Milk drinking: in our genes?

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(PhysOrg.com) -- A new study led by UCL scientists has found that current genetic data cannot explain why vast swathes of the world can digest milk.

The ability to digest the milk sugar lactose - also known as lactase persistence - is a selectively advantageous and recent evolutionary genetic trait, which emerged about 7,500 years ago in Europe and probably later in other parts of the world. This means that, once weaned, people in most parts of the world (large parts of Africa, most of Asia, and Oceania) cannot digest milk for the rest of their life.

However, the study published in BMC Evolutionary Biology, shows that the four genetic mutations currently associated with the ability to digest

milk cannot explain why many people in western and southern Africa, south eastern Europe, the Middle East, and southern and central Asia are able to digest milk. It also suggests that other genetic variants leading to the ability to digest milk exist, but have not yet been discovered.

Study co-author Dr Yuval Itan (UCL Genetics, Evolution and Environment) explains more fully what this study reveals.

Q: Can you explain 'lactase persistence' in lay terms?

Lactase is the enzyme that breaks the lactose into two units that are digestible. It is produced by a gene and is active in human (and most mammalian) infants. Most human (and mammalian) adults cannot digest the sugar milk lactose after weaning. The minority of the human world population, mostly those from pastoralist background, is lactase persistent.

Lactase persistence is an evolutionary new trait, as shown by a collaborative study demonstrating that lactase persistence was absent in ancient Neolithic bones in Europe. A recent UCL study 'The origins of lactase persistence in Europe' showed that lactase persistence and dairying gene-culture co-evolution was likely to have originated in Europe about 7,500 years ago, in Central Europe/Northern Balkans, in the Linearbandkeramik culture.

However, the trait was likely to have evolved independently in Africa and in the Middle East since in these regions there are different associated genetic variants. Lactase persistence has a very high selective advantage, probably because of the various nutritional benefits that it gave during the harsh Neolithic times.

What were the aims of this study and how does it support previous studies undertaken in recent years?

Human lactase persistence has been thoroughly investigated in recent decades. Recent advances in genetics allowed specific genetic mutations to be associated with the lactase persistence trait.

However, we noticed that for some populations genetic data is insufficient to explain lactase persistence, mostly due to under prediction of the trait. For example, the Wolof ethnic group in Senegal have 51% frequency of lactase persistence, but when predicting their lactase persistence by their lactase persistence associated genetic data, it would appear that their lactase persistence is 0%. This suggested that there are more genetic variants associated with the ability to digest milk sugar that are waiting to be discovered.

The main objectives of this study were to investigate the correlation between the lactase persistence trait and its genotypes (the various mutations that explain the trait) worldwide in order to identify regions where lactase persistence genetic data is currently insufficient to explain lactase persistence, and in that way to suggest regions where genetic studies on local populations are likely to reveal new lactase persistence associated genetic variants.

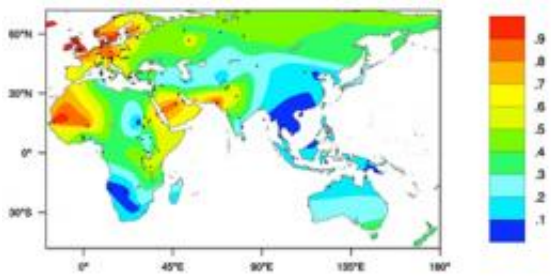
We used and extended the database of lactase persistence that was first described in the UCL study 'Lactose digestion and the evolutionary genetics of lactase persistence' and extended the novel statistical correlation methodology that was developed for another UCL study to check in which geographic regions the lactase persistence phenotype can be explained by the lactase persistence genotypes. We have presented the GLAD (Global Lactase Persistence Association Database), as well as visually depicting in maps the Old World distribution of lactase persistence phenotype, genotypes and the correlation between the two.

What was the most surprising aspect of the research?

The vast extent of the regions where genetic data cannot explain lactase persistence was surprising. A good example is West Africa where genetic and phenotypic data of lactase persistence is available and there is a lack of correlation between the two. This is also very exciting, because performing further genetic studies in this region is likely to reveal new lactase persistence genetic variants that are yet unknown.

What does this study tell us about lactase persistence in global terms?

This study shows the frequency of lactase persistence across hundreds of different populations around the world, and it can be clearly seen in the map (see Figure 1 below). It also tells us to exercise caution when attempting to use genetic data only for predicting lactase persistence in individuals across the world - as we have shown, it will be a long time before genetics can fully replace physiological tests of lactase persistence (such as breath hydrogen or blood glucose), because in various populations the genetic variants associated with lactase persistence have not yet been characterised.



The frequency of lactase persistence across world populations

How do your findings relate to the notion that milk drinking is healthy?

In my personal opinion, it seems that milk drinking is shown in the media as the healthy state of adults and children, while the fact is that the majority of humans are healthy individuals who cannot digest milk, and this shouldn't be conceived as a problem.

Milk-related or processed milk products (such as yogurt and cheese) usually have a lower proportion of lactase than raw milk, so in some cases, individuals that are non-lactase persistent could digest them, and a clinician or a nutrition expert could give a good individually tailored advice on this matter.

Based on the conclusions of this study I would recommend that individuals who want to test if they are lactase persistent should take the traditional physiological tests rather than the genetic test, because it is currently not sufficient for many populations.

More information: Paper link: www.biomedcentral.com/1471-2148/10/36

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