Lactase insufficiency (lactose intolerance) is a condition that is characterized by the inability of the child's organism or adult person to digest lactose because of inadequate lactase enzyme production in the intestine. Genotyping of polymorphisms -13910C / T and -22018G / A of LCT lactase gene is considered as a genetic test in Finland and other European countries. T-allele in position -13910 bp. and the G-allele at position 22018 bp. The lactase gene (LCT) is associated with lactose intolerance syndrome, as they are mutant variants of the wild-type alleles -13910C and -22018A. Lactose intolerance is controlled as an autosomal dominant trait and is related to a population that traditionally practices domestication of cattle and in populations where dairy products form the bulk of the daily diet. Intolerance to lactose and milk can lead to a decrease in calcium intake and, thus, an increase in risk factors for osteoporosis.

The aim of our study is to estimate the frequency of lactose intolerance and to determine the genetic polymorphisms associated with the syndrome of lactose intolerance among the population of Kazakhstan.

Materials and methods: The study included school children of NIS of Semey and students of ENU, Astana, Kazakh nationality, aged 15 to 20 years. All participants were questioned regarding symptoms of intolerance to dairy products, a lactose tolerance test were done. Buccal epithelium was taken and genomic DNA was extracted from buccal epithelium cells. Polymorphism of the LCT-13910C/T and -22018G/A lactase gene was determined by PCR and direct sequencing. An analysis of the phenotype-genotype association was conducted to assess the frequency and characteristics of the syndrome of lactose intolerance among Kazakhs.

Results. The study included 90 people. All participants were tested for tolerance to lactose. 27.8% (25/90) of participants demonstrated lactase resistance, expressed as an increase in blood glucose after taking lactose, whereas 72.2% (65/90) showed a slight increase in glucose in the blood, indicating an inadequate function of the enzyme lactase. The mean blood glucose concentration and standard deviations for both lactase persistence and lactose intolerance to (0 min) and after taking lactose (20, 40, 60 and 90 minutes) were higher when measured after 20 minutes. For patients with lactase persistence, the mean difference for blood glucose after lactose intake was 2.02±0.8 mmol/L, whereas for patients with lactase deficiency, the glucose difference level was 0.7±0.35 mmol/L. DNA samples from all 90 subjects were genotyped for the polymorphisms -13910 C/T and -22018G/A of the LCT gene. Polymorphism -22018G/A did not correlate with the preservation of lactase, which makes polymorphism -13910 C/T the only SNP associated with lactase in the population of the Kazakh population. Analysis of the phenotype-genotype association made it possible to calculate lactase persistence and lactase deficiency rates and their relationship to the corresponding alleles of the -13910 C/T polymorphism of the LCT gene. Thus, 25 (27.8%) of the total number of 90 participants had a mutant type T-allele and demonstrated an increase in glucose level after consumption of milk (persistence of lactase), while 65 (72.2%) were carriers of the wild-type C-allele and not showed changes in blood sugar levels after consumption of milk (lactase deficiency). In 24.4% (24/90) of the examined subjects, the CT/TT genotype was identified at position 13910 bp upstream of the LCT gene sequence, whereas the wild type CC genotype was detected in 75.6% (68/90) subjects. The CT/TT genotype corresponds to lactase persistence, and CC is a wild-type genotype that corresponds to lactase deficiency, was found in 63.3% of those surveyed who showed symptoms of lactose intolerance. Thus, 65 (72.2%) patients had lactose intolerance syndrome and wild type CC genotype was detected in 75.6% (68/90). Associations of the phenotype of lactose intolerance with genetic variants of polymorphism -13910 C/T of the LCT gene were noted.