

Founder mutations: evidence for evolution?

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Sometimes a mutation arises which is not eliminated by natural selection and is carried through time and space in that person's descendants for thousands of years, and in individuals dispersed over thousands of miles. This long-gone ancestor is known as the 'founder' of this population, and his or her genetic legacy is called a 'founder mutation'.¹

The victims of many genetic diseases die before reproducing, so natural selection stops the mutant genes from reaching future generations. In other cases, mutations may be neutral and/or recessive and they survive and pass on in single copies because it is only when the host is homozygous (both parents pass a copy to the child) that the condition may be detrimental. In a few rare cases, founder mutations have been discovered to spare their carriers by rendering some beneficial advantage. Is this evidence for 'molecules to man' evolution?

The International HapMap Project

The simplest kind of mutation is a change in a single nucleotide (called a point mutation). When we look across the genomes of whole populations of people, such mutations show up as 'single nucleotide polymorphisms' (SNPs)—that is, there are differences at that point in that particular gene. Those SNPs that are found to be statistically correlated are likely to be carried on the same chromatid (single strand of DNA) and are called 'haplotypes'. Haplotype information is extremely valuable in investigating the genetics behind common diseases because they can act as tracers for diseases associated with the mutations. For this reason they are being collected on a large scale by the International HapMap Project.² There are about 10 million SNPs in the human

genome. Not all of them are the result of mutation, for God has built into our genetic systems a natural ability to produce variation. But let's look at some examples.

Sickle cell anemia

One of the best known examples of a founder mutation is sickle cell anemia (figure 1), which occurs in regions infested with malaria in Africa and the Middle East. Two copies of a sickle cell gene doom the bearer to painful disease and a shortened life span. But a single copy of the faulty gene can help the carrier survive malarial infection. This is because some red blood cells become faulty, but not enough to cause disease, and when these cells become infected with the malaria parasite they rupture, preventing the parasite from completing its life cycle.³ This reduces the number of malaria parasites in the blood and gives the carrier a slight advantage over non-carriers and they tend to be naturally selected to survive.

The sickle cell mutation today can be found in five different haplotypes (figure 2), leading to the conclusion that the mutation appeared independently five times in five different founders.¹ But this 'beneficial' mutation can in no way be construed as a gain of new genetic information that might support 'molecules to man' evolution. It causes deformity and malfunction of hemoglobin molecules, and its 'benefit' is the result of the *interaction of two horrible diseases*.

Hereditary Hemochromatosis

Another founder mutation is Hereditary Hemochromatosis.¹ This is caused by a mutation on the HFE gene on Human Chromosome 6. In the

homozygous condition, the person's iron regulation system (figure 3) is disrupted and the body continues to absorb iron from food as if it was suffering from an iron deficiency. As a result, abnormal amounts of iron build up in various tissues and cause a wide range of disease symptoms, leading to multiple organ damage and then failure through unnatural iron deposition and overload, and ultimately leading to death.⁴

Studying the prevalence of this haplotype in various populations has led to the conclusion that the founder lived in Central Europe between 60 and 70 generations ago and was probably of Celtic origin. This mutation has been carried through space and time in that European's descendants to now include some 22 million Americans possessing at least one copy of the gene.

Why has such a debilitating condition survived for so long? It has been suggested that poor diet (i.e. low in iron) accompanied the spread of the mutation and carriers of only one copy of the gene were able to absorb more iron and were thus at a selective nutritional advantage over non-carriers.¹ However, once again there is no evidence of new information being added by the mutation. It disrupts

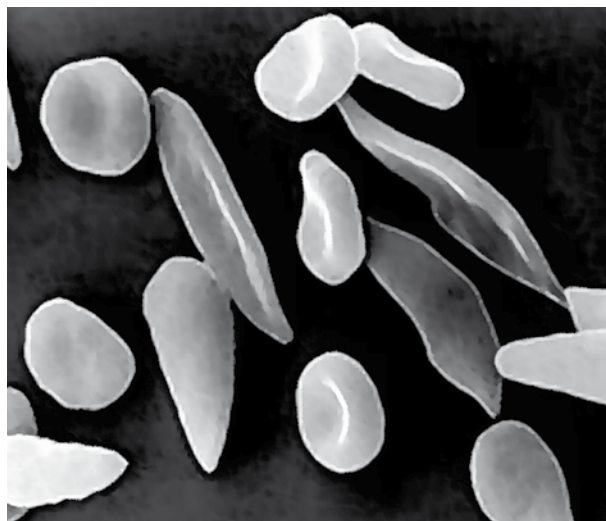


Figure 1. In sickle cell anemia, a mutation in the HBB gene, which codes for a hemoglobin subunit, changes amino acid glutamate to valine at position 6. This encourages binding between hemoglobin molecules and its polymerisation, deforming red blood cells into a sickle shape.

After wikipedia.org

the body's normal iron regulation mechanisms, and the advantage comes at the cost of disease and early death.

Factor V Leiden

Factor V Leiden is a mutation that disables the body's ability to dissolve blood clots in veins. As a result, when clotting begins, it continues

unchecked and can cause deep vein thrombosis, pulmonary embolism, miscarriage or stillbirth. This Founder mutation occurs in 4% of Europeans and a recent discovery may explain its persistence. In 2003, Bryce A. Kellin and his colleagues at the blood centre of South-East Wisconsin and the Medical College of Wisconsin

demonstrated that carriers of this mutation are more resistant to the lethal effects of bacterial infections in the blood stream than are non-carriers.¹ Since bacterial infection was a huge threat to survival in the pre-antibiotic past, and is still a cause of death today, the mutation seems to provide a competitive advantage—albeit at the cost of other disorders.

Once again, however, no new genetic information has been created. The amazingly complex blood-clotting mechanism is corrupted and the competitive advantage comes from an *interaction of diseases*—once again!

Conclusion

The study of founder mutations makes it possible to trace human migration pathways and survival patterns. But they are all examples of a breakdown and loss of the body's originally functional information, giving rise to a selective advantage only in a particular set of adverse conditions. In no instance is there an 'evolution' to a better and more advanced condition, with gain of new information and new capabilities. Indeed, the entire focus of the International HapMap Project is upon disease and disease prevention and cure—there is no evidence in their study of mutations that they cause 'health'. We could hardly wish for better evidence that mutations produce nothing new and have no power to transform microbes into microbiologists.

References

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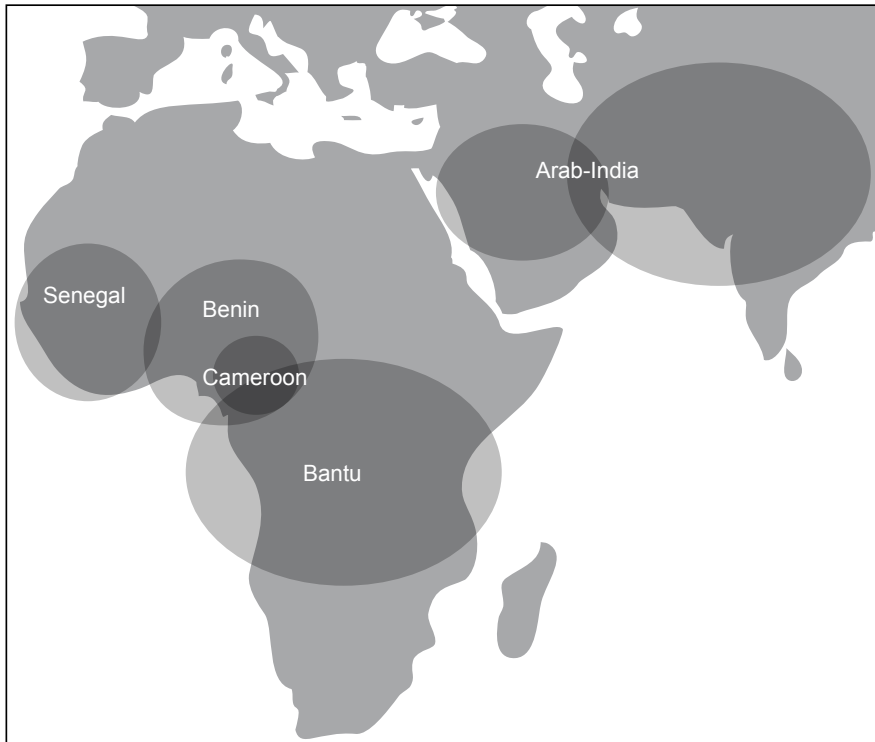


Figure 2. Global geographic distribution of five distinct haplotypes of sickle cell disease. (After Drayna¹).

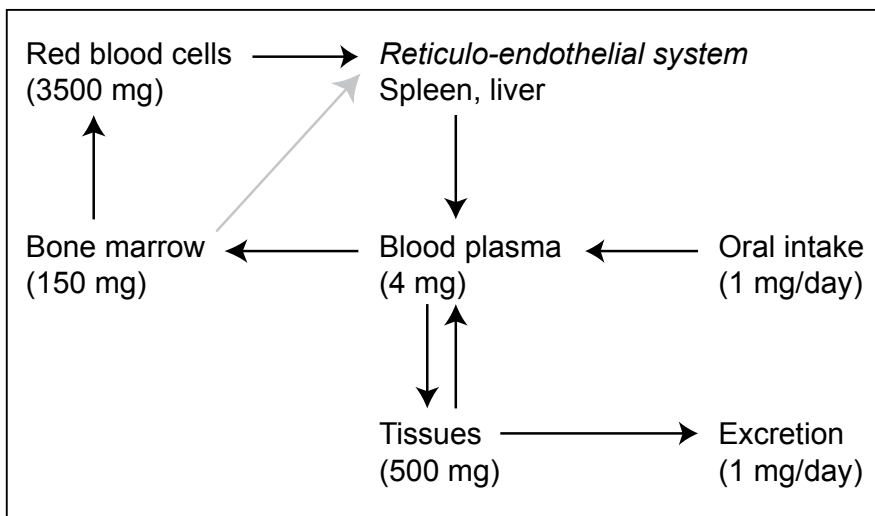


Figure 3. Human iron regulation system.