**Kingdom of Saudi Arabia**
**Taibah University – Madinah**
College of Applied Medical Sciences
Department of Clinical Nutrition

# INHERITED METABOLIC DISORDERS (CLN 426) 1437-1438H

## A- COURSE INFORMATION:

| Course Code | Course Title | Credit Units | | | Study Level | Pre-requisites |
|---|---|---|---|---|---|---|
| | | Total | Theory | Practical | | |
| CLN-426 | Inherited metabolic disorders | 3 | 2 | 2 | 8th | CLN-212 |

| Course Coordinator | Extension | Email Address |
|---|---|---|
| Prof: Sahar Ali Ibrahim Hammouda | 8690 | sahammouda@taibahu.edu.sa |

## B- COURSE DESCRIPTION:

This course deals with the basics of dietary management of inborn errors of metabolism (amino acid, carbohydrate, fatty acid, urea cycle disorders & minerals). Students will learn how to manage their food and nutrients intake during health and disease staring from the first days of life throughout the life cycle (inborn errors in infancy, childhood, adults & pregnancy).

## C- COURSE OBJECTIVES:

1- describe the scientific basis of genetics and methods of inheritance of diseases.
2- Define inborn errors of metabolism.
3- Distinguish between different biochemical abnormalities
4- Discuss the biochemical error, consequences and complications of consuming regular diet and clinical presentation in infancy, older children and adults.
5- Explain the principals of nutritional support in inborn errors of metabolism.
6- Prepare nutrition management plan for infants, children and adults with inborn errors of metabolism

## D- THEORY TOPICS:

| Week | Theory Topic | Hours |
|---|---|---|
| 1 | Principals of human genetics<br>Mode of inheritance | 4 |
| 2 | Pathophysiology of impaired metabolism, Principal of nutrition support. Diagnosis of inborn errors of metabolism, guidelines for nutritional therapy of inborn errors of metabolism. | 2 |
| 3 | Newborn screening | 2 |
| 4 | Nutrition support in amino acid disorders (phenylketonuria) | 2 |
| 5 | Nutrition support in amino acid disorders (tyrosinemia, homocystinurea) | 2 |
| 6 | Urea cycle disorders | 2 |

| | | |
|---|---|---|
| 7 | Organic academia, | 2 |
| 8 | Defects in carbohydrate metabolism: galactosemia, | 2 |
| 9 | Defects in carbohydrate metabolism: hereditary fructose intolerance, glucose/ galactose malabsorption. | 2 |
| 10 | Glycogen storage diseases | 2 |
| 11 | Disorders of fatty acid oxidation | 2 |
| 12 | Mitochondrial disorders | 2 |
| 13 | Disorders related to mineral metabolism (copper & zinc) | 2 |
| 14 | Disorders related to vitamin metabolism and vitamin responsive metabolic disorders | 2 |
| 15 | The ketogenic diet | 2 |

**E- PRACTICAL SESSIONS:**

| Week | Practical Session | Hours |
|---|---|---|
| 1 | Theory | |
| 2 | Nutrition support in amino acid disorders (phenylketonuria) in infants | 2 |
| 3 | Nutrition support in amino acid disorders (phenylketonuria)in adults & pregnant | 2 |
| 4 | Nutrition support in amino acid disorders (tyrosinemia, homocystinurea) | 2 |
| 5 | Urea cycle disorders | 2 |
| 6 | Organic academia, | 2 |
| 7 | Defects in carbohydrate metabolism: galactosemia, | 2 |
| 8 | Defects in carbohydrate metabolism: hereditary fructose intolerance | 2 |
| 9 | Glycogen storage diseases | 2 |
| 10 | Disorders of fatty acid oxidation | 2 |
| 11 | Mitochondrial disorders | 2 |
| 12 | Disorders related to mineral metabolism (copper & zinc) | 2 |
| 13 | Disorders related to vitamin metabolism and vitamin responsive metabolic disorders | 2 |
| 14 | The ketogenic diet | 2 |
| 15 | | |

**F- ASSESSMENT TASKS:**

| # | Type of assessment task | Week | Total Grades |
|---|---|---|---|
| 1 | Exam 1 | Week 5 | 10% |
| 2 | Mid term | Week 9 | 20% |

| | | | |
|---|---|---|---|
| 3 | Exam 2 | Week 11 | 10% |
| 4 | Final practical | Week 15 | 20% |
| 5 | Final theory | Week 16 | 40% |

**G- LEARNING RESOURCES:**

1- Required textbook:

Nutrition Care Manual and Pediatric Nutrition Care Manual (www.nutritioncaremanual.org) Phyllis B. Acosta: Nutrition Management of Patients with Inherited Metabolic Diseases Publisher: Jones and Bartlett Publishers; 1st ed (2009); ISBN-10: 0763757772; ISBN-13: 978-0763757779

2- Essential references:

Patricia Samour: Handbook of Pediatric Nutrition, 3rd ed. Publisher: Jones and Bartlett Publishers, Inc. (2005); ISBN-10: 0763783560; ISBN-13: 978-0763783563

**Notes:**

- You will submit 4 cases as a final individual evaluation therefore group work in this final task is not allowed, you can refer to your notes during discussion and the reference books . Cases include cancer, ICU patients & burn). Grade distribution for each case will be as follows:

| # | NCP | Total Grades |
|---|---|---|
| | Introduction of the case with brief medical history and diagnosis in few lines e.g A 56 years old man with 2 month history of coughing, sore throat, and progressive difficulty swallowing first to solid food (and then to liquids. He is currently complaining of nausea and early satiety. A biopsy revealed adenocarcinoma of the distal esophagus. | .5 |
| | comprehensive assessment of nutrition status and risks and includes the following major categories of data collection: | |
| | Patient history: Medication and supplement history, personal history, e.g He was prescribed chemotherapy with 5-fluorouracil and cisplastin, along with chest irradiation over the lower esophagus and the stomach and an esophagectomy is planned. ....etc | .5 |
| | Food and nutrition history (diet history), which includes food consumption, nutrition and health awareness, physical activity and exercise, and food availability.<br>Analysis of diet using special program or food composition tables is required with comments on the diet e.g diet is high in simple sugar and trans fat, iron intake is less than 50% of requirements.....etc | .5 |
| | Nutrition-focused physical examination: Review of systems, including general conditions and physical appearance, gastrointestinal, musculoskeletal, skin, extremities, and other systems e.g severe pallor, loss of subcutaneous | .5 |

| | | |
|---|---|---|
| | muscles over the chest and abdomen, ,..etc negative signs should be included when relevant e.g no edema (in cases of malnutrition and hypoalbuminemia) | |
| | Anthropometric measurements: Height, weight, body mass index (BMI), weight change,...etc with interpretations e.g loss of 30% of body weight over the last 4 months =severe malnutrition | .5 |
| | Biochemical data: Laboratory data with interpretation e.g albumen 1.5mg/dl = severe hypoalbuminemia | .5 |
| | Nutrition Diagnosis (PES statement) should be Clear, concise, Specific, Related to one problem, Accurate – related to one etiology, Based on reliable, accurate assessment data. address all nutrition problems and clearly differentiate medical diagnosis from nutrition diagnosis (it is not allowed to write medical problems in place of nutritional problems) | 1.5 |
| | Nutrition Intervention: put objective for each category<br>1. Food and/or nutrient delivery (ND): including clear objectives for each of:<br>Meals and snacks with complete diet planning and analysis<br>Enteral/parenteral nutrition<br>Medical food supplements<br>Vitamin and mineral supplement<br>Bioactive substance supplement<br>Feeding assistance<br>Feeding environment<br>Nutrition-related medication management<br>2. Nutrition education (E)<br>3. Nutrition counseling (C)<br>4. Coordination of nutrition care (RC) | 4 |
| | Prioritization of the nutrition diagnoses as to each problem's severity or importance | .5 |
| | Interventions should be specific: —What? —When?—Where?—How?<br>e.g increase HB level to a minimum of 12 g/dl in 6 weeks<br>iron supplements 60 mg once /day with fruit juice | .5 |
| | Nutrition Monitoring and Evaluation | .5 |

- you are expected to critically think in the following ways:

1. Determine appropriate data to collect
2. Determine the needs for additional information
3. Select assessment tool that match the situation
4. Apply assessment in valid and reliable way
5. Distinguish relevant from irrelevant

6. Distinguish important from non important
7. Validate the data