1. Module identification code.	
Name of the institution:	Universidad Autónoma de Nuevo León
Name of school:	School of Medicine
Name of the degree program:	Clinical Chemistry
Name of the course (learning unit):	Genetics
Total number of class hours - theory and/or practice:	40
Class hours per week :	2
Independent study:	20
Course modality:	Face to face instruction
Module level:	5th semester
Core/elective module:	Core
Curriculum area:	ACFP-F
UANL credits points:	2
Create date:	January 15th, 2017
Date of last amendment made:	July 20th, 2024

Person(s) responsible for design and amendment of the module:	PhD. Ma. del Roble Velasco Campos PhD. Geovana Calvo Anguiano MD. Luis Daniel Campos Acevedo

2. Presentation:

This module (learning unit is developed in three phases:

The first phase focuses on understanding the types of cell division, mitosis and meiosis, gametogenesis, as well as the mechanisms behind chromosomal abnormalities, so that the laboratory methods used for the identification of chromosomal diseases can subsequently be described.

In the second phase, two specialized topics are analyzed: 1) The biochemical basis of non-invasive prenatal diagnosis, which establishes the parameters used in the laboratory, and invasive prenatal diagnosis, including cytogenetic and molecular methodologies. 2) Inborn errors of metabolism, where, based on their biochemical foundations, laboratory techniques for their diagnosis will be identified, concluding with the PIA (Integrative Assessment Project), which includes a group research project on an assigned genetic disease that presents the disease background and the appropriate methodology for its diagnosis.

In the third phase, the mechanisms of inheritance transmission are analyzed, which can be classified into two groups: Mendelian, which includes autosomal dominant inheritance, autosomal recessive inheritance, X and Y-linked inheritance, and multifactorial inheritance; and Neo-Mendelian, which includes mitochondrial inheritance, triplet expansion, uniparental disomy, and epigenetics. Molecular methods will also be identified, along with their foundations and advantages in the diagnosis of hereditary diseases included in this phase.

3. Purpose(s)

This learning unit aims to contribute to both professional and human formation by promoting the understanding of the conceptual foundations of Genetics. This will enable graduates to substantiate many of the analytical procedures in clinical laboratories, aiding in the diagnosis of genetic or chromosomal disorders, monogenic diseases, complex diseases, and metabolic disorders across different matrices.

Regarding general competencies, students will be able to use their native language effectively in both oral and written forms, with relevance, timeliness, and ethics, adapting their message to the context to convey their ideas when describing genetic foundations and applying new technologies in the laboratory. They will also develop an attitude of commitment and respect towards the diversity of social and cultural practices that affirm the principle of integration while solving integrative research assignments; and achieve the adaptability required in social and professional environments by working in various situations and types of classrooms.

Furthermore, students will review literature in English as a second language, developing language skills from everyday contexts to professional settings in a globalized environment. There is a connection with learning units from previous semesters, such as Cell Biology, where knowledge of cell structure and function is applied; Morphological Sciences, utilizing the foundations of early development and gametogenesis; Pathology, analyzing the origins and genetic factors of diseases; and Biochemistry, applying knowledge of molecular properties. The learning units on Clinical Pathology and Cytogenetic Diagnostic Tools provide a foundation for understanding genetically derived diseases.

4. Competencies of the graduate profile:

General competencies that this learning unit contributes to:

Instrumental Competencies:

4. To master their mother tongue orally and in writing with correctness, relevance, timeliness and ethics, adapting their message to the situation or context, for the transmission of ideas and scientific findings.

Personal and Social Interaction Competencies:

9. To maintain an attitude of commitment and respect towards the diversity of social and cultural practices that reaffirm the principle of integration in the local, national and international context in order to promote environments of peaceful coexistence.

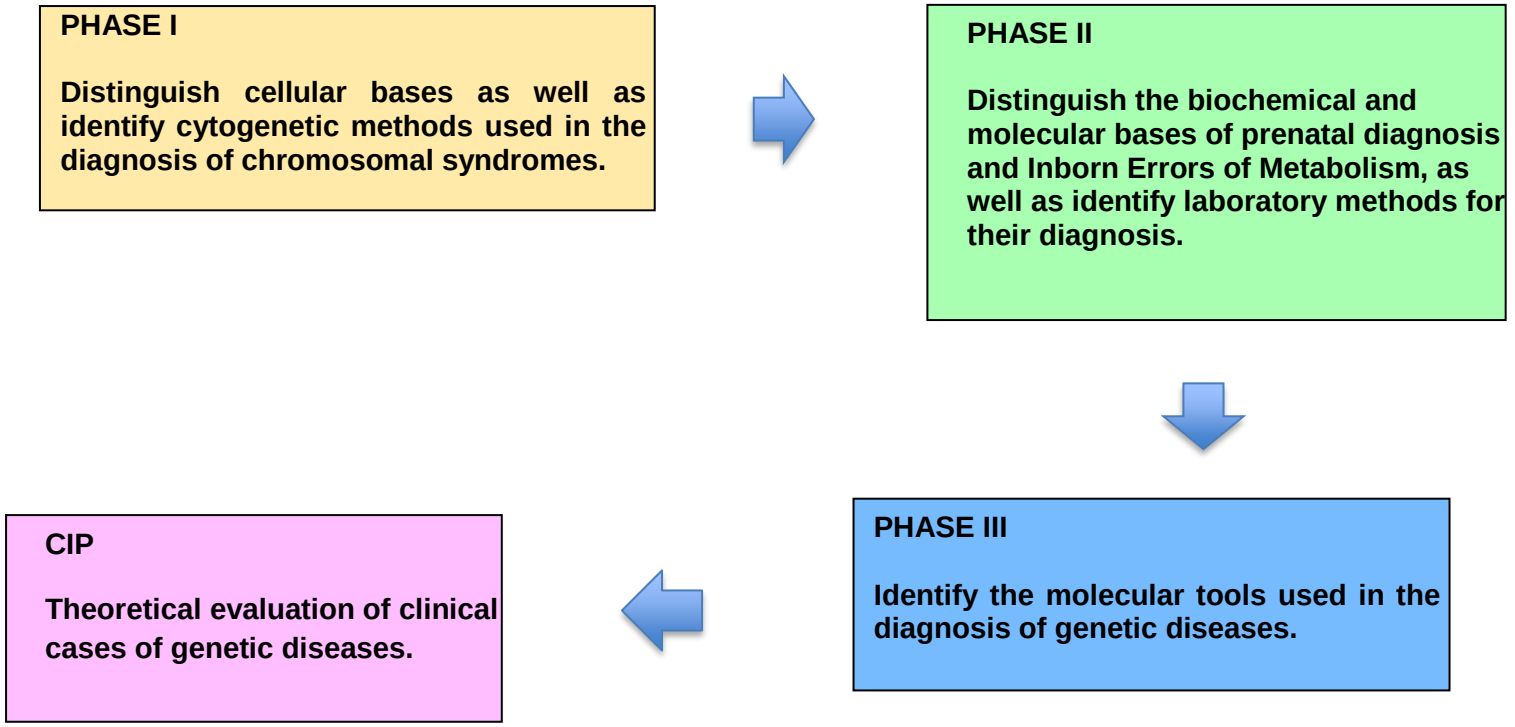
Integrative Competencies:

15. To achieve the adaptability required by the uncertain social and professional environments of our time to create better living conditions.

Specific competencies of the graduate profile that this learning unit contributes to:

To interpret the results of analyses based on established criteria that allow timely and pertinent decision-making in clinical, toxicological, chemical, food, forensic, and environmental diagnosis.

5. Graphical representation:



6. Structuring in stages or phases:

Stage 1: Chromosomal Bases of Inheritance

Component(s) of the competence: To identify the most common chromosomal abnormalities using appropriate cytogenetic methods for the diagnosis of chromosomal syndromes.

Learning evidence	Performance criteria	Learning activities	Contents	Resources
Evidence 1: Questionnaire for identifying karyograms in microphotographs	Identify the chromosomes according to their morphological characteristics and the chromosomal complement; at the end of the class, answer a questionnaire.	The teacher, with the support of a PowerPoint presentation, explains the program and its relationship with other learning units and presents the framework of the topic. The student answers the questions of a diagnostic questionnaire on prior knowledge before the start of the first content. The student individually participates actively in the sessions by	- Introduction to Genetics History, evolution, and impact on biomedical sciences Cell cycle Mitosis Meiosis Gametogenesis - Chromosomal abnormalities: Classification and formation mechanisms: nondisjunction, lagging anaphase. Most common numerical chromosomal abnormalities: Down syndrome, Edwards syndrome, Patau syndrome,	- Analytical program of the Learning Unit -web page: http://genetica-uanl.mx/ - Articles on the Moodle platform and teams Other open-access digital media - Multimedia material of the topics presented in each of the sessions.

Learning evidence	Performance criteria	Learning activities	Contents	Resources
		analyzing the reviewed contents. The student individually prepares a comparison chart on the differences between mitosis and meiosis and submits it through the Moodle platform(**Accredited activity # 1**). The student responds to a written evaluation on the topics covered in this stage. (**Accredited activity # 2: First written evaluation**)	Turner syndrome, Klinefelter syndrome. Most common structural chromosomal abnormalities: Deletions and microdeletions, Cri-du-Chat syndrome, Wolf-Hirschhorn syndrome, 22q11 deletion syndrome, Williams syndrome, Smith-Magenis syndrome, and 1p36 deletion. Tools for studying chromosomal abnormalities: Karyotype: types of samples, cell culture technique, staining Hibridación in situ fluorescente (FISH)	Textbooks: Solari AJ. (2004) Chapter 1-2. Paniagua R. (2003). Chapter 7. Turnpenny P y Elard S. (2020), Chapter 3

Stage 2: Prenatal Diagnosis and Inborn Errors of Metabolism
Component(s) of the competence: To distinguish the laboratory parameters characteristic of prenatal diseases and inborn errors of metabolism to contribute to the diagnosis of these diseases

Learning evidence	Performance criteria	Learning activities	Contents	Resources
2nd. Infographic on prenatal diagnosis methods 3rd. Comparative chart on techniques for the diagnosis of	Create an infographic on the applications of the most suitable laboratory methods for prenatal diagnosis. Prepare it as an electronic document and upload it to the Moodle platform within the established timeframe. Include a cover page with identification details. Write in manuscript format with correct spelling and coherence Identify the metabolic disorder. Establish the most suitable biochemical laboratory method for its diagnosis and upload it to the Moodle platform.	The teacher, with the support of a PowerPoint presentation, presents the framework of the topic with graphic material. The student answers the interspersed questions during the presentations about the assigned topics. The teacher, with the support of a PowerPoint presentation, presents the framework of the topic with graphic material. The student reviews the topic they will address in each session according to the established schedule.	Prenatal Diagnosis Non-invasive methods: ultrasound, maternal biochemical markers, fetal cells in maternal blood. Available preimplantation diagnostic tests (polar body biopsy, blastomere biopsy, and blastocyst biopsy) Prenatal Diagnosis Invasive methods: Cytogenetics (amniotic fluid) Invasive procedures (amniocentesis, cordocentesis, chorionic villus sampling, fetoscopy) Circulating DNA in maternal blood Errores Innatos del Metabolismo Colorimetric tests Biotinidase deficiency Glucose-6-phosphate dehydrogenase deficiency Aminoacidopathies, Organic acidemias	- Moodle platform web page: http://genetica-uanl.mx/ Multimedia material of the topics presented in each of the sessions Textbooks: Turnpenny P y Elard S. (2009), Chapter 21 Genetics and Genomics In Medicine, Thompson & Thompson (2016), Chapter 18

Learning evidence	Performance criteria	Learning activities	Contents	Resources
Inborn Errors of Metabolism		The student answers the interspersed questions during the presentations about the assigned topics during the sessions in MS Teams. The student responds to a written evaluation on the topics covered in this stage. (**Accredited activity #3 Second written evaluation)**	Fatty acid oxidation disorders, Galactosemia Congenital Hypothyroidism and Congenital Adrenal Hyperplasia. Mucopolysaccharidosis Confirmatory tests for the diagnosis of metabolic disorders.	- Moodle platform web page: http://genetica-uanl.mx/ Multimedia material of the topics presented in each of the sessions Textbook: Turnpenny P y Elard S. (2020), Chapter 11

Stage 3: Mechanisms of inheritance transmission.

Component(s) of the competence. To analyze laboratory methods based on their molecular foundations for the diagnosis of the most common genetic diseases

Learning evidence	Performance criteria	Learning activities	Contents	Resources
4th Comparative table on methodologies used in molecular diagnosis	Include a cover page with identification details. Write in manuscript format with correct spelling and coherence. Submit the comparative chart on time	The student reads the topic to be covered in each session in advance, according to the established schedule (outside the classroom). In the session, the teacher presents and explains the topic, initiating a discussion forum with questions directed at the students, using examples related to the topic. At the end of the class, the student individually answers a questionnaire on concepts, in a timely manner (**Accredited activity #4**). The student answers a written assessment on the topics reviewed in	Mendelian inheritance patterns: Autosomal dominant (Tuberous sclerosis, Neurofibromatosis, Marfan syndrome) and autosomal recessive (Cystic fibrosis, Sickle cell anemia, Spinal muscular atrophy). Multifactorial inheritance: Inheritance patterns linked to sex chromosomes (Duchenne and Becker muscular dystrophy, Hemophilia A, Rett syndrome). Hereditary cancer Neo-Mendelian:Herencia mitocondrial, Triplet expansion: (Huntington's disease and Fragile X syndrome) Epigenetics:	Moodle platform web page: http://genetica-uanl.mx/ Power Point presentations (teacher). Textbook: Solari AJ. (2004). Chapters 9 and 10 Genetics and Genomics In Medicine, Thompson & Thompson (2016),

Learning evidence	Performance criteria	Learning activities	Contents	Resources
		this stage (**Accredited activity # 5 Third written evaluation)**	Tools used in molecular diagnosis (applications of Comparative Genomic Hybridization Arrays (aCGH), PCR and PCR variants, MLPA, and types of sequencing (Sanger sequencing and Next-Generation Sequencing (NGS)).	Chapter 4, 5, 11 y 12 Turnpenny P y Elard S. (2020).
		Accredited activity # 6: Electronic presentation by team on an assigned topic that includes genetic methodologies for its diagnosis.		

7. Summative evaluation:

Phase 1		Phase 2		Phase 3		Weighing
Evidence 1 Questionnaire for the identification of karyograms in microphotographs.	5%	**Evidence 2** Infographic on prenatal diagnostic methods.	5%	**Evidence 4** Comparative chart on methodologies used in molecular diagnosis	5%	20%
		Evidence 3 Comparative chart on techniques for the diagnosis of Inborn Errors of Metabolism.	5%			
Accredited activity 1, Comparative chart on the differences between mitosis and meiosis.	2%			**Accredited activity 3,** concept questionnaire	2%	4%
Accredited activity 2, First written evaluation	12%	**Accredited activity No.4,** Second written evaluation.	12%	**Accredited activity 5,** Third written evaluation	12%	46%
				Accredited activity 6, Electronic presentation by team	10%	
CIP					30 %	30 %
TOTAL						**100%**

8. Course integrative project/product.
Theoretical evaluation of clinical cases of genetic diseases in which the ability to select the appropriate technique is demonstrated and the results of the selected genetic tests are interpreted.

9. References:
Textbooks: Turnpenny P y Elard S. (2020). Emery's Elements of Medical Genetics and Genomics, 16° edition. México. Editorial Elsevier. Solari AJ. (2004). Human Genetics. Fundamentals and Applications. 4a edition. México, Editorial Médica Panamericana. Gersen S.L. Keagle M.B. (2013). The principles of Clinical Cytogenetics. Second edition, Editorial Springer. Nussbaum, McInnes and Willard (2016). Genetics and Genomics In Medicine, Thompson & Thompson Seventh Edition, Editorial Elsevier. Tom Strachan & Andrew Read. (2019). Human Molecular Genetics, 15th edition. Paniagua R. (2017). Cell and Molecular Biology. 4 Edition. España. Editorial Mc Graw Hill David L. Valle, Stylianos Antonarakis, Andrea Ballabio, Arthur L. Beaudet, Grant A. Mitchell (2019) The Online Metabolic and Molecular Bases of Inherited Disease. United States. Editorial McGraw-Hill Hereditary diseases: https://www.ncbi.nlm.nih.gov/omim