

The term gene was coined in 1909 by Wilhelm Johannsen to designate theoretical unit of genetic analysis. Concept of the gene has changed over time due to progress in genetics and its experimental practice. Currently used molecular concept is based on linear correspondence between DNA sequence and its product (Waters 2007). It also describes gene as discrete physical structure – DNA segment with clearly defined boundaries. But the investigation of wide range of genomes has put the molecular concept of gene into doubt.

The aim of PhD thesis is to specify the term gene by employing mechanical, semantic and hermeneutic interpretation (Payne 2002, 101-124). It summarizes history of the gene and presents some of the biological findings that undermine the molecular concept. In biological context the gene is always connected to DNA molecule. But it can be shown that DNA functions only as a mediator or carrier, and the substance of gene is not material but it has a character of information. Therefore in the semantic view the gene is information containing instruction for development of phenotypic trait which is specified by medical or other theoretical interest. Hermeneutical interpretation of the gene can be point out in clinical genetics. Here the gene is also information containing instruction for development of the phenotypic trait. In some cases the information can be read during genetic testing. Gene information has to be then translated during the process of genetic counseling into the information understandable for patient. Only translation of the gene information into patient's language is relevant for his health decisions. Clinical geneticist has an interpreter role in process of genetic testing.