

# HMM in crosses and small pedigrees

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# Discrete-time Markov chains

Consider a sequence of random variables  $X_1, X_2, X_3, \dots$  with common finite **state space**  $S$ . This sequence forms a **Markov chain** if for all  $t$ ,  $\{X_1, \dots, X_{t-1}\}$  and  $\{X_{t+1}, X_{t+2}, \dots\}$  are **conditionally independent** given  $X_t$ , equivalently,

$$pr(X_t | X_{t-1}, X_{t-2}, \dots) = pr(X_t | X_{t-1}).$$

The matrix  $p(i, j; t) = pr(X_t = j | X_{t-1} = i)$  is the **transition matrix** at step  $t$ .

When  $p(i, j; t) = p(i, j)$  for all  $i$  and  $j$ , independent of  $t$ , we say the Markov chain is **time-homogeneous**, or has stationary transition probabilities. Many of the chains we'll be meeting will be inhomogeneous, and  $t$  will be in space, not time.

There are plenty of good books on elementary Markov chain theory, **Feller vol 1** being my favourite, but they mostly concentrate on asymptotic behaviour in the homogeneous case. For the time being we don't need this, or much else from the general theory, apart from the fact that multi-step transition matrices are **products** of 1-step transition matrices (**Exercise**).

# Hidden Markov Models (HMM)

If  $(X_t)$  is a Markov chain, and  $f$  is an **arbitrary function** on the state space, then  $(f(X_t))$  will **not** in general be a Markov chain.

**Exercise:** Construct an example to demonstrate the last assertion.

It is sometimes the case that associated with a Markov chain  $(X_t)$  is another process,  $(Y_t)$  say, whose terms are **conditionally independent given the chain**  $(X_t)$ . This happens with so-called semi-Markov chains.

Both functions of Markov chains and this last situation are covered by the following useful definition, based on the work of L. E. Baum and colleagues around 1970. A bivariate Markov chain  $(X_t, Y_t)$  is called a **Hidden Markov Model** if a)  $(X_t)$  is a Markov chain, and b) the distribution of  $Y_t$  given  $X_t, X_{t-1}, X_{t-2}, \dots$  depends only on  $X_t$  and  $X_{t-1}$ . In many examples, this dependence is only on  $X_t$ , but in some, it can extend beyond  $X_{t-1}$ , and/or include  $Y_t$ . Once you see how the defining property is used in the calculations, you will get an idea of the **possible extensions**.

**Exercise.** Explain how functions of Markov chains are always HMM. 3