Module Title:
Neonatal Screening for Congenital Diseases

Module Code:
AM0312

Maximum Number of Students:
15

Total ECTS Credits
2

Notional Learning Hours
(a) Contact Time -   10 h **(b) Private Study -**   40 h

Format of Teaching:

Lectures	10 h
Laboratories or Practicals	0 h
Other	0 h

Teaching Strategy:
Please show how the contact hours are to be allocated in terms of the type of class involved.

 Formal lectures in 120 min timetable.

Convener:
Susana Etcheverry

University / Department:
National University of La Plata, Biological Sciences Department

Language of Tuition:
English

Module Description - The Purpose or Aims:
1 To motivate students about the importance of Newborn Screening as part of Health Prevention Programs, with emphasis in the organization, components and structure of a Newborn Screening Laboratory, the selection criteria for diseases to be screened, and the implementation of Quality Systems in Neonatal screening laboratories. 2. Introduction to the clinical and the biochemical bases and methods for detection of the most frequent congenital diseases: PKU and Congenital hypothyroidism 3. Brief mention to other pathologies currently involved in Newborn Screening Programs

Specific Learning Outcomes for this module: (contributing to general learning outcomes GLO 1 – GLO 10)

At the end of the module the learner is expected to be able to:

1. Understand clearly the importance of Newborn Screening Program in the frame of a prevention health system.
2. Highlight the importance of Quality Systems in Newborn screening Laboratories.
3. Recognize the elements required in the structure of a Newborn Screening Laboratory and the components of a Quality System.
4. Identify correctly the criteria for including pathologies in a Newborn Screening Program.
5. Understand clearly the clinical and biochemical bases of PKU and Congenital hypothyroidism
6. To develop students abilities to address the methodological strategies for the detection of other pathologies subjected to Newborn Screening

Summary of Course Content:

This module aims to motivate the students to understand the need of a Newborn Screening Program in the frame of a Public Health Systems and also to emphasize the importance of Quality Systems in the laboratories.
The identification of the variables in the pre-analytical and post analytical processes; method validation and the development of an integral Quality System
The application, in detail, of the previous concepts to the Newborn Screening for PKU and Congenital Hypothyroidism
Brief description of clinical and biochemical bases of other pathologies included in Newborn Screening Programs

Transferable Skills Taught:

IInformation Tecnhology:

      Web searching for Procedure Manuals of different Newborn Screening Programs
Communication:
      Communication with use of terminology based on **Lexicon** version 8
      http://www.isns-neoscreening.org/pdf/Lexicon8.pdf

Assessment Methods:

Students will be organized in five groups of three persons and:

I) . Each group will give a short seminar (30 min) for the whole class related to the NS of one of the following congenital diseases, included in the bibliography
1. *Galactosemia*
2. *Congenital Adrenal Hyperplasia*
3. *Maple Syrup Urine Disease*
4. *Biotinidase Deficiency*
5. *Cystic Fibrosis*

Probably discussion of a short simple paper could be included.

or

II) A short written presentation of one of the above topics performed by each group of three students

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Assessment Criteria:

Threshold Student has Newborn screening (NS) concepts but is not able to plan or interpret a NS Program

Good: has the criteria for NS but with limitations in methods validation or the implementation of a NS Program

Very good: has the criteria for NS, methods validation and implementation of a NS Program but he is not able to implement an Integral Quality Control System

Excellent: has the criteria for NS, methods validation and he is able to develop a complete Newborn Screening Program

Resource Implications of Proposal and Proposed Solutions:
Details on any resources required and should be included. Please also list e.g core texts; recommended reading material; equipment; films etc.

Lecture notes will be available for students. These will eventually be included in the course textbooks.
Recommended reading:

-International Atomic Energy Agency (IAEA): Screening of Newborns for Congenital Hypothyroidism. Guidelines for Developing Programs. International Atomic Energy Agency Ed. , Vienna, 2005.
 -CLSI: Blood Collection on Filter Paper for Newborn Screening Programs. Aproved Standards.-Fifth edition. CLSI Document LA4-A5, Vol 27 (20), 2007.
-Therrell BL, Panny SR, Davidson A, Eckman J, Hannon WH, Henson MA, Hillard M, Kling S, Levy HL, Meany FJ, MCCabe ERB, Mordaunt V, Pass K, Shapira E and Tuerck J. U.S. Newborn Screening System Guidelines: Statement of rhe Council of Regional Networks for Genetics Services, Screening 1: 135-147, 1992
- Pass KA, Lane PA, Fernhoff PM, Hinton CF, Panny SR, Parks JS, Pelias MZ, Rhead WJ, Ross SI, Wethers DL and Elsas LJ; "U.S. Newborn Screening System Guidelines II: Follow-up of Children, Diagnosis, Management, and Evaluation Statement of the Council of Regional Networks for Genetic Services (CORN)". Journal of Pediatrics 137, S1-S46, 2000.
-Guthrie R. Organization of a Regional Newborn Screening Laboratory, in: Neonatal Screening for Inborn Errors of Metabolism. Bickel H, Guthrie RHammersen G Eds, , Springer-Verlag, Berlin Heidelberg new York 259-270, 1980

Pre-Requisites:

Students may have knowledge of biochemistry, genetics, analytical methods, and quality control