# Exam Statistical Genomics

Date: Thursday 30 January, 2014

Time: 9.00-12.00

Place: Bernoulliborg Building, Room 5161.0222

Progress code: WISG-09

Rules to follow:

• The number of points per question are indicated within a box. Ten points are free.

• Do not forget to fill in your name and student number.

• We wish you success with the completion of the exam!

### START OF EXAM

- 1. Kullback-Leibler divergence, likelihood and deviance. 20 The Kullback-Leibler divergence is defined as  $I(f;g) = E_f \log \frac{f(X)}{g(X)}$ . We consider a graphical log-linear model. Let n be a table of counts, where n(x) is a particular cell-count. Let  $N = \sum_x n(x)$  be the total number of observations.
  - $\boxed{10}$  Show that the log-likelihood for a table n can be written as

$$l(p;n) = l(\frac{n}{N};n) - N \times I(\frac{n}{N};p),$$

where p and n/N are interpreted as cell probabilities.

• 10 Consider a particular graphical log-linear model M. Show that the deviance can be written as

$$Dev(M) = 2\sum_{x} n(x) \log \frac{n(x)/N}{\hat{p}^{M}(x)},$$

where  $\hat{p}^M$  is the maximum likelihood estimator for p under M.

2. Binary log-linear model. 35

We study the three year survival  $(X_3)$  of 474 breast cancer patients according to nuclear grade  $(X_2)$  and diagnostic centre  $(X_1)$ .

- (a) 5 Derive the MLE of  $p(X_1 = 1, X_2 = 1, X_3 = 1)$  under the saturated model.
- (b)  $\boxed{5}$  Derive the MLE of  $p(X_1=1,X_2=1,X_3=1)$  under the model 1.2+1.3.

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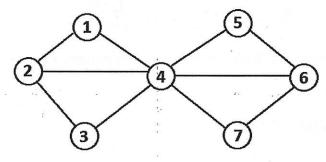
	$X_2$	=0	$X_2$		
~	$X_3=0$	$X_3 = 1$	$X_3 = 0$	$X_3 = 1$	
$X_1 = 0$	.35	59	<i>A</i> 7	112	253
$X_1 = 1$	42.	(77)	26	(76)	221
total	77	136	73	188	474

- (c)  $\boxed{10}$  We can decompose the log-density  $\log p$  in a log-linear way using u-terms. Derive the MLE of the  $u_1$  term under the model 1.2 + 1.3.
- (d) 10 We want to test whether we can exclude the link (2,3) from the saturated model. Determine the edge exclusion deviance and test whether you can delete it at a 5% significance level. A chi-squared table can be found at the end of the exam.
- (e)  $\boxed{5}$  Argue whether the model 1.2 + 1.3 + 2.3 is graphical and/or hierarchical.

## 3. Gaussian graphical model (1). 15

The sample variance matrix based on  $\overline{N} = 50$  observations from a Gaussian graphical model is

Calculate the following 3 elements of the MLE of the variance covariance matrix  $\Sigma$  associated with the following conditional independence graph:



- (a)  $\boxed{5} \hat{\Sigma}_{12}$ ?
  - (b)  $5 \hat{\Sigma}_{13}$ ?
  - (c)  $[5] \hat{\Sigma}_{17}$ ?

4. Gaussian graphical model (2). 20

A sample covariance matrix for a sample from a Gaussian graphical model N(0, V) of size N = 10 is given as

$$S = \begin{pmatrix} 2 & 1 & 1 \\ 1 & 2 & 1 \\ 1 & 1 & 2 \end{pmatrix}$$

(a) 10 Use the deviance  $Dev(M_1)$  for  $M_1 = X_3 \perp (X_2, X_1)$  to test whether  $M_1$  fits the data.

**Hint:** You can use the fact that |S| = 4 and that the determinant of the top submatrix of S is  $|S_{12,12}| = 3$ . Moreover, the inverse of a  $2 \times 2$  matrix, is given by

$$\begin{pmatrix} a & b \\ c & d \end{pmatrix} = \frac{1}{ad - bc} \begin{pmatrix} d & -b \\ -c & a \end{pmatrix}.$$

(b) 10 Use (for example) the fact that

$$Inf(X_3 \perp X_2 | X_1) = -\frac{1}{2} \log \frac{|V| |V_{11}|}{|V_{12,12}| |V_{13,13}|}$$

to calculate the deviance  $\mathrm{Dev}(M_2)$  for  $M_2=X_3\perp X_2|X_1$  to test whether  $M_2$  fits the data.

If you want to use directly the edge exclusion deviance, then that's fine too.

#### END OF EXAM

### Chi-squared table.

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$\nu \setminus \alpha$	0.995	0.99	0.975	0.95	0.05	0.025	0.01	0.005
1	0.000	0.000	0.001	0.004	3.841	5.024	6.635	7.879
2	0.010	0.020	0.051	0.103	5.991	7.378	9.210	10.597
3	0.072	0.115	0.216	0.352	7.815	9.348	11.345	12.838
4	0.207	0.297	0.484	0.711	9.488	11.143	13.277	14.860

Table 1: Values of  $\chi^2_{\alpha,\nu}$ : entries correspond to values of x, such that  $P(\chi^2_{\nu} > x) = \alpha$ , where  $\chi^2_{\nu}$  correspond to a chi-squared distributed variable with  $\nu$  degrees of freedom.