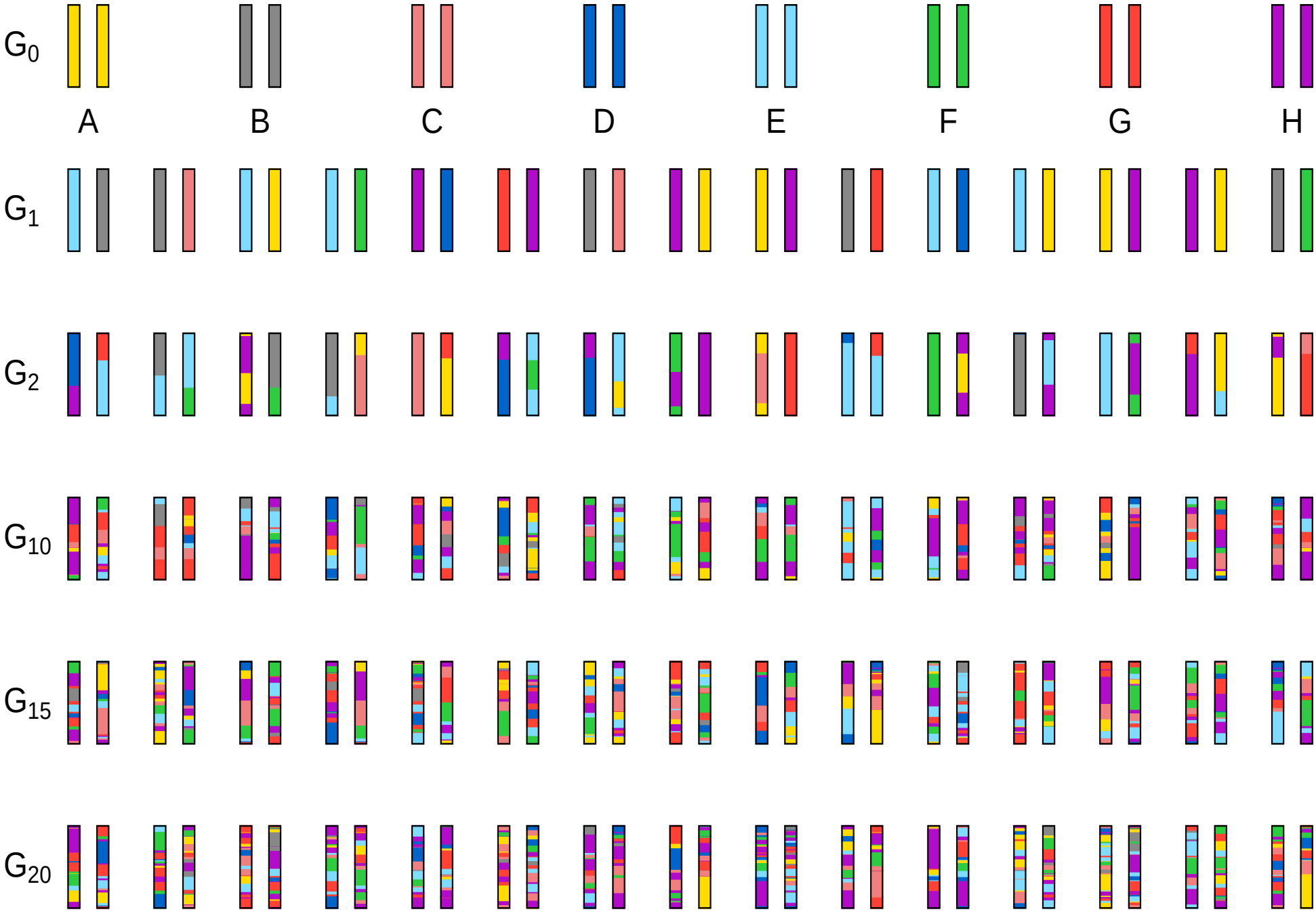
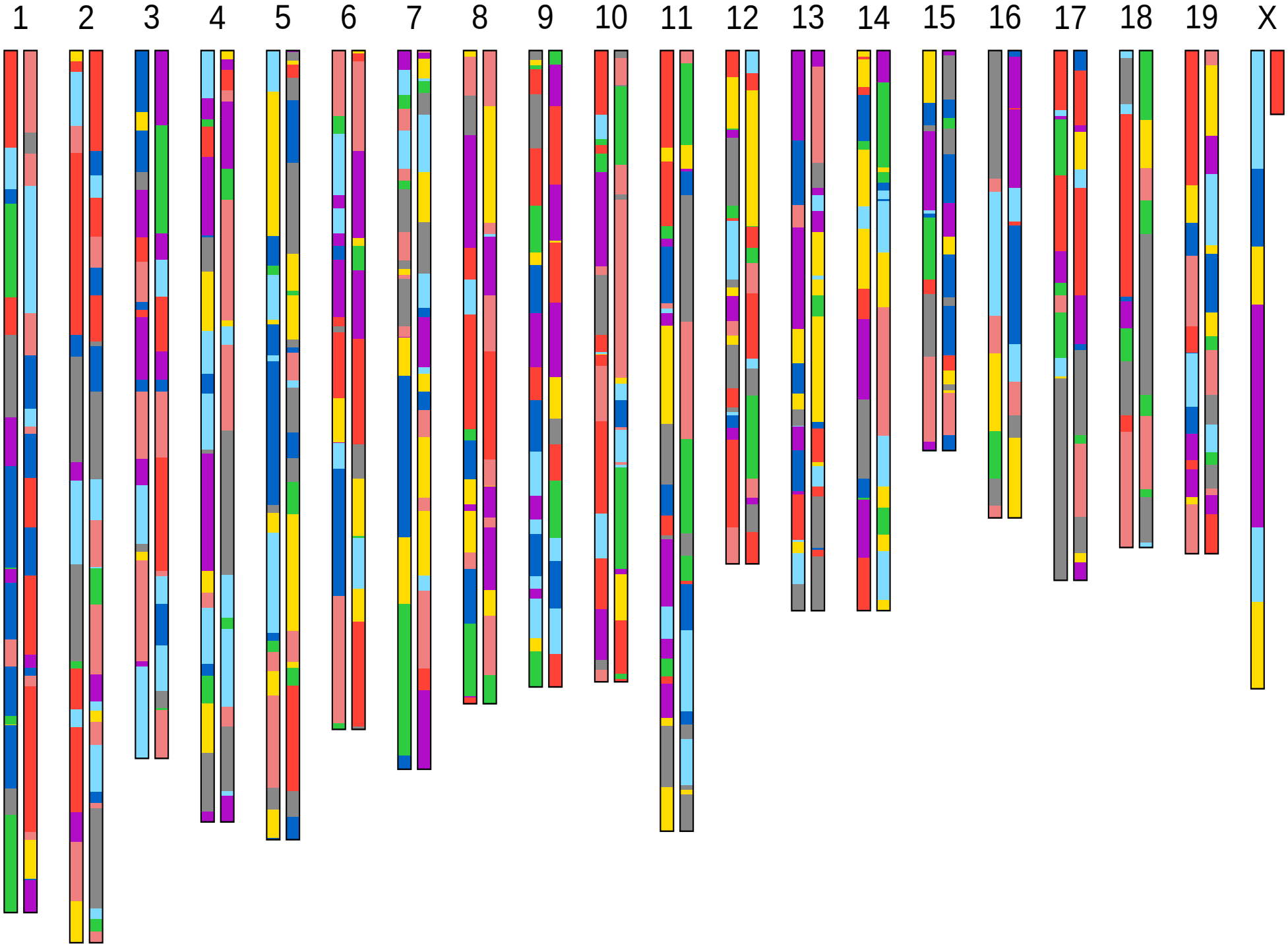
Genotype Reconstruction for Diversity Outbred Mice

A comparison of `r/qtl2` and `DOQTL`

John Spaw[1]
Karl Broman[2]

`github.com/JohnPSpaw`[1]
`github.com/kbroman`[2]


G0
A
B
C
D
E
F
G
H
G1
G2
G10
G15
G20


1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
X

Hidden Markov Model

Hidden Markov Model

Probabilities are generated from:

Hidden Markov Model

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1. Transition Model

Hidden Markov Model

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1. Transition Model
2. **Emission Model**

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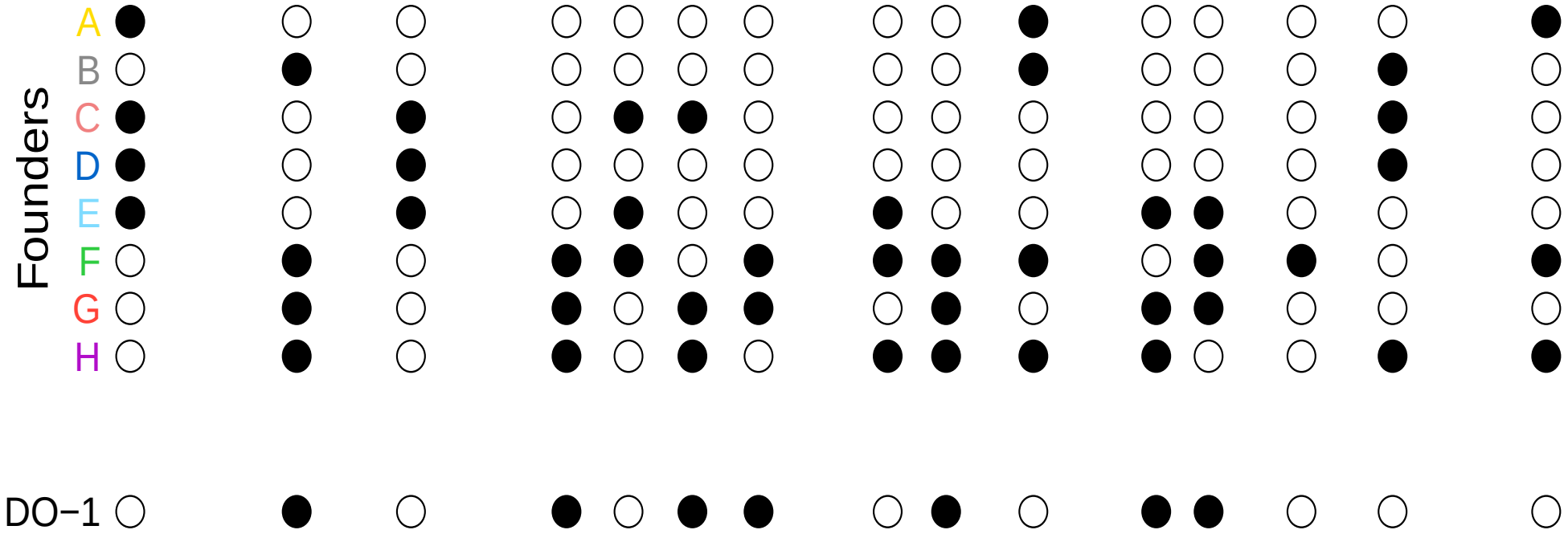
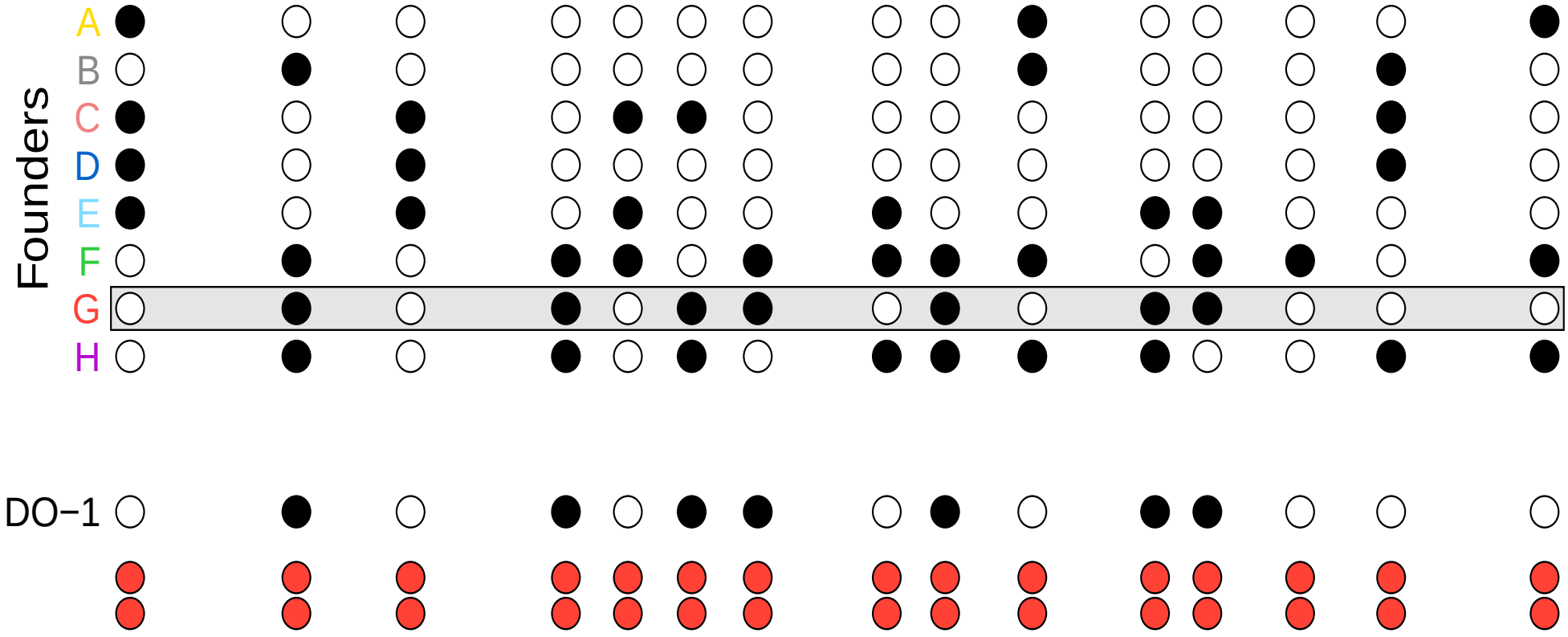
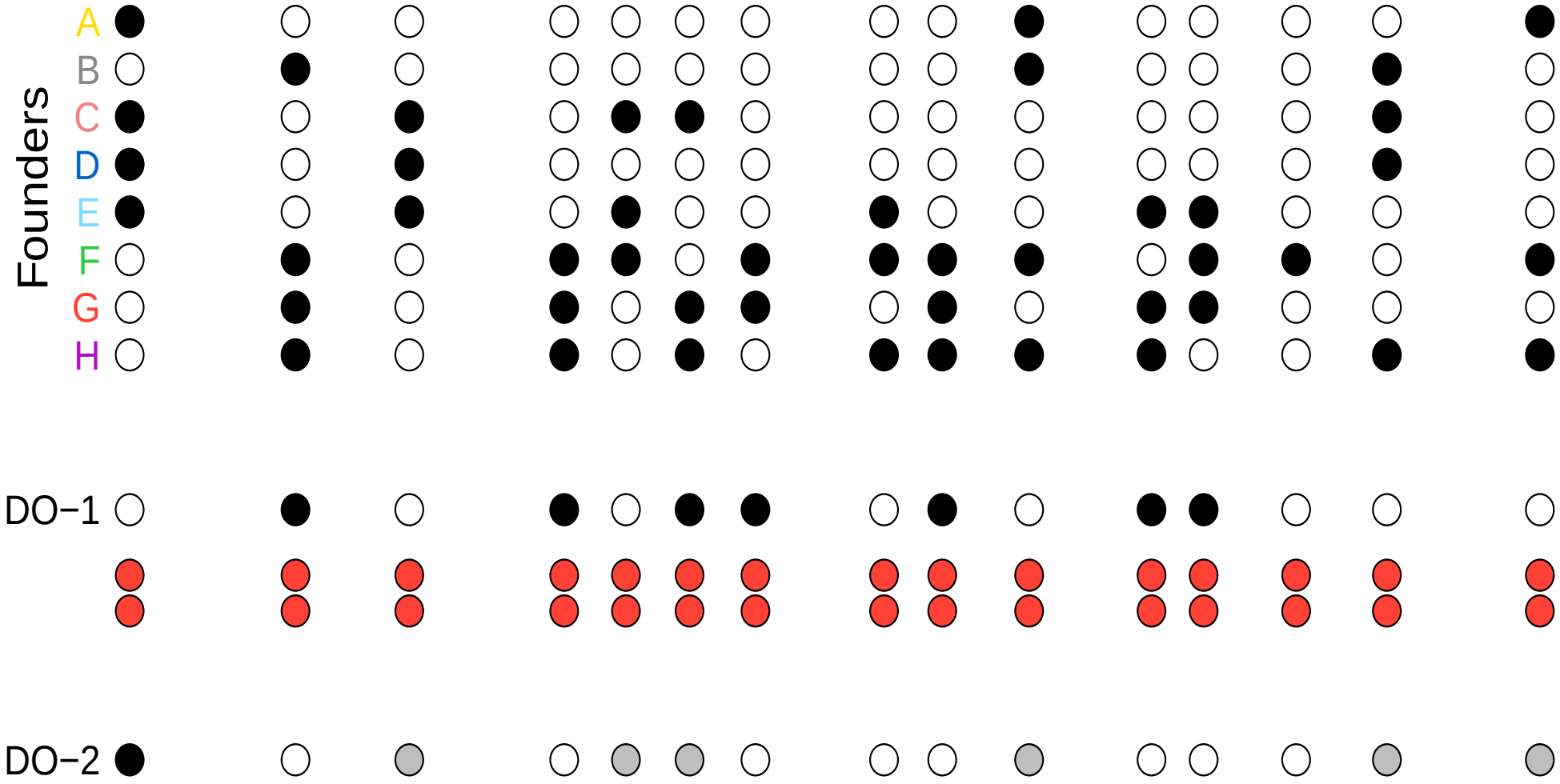
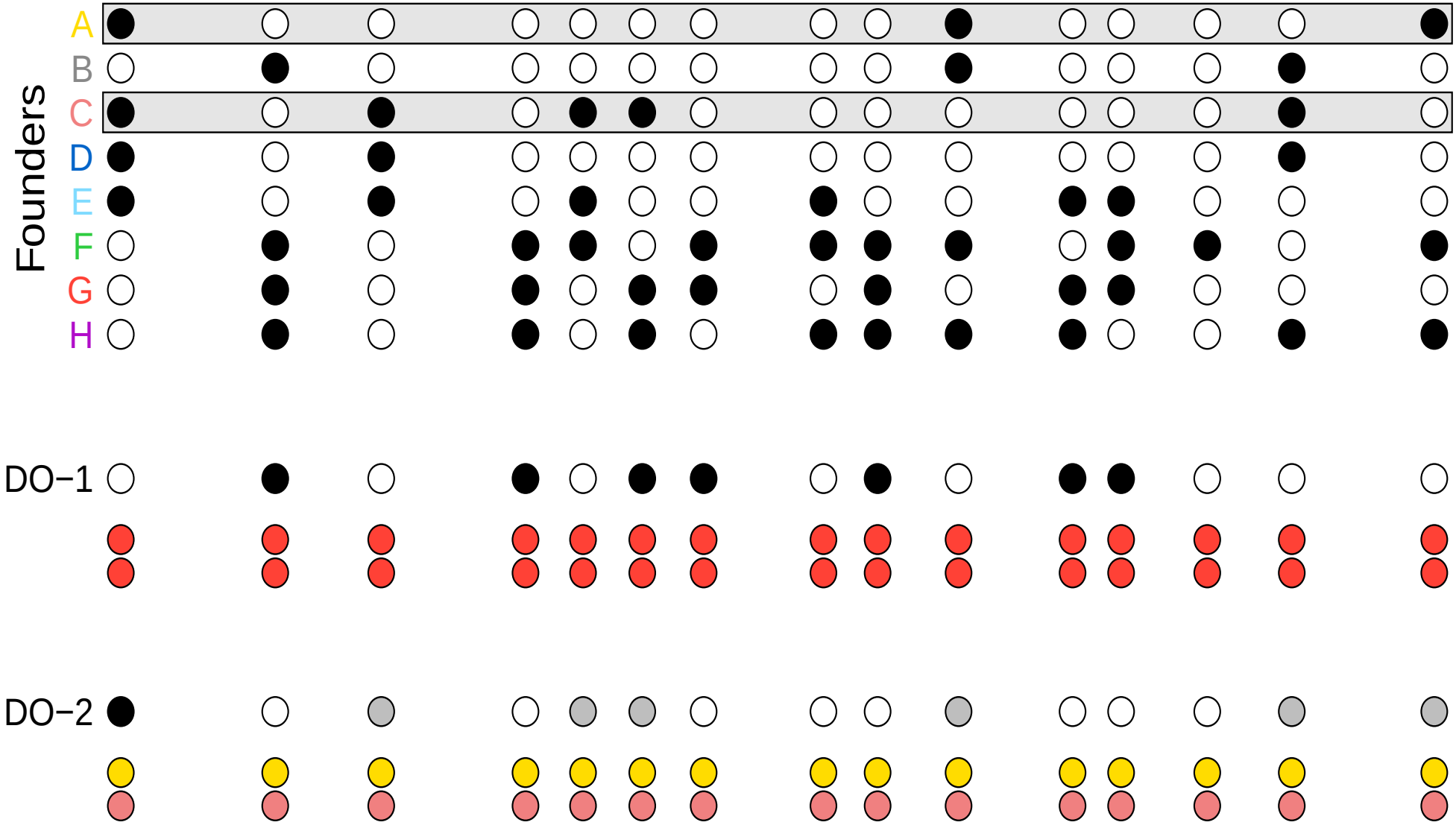
2. **Emission Model**

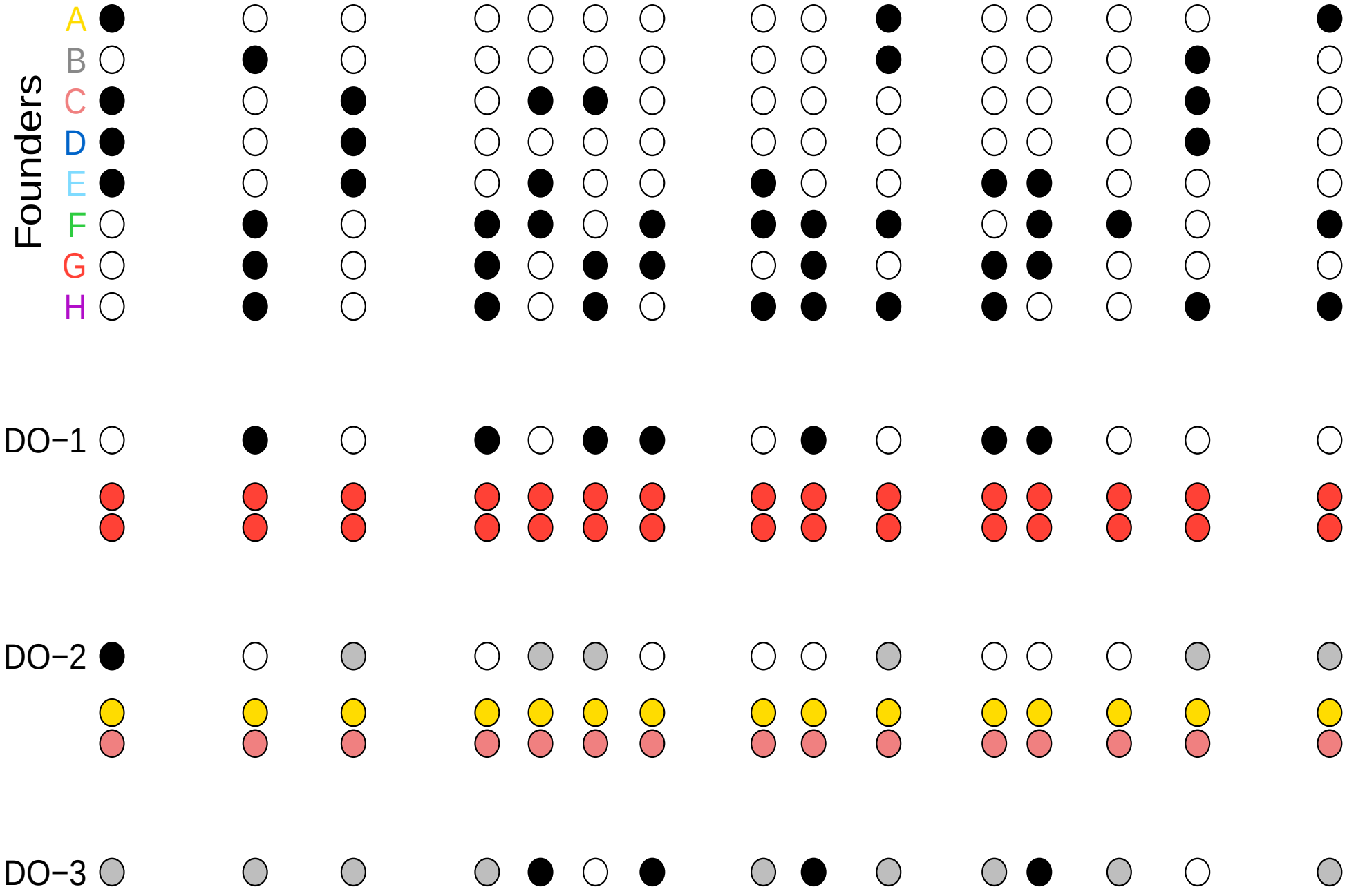
Conditional probability of observed data given underlying diplotype state

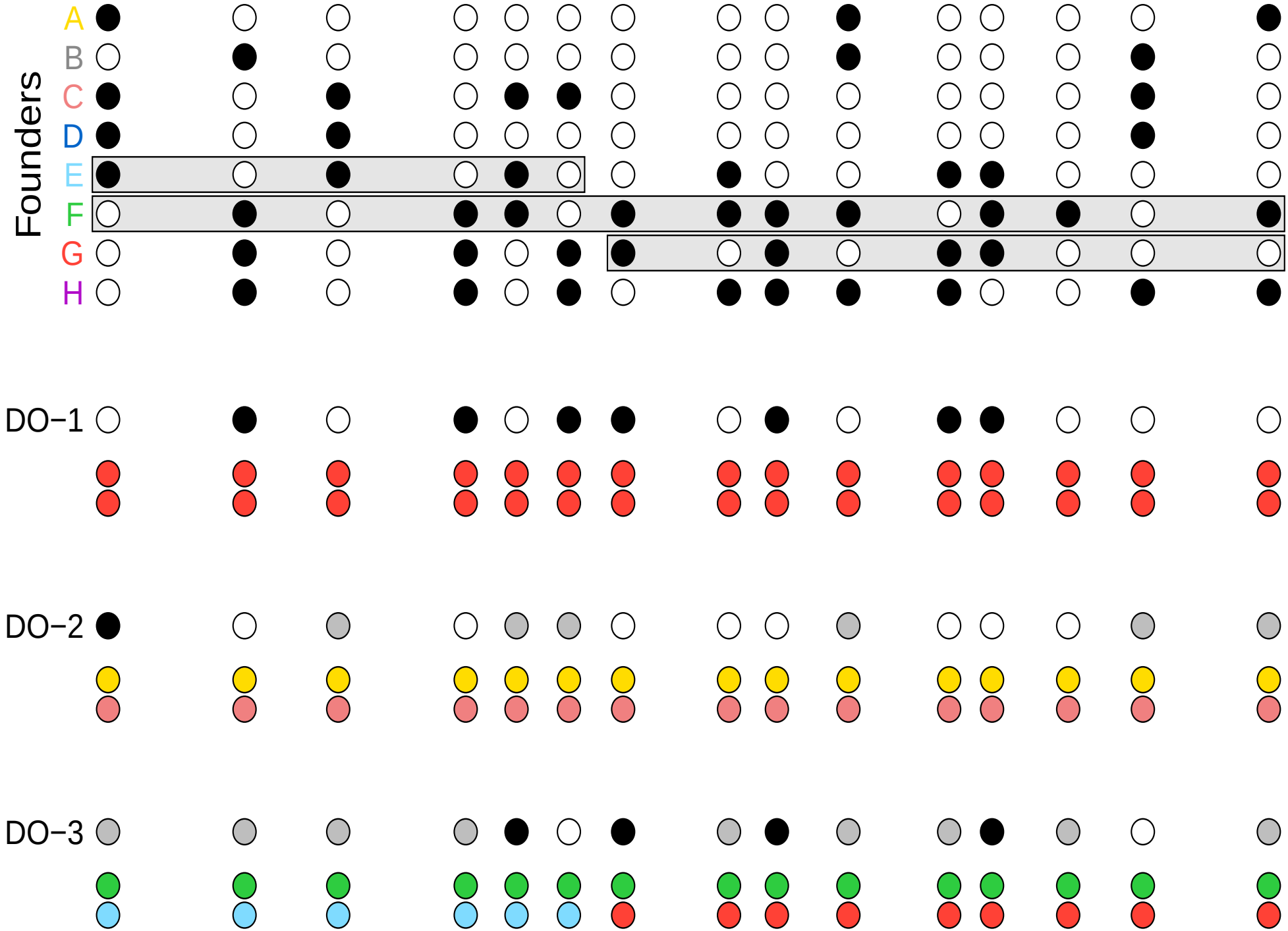

Founders
A
B
C
D
E
F
G
H
DO−1


Founders
A
B
C
D
E
F
G
H
DO−1


Founders
A
B
C
D
E
F
G
H
DO−1
DO−2


Founders
A
B
C
D
E
F
G
H
DO−1
DO−2


Founders
A
B
C
D
E
F
G
H
DO−1
DO−2
DO−3


Founders
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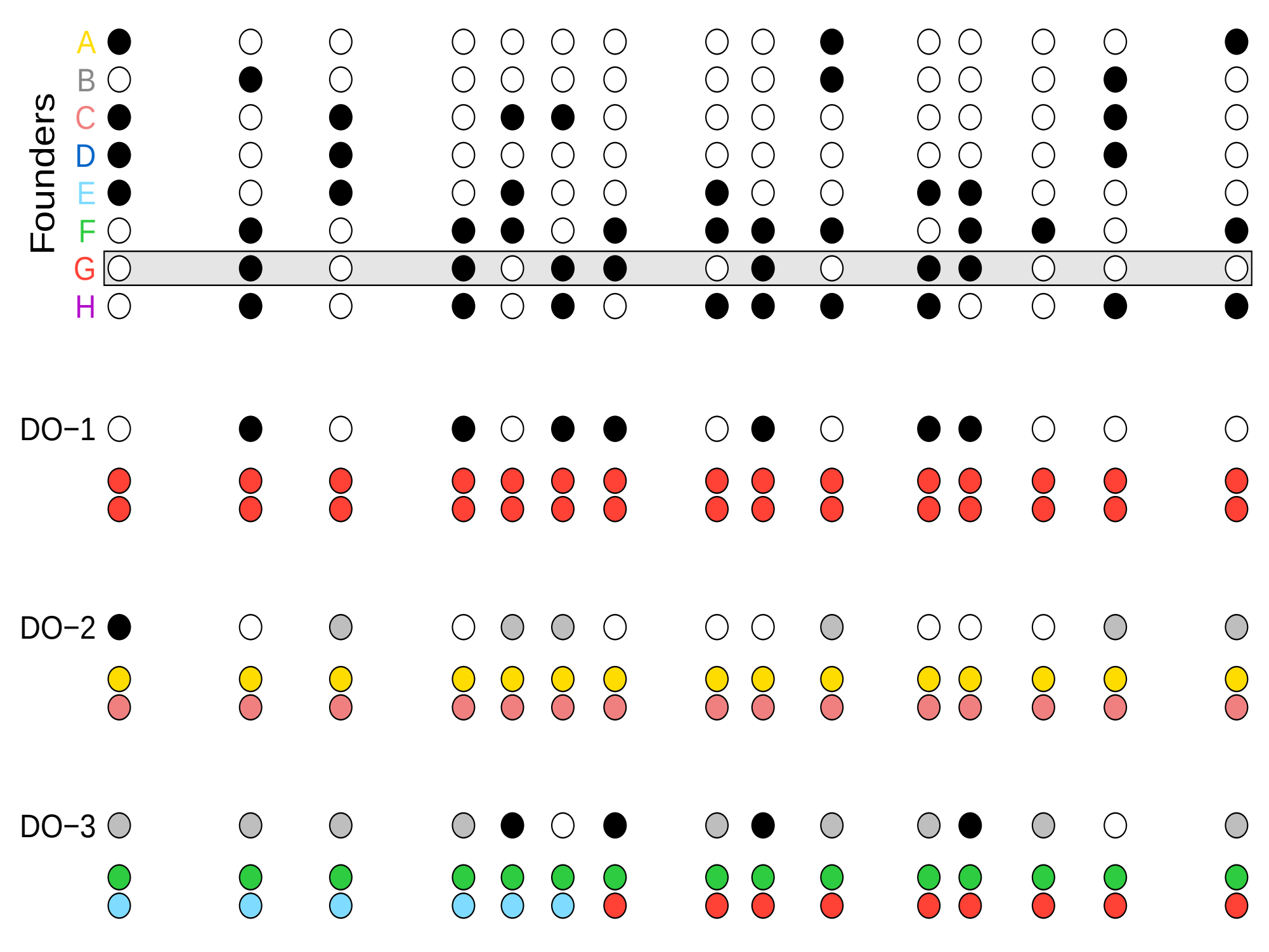
B
C
D
E
F
G
H
DO−1
DO−2
DO−3


Founders
A
B
C
D
E
F
G
H
DO−1
DO−2
DO−3

Data

Two *large* 3D arrays of emission probabilities

- `r\qtl2` (Broman)
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$500 \times 120,000 \times 36$ (Individual $\times$ Markers $\times$ Haplotype)

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How are they different (or the same)?

Inferred Haplotypes

INSERT INFERRED HAPLOTYPE IMAGE (BARS)

Measure of distance

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For each individual at each marker:

Compute *sum of absolute differences*

$$\sum_{i=1}^{36} |p_{1,i} - p_{2,i}|$$

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Each entry represents distance between methods