Module name	Molecular basis of genetic diseases diagnostics
Module code	B-BA.BTM.060
ISCED code	0511: Biology
Study cycle	II^{o}
Semester	summer
Responsible for this module	Małgorzata Marczak Department of Genetics and Microbiology email: malgorzata.marczak@poczta.umcs.lublin.pl Barbara Michalec-Wawiórka Department of Molecular Biology email: basiam@hektor.umcs.lublin.pl
Language of instruction	English
Prerequisites	completed courses in genetics and genetic engineering
ECTS	5
ECTS points hour equivalents	Contact hours (work with an academic teacher) – 60 - lectures: 30 - labs: 30 Non-contact hours (students' own work) – 65 - preparation for partial tests and exam: 25 - preparation for labs: 20 - literature study: 20 Total number of ECTS points for the module – 5
Learning outcomes verification methods	Lecture: - written examination (W1-W4, U1-U2, K1-K3) Laboratory: - partial tests (W1-W4, U1-U4, K1-K4)

Course full description	**The lecture covers the following topics:** types of inheritance of genetic diseases and risk level determination; molecular and cytogenetic methods used in the diagnostics of genetic diseases; monogenic and complex diseases; epigenetic diseases (basics of epigenetics, mechanisms of epigenetic disorders, epigenetic regulation of gene expression in the CNS during cell development and differentiation and in pathological states, methods of examination of epigenetic changes, epitherapies and projects to explore the human epigenome); the molecular basis of ribosomopathies: symptoms, diagnostics and therapies of selected ribosomopathies, the ribosomopathy paradox; mitochondrial diseases; cancer; interpretation and evaluation of genetic tests; latest achievements of genetics used in medical diagnostics and therapy; ethical aspects of molecular diagnostics. **The laboratory covers the following issues:** genetic diagnostics, indications for performing a genetic test, genetic test variants; molecular pathology (differences between dominant and recessive diseases at the molecular level, mutations such as loss of function, gain of function, acquisition of new properties and mutations associated with heterochronic or ectopic gene expression) in recessive and dominant inheritance; selected examples of monogenic diseases and molecular mechanisms leading to dysfunction; phenomenon of heterogeneity in genetic diseases - sources and importance for the successful diagnosis of genetic disease; algorithm in diagnostics: from phenotype to identification of the molecular basis of the disease; examples of molecular diagnostics results; fundamentals of chromosome syndrome diagnostics; modern techniques for identifying proteins - potential markers or pathogenesis.
Bibliography	R.L. Nussbaum, McInnes R.R., Willard H.F. Genetics in Medicine. Elsevier, 2007.

Learning outcomes	**Knowledge** W1. Student knows and understands various types of inheritance of genetic diseases and understands the level of risk in these diseases W2. Student knows and understands the differences between genetic diseases at the molecular level and the sources of heterogeneity in genetic diseases W3. Student knows and understands the possibilities of using the achievements of genetics in medical diagnostics W4. Student knows and understands modern techniques used in genetic diagnostics **Skills** U1. Student is able to propose a diagnostic procedure: from phenotype to molecular recognition U2. Student is able to choose the appropriate molecular techniques to verify clinical diagnosis or screening tests U3. Student is able to perform basic diagnostic tests in the field of detection of point mutations and screening tests U4. Student is able to interpret the result of molecular analysis in a specific diagnostic context **Social competence** K1. Student is ready to critically evaluate the value of diagnostic tests K2. Student is ready to recognize the importance of the heterogeneity of genetic diseases for the success of diagnosis K3. Student is ready to recognize the importance of molecular diagnostics in the diagnosis of genetic diseases K4. Student is ready to behave ethically in molecular medical diagnostics
Teaching methods	Lecture: information lecture, multimedia presentation, animation, discussion, case study Labs: experiment, direct observation, discussion