INSTRUCTIVO PARA LLENADO

- No deben eliminarse los títulos en mayúsculas y negrita.
- Los ** solo indica la existencia de un título/sección, pero no se ve en la plataforma.
- Los ++ corresponde a la sección donde se agregan diagnósticos adicionales.
- Los ## indica secciones rellenables en la ficha (título + contenido).
- Que no se debe alterar el orden de las secciones.
- El contenido editable corresponde al texto que aparece en cursiva azul.
- Todo párrafo debe terminar con un punto.

** TÍTULO DE LA COMPETENCIA

GENETIC TESTING AND COUNSELLING

** DIAGNÓSIS / SITUACIÓN 1

GENETIC TESTING FOR MONOGENIC DISORDERS

** DIAGNÓSIS / SITUACIÓN 2

AUTOSOMAL DOMINANT MODY2

** DIAGNÓSIS / SITUACIÓN 3

AUTOSOMAL RECESSIVE CONGENITAL ADRENAL HYPERPLASIA

** DIAGNÓSIS / SITUACIÓN 4

ANDROGEN INSENSITIVITY SYNDROME

COMPETENCIES TO ACQUIRE

GENERAL.

Genetic testing and counseling for monogenic hereditary disorders, providing clinicians and family members with information about 1) the consequences of the disorder, 2) the probability of it developing, 3) the probability of transmitting pathogenic variants, 4) possible ways to prevent it, 5) possible ways to treat it.

SPECIFIC.

Diagnosis and decision making in genetic counseling for a family with a hereditary disease.

Creating a family tree of a family with a hereditary disease and identifying individuals susceptible to developing the disease or to being carriers of the disease.

Indicating the relevant tests and analyses.

Communicating with clinicians and/or family members about the diagnosis and the options for prevention, treatment, and reproduction.

Use verbal and nonverbal communication skills in the physician-patient relationship.

Teamwork.

BACKGROUND KNOWLEDGE

Monogenic hereditary patterns.

Symbols used in constructing a pedigree.

How to interpret genetic reports and blood tests (basic clinical parameters).

SUGGESTIONS FOR STUDY MATERIAL

Presentations in the virtual campus about genetic diagnosis and the clinical characteristics of the three disorders.

SCENARIO

Doctor's office with a table and chairs on both sides. Access (on paper or by computer) to the clinical history and reports of findings on genetic and/ or laboratory tests relevant to each diagnosis, blank sheets of paper to facilitate explanations for patients (e.g.. pedigree).

MOSAICO TEMPLATES

Students should receive the different templates in the room where they design the cases (one copy for each table): Case design template. Students' template for assessing other groups, Teacher's template for assessing the competency being taught.

DIFFERENTIAL DIAGNOSES

Maturity onset diabetes of the young (MODY2).

Congenital adrenal hyperplasia (CAH).

Androgen insensitivity syndrome.

CASE DOCUMENTS

Same for the 3 cases.

Clinical guide, results of genetic tests, results of laboratory tests.

Same for the 3 cases.

SUPPLIES

Same for the 3 cases.

Doctor's office supplies Manikin of a baby.

Same for the 3 cases.

MOST COMMON ERRORS

Recommending the clinician or family members the most appropriate therapeutic option according to the physician's own personal criteria.

Focusing excessively on the disorder being studied and not focusing enough on other possible hereditary disorders that the family might have.

Focusing excessively on the genetic disorder without attending to the psychological needs of the people being attended.

Failure to identify family members in the family tree who are at risk of developing or transmitting the hereditary disorder.

Disorganization and lack of leadership.

Inappropriate language (too technical).

Lack of eye contact with the patient.

TIPS

Devise a rotation scheme for the cases to maximize group circulation.

The recommended order of acting out the cases is MODY2 - CAH-Androgen insensitivity syndrome.

Train a student in the designing subgroup how to act when arriving at the doctor's office and when receiving the specific medical information provided. The disorders treated in this workshop are difficult for patients to accept, so the patient should be upset, worried, and anxious. The patient should also express unfamiliarity with the diseases and with medical vocabulary.

Use an intercom or cell phones to connect the professor facilitating the simulation with the aide in simulation.

TECHNOLOGY

An intercom or cell phones to connect the professor facilitating the simulation with the aide in simulation.

The MOSAICO web platform can be used in the design or other phases of the process.

EXPERT ADVICE

Make sure that the communication between the professor and the aide in the scenario works properly before starting to act out the scenes.

Make sure that the observers can see and hear everything in the scenario and that they receive the results of the genetic and laboratory tests at the same time as the treating group complementary.