

Abstract

Lactase persistence (LP) is genetically determined ability to digest lactose (milk sugar) in adulthood. Lactose is digested by the intestinal enzyme – lactase. In mammals, production of lactase declines after the weaning period. In case of the LP, the lactase is produced during whole life and thus it is possible to drink larger amounts of fresh milk without digestive problems. However, LP is documented only in some human populations. Its worldwide distribution is unequal, the highest rates are found in northern Europe and in pastoralists living in arid regions of Africa and Arabia. The origin of this trait is related to the emergence of agriculture 10,000 years ago, and the beginnings of milk production. In some populations, there is a strong signal of positive selection for the LP caused by various factors depending on various living conditions. The LP is associated with single nucleotide polymorphisms (SNPs) located approximately 14 kb upstream from lactase gene. Nowadays we know five different SNPs which cause the LP and they have probably different origin and have expanded independently.

One of the possible centres of the LP origin is the Arabian Peninsula, where the LP variant –13,915*G probably first appeared and spread. In this thesis, occurrences of different LP mutations were investigated in the populations of southern Arabia and African Arabs. In all analysed datasets, the presence of the –13,915*G has been confirmed. The frequency of the –13,915*G in southern Arabia was higher than 50% in most of analysed datasets. Higher incidence of this mutation (44.5–76%) was recorded in the African Arabs who raise camels, compared to the African Arabs, who switched to the cattle farming or sedentary agriculture (1.4 to 25.5%). The results support the hypothesis formed in previous studies suggesting that the –13,915*G was at first spread in Arabia probably in connection of the camel domestication. The estimating of the age and expansion of the –13,915*G is consistent with the migrations from Arabia to Africa which started in the 7th century AD.