



# GENETIC BRITAIN

Mapping the country's genes with the help of mathematics

genetic marker

chromosome

Punnett Square

frequency

probability

Hardy-Weinberg Law

statistical

Scientists at the University of Oxford have constructed a map of the UK showing different patterns in the country's genetic make up.

This will help reveal the likelihood of different populations being at risk from certain genetic diseases, along with tracing back ancient migration into the country.

To make the map, researchers collected 4500 blood samples from across the nation and recorded *genetic markers* at 600,000 different positions along participants' *chromosomes* – our DNA is constructed from 23 pairs of these chromosomes. One person's markers can then be compared to another's.

However, analysis of 600,000 markers for each of the 4500 participants is a big undertaking. Scientists have to be confident they've correctly measured the markers and that's where mathematics comes in.

We inherit our genetic information from our parents – half from our mother, half from our father. If Mum and Dad both have the genetic marker AB, you can inherit one of A or B from each parent, each with a *probability* of  $\frac{1}{2}$  just like heads or tails when tossing a coin. A table called a *Punnett Square* can be drawn to show the four possible outcomes, which each occur with the same probability of  $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ .

		Mum	
		A	B
Dad	A	AA	AB
	B	BA	BB

So, you can either be AA, AB, BA, or BB. But not everyone's parents have the same combination of markers. If both parents are BB then their child can't inherit an A. If A appears in the blood samples with a *frequency* of P, and B with a frequency Q (where  $P + Q = 1$ ), the probability of the participants of the study having inherited AA is  $P \times P$ . The table below shows the frequency with which you'd expect to find the different combinations in the blood samples assuming the As and the Bs combine at random.

Combination	Frequency
AA	$P^2$ (since $P \times P = P^2$ )
AB/BA (the same)	$2PQ$ (since $PQ + QP = 2PQ$ )
BB	$Q^2$ (since $Q \times Q = Q^2$ )

This illustrates the *Hardy-Weinberg Law*, which says that, if you assume mating happens at random, the proportions of the different combinations in the blood samples should be as above. The researchers can check, using some simple *statistical* tests, to see whether the relative frequencies of the combinations of each marker match this law. If a particular marker doesn't then they know that marker's results are unreliable and that it should not be used for any further analysis. For the map researchers ended up rejecting around 10% of markers because they didn't follow the Hardy-Weinberg Law closely enough. The actual construction of the map uses sophisticated statistical methods running on high performance computers.



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