

Metabolic integration and disorders

(CHS 344) – 2 credits

Second Semester, 2014-2015/1435-1436H

Lecturer:

Dr. Sadeem Aljammaz

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The best way to reach me is by email, I do my best to answer within one working day. Always include your name in your email; I will not answer emails without student names.

Time and location:

Group **39136**: Sunday 8am to 11am. Room S 1.

Group **26324**: Thursday 8am to 11am. Room S 31.

Course description:

In this course the student will learn about some inborn errors of metabolism and the importance of newborn screening. The course will cover the clinical presentation, etiology, prognosis, and the nutritional management of each disorder.

Textbook:

Fernandes, J., Saudubray, J.M., van den Berghe, G., & Walter, J. H. (Eds.) (2006). *Inborn Metabolic Diseases: Diagnosis and Treatment*. (4th Ed). Heidelberg, Germany: Springer.

Classroom policies and expectations:

- **Assignments:** To be handed in one week after being assigned; during the following class. It will not be accepted if handed in later.
- **Attendance:** regular on-time attendance is expected, continued lateness will be considered absence.
- **Participation:** in class discussions are strongly encouraged. Not participating at all in class discussions will affect your participation grade. One mark will be deducted for unexplainable laughs or side talks.
- **Cell phones** must be kept on silent mode;
 - Texting and talking on the phone during class will not be tolerated.
 - If you need to answer an important call, you may step out of the room and come back when finished.
- **Ear phones** are not allowed during class.
- **Laptops and iPads** may be brought to class to take notes as long as typing is not disturbing other students:
 - This policy can change if there are complaints by your classmates.
 - You may only use your laptop for note taking – other uses are not allowed of course.
- **Cheating and plagiarism** are not accepted of course, nor tolerated.
- If you have questions about the cases or the assignments, please feel free to ask at any time during class, as the answers will most likely benefit your classmates as well.

Midterms and Final exam:

They will assess the information and knowledge you have learnt in the course. They'll be based on class discussions, book chapters, and your notes.

There will be **NO** makeup exams if you missed the scheduled exam.

PLEASE NOTE:

- During all exams abayas and cell phones are not allowed.
- If you are more that 10-minutes late from the start of the exam, you will lose a mark for lateness and disruption to your classmates.

Grading structure:

1 st Midterm	20
2 nd Midterm	20
Quizzes	10
Attendance and assignments	8
Discussion and participation at class	2
Final	40
Total	100

Course Syllabus:

List of topics	Number of weeks
Introduction and newborn screening	1
Classification of inborn errors of metabolism and present treatments and new trends	1
Disorders of carbohydrate metabolism	2
1st Midterm. Week 5: Monday or Tuesday 12pm.	-
Aminoacidopathies: hyperphenylalaninaemias, tyrosinaemias, and urea cycle disorders	4/5
2nd Midterm. Week 11: Monday or Tuesday 12pm.	-
Aminoacidopathies: Branched-chain organic acidemias	2/3
Disorders of mitochondrial fatty acid oxidation	1
Practical final exam. Week 16.	-
Final. Week 17/18.	-