Primary hypolactasia

- **Congenital hypolactasia or congenital lactase deficiency** is determined by an autosomal recessive alteration of the region responsible for regulating the lactase gene. It is characterised by an important reduction in lactase from birth which continues during the patient’s lifetime. The diagnosis is based on the selective and permanent deficit of the lactase activity and histological normality of the intestinal mucosa. It is a rare disorder, with very few cases reported on a worldwide scale. If not diagnosed and treated quickly, it is fatal (1).

- **Acquired primary hypolactasia or primary lactase deficiency** is correlated with the development of certain polymorphisms in the region responsible for regulating the lactase gene which appear to be associated with lactase persistence. Adult-type hypolactasia is the most common type. This type of hypolactasia is progressive and develops a few years after birth. Its prevalence varies, depending on the ethnic origin, and the time or age at which the enzymatic activity starts to decline seem to be genetically determined by ethnic factors (2).

Epidemiology of acquired or primary hypolactasia

Generally speaking, the epidemiological studies conducted are focused above all on ascertaining the epidemiology of hypolactasia or lactose malabsorption due to a lactase deficit.

In Europe, the area with the lowest incidence of hypolactasia is north-west Europe, the region near the North Sea and parts of the south-western area of the Baltic. The area in which this condition is most prevalent is southern Europe and parts of eastern Russia. In Greece, it has been determined that 38-45% of the population suffers from hypolactasia, with this figure being 18-52% in Italy and 37-47% in France (Figure 1).

Populations which are accustomed to drinking milk from animals have fewer cases of hypolactasia. In this regard, on comparing the areas of the world that “consume milk” and those that do “not consume milk” with the lactase persistence and hypolactasia map, it can be seen that the areas where milk is not consumed are those where there is a deficit of the enzyme and vice versa (3).

Figure 1. Prevalence (%) of adult-type hypolactasia in different European countries (4).

In addition, several studies have been conducted in Spain regarding the prevalence of hypolactasia in different regions. A prevalence of 28% has been found in Valencia (5), of 32.5% in Galicia (6), of 13-15% in Barcelona (7), of 36% in a study conducted on a small village of elderly people in Madrid (8) and of 17% in a population of university students in Navarra (9).


3. Rodríguez Martínez D, Pérez Méndez LF. Intolerancia a la lactosa. Rev Esp Enferm Dig (Madrid) 2006; 98(Suppl 2):143.


