Folate or vitamin B9 is an essential water-soluble nutrient that takes part in important cellular processes. These include amino acid metabolism, methylation, and nucleotide synthesis, the last two of which play a key role in early embryonic development. It was approximately 30 years ago when folic acid supplementation was confirmed to help prevent embryonic neural tube defects and since then an intake of 400 µg of folate a day has been recommended to all women of childbearing age. Despite this, folate deficiency is a relatively common phenomenon and during pregnancy can lead to congenital malformations and pregnancy complications. This bachelor thesis summarizes the latest findings on the association of neural tube defects and miscarriages with single-nucleotide polymorphisms in genes encoding folate metabolism enzymes in humans, focusing mainly on the gene encoding the enzyme 5,10-methylenetetrahydrofolate reductase (MTHFR). Furthermore, this thesis deals with potential mechanisms of the folate’s protective effect which could participate in the prevention of neural tube defects and are studied mainly on model organisms.

**Key words:** folate, embryo, neural tube defects, MTHFR, homocysteine, miscarriage, single-nucleotide polymorphisms