UANL
UNIVERSIDAD AUTÓNOMA DE NUEVO LEÓN

FACULTAD DE MEDICINA

MODULE DESCRIPTION (ANALYTICAL PROGRAM).

1. Module Information Code:	
• Name of the Institution and School	Universidad Autónoma de Nuevo León, School of Medicine
• Name of the Learning Unit	Genetics
• Total classroom hours for theory and/or practice.	51 hours
• Total extra classroom hours	39 hours
• Course Modality	Schooled
• Type of academic period in which the module is offered	3rd Semester
• Type of Learning Unit in the Curriculum	Compulsory
• Curriculum area:	ACFB
• UANL credit points	3
• Date of module creation:	September 30, 2014
• Date of last amendment:	August 10, 2020
• Person(s) responsible for the module design and amendments:	Dra. med. Laura Elia Martínez Garza, Dra. Graciela Arelí López Uriarte.

2. Introduction:

This learning unit is divided into five phases and in each one of them the following will be addressed: in the first phase the bases of heredity are reviewed as well as its transmission mechanisms, the structure of the human genome and its relevance for the practical application of this basic knowledge to health and genetic disease.

In the second phase, the human karyotype is known with the aim of identifying the main diseases of the chromosomes.

In the third phase, the mechanisms of inheritance with a classical or Mendelian pattern are described through the resolution of clinical cases with pathologies with autosomal dominant, recessive and sex-linked inheritance, as well as their graphic representation through the family trees. It is also based on genetic counseling and the segregation of the alleles by means of the elaboration of Punnett's pictures.

In the fourth phase, the differences presented by some pathologies with a non-classical or neo-Mendelian pattern are analyzed through the discussion and solution of clinical cases.

Finally, in the fifth phase, those pathologies with a complex or multifactorial hereditary pattern and other genetic pathologies or with a genetic component are integrated with a focus on different areas of medical specialization such as pediatrics, gynecology and obstetrics, internal medicine and oncology, highlighting the importance of the interrelations of the multiple disciplines associated with genetics, supported by the use of the diagnostic tools of the laboratories of cytogenetics, biochemical genetics and molecular biology, to offer diagnosis and comprehensive management.

The Learning Integrator Product evaluates, through the solution and development of a clinical case, what is reviewed and analyzed during the learning unit and uses the skills, knowledge and attitudes developed during the unit.

3. Purpose(s)

The development of new technologies that are increasingly specific and precise has allowed the knowledge of new genes, as well as their regulation and expression in various tissues or cells in particular; today, through complex diagnostic and therapeutic methodologies, it is possible to provide a management to certain genetic diseases, thus generating additional fields to act with genetic patients.

To use this knowledge and make it useful in the training of the surgeon and midwife, it is important to establish the clinical characteristics, cytogenetic and molecular variants, risk factors and epidemiological aspects to integrate the diagnosis of the most frequent genetic diseases, as well as to know the genetic component of those multifactorial pathologies even more common than the purely genetic ones.

This Learning Unit is specifically related to Developmental Biology, Cell and Tissue Biology, Biochemistry, as well as Molecular Biology, which provide the concepts of human development as well as the structure and function of the normal human organism that serve as the basis for understanding genetic pathology. It contributes to the general competences favouring the autonomous learning and the handling of information and communication technologies by structuring activities in electronic and written environments with critical judgement. It also prepares and lays the foundations for the clinical integration of diseases with a chronic degenerative genetic component, pediatric and the field of gynecology and obstetrics. In addition, it promotes an attitude of respect and professionalism when analyzing specific clinical cases or situations that arise in the local area, as well as in other contexts.

With regard to the specific competences of the profession, it contributes to the use and application of the scientific method through the analysis of clinical cases using deductive reasoning in order to integrate a clinical diagnosis. Likewise, it promotes ethics and professionalism by exemplifying situations susceptible to discrimination, raising awareness through case analysis, of the value of medical care regardless of the patient's condition. Collaborative work is also promoted by using teaching and learning strategies of this type during the sessions and encourages participation in continuing medical education activities as well as charity.

4. Competences of the graduate profile

a) General competences contributing to this learning unit.

Instrumental skills:

1. Apply autonomous learning strategies in the different levels and fields of knowledge that allow them make appropriate and relevant decisions in the personal, academic and professional fields.

3. Use the information and communication technologies as access tools to information and its transformation in knowledge, as well as for learning and collaborative work with cutting-edge techniques that allow its constructive participation in society.

4. Dominate their native language in oral and written form with correctness, relevancy, opportunity and ethics adapting its message to the situation or context, in order to transmit of ideas and scientific findings.

Personal and social interaction skills

9. 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 with the purpose of promoting environments of peaceful coexistence.

Integrative skills

14. Resolve personal and social conflicts in accordance with specific techniques in the academic field and their profession for the proper decision making.

b) Specific competences of the graduate profile that contributes to the learning unit

Scientific basis of medicine

1.- Use the medicine scientific fundaments considering economical, psychological, social, cultural and environmental factors which contribute to the development and evolution of a disease for decision-making and medical actions.

Professional Clinical Practice

2.- Solves clinical problems through deductive reasoning, interpretation of findings and definition of their nature with the aim of making decisions and determine action principles of the medical practice to follow in a responsible way, impacting individual and collective health.

3.- Evaluate the development and evolution of the disease through the analysis of biomedical information and related physical, social and cultural factors, promoting health education and encouraging preventive medicine.

4.- Manages properly patients with the most frequent diseases from a biopsychosocial perspective, through the application of knowledge, technical procedures and basic diagnostic, based on clinical guides and attention protocols in order to solve the main health problems from the Primary Health Care level from individuals and the community.

Critical thinking and research

7.- Applies the scientific method for the resolution of medical problems with an innovative, analytic and self-critical attitude for preventing, diagnosing and treating diseases.

Professional values and ethics

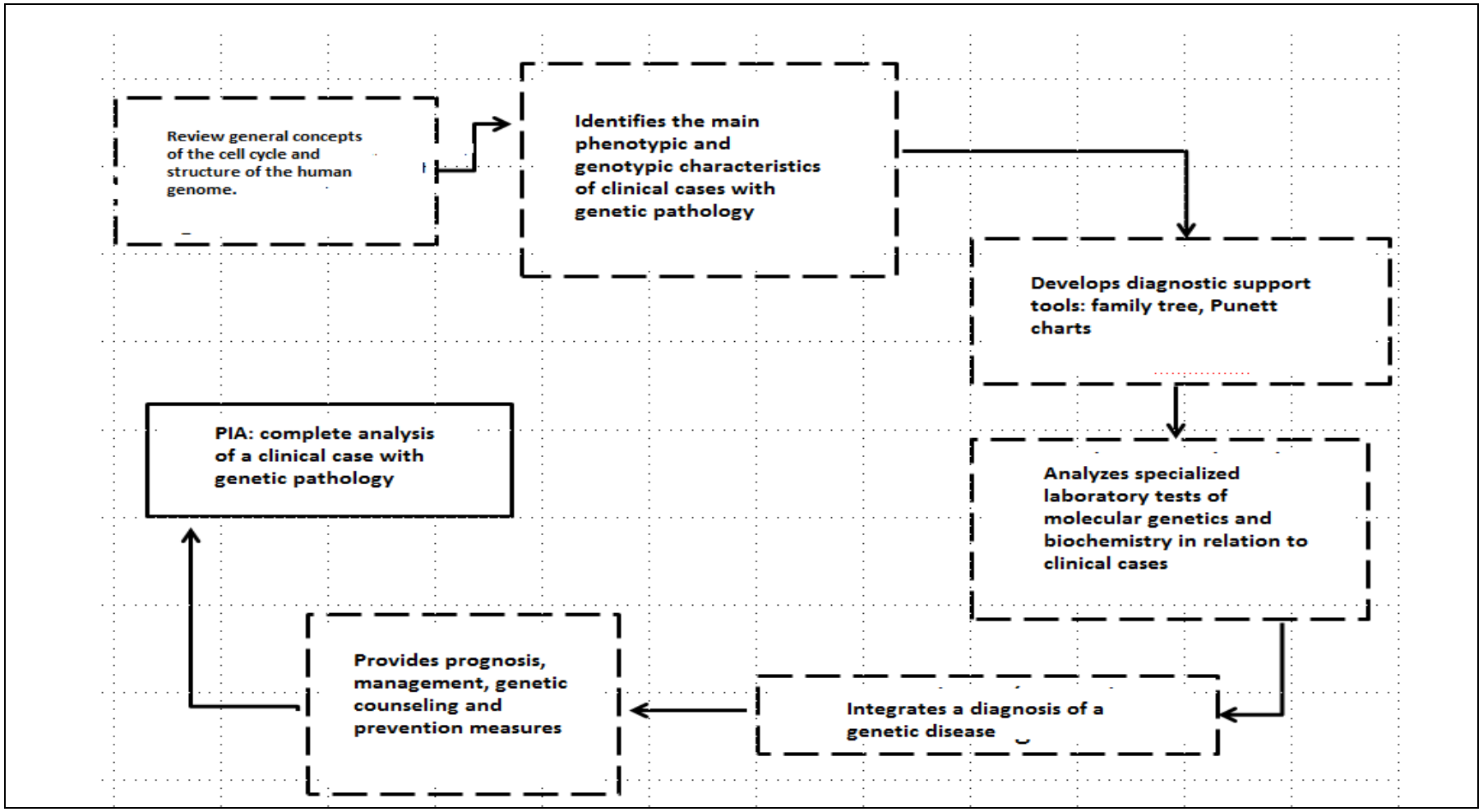
8.- Integrates professional values and ethics into his medical practice, making no difference due to gender, race, political or sexual preference, religious beliefs, activities developed, disabilities or socioeconomic level, promoting social inclusion and contributing to the population's well-being, their life quality and human development.

9.- Respects the patient's integrity keeping the patient's medical information as an essential part of their professional secret in order to preserve his rights.

Communication

11.- Applies effective communication principles, establishing a respectful and sympathetic relationship with the patient, relatives, the community and other health professionals in order to use the information properly.

5. Couse Roadmap:


Review general concepts of the cell cycle and structure of the human genome.
Identifies the main phenotypic and genotypic characteristics of clinical cases with genetic pathology
Develops diagnostic support tools: family tree, Punett charts
Analyzes specialized laboratory tests of molecular genetics and biochemistry in relation to clinical cases
Integrates a diagnosis of a genetic disease
Provides prognosis, management, genetic counseling and prevention measures
PIA: complete analysis of a clinical case with genetic pathology

Phase 1. General framework, bases of inheritance and transmission mechanisms.

Component(s) of the competence:
Analyze the molecular bases of heredity, its transmission mechanisms as well as the structure of the human genome relating it to health and disease to diagnose and refer cases with genetic pathology.

Evidence of student learning	Performance Criteria	Learning activities	Content	Resources
• Written report of the search for a disease in OMIM • Graphical representation of a case segregation analysis	• Describes the clinical characteristics of the assigned genetic pathology. • Obtains the information from the OMIM database. • Make a copy of the electronic consultation screen and attach it to the written report. • Write a handwritten summary of the genetic factors involved in the pathology, with an extension of one page. • Delivery in time and form the report. • From a Cariogram of a clinical case with numerical or structural chromosomal alteration. • Explain with a drawing how non-disjunction (numerical) occurs,	The professor exposes and explains the generalities. Based on the concepts reviewed, the student, individually, performs chromosome segregation of the most frequent examples of numerical and structural aberrations, either of autosomal or sexochromosomes. The student identifies the human chromosomes in metaphase.	**Conceptual Content** • Basic Concepts • Aspects of inheritance cytogenetics: karyotype and cytogenetic alterations (numerical, structural, autosomal, sex chromosome) • Organization of the human genome • Diagnostic tools for chromosomal aberrations **Procedural Content** Searching for information in databases Identification of the chromosomes in a Karyotype **Attitudinal Content**	School of Medicine classrooms. Internet for database access: OMIM, PubMed. Metaphase printing of lymphocytes for karyotype analysis. Computer with classroom projector. Power Point presentations students/teacher. Textbook. Reference book. Scientific article of support.

• Lymphocyte karyogram	and the possible gametes formed according to the possible forms of segregation (structural: two examples, translocation 13;14 in the mother, translocation 21;21 in the father). • Delivery in time and form the report. • Sort the chromosomes according to their groups (A-G). • Points out the position of the centromere of each chromosome. • Indicates the type of staining used for that study. • Write the chromosome formula (karyotype). • Interprets the analyzed karyogram: euploid female, male with regular trisomy 21, female with translocation trisomy 13, etc, as appropriate for the karyotype. • Delivery in time and form the report.		Discipline in the methods of searching and analyzing information.	

Phase 2. Chromosomal alterations.

Component(s) of the competence:
Understand the alterations in the mechanisms of heredity transmission based on clinical study and cytogenetic analysis for the identification of numerical, structural, autosomal and sex chromosome diseases.

Evidence of student learning	Performance Criteria	Learning activities	Content	Resources
Resolution of a clinical case of a numerical or structural chromosomopathy.	1. Identify the list of problems of the case presented. 2. Requests cabinet and laboratory studies to support their diagnosis. 3. Describes the etiology of the disease: trisomy 21, Turner syndrome, etc. As appropriate. 4. Explains the medical follow-up that the case presented should have, important in the timely detection of possible complications. 5. Offers the risk of recurrence. 6. Delivery in time and form the report.	The professor presents a clinical case and illustrates it with photographs. By means of the integration of the clinical picture and laboratory studies, the students solve the questionnaire that is given by teams, directed to integrate diagnosis (clinical analysis, general and genetic laboratory studies, cabinet studies, management, prognosis and advice). The students, in teams, present an example of each pathology contained in this phase.	**Conceptual Content** Syndromes by numerical, autosomal and sexochromosomal aberrations: Down, Edwards, Patau. Turner, Klinefelter, Y and X polysomia. Structural, autosomal sex chromosome abnormalities: Cri-du-chat, Wolff-Hirschhorn, Williams, Di George, Smith Magenis, 1p36 deletion. **Procedural Content** Analysis and resolution of cases on the alterations of the mechanisms of inheritance transmission. Synthesize information for subsequent oral	School of Medicine classrooms. Computer with classroom projector. Power Point presentations students/teacher. Textbook. Print out of clinical case with questions.

			presentation with the support of different didactic tools. **Attitudinal Content** Responsibility for delivering the requested material and presenting your oral class. Confidentiality of the patients' clinical data.	

Phase 3. Classic or Mendelian Heritage Patterns				
Component(s) of the competence: Identify the characteristics of Mendelian inheritance through analysis of clinical cases and molecular diagnostic tools to establish the pattern of inheritance and its diagnosis, determine its management, prognosis and genetic counseling.				
Evidence of student learning	**Performance Criteria**	**Learning activities**	**Content**	**Resources**
• Family trees: one personal and another with an example of Mendelian pathology.	1. Employs appropriate symbolism for the gender of individuals. 2. Draw properly the relationship lines in the genealogy. 3. Identifies generations and numbers of individuals. 4. Point out the purpose. 5. Identifies inheritance pattern.	The professor chooses a clinical case, with clinical photographs and laboratory results. The professor delivers a questionnaire aimed at integrating diagnosis (clinical analysis, general and genetic laboratory studies, cabinet studies, management, prognosis and counseling).	**Conceptual Content** • Autosomal dominant inheritance and its variants: osteogenesis imperfecta, neurofibromatosis, tuberous sclerosis, familial hypercholesterolemia, Ehlers-Danlos • Autosomal recessive inheritance and its variants: cystic fibrosis, sickle cell anemia, spinal muscular	School of Medicine classrooms. Computer with classroom projector. Power Point presentations students/teacher. Textbook. Supporting article Print out of clinical case with questions.

• Resolution of a clinical case with monogenic disease.	6. Delivery in time and form the report. 1. Identifies the list of problems of the case presented. 2. Requests cabinet and laboratory studies to support their diagnosis, including genetic studies. 3. Describes the name of the gene, its locus, the encoded protein and its cellular function. 4. Explains the medical follow-up that the case presented should have: importance in timely detection of possible complications. 5. Offers the risk of recurrence. 6. Delivery in time and form the report.	The student, individually, resolves the clinical case shown. The students in teams, present an example of each pathology contained in this phase.	atrophy, epidermolysis bullosa, oculocutaneous albinism • Sex-linked inheritance: hemophilia A and B, Duchenne and Becker, X-linked ichthyosis, incontinentia pigmenti, familial hypophosphatemic rickets **Procedural Content** Elaboration of Punnet charts to support your genetic counselling on the risk of recurrence. Discuss in an orderly and respectful way the clinical cases and examples projected in class Synthesize information for subsequent oral presentation with the support of different didactic tools. **Attitudinal Content** Responsibility for delivering the requested material and presenting your class orally Teamwork.	

• First partial exam.			Confidentiality of the patients' clinical data. Respect for the opinion of others	

Phase 4. Non-Classical Heritage.				
Component(s) of the competence: Identify the characteristics of non-classical inheritance through analysis of clinical cases and molecular diagnostic laboratory studies to establish the pattern of inheritance, determine its management, prognosis and genetic counseling.				
Evidence of student learning	**Performance Criteria**	**Learning activities**	**Content**	**Resources**
Written report of the analysis and interpretation of the molecular biology laboratory result. Resolution of a clinical case of non-classical inheritance.	Solve exercises to identify the mutation represented by the specific study. Describes the techniques of: • Agarose gel from a specific allele PCR. • Chromatogram of a SANGER or automated sequencing.	The professor chooses a clinical case, illustrates it with photographs, and delivers a questionnaire aimed at integrating diagnosis (clinical analysis, general and genetic laboratory studies, cabinet studies, management, prognosis and counseling). The professor encourages class discussion of the clinical scenario presented. The professor coordinates and moderates the class discussion on genetic counseling in each case.	**Conceptual Content** • Genetic diseases by mosaicism (somatic and germinal: Ito's hypomelanosis), • Mitochondrial Inheritance: Leber's Optical Atrophy, MELAS, MERRF • Diseases caused by the expansion of microsatellites: Huntington's disease, myotonic dystrophy, Friedreich's ataxia • Pathologies by uniparental disomy (DUP) and imprint: Prader-Willi/Angelman,	School of Medicine classrooms. Computer with classroom projector Power Point presentations students/teacher Textbook Reference book Print out of clinical case with questions Print out of molecular biology laboratory study

	1. Identifies the list of problems of the case presented. 2. Requests cabinet and laboratory studies to support the diagnosis including genetic studies. 3. Describes the etiology of the disease: repeated expansion, oligogenic, uniparental disomy, as appropriate. 4. Describes the name of the gene, its locus, the coded protein and its cellular function of the pathology presented in the case. 5. Explains the medical follow-up that the case presented should have: importance in timely detection of possible complications. 6. Offers the risk of recurrence. 7. Delivery in time and form the report.	The student in teams, exposes an example of each pathology contained in this phase. The student analyzes the clinical data presented in class, and discusses possible diagnoses and genetic counseling.	Silver-Russell, Beckwith-Wiedemann • Oligogenic inheritance: Bardet Biedl, retinitis pigmentosa. **Procedural Content** Synthesize information for subsequent oral presentation with the support of different didactic tools. Analysis and interpretation of the molecular biology laboratory result. Discuss in an orderly and respectful way the clinical cases and examples projected in class. **Attitudinal Content** Teamwork. Responsibility for delivering the requested material and presenting your oral class. Confidentiality of the patients' clinical information. Respect for the individual with various diseases.	

Phase 5. Genetics and its application to specialized medicine.

Component(s) of the competence:
Integrate the advances in genetics and genomic medicine in the diagnosis of pathologies frequently presented in clinical practice for their reference and interdisciplinary management.

Evidence of student learning	Performance Criteria	Learning activities	Content	Resources
Dysmorphological evaluation of clinical photographs.	• Describe using the appropriate terms the picture of the patient shown in the class. • Recognizes if it is an isolated or multiple defect. • Defines if it is a malformation, deformation, disruption or dysplasia. • Identifies if it is a syndrome, sequence or association • Describes the alteration(s) identified in the exercise, noting their frequency, risk factors, method of diagnosis, medical follow-up and genetic counseling given to the family. Primary, secondary and tertiary prevention methods. • Delivery in time and form the report.	The professor chooses a clinical case, illustrates it with photographs, and delivers a questionnaire aimed at integrating diagnosis (clinical analysis, general and genetic laboratory studies, cabinet studies, management, prognosis and counseling). The student in teams, exposes an example of each pathology contained in this phase. Debate is encouraged in relation to differential diagnoses, the interrelationship between different medical specialties, the importance of the genetic approach to patients with common diseases.	**Conceptual Content** <u>Genetics and Pediatrics</u> - Multifactorial inheritance children. Association, ligation, functional and positional cloning studies. Threshold model. Examples of diseases (NTCD, congenital heart disease, LPH, hip dysplasia) - Congenital defects: malformation, disruption, deformation, dysplasia, syndrome, sequence. Environmental agents: teratogens such as alcohol, maternal diseases, anticonvulsants Prevention strategies. - Neurodevelopmental disorders: genetic component of intelligence, normal	School of Medicine classrooms. Computer with classroom projector. Power Point presentations for students/teacher Dysmorphology photographs Textbook. Reference books. Supporting articles.

Analysis of an expanded neonatal sieve or urine sieve result report. 2nd partial exam	• From a result report with a brief clinical case: • Identifies what type of test was performed. • Mention with which type of metabolic diseases the shown technique can be used. • Identifies the factors that cause a false positive and false negative result. • Delivery in time and form the report.		developmental milestones and assessment scales. Fragile X syndrome, Rett. Fetal alcohol syndrome, perinatal infections. - High/low size: definitions, genetic component of size. Diseases: Achondroplasia, Noonan, child of diabetic mother, Marfan and Sotos. - Inborn errors of metabolism: classification of diseases. Examples of metabolic diseases: Phenylketonuria, maple syrup urine disease, galactosemia, b-oxidation disorders, OTC deficiency, MPS and Glycogenosis. <u>Genetics and Obstetrics-Gynecology:</u> - Disorders of sexual differentiation: normal process of sexual differentiation in men and women, classification of	

			disorders of sexual differentiation, examples of diseases: congenital adrenal hyperplasia, androgen insensitivity, 5-a-reductase deficiency, gonadal dysgenesis - Prenatal and pre-conception diagnosis: non-invasive and invasive tests. PGD. <u>Genetics and Internal Medicine</u> Adult multifactorial inheritance: Chronic-degenerative diseases (Parkinson's, Alzheimer's, MD, obesity, coronary disease) Cancer Genetics Risk factors. Familial and hereditary cancer. Responsible genes. Hematolymphatic cancer, retinoblastoma, breast and ovarian cancer Li-Fraumeni. Polyposis and non-polyposis colon cancer	

			Procedural Content Analysis of a report on the results of the expanded neonatal sieve or urine sieve Synthesize information for subsequent oral presentation with the support of different didactic tools Discuss in an orderly and respectful way the clinical cases and examples projected in class **Attitudinal Content** Teamwork. Respect for patient and family decisions Respect for the individual with rare diseases and birth defects Justice with respect to access to specialty patient care Humanism in daily medical care, translated into empathetic attitudes towards the patient.	

7. Summative Evaluation

- Written report of the search for a disease in OMIM ……..4%
- Punnett squares…… ……4%
- Lymphocyte kariogram: ……4%
- Resolution of a clinical case of a numerical or structural chromosomopathy ……..….4%
- Family trees: one personal and another with an example of Mendelian pathology ……4%
- Resolution of a clinical case with monogenic disease ……..4%
- Written report of the analysis and interpretation of the molecular biology laboratory result ……...4%
- Resolution of a clinical case of non-classical inheritance…… 4%
- Dysmorphological evaluation of clinical photographs ……..4%
- Analysis of report result containing an extended neonatal sieve or urine sieve……4%
- First partial exam ……....20%
- Second partial exam…….....20%
- CIP ……..….20%
- Total……100%

8. Course Integrative Product

PIA: resolution of clinical cases of a genetic disease.

References

Textbook:

- Del Castillo, V., Uranga, R., Zafra, G. (2012). Genética clínica. México, D.F.: Manual Moderno.

Reference books:

- Turnpeny, P., Ellard S. (2009). Emery: elementos de Genética Médica. Barcelona, España: Elsevier, Churchill Livingstone.

Support articles:

- Bennett R., French, K., Resta, R. & Doyle, D. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counselors, 17, pp.424-433.

- Del Valle Torrado, M. (2009). Evaluación etiológica del retardo mental de origen genético. Algoritmo diagnóstico y nuevas técnicas moleculares. Arch Argent Pediatr, 107, pp.246-255.
- Prieto, M., Guitian, C. (2009). Guía de uso de Medline/PubMed. noviembre, 2009, de Fisterra Sitio web: www.fisterra.com
- Rey, R., Grinspon R. (2011). Normal male sexual differentiation and aetiology of disorders of sex development. Best Practice & Research Clinical Endocrinology & Metabolism, 25, pp.221-238.
- Sabin M., Werther G., Kiess, W. (2011). Genetics of obesity and overgrowth syndromes. Best Practice & Research Clinical Endocrinology & Metabolism, 25, pp.207-220.
- Seaver L, Irons M. (2009). ACMG practice guideline: Genetic evaluation of short stature. Genetics in Medicine, 11, pp.465-470.
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Electronic Sources:

- Enfermedades hereditarias:
  http://www.ncbi.nlm.nih.gov/Omim/searchomim.html
- Citogenética:
  http://www.pathology.washington.edu/galleries/cytogallery
  http://www.citi2.fr/GENATLAS
  http://www.infobiogen.fr/services/chromcancer
- Genómicas:
  http://www.ncbi.nlm.nih.gov/blast

  http://www.ncbi.nlm.nih.gov/entrez

  http://www.ncbi.nlm.nih.gov/genemap
- Mutaciones:
  http://wwwuwcm.ac.uk/search/mg/allgenes
- Mitocondriales:
  http://www.mips.biochem.mpg.de/proj/medgen/mitop

APPENDIX.

ASSESSMENT AND WORKLOAD

Module workload		Number of hours	Percentage
Contact hours	Class-based instruction	38h (74.50%)	56.66%= 51 horas
	Workshops (resolution of clinical cases and other written activities)	10h (19.60%)	
	Exam taking	2h (3.92%)	
	Course integrative product (CIP)	1h (1.96%)	
Independent study	Study	33h (84.61%)	43.33%= 39 horas
	Exam preparation	6h (15.38%)	
Total hours of the workload: 30 hours X 3 credits UANL/ECTS*		90 h	

*European Credit Transfer and Accumulation System
1 UANL credit = 30 hours

NOTE: Rubrics, checklists and evaluation formats are elaborated by using the performance criteria described in each stage of the module.

SUPLEMENTO COVID-19

Siguiendo las recomendaciones de la Secretaría de Salud del país y la Rectoría de la Universidad, ante la coyuntura de salud COVID-19, la organización de la docencia desde marzo del 2020, seguirá un modelo híbrido, donde la docencia se ajustará a los horarios aprobados por la Secretaría de Salud siguiendo un modelo de Presencialidad / No presencialidad en la medida en que las circunstancias sanitarias y la normativa lo permitan. Los estudiantes asistirán a las clases de manera no presencial mediante la transmisión de las mismas de manera síncrona/asíncrona vía “on line”.