MODELLING GENETIC EPISTASIS BETWEEN SELECTED CANDIDATE GENES FOR MILK PRODUCTION TRAITS IN JERSEY CATTLE

G18.8 (abstract # 0943)

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ANIMALS

- **192 Jersey cows**

TRAITS

- **production traits:** milk-, fat-, protein- yield, fat-, protein- content
- **305-day lactation yields**
- **3 parities**

GENES

- **leptin receptor (LEPR)** **BTA03**

  T945M

- **leptin gene (LEP)** **BTA04**

  Y7F, R25C, A80V, C/T sub. -963

- **diacylglycerol acyltransferase 1 (DGAT1)** **BTA14**

  K232A

- **butyrophilin (BTN)** **BTA23**

  F16Y, P35Q, K468R

- **no missing genotypes**

GENERAL MODEL

$$\mathbf{y} = \mathbf{X}_1\boldsymbol{\beta} + \mathbf{X}_2\mathbf{q} + \mathbf{Z}_1\boldsymbol{\alpha} + \mathbf{Z}_2\mathbf{p} + \mathbf{e}$$

$$\boldsymbol{\beta} = \begin{bmatrix} birth \quad year \\ parity \\ calving \quad year * season \end{bmatrix}$$

$$\mathbf{q} = \begin{bmatrix} LEPR \\ LEP \\ DGAT\ 1 \\ BTN\ 1A1 \end{bmatrix}$$

$$\alpha \sim N\left(0, A\sigma_\alpha^2\right) \qquad \sigma_\alpha^2 = 30\%\sigma_y^2$$

$$p \sim N\left(0, I\sigma_p^2\right) \qquad \sigma_p^2 = 20\%\sigma_y^2$$

$$e \sim N\left(0, I\sigma_e^2\right) \qquad \sigma_e^2 = 70\%\sigma_y^2$$

PARAMETERISATION OF GENE EFFECTS

$$\boxed{\mathbf{y} = \mathbf{X}_1\boldsymbol{\beta} + \mathbf{X}_2\mathbf{q} + \mathbf{Z}_1\boldsymbol{\alpha} + \mathbf{Z}_2\mathbf{p} + \mathbf{e}}$$

$$\mathbf{X}_a \in \left\{2P(2),\ \ P(2) - P(1),\ \ -2P(1)\right\}$$

$$\mathbf{X}_d \in \left\{\frac{1}{P(11)},\ -\frac{2}{P(12)},\ \frac{1}{P(22)}\right\}$$

$$\mathbf{X}_{\text{int}} \in \left\{x_{ai}x_{aj}\right\}$$

Cockerham, 1954; Kao and Zeng, 2002

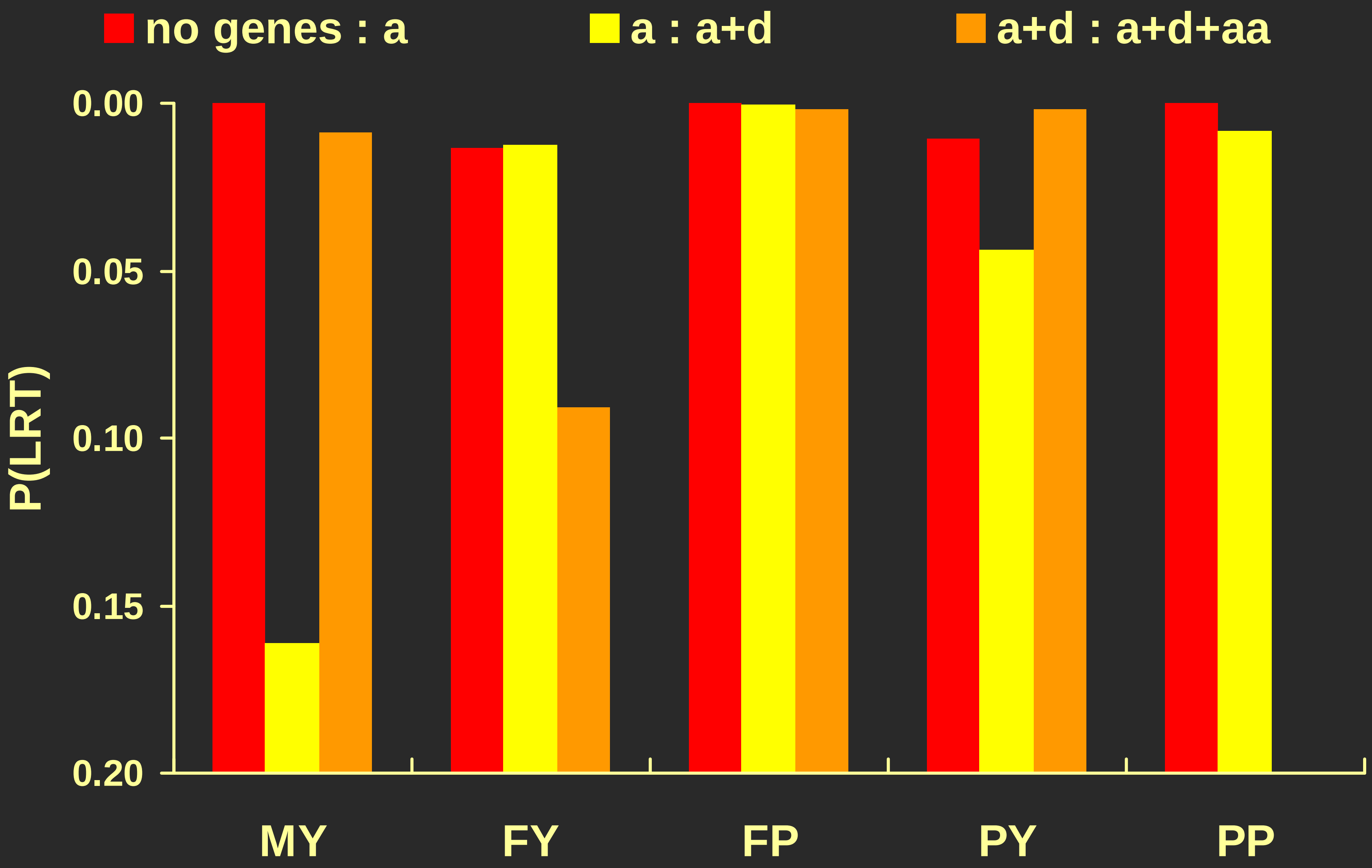
PARAMETERISATION OF GENE EFFECTS

$$\boxed{\mathbf{y} = \mathbf{X}_1\boldsymbol{\beta} + \mathbf{X}_2\mathbf{q} + \mathbf{Z}_1\boldsymbol{\alpha} + \mathbf{Z}_2\mathbf{p} + \mathbf{e}}$$

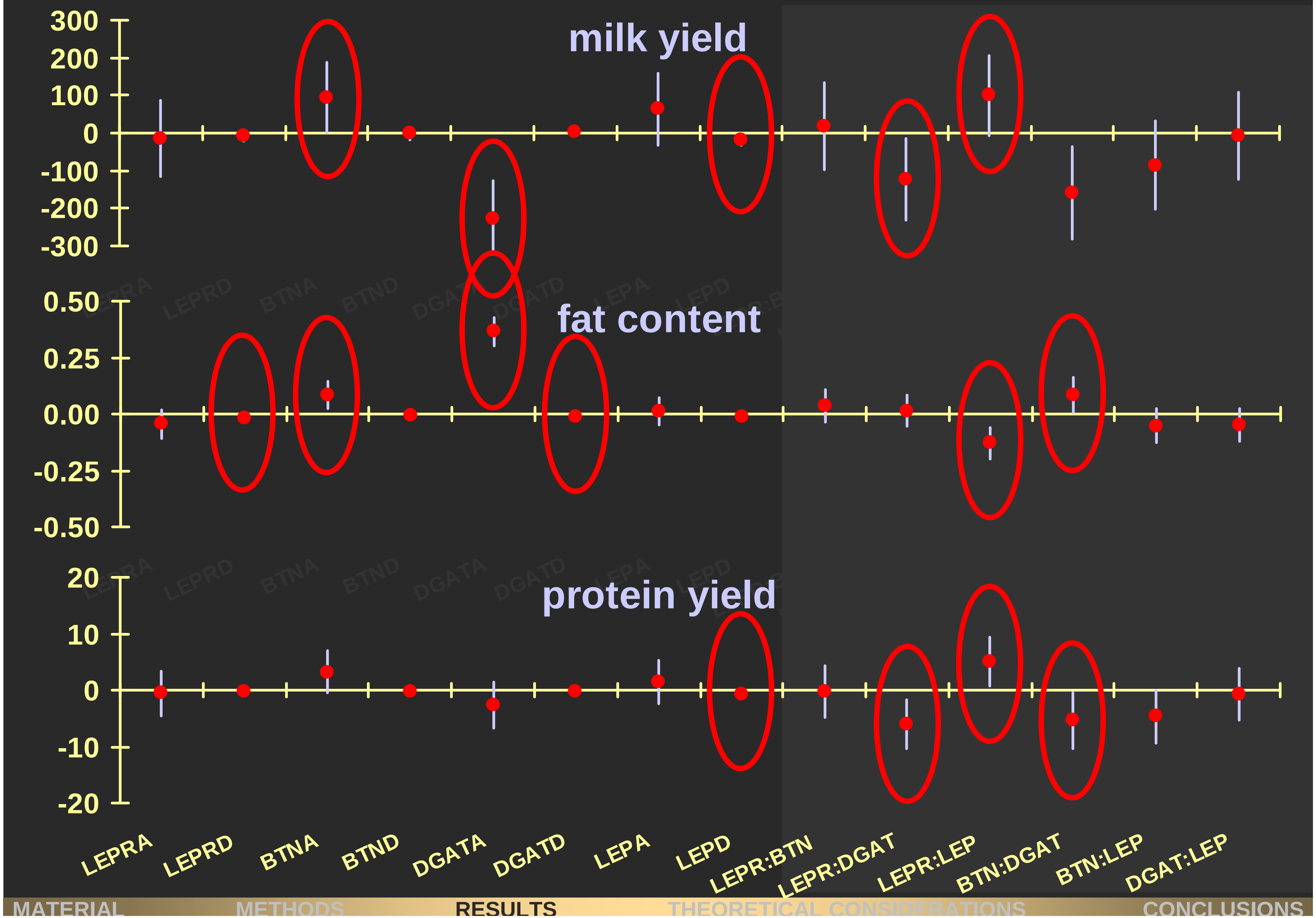
- no genes $\xcancel{\mathbf{q}}$
- additive $\mathbf{q}^T = \left[\sum_{i=1}^{4} a_i \right]$
- additive and dominance $\mathbf{q}^T = \left[\sum_{i=1}^{4} a_i + \sum_{i=1}^{4} d_i \right]$
- additive, dominance and additive*additive

$$\mathbf{q}^T = \left[\sum_{i=1}^{4} a_i + \sum_{i=1}^{4} d_i + \sum_{i=1}^{4} \sum_{j=1}^{4} aa_{ij} \right]$$

MODEL COMPARISON



GENE EFFECTS ESTIMATES



MODEL PARAMETERISATION

$$\boxed{\mathbf{y} = \mathbf{X}_1\boldsymbol{\beta} + \mathbf{X}_2\mathbf{q} + \mathbf{Z}_1\boldsymbol{\alpha} + \mathbf{Z}_2\mathbf{p} + \mathbf{e}}$$

Kao & Zeng F2 model

$$a_i = \frac{1}{2}\left(G_{i11} - G_{i22}\right) \qquad d_i = 2G_{i12}$$

$$\mathbf{X}_a \in \{1,\ 0,\ -1\} \qquad \mathbf{X}_d \in \left\{-\frac{1}{2},\ \frac{1}{2},\ -\frac{1}{2}\right\} \qquad \mathbf{X}_{\text{int}} \in \{x_{ai}x_{aj}\}$$

Kao & Zeng F∞ model

$$a_i = \frac{1}{2}\left(G_{i11} - G_{i22}\right) \qquad d_i = G_{i12} + \frac{1}{2}d_j$$

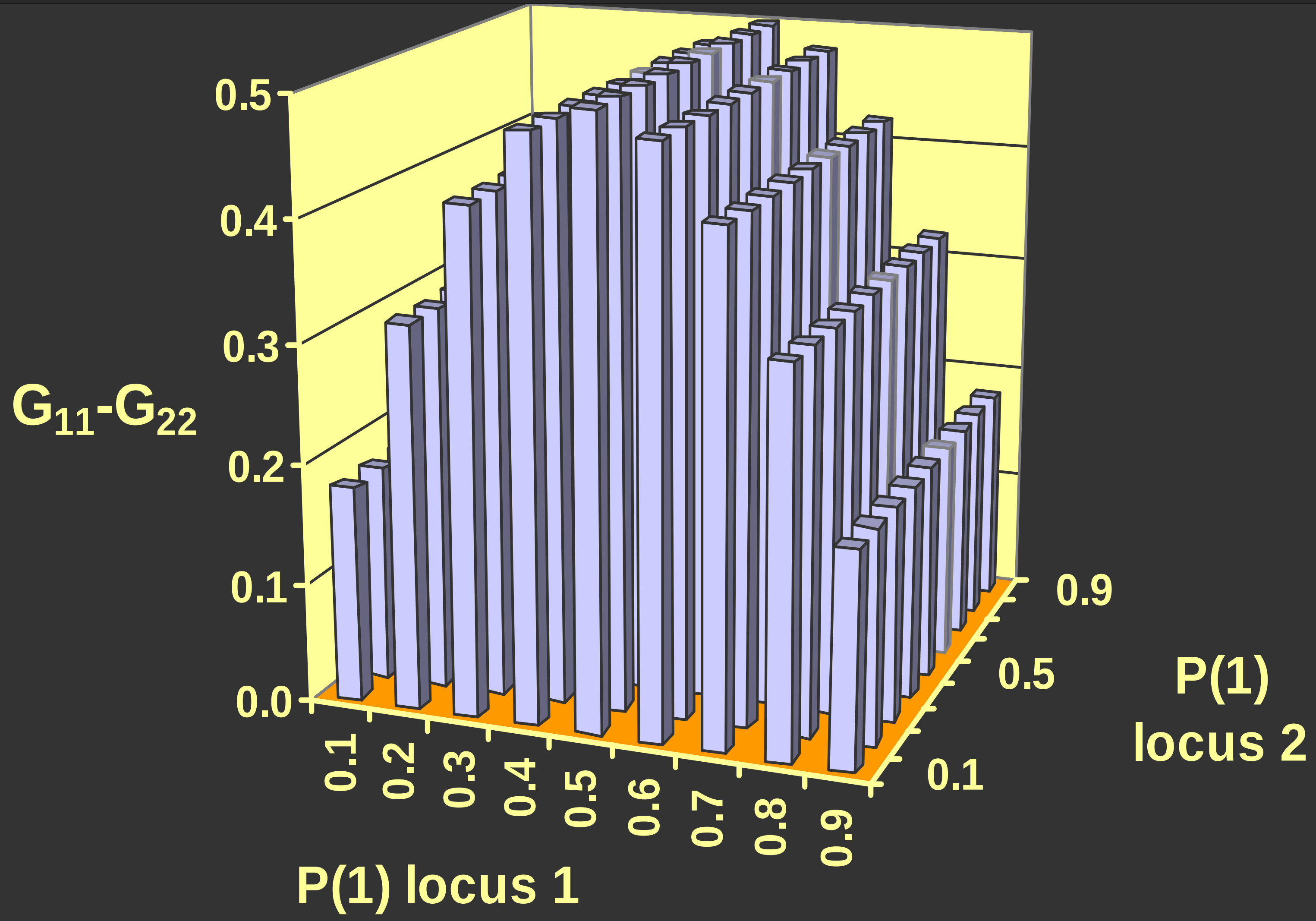
$$\mathbf{X}_a \in \{1,\ 0,\ -1\} \qquad \mathbf{X}_d \in \{0,\ 1,\ 0\} \qquad \mathbf{X}_{\text{int}} \in \{x_{ai}x_{aj}\}$$

Cockerham model

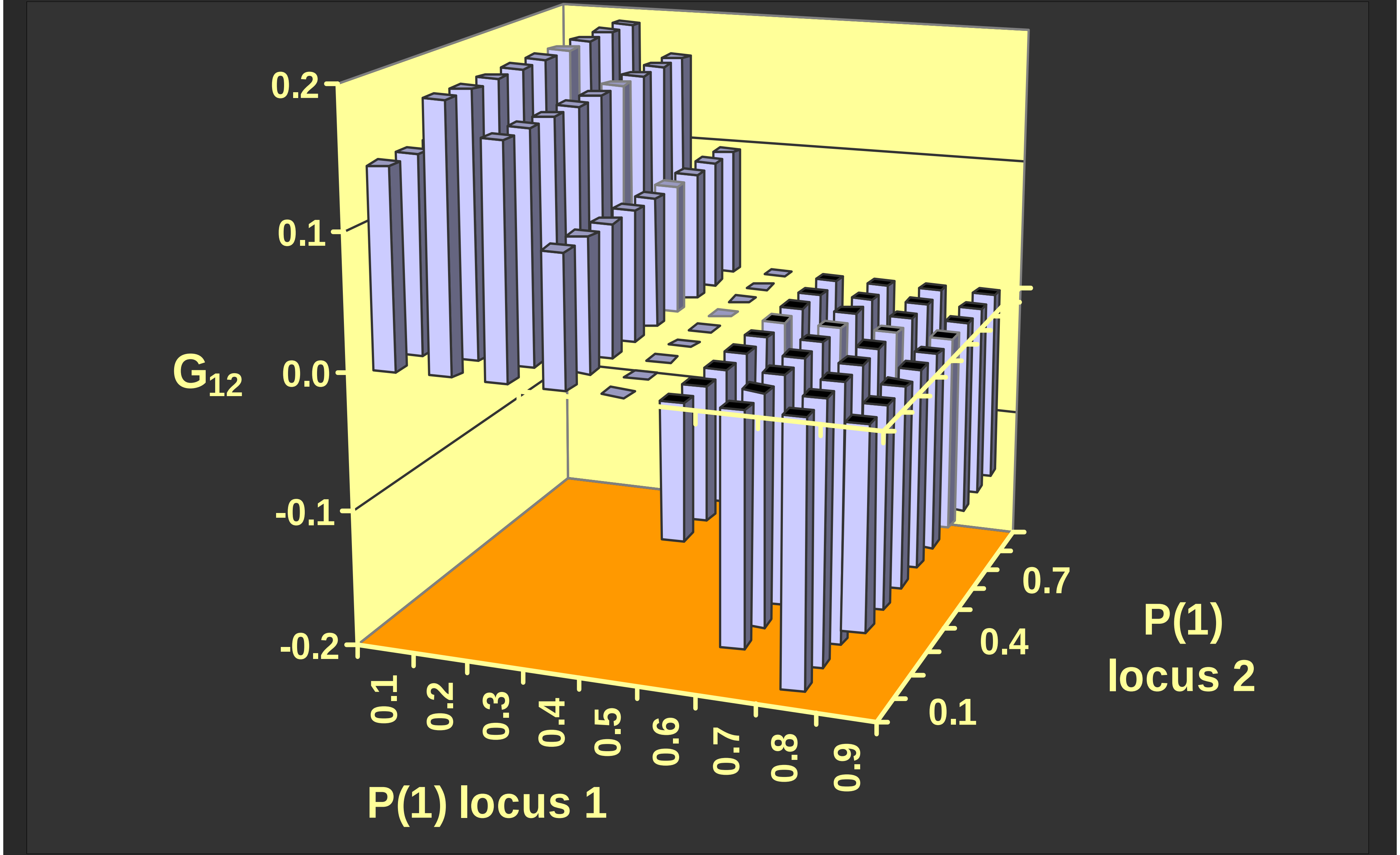
$$a_i = \frac{\left(G_{i11} - G_{i22}\right)}{P(12)} \qquad d_i = \left[P(1)P(22) - P(2)P(11)\right]a_i - \frac{G_{i12}}{2}$$

$$\mathbf{X}_a \in \{2P(2), P(2) - P(1), -2P(1)\} \qquad \mathbf{X}_d \in \left\{\frac{1}{P(11)}, -\frac{2}{P(12)}, \frac{1}{P(22)}\right\} \qquad \mathbf{X}_{\text{int}} \in \{x_{ai}x_{aj}\}$$

MODEL PARAMETERISATION



MODEL PARAMETERISATION



CONCLUSIONS

1. Model selection shows significant epistasis for:

- milk yield → DGAT1-LEPR and DGAT1-BTN
- fat content → LEP-LEPR and DGAT1-BTN
- protein yield → LEP-LEPR, DBAT1-BTN, DGAT1-LEPR

2. Using Cockerham's model

- estimates of additive effects - biased upwards
- correction → 0.5 * P(12)
- estimates of dominance effects - difficult to interpret
- estimates of main effects of 1 locus - independent of the other loci