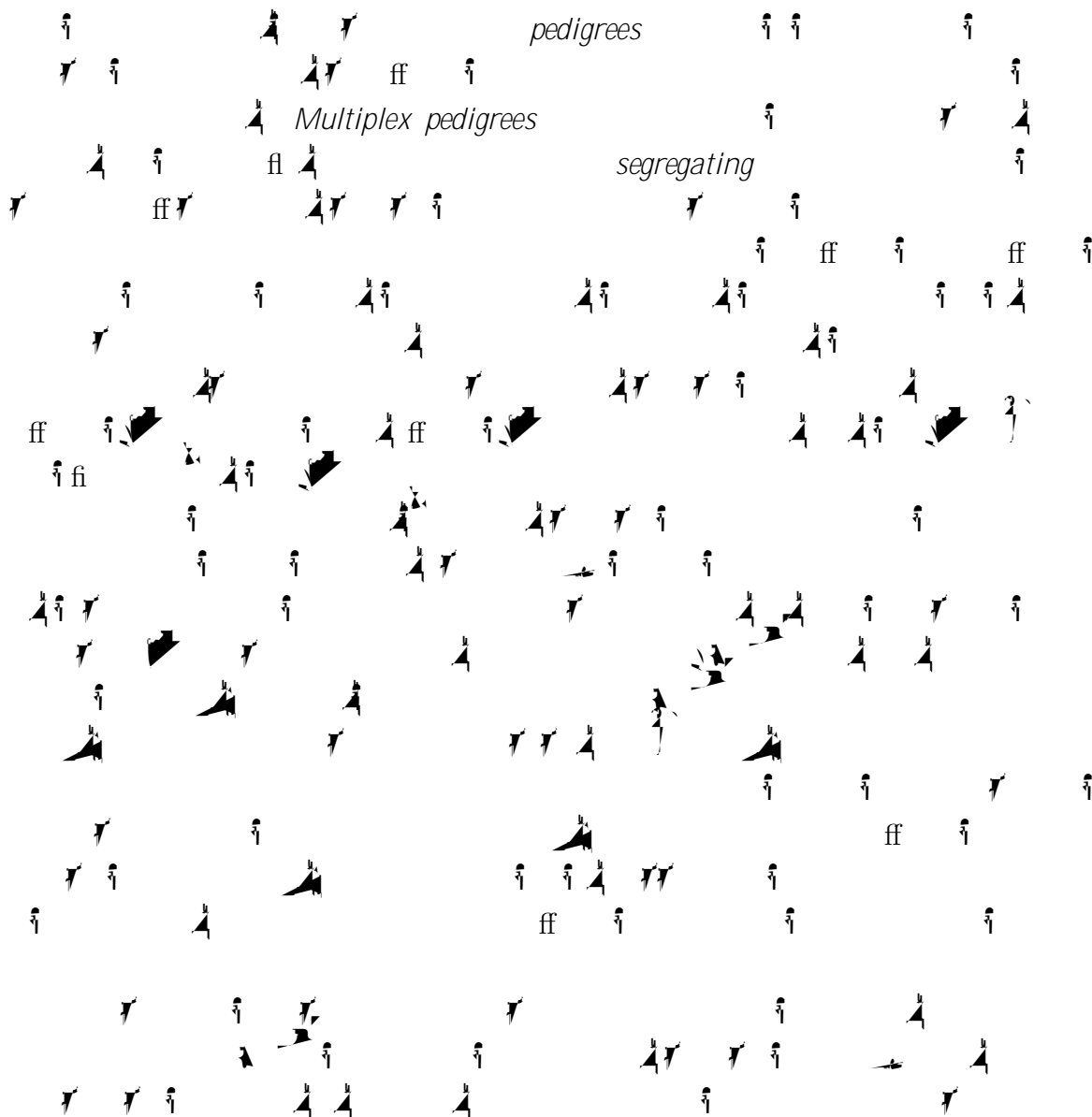
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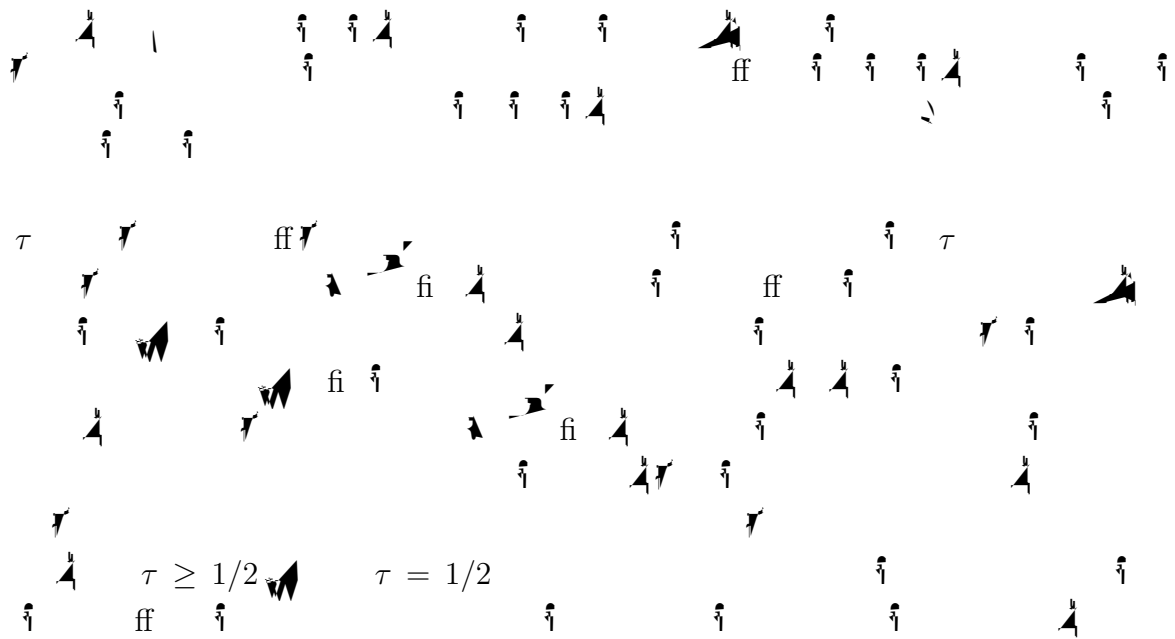
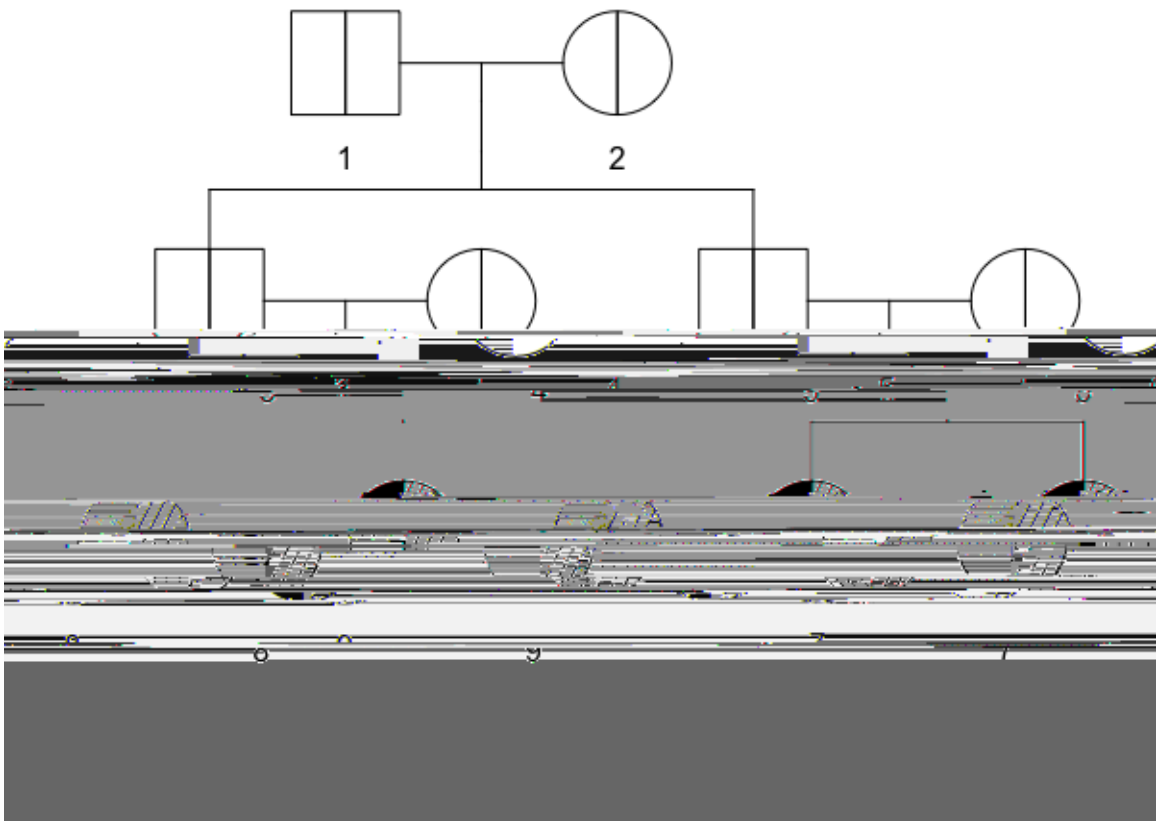
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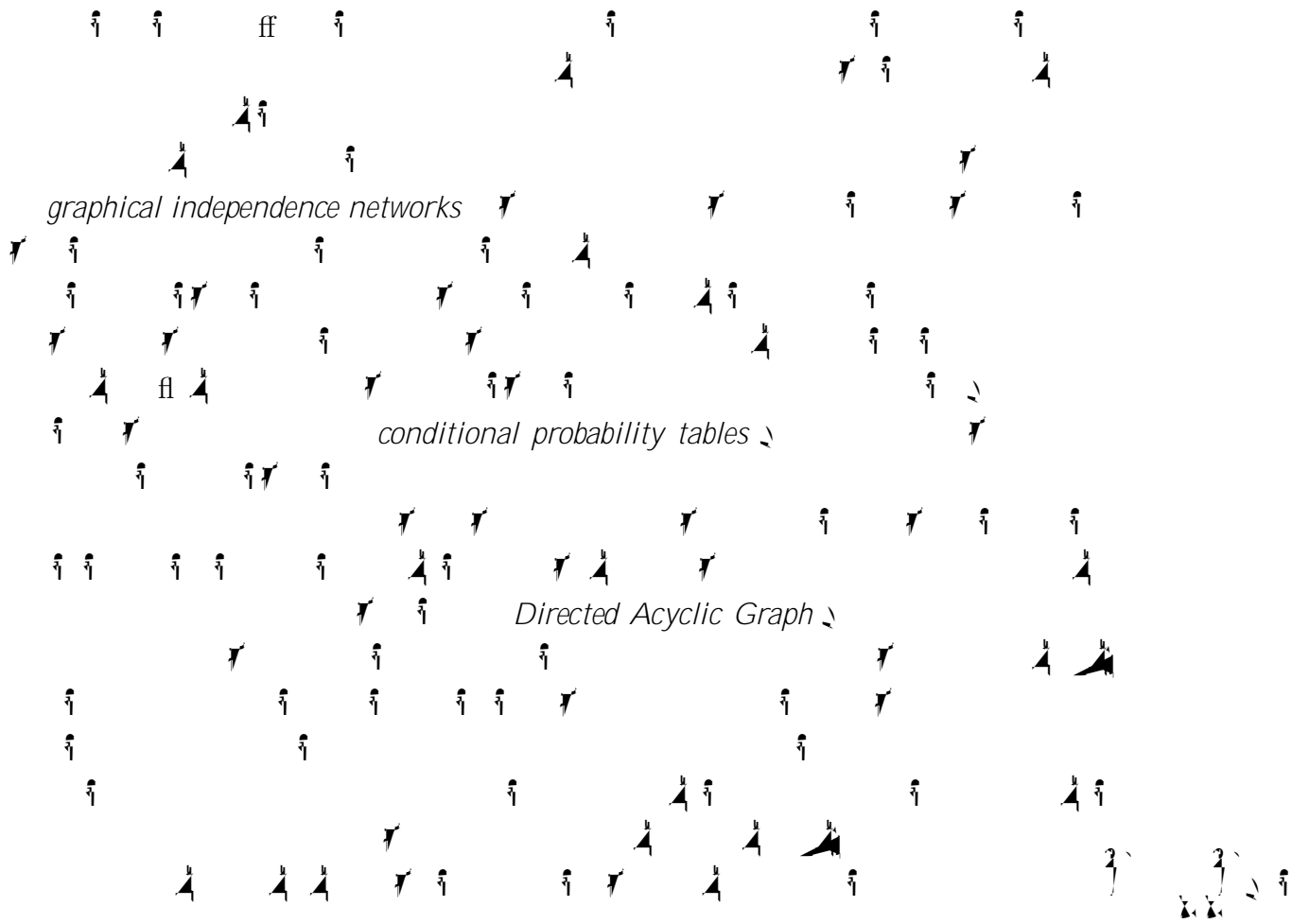


Chapter 1

Introduction and background







Chapter 2

Likelihood

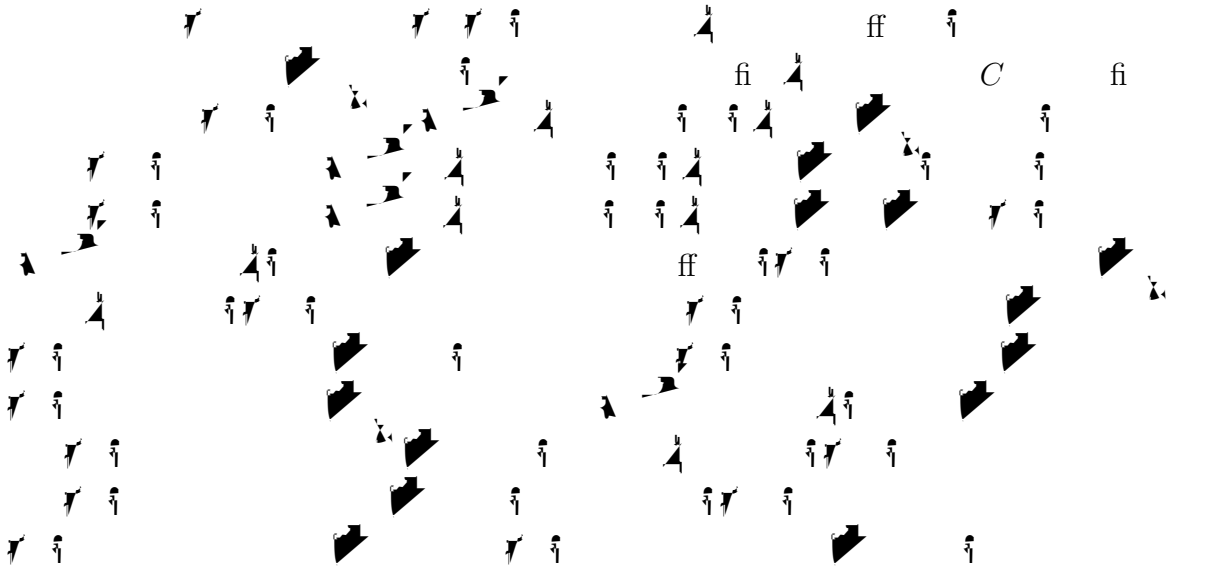
2.1 Formulas



$$\begin{aligned}
 P(C | \cup_i F_i) &= \frac{P(C, \cup_i F_i)}{P(\cup_i F_i)} \\
 &= \frac{\sum_i P(C, F_i)}{\sum_i P(F_i)} \\
 &= \sum_i P(C | F_i) \frac{P(F_i)}{\sum_j P(F_j)}.
 \end{aligned}$$

$$P(F_1) = P(F_2) = P(F_3) = \dots$$

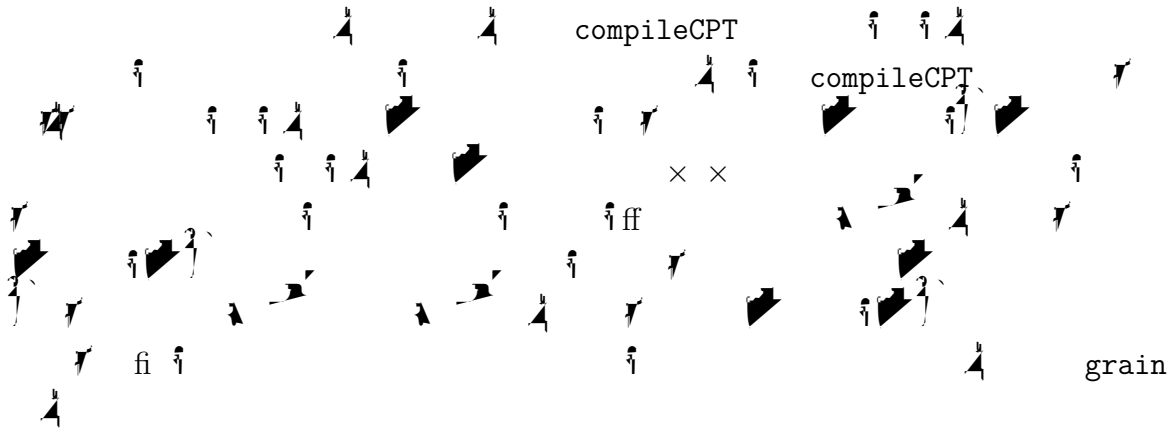
$$P(C|F_i) \text{ path counting}$$



$$P(C = (1, 1, 1)|F_1) = \tau^{2+1+2} \quad P(C = (1, 1, 0)|F_1) = \tau^{2+1+1}(1 - \tau)$$

$$P(C = (1, 1, 0)|F_1)$$

2.2 Implementation



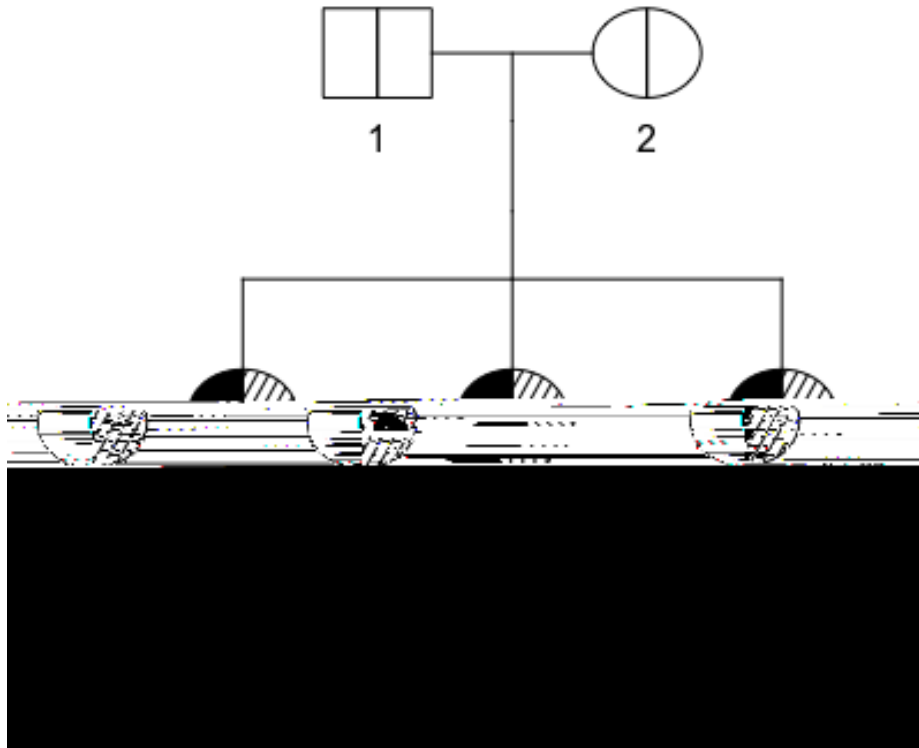
2.2.2 Likelihood function

```

likehd()
fi pedfile kinship2 tau config
pedfile tau C
config C
ff C = (1,0,1) BNcreate()
P(C|i)
P(C|
(
(
#
(
#
(

```


`ff` `likehd()`
path-counting

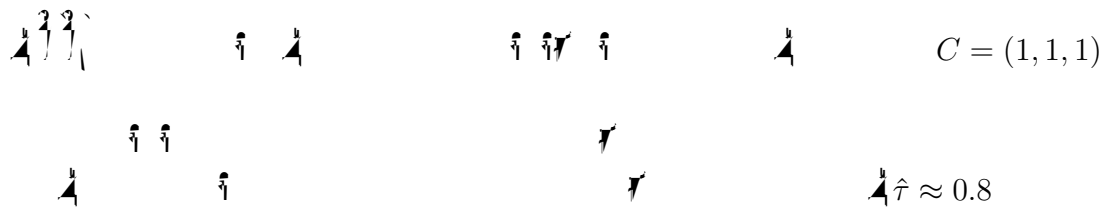
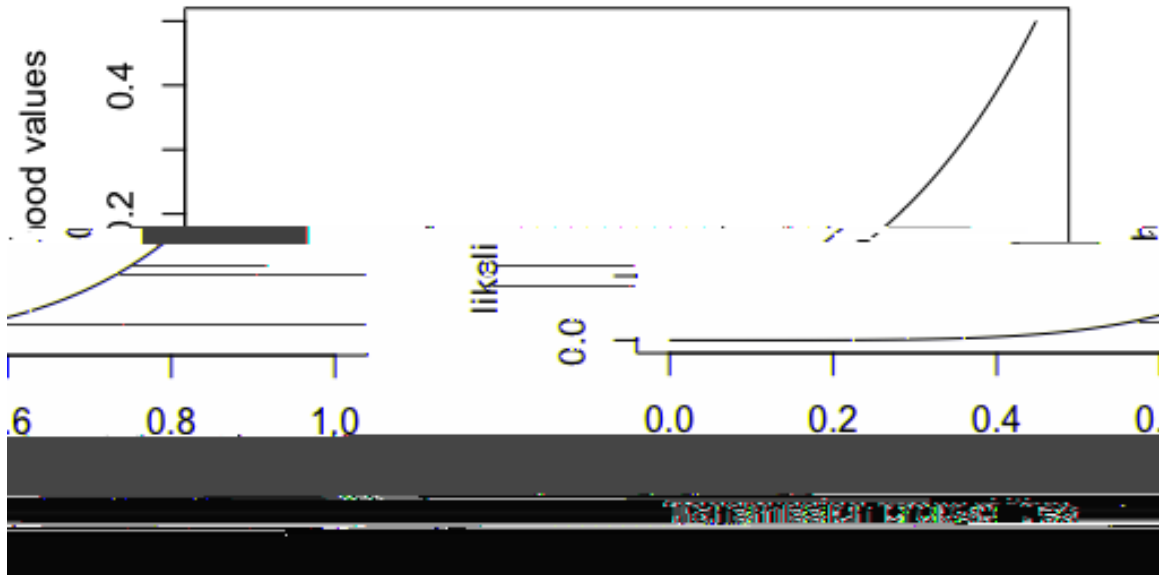


$\tau = 3/4$ `likehd()` 0.421875 $C = (1, 1, 1)$ $\frac{1}{2}(\tau^3 + \tau^3) = 0.421875$
 $C = (1, 0, 1)$ `likehd()` 0.140625 $\frac{1}{2}(\tau^2(1 - \tau) + \tau^2(1 - \tau)) = 0.140625$
`likehd()` $\tau = 3/11$ 4034 $C = (1, 1, 1)$

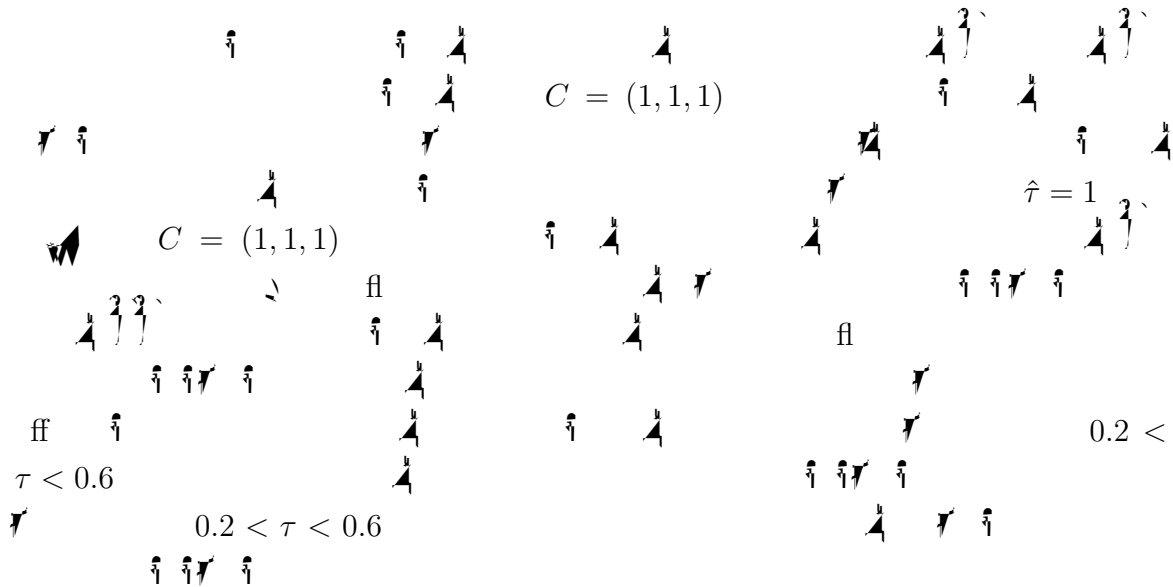
$C = (1, 1, 0)$ $\tau = 3/4$
`likehd()` .0396
 $\frac{1}{2}\tau^4(1 - \tau) = \frac{1}{2} \left(\frac{3}{4}\right)^4 \left(1 - \frac{3}{4}\right) = 0.0396$

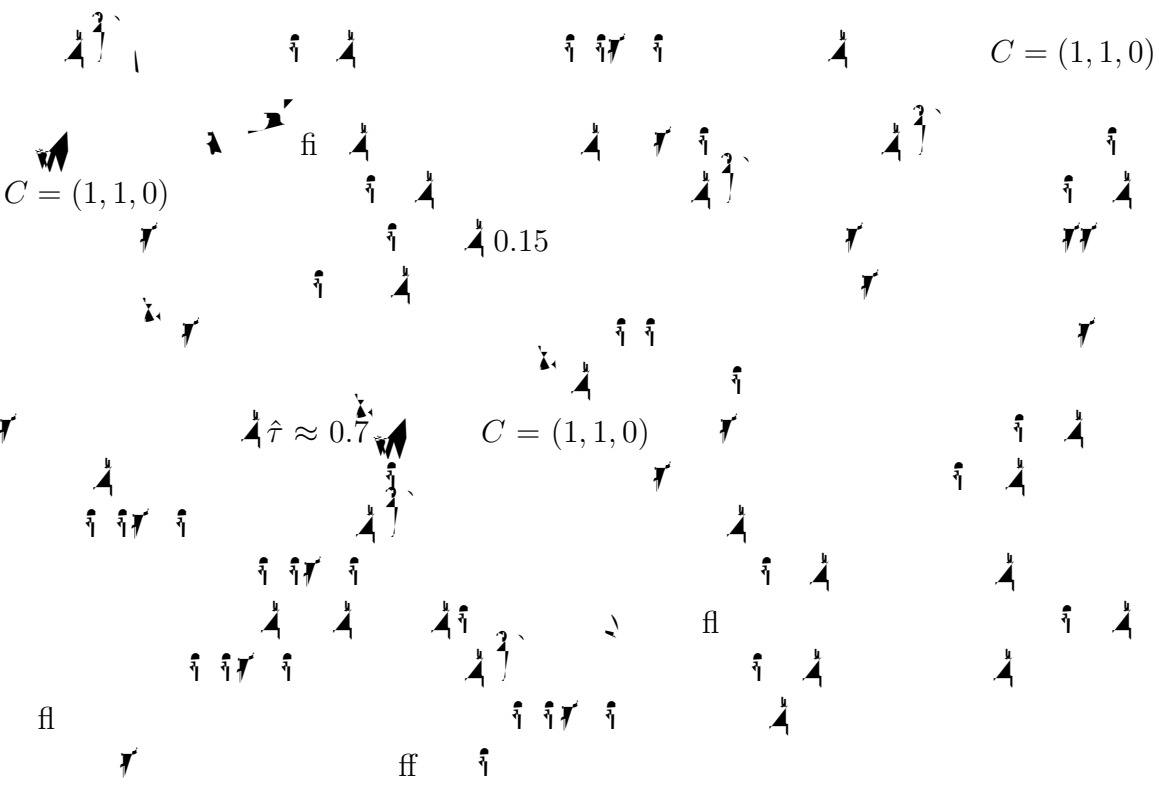
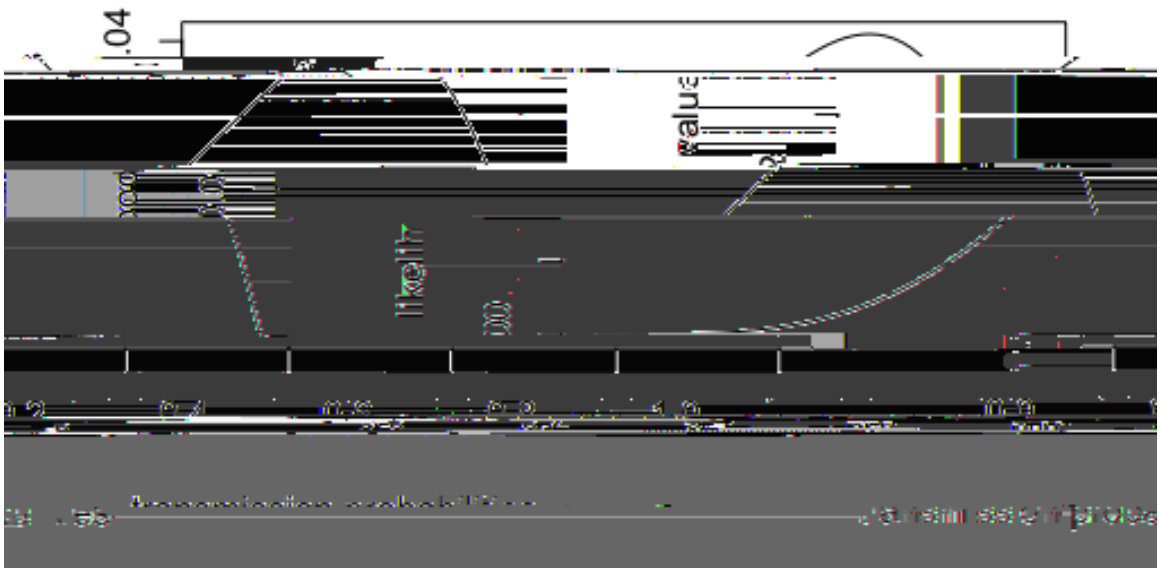
$C = (1, 1, 1)$ $\tau = 3/4$
 0.421875 0.1187

fi4

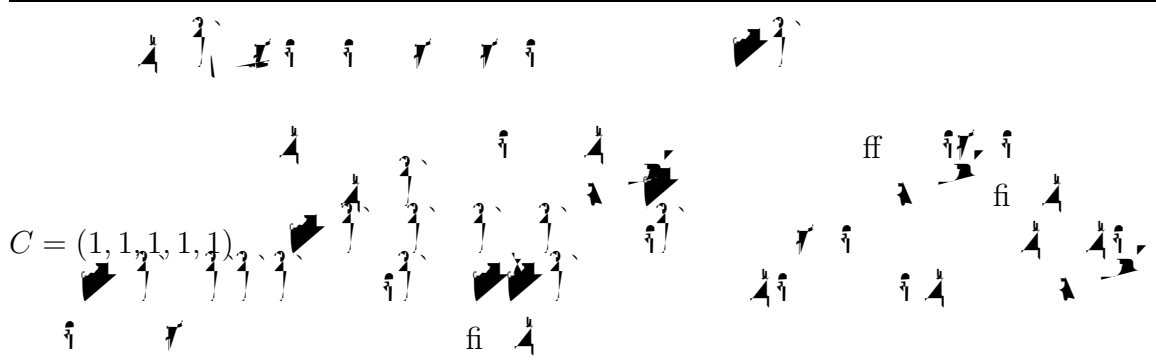
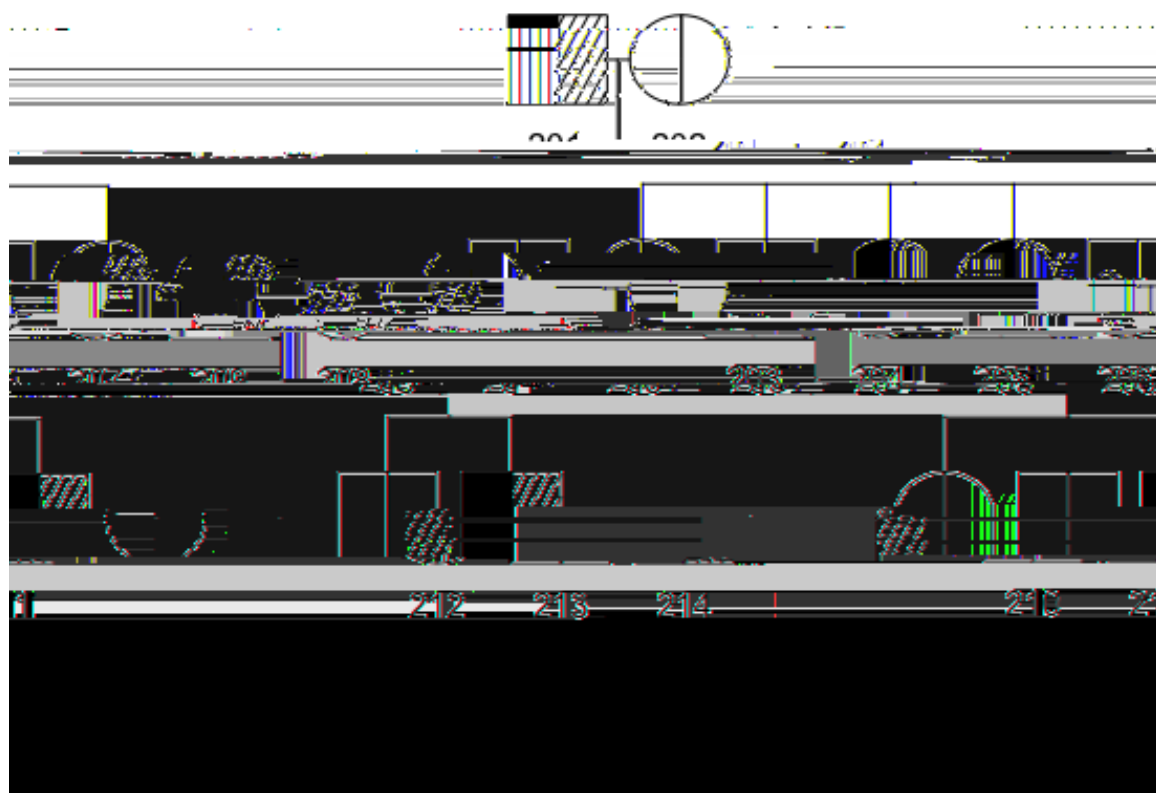
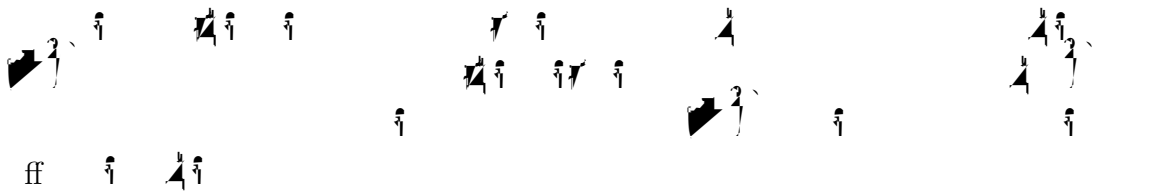


2.3.2 Nuclear Pedigree





3.2 Second pedigree



$$C = (1, 1, 1, 1, 1)$$

$$P(C = (1, 1, 1, 1, 1) | F_{201}) = 1 \times \tau \times \tau \times \tau^2 \times \tau^2 = \tau^6.$$

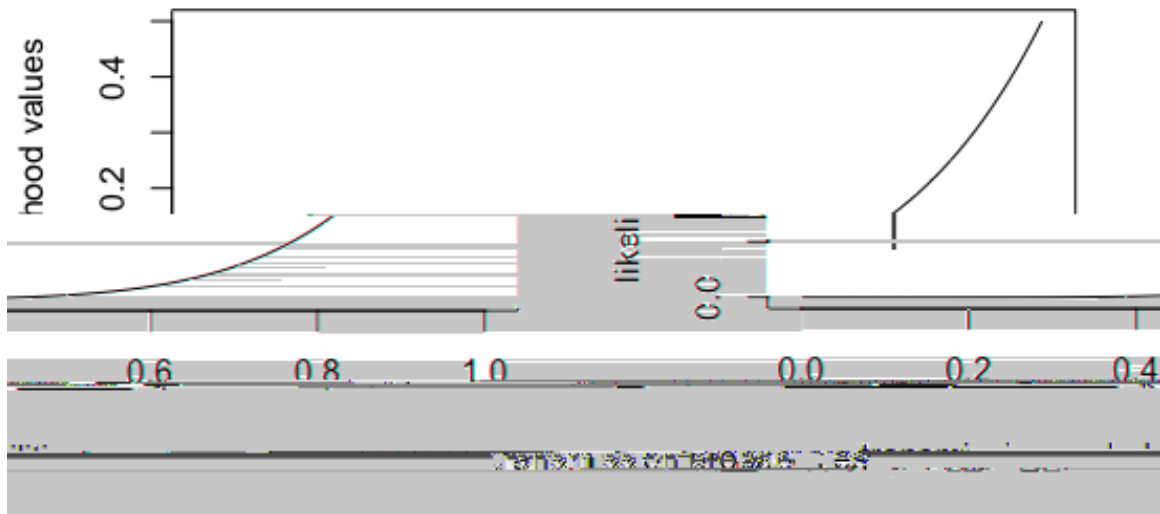
fi

$$P(C = (1, 1, 1, 1, 1) | F_{202}) = 0 \times \tau \times \tau \times \tau^2 \times \tau^2 = 0.$$

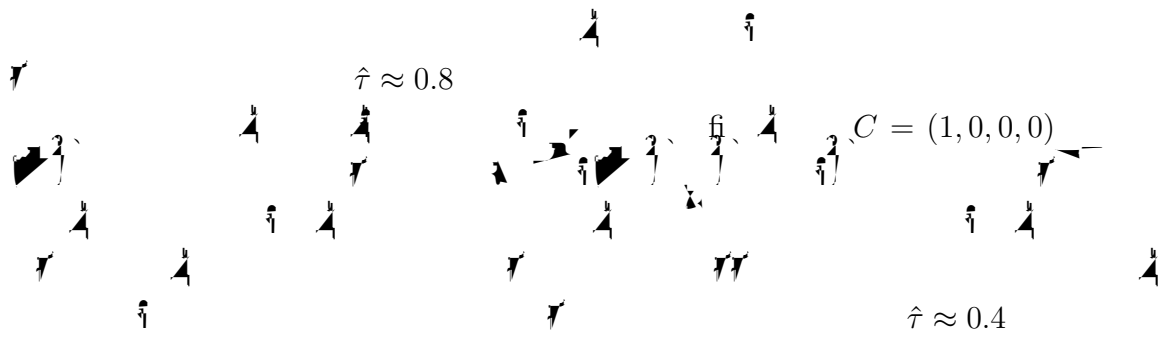
$P(C = (1, 1, 1, 1, 1) | \cup_i F_i) = \frac{1}{4}\tau^6.$
 0.00390625 $\tau = 1/2$ fi i likehd()
 $C = (0, 1, 1, 1, 1)$
 $P(C = (0, 1, 1, 1, 1) | F_{201}) = 0$
 $P(C = (0, 1, 1, 1, 1) | F_{202}) = \tau \times \tau \times \tau^2 \times \tau^2 = \tau^6$
 $P(C = (0, 1, 1, 1, 1) | F_{203}) =$
 $P(C = (0, 1, 1, 1, 1) | F_{209}) = 0$
 $P(C = (0, 1, 1, 1, 1) | \cup_i F_i) = \frac{1}{4}\tau^6$
 $\tau = 1/2$ fi i likehd() 0.00390625

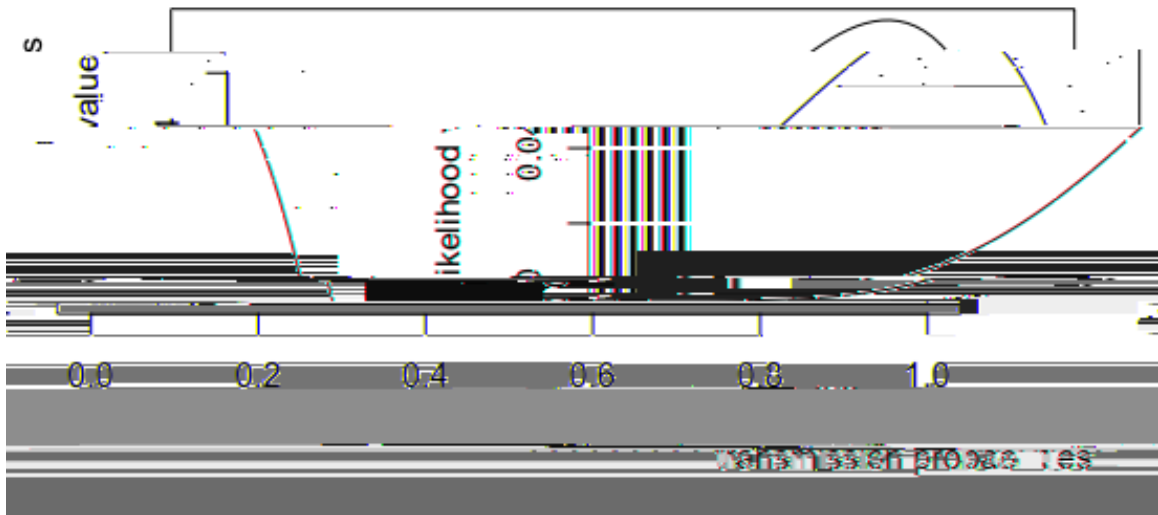
3.3 Likelihood Curves

fi $C = (1, 1, 1, 1)$ fi ff i

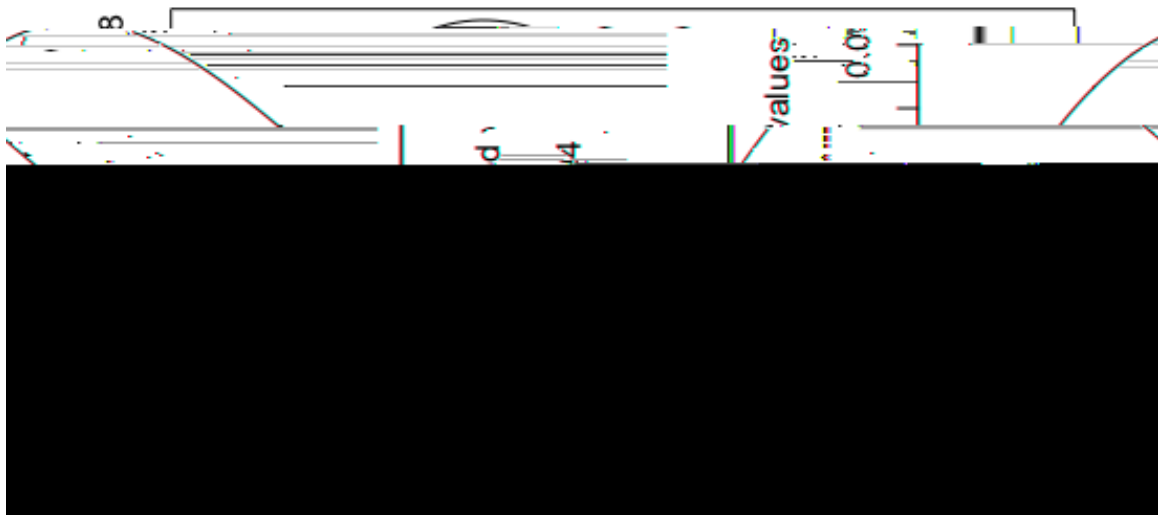


$C = (1, 1, 1, 1)$





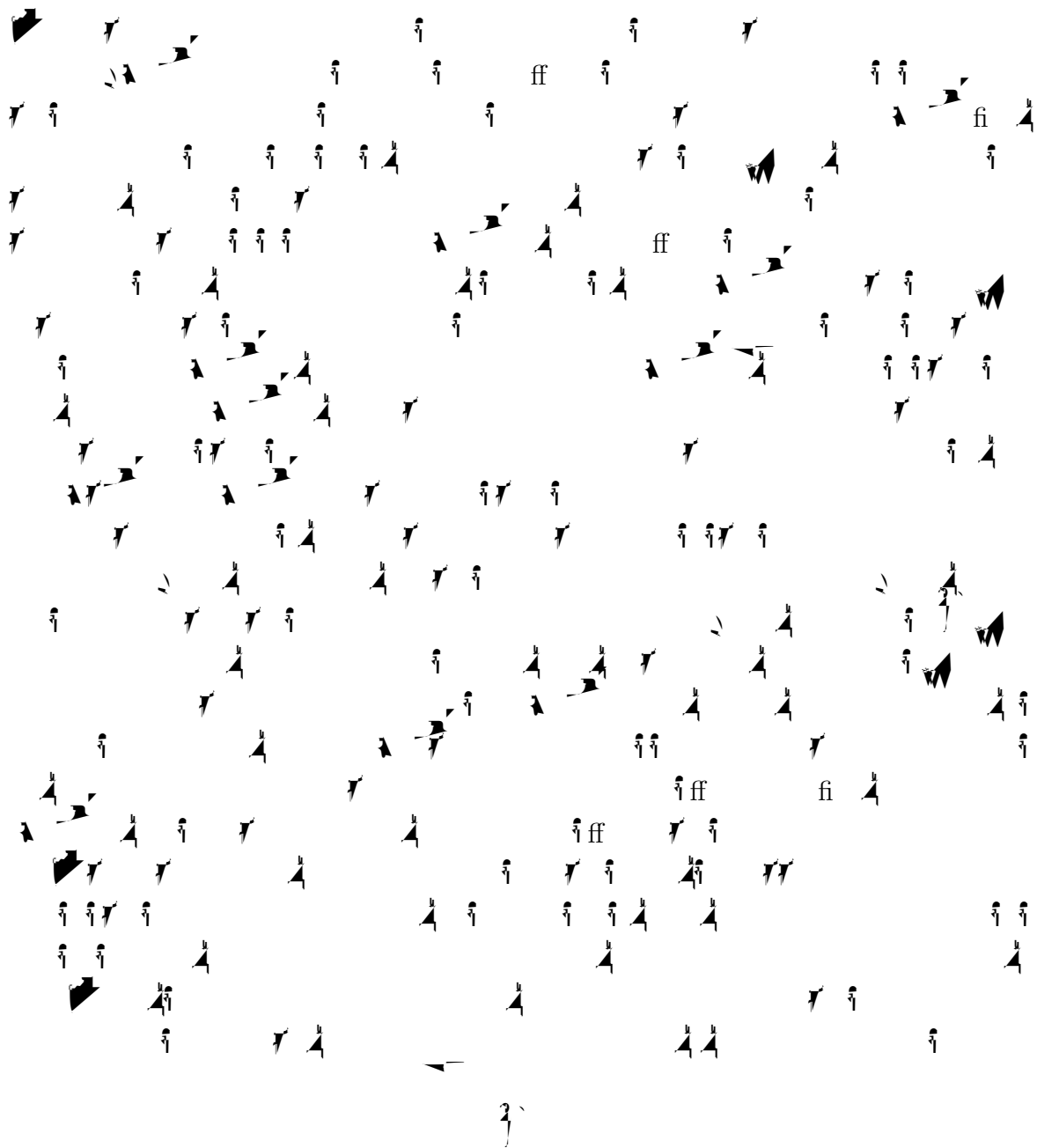
↑ ↓ ↑ ↓ ↑ ↓ ↑ ↓ ↑ ↓ $C = (1, 1, 0, 1)$



↑ ↓ ↑ ↓ ↑ ↓ ↑ ↓ ↑ ↓ $C = (1, 0, 0, 0)$

Chapter 4

Conclusions



Bibliography

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Journal of Nursing

Nat Methods

Introduction to Algorithms for Data Mining and Machine Learning

Bayesian Networks: An Introduction

Hum Hered

Journal of Statistical Software

American

The American

Appendix A

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