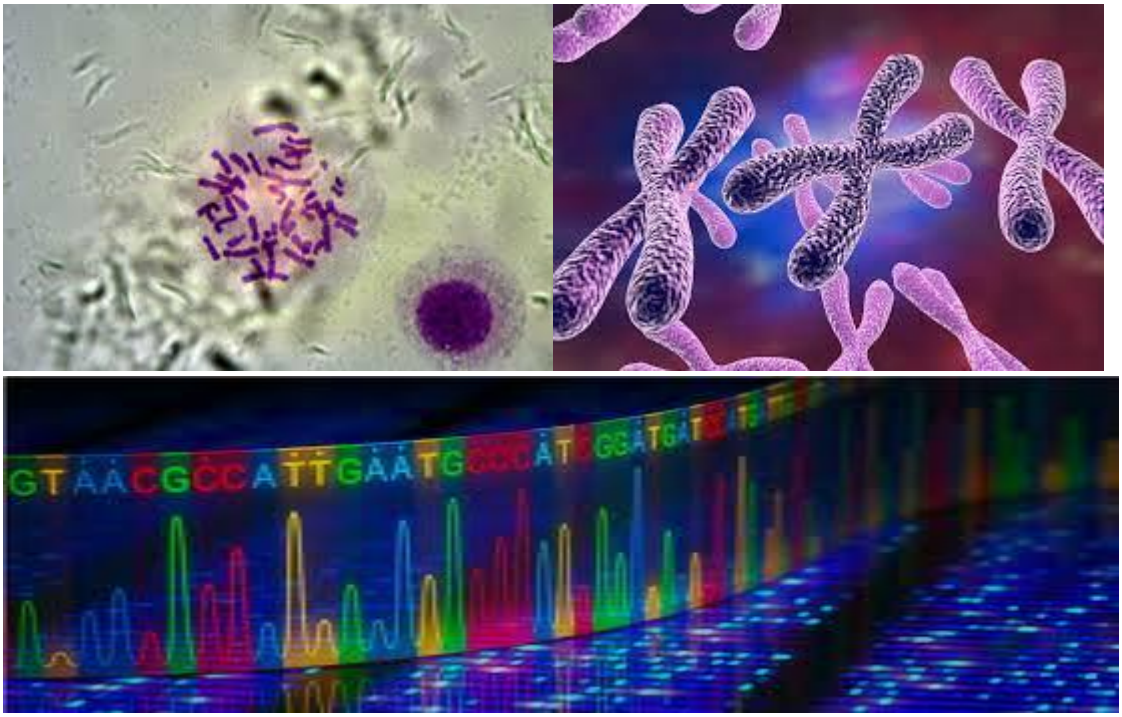
Kingdom of Saudi Arabia
Ministry of Higher Education
Northern Border University (NBU)
College of Medicine
Department of Biochemistry

المملكة العربية السعودية
وزارة التعليم
جامعة الحدود الشمالية
كلية الطب
قسم الكيمياء الحيوية

# STUDY GUIDE

## Medical Genetics

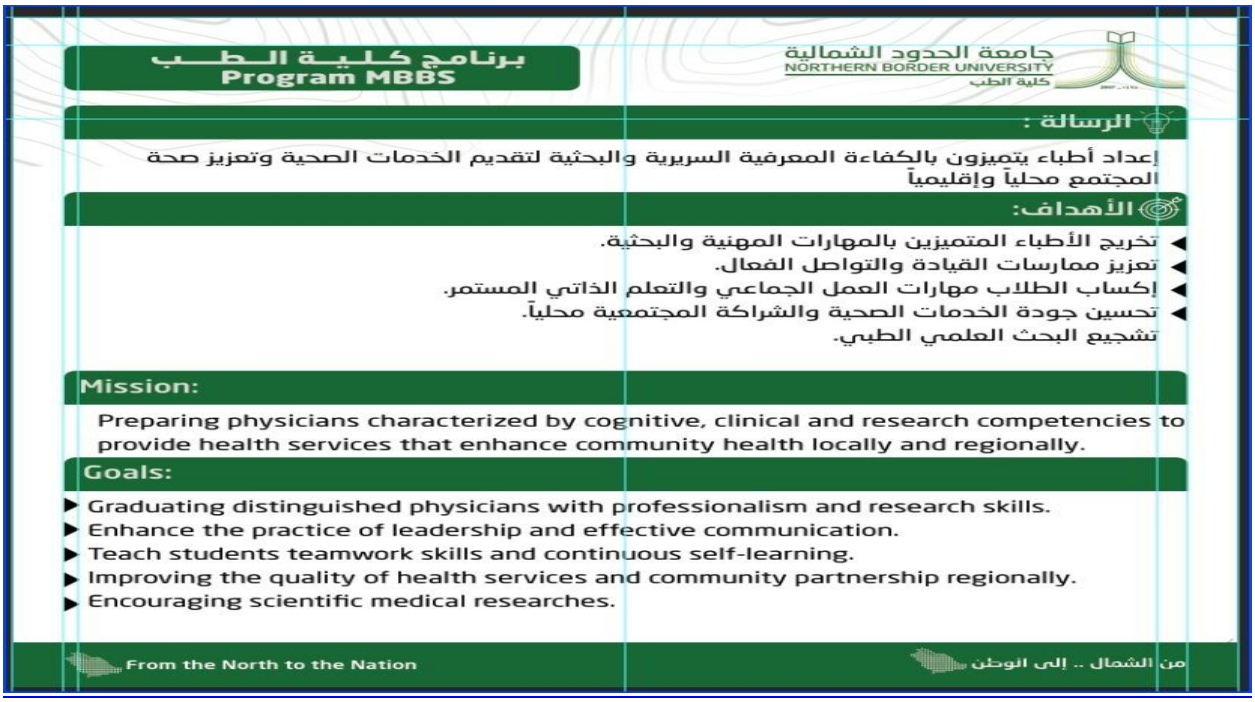
## 1211411




**Course coordination**

Female section **Dr. Naglaa Mokhtar** (Naglaa.Ibrahim@nbu.edu.sa)

Male section **Dr. Abdelnaser Badway** (abdelnaser.ali@nbu.edu.sa)


برنامج كلية الطب
Program MBBS
جامعة الحدود الشمالية
NORTHERN BORDER UNIVERSITY
كلية الطب
الرسالة :
إعداد أطباء يتميزون بالكفاءة المعرفية السريرية والبحثية لتقديم الخدمات الصحية وتعزيز صحة المجتمع محلياً وإقليمياً
الأهداف:
تخريج الأطباء المتميزين بالمهارات المهنية والبحثية.
تعزيز ممارسات القيادة والتواصل الفعال.
إكساب الطلاب مهارات العمل الجماعي والتعلم الذاتي المستمر.
تحسين جودة الخدمات الصحية والشراكة المجتمعية محلياً.
تشجيع البحث العلمي الطبي.
Mission:
Preparing physicians characterized by cognitive, clinical and research competencies to provide health services that enhance community health locally and regionally.
Goals:
Graduating distinguished physicians with professionalism and research skills.
Enhance the practice of leadership and effective communication.
Teach students teamwork skills and continuous self-learning.
Improving the quality of health services and community partnership regionally.
Encouraging scientific medical researches.
From the North to the Nation
من الشمال .. إلى الوطن

## Course Identification

| | |
|---|---|
| **1. Credit hours** | 1 |
| **2. Level/year at which this course is offered** | 4th year – $10^{th}$ ,$11^{th}$ , $12^{th}$ semester |
| **3. Pre-requisites for this course** | **Pass in $2^{nd}$ and $3^{rd}$ year** |

## Course contributors names

1- Prof. Manal Said Fawzy
2- Dr. Abdelnaser Badway
3- Dr. Naglaa Mokhtar

## Actual Learning Hours (Copy and paste the table from courses specification)

| No | Activity | Learning Hours |
|---|---|---|
| **Contact Hours** | | |
| **1** | **Lecture** | **11** |
| **2** | **Laboratory/Studio** | **2** |
| **3** | **Tutorial** | **1** |
| **4** | **Others (specify)** C/P &PBL | **2** |
| | **Total** | **16** |

## Course Objectives (Copy and paste the table from courses specification)

### 1. Course Description

This course is intended to help the students to gain the basic knowledge about chromosome structure, organization and function. The course discusses the patterns of inheritance and explains the types of mutations and their effect. The course illustrates clinical features of common chromosomal abnormalities and describes cytogenetic diagnostic techniques and their applications to genetic disorders. The course focuses on how constitutional and acquired genetic alterations can lead to the development of malignant neoplasms and the course also, summarize conventional approaches for treatment of genetic diseases and the general status of gene-based therapies

### 2. Course Main Objectives

1. Recall sufficient knowledge about gene structure, organization, function, control and segregation.
2. Explain patterns of inheritance and characteristics of autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive traits.
3. Recognize the types of mutations and their contribution to human variation and disease.
4. Outline the clinical features of common numerical, structural, and mosaic chromosomal abnormalities.
5. State common molecular and cytogenetic diagnostic techniques and their applications to genetic disorders.
6. Explain how constitutional and acquired genetic alterations can lead to the development of malignant neoplasms and how identification of these changes can be used in the diagnosis, management and prevention of malignancy.
7. Compare the alternative approaches and goals of different screening programs for genetic diseases in newborn infants, pregnant women, and adults, including their ethical issues.
8. Summarize conventional approaches for treatment of genetic diseases and the general status of gene-based therapies.
9. Recognize how DNA and RNA are extracted and subsequent usage in downstream techniques.

## Course Learning Outcomes (Copy and paste the table from courses specification)

| CLOs | | Aligned PLOs |
|---|---|---|
| 1 | **Knowledge and Understanding** | |
| 1.1 | Describe the structure of gene and chromosome, and their biological functions, in health and disease | K1 |

| CLOs | | Aligned PLOs |
|---|---|---|
| **2** | **Skills:** | |
| **2.1** | Interpret the results of clinical, laboratory, and radiological findings for proper problem solving and decision making | S1 |
| 2.2 | Interpret DNA, RNA and protein analytical techniques for genetic diseases in newborn infants, pregnant women, and adults, including their ethical issues | S2 |
| 2.3 | Communicate effectively with peers and instructors | S6 |
| **3** | **Values:** | |
| 3.1 | Demonstrate ethical attitude with colleagues and facilitators | V1 |
| 3.2 | Operate self-learning from updated medical information from different approved sources in the web. | V2 |
| 3.3 | Demonstrate the capacity for self-reflection and personal development | V2 |

## Course Content (Copy and paste the table from courses specification)

| No | List of Topics | Contact Hours |
|---|---|---|
| 1 | • Structure of chromosomes<br>• Mitosis and Meiosis Karyotyping<br>• Numerical chromosomal abnormalities<br>• Methods of inheritance | 2 |
| 2 | Nucleic acids Biochemistry | 2 |
| 3 | • Types of mutations<br>• Clinical application (Hemoglobinopathies<br>• Genotypes and Phenotypes | 1 |
| 4 | • Restriction Endonucleases and Vectors<br>• Gene cloning | 2 |

| | | |
|---|---|---|
| | • Transgenic animals, gene therapy and cell-based therapy | |
| 5 | • Preparation of different probes<br>• Applications of probes in<br>- Forensic medicine<br>- Prenatal diagnosis<br>- Genetic screening in infants and pregnant women<br>- Pedigree analysis | 2 |
| 6 | • Genes of cancer (Oncogenes and tumor suppressors)<br>• Mechanism of cancer<br>• Apoptosis and cell death | 2 |
| 7 | Practical: Southern and northern blotting<br>DNA sequencing and microarrays<br>Chromosomal culture | 2 |
| 8 | Tutorial : Gene therapy - transgenic animals – gene knockout animals | 1 |
| 9 | PBL: Chromosomal abnormalities diseases, e.g . Down syndrome. | 1 |
| 10 | C/P: DNA fingerprinting and paternity problems | 1 |
| **Total** | | 16 |

## Teaching strategies and Assessment Methods for Students (Copy and paste the table from courses specification)

| Code | Course Learning Outcomes | Teaching Strategies | Assessment Methods |
|---|---|---|---|
| **1.0** | **Knowledge and Understanding** | | |
| **1.1** | Describe the structure of gene and chromosome, and their biological functions, in health and disease | Direct instructional (Lectures)<br>Interactive (tutorial, PBL, CP) | - Written exams included MCQs & SAQs.<br>PBL Checklist<br>CP checklist |
| **2.0** | **Skills** | | |
| **2.1** | Interpret the results of clinical, laboratory, and radiological findings for proper problem solving and decision making | Interactive (tutorial, PBL, CP) | PBL Checklist<br>CP checklist |
| **2.2** | Interpret DNA, RNA and protein analytical techniques for genetic diseases in newborn infants, pregnant women, and adults, including their ethical issues | Lab based strategies (demonstration, Direct instruction, cooperative) | OSPE |
| **2.3** | Communicate effectively with peers and instructor | Interactive (tutorial, PBL, CP) | CP checklist |
| **3.0** | **Values** | | |
| 3.1 | Demonstrate ethical attitude with colleagues and facilitators | Interactive (tutorial, PBL, C | PBL Checklist<br>CP checklist |
| 3.2 | Operate self-learning from updated medical information from different approved sources in the web. | Interactive (tutorial, PBL, CP) | PBL Checklist<br>CP checklist |

| Code | Course Learning Outcomes | Teaching Strategies | Assessment Methods |
|---|---|---|---|
| 3.3 | Demonstrate the capacity for self-reflection and personal development | Interactive ( PBL) | PBL Checklist |

## Assessment Tasks for Students (Copy and paste the table from courses specification)

| # | Assessment task* | Week Due | Percentage of Total Assessment Score |
|---|---|---|---|
| Quiz | 1st | 20% | Quiz |
| C/P | 1st | 10% | C/P |
| PBL | 1st | 10% | PBL |
| OSPE | End of module | 20% | OSPE |
| Final exam | End of module | 40% | Final exam |

## Course blueprint (% of total summative marks in blueprint is to be given in the range)

| Topics | Teaching strategies | Assessment methods | Knowledge and Understanding | | | Skills | | | values | | | % of total contact hours | % of total summative marks |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | K1 | K2 | … | S1 | S2 | S 6 | V1 | V2 | … | | |
| • Structure of chromosomes<br>• Mitosis and Meiosis Karyotyping<br>• Numerical chromosomal abnormalities<br>• Methods of inheritance | Direct instructional (Lectures) | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 12 |
| Nucleic acids Biochemistry | Direct instructional (Lecture | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 12 |

| Topics | Teaching strategies | Assessment methods | Knowledge and Understanding | | | Skills | | | values | | | % of total contact hours | % of total summative marks |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | K1 | K2 | … | S1 | S2 | S 6 | V1 | V2 | … | | |
| | s) | | | | | | | | | | | | |
| • Types of mutations<br>• Clinical application (Hemoglobinopathies)<br>• Genotypes and Phenotypes | Direct instructional (Lectures) | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 12 |
| • Restriction Endonucleases and Vectors<br>• Gene cloning<br>• Transgenic animals, gene therapy and cell-based therapy | Direct instructional (Lectures) | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 12 |
| • Preparation of different probes<br>• Applications of probes in<br>- Forensic medicine<br>- Prenatal diagnosis | Direct instructional (Lectures) | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 11 |

| Topics | Teaching strategies | Assessment methods | Knowledge and Understanding | | | Skills | | | values | | | % of total contact hours | % of total summative marks |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | K1 | K2 | … | S1 | S2 | S 6 | V1 | V2 | … | | |
| - Genetic screening in infants and pregnant women<br>• Pedigree analysis | | | | | | | | | | | | | |
| • Genes of cancer (Oncogenes and tumor suppressors)<br>• Mechanism of cancer<br>• Apoptosis and cell death | Direct instructional (Lectures) | Written exams (MCQs & SAQs) | K1 | | | | | | | | | 2 | 11 |
| Southern and northern blotting | Laboratory-based strategy) | Written exams (MCQs & SAQs) | | | | S 1 | S 2 | | | | | 1 | 10 |
| DNA sequencing and microarrays<br>Chromosomal culture | Laboratory-based strategy) | OSPE | | | | S 1 | S 2 | | | | | 1 | 10 |
| Tutorial : Gene therapy - transgenic animals – gene knockout animals | Interactive | | | | | S 1 | | | | | | 1 | 0 |

| Topics | Teaching strategies | Assessment methods | Knowledge and Understanding | | | Skills | | | values | | | % of total contact hours | % of total summative marks |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | K1 | K2 | … | S1 | S2 | S 6 | V1 | V2 | … | | |
| PBL: Chromosomal abnormalities diseases, e.g . Down syndrome. | Interactive | PBL Checklist | K1 | | | S 1 | | **S 6** | **V 1** | **V 2** | | 1 | 5 |
| C/P: DNA fingerprinting and paternity problems | Interactive | CP checklist ) | K1 | | | | | **S 6** | **V 1** | **V 2** | | 1 | 5 |

## Learning Resources (Copy and paste the table from courses specification)

| | |
|---|---|
| ¤ **Required Textbooks** | Emery's Elements of Medical Genetics (12th Ed.) 2005 by Peter D. Turnpenny, Sian Ph.D. Ellard, Churchill Livingston, New York, USA |
| **Essential References Materials** | Thompson & Thompson Genetics in Medicine (6th Ed) 2004. by Robert L. Nussbaum, Roderick R. McInnnes, Huntington F. Willard W.B. Saunders Company, London, UK, |
| **Electronic Materials** | Web Sites<br>• http://www.genome.gov<br>• • http://www.geneclinics.org. |
| **Other Learning Materials** | Recommended Books and Reference Material<br>• Public Health Genomics. Journal Abbreviation: Public Health Genomics. www.karger.com/PHG ISSN 1662-4246 (Print) e-ISSN 1662-8063 (Online)<br>• http://www.genetics.org/<br>Other learning material such as computer-based programs/CD, professional standards/regulations<br>•http://www.genome.gov/Pages/Education/DNADay/Animations/MakingSNPsMakeSense.html |

## Related check lists

PBL √
Assignment
Clinical skills checklist
Presentation checklist √
Project checklist
Workshop checklist
(Checklist must be aligned with the learning outcomes)

# Assessment of Student in PBL MBBS 4rd Year 1443/1444

**Facilitator's name:** **Group number:** ..... **Module: Medical genetics**

The assessment items are grouped under two main headings
(Total marks to give for each case = 6 marks)

**A. Learning and cognitive skills:**

| No | Ability of the student to | Marks | Marks Obtained |
|---|---|---|---|
| 1. | Attend to PBL session on time | 1 | |
| 2. | Take active roles such as scribing/ becoming a leader | 1 | |
| 3. | Identify New/difficult words in the case | 1 | |
| 4. | Participate effectively during the session (Sharing appropriate ideas and information) | 1 | |
| 5. | Interpret the case and the related findings | 1 | |
| 6. | Generate learning issues | 1 | |
| | **Total marks for Session 1** | **6** | |

**B. Interaction and participation to group function:**

| No | Ability of the student to | Marks | Marks Obtained |
|---|---|---|---|
| 1. | Build discussion and Maintain good group dynamics | 1 | |
| 2. | Present his/her learning issue effectively | 1 | |
| 3. | Prepare well with all the identified learning issues | 1 | |
| 4. | Manage presentation time well | 1 | |
| 5. | Communicate effectively and discuss collaboratively with other members | 1 | |
| 6. | Ask and answer questions relevant to the topic | 1 | |
| | **Total marks for Session 2** | **6** | |

## Students Score's for PBL Cases in the Medical genetics module

| No | Students names | Students ID | Marks in PBL1a (out of 6) | Marks in PBL1b (out of 6) | Marks in PBL1 (out of 12) | Total score (Out of 10 marks) |
|---|---|---|---|---|---|---|
| 1. | | | | | | |
| 2. | | | | | | |
| 3. | | | | | | |
| 4. | | | | | | |
| 5. | | | | | | |
| 6. | | | | | | |
| 7. | | | | | | |
| 8. | | | | | | |
| 9. | | | | | | |

**Any Comments:**

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**Facilitator's Name:** **Signature:** **Date:**

## CP Evaluation Rubric

| | Good = 3 (Exemplary) | Fair = 2.5 (Competent) | Needs improvement = 2 (Developing) | Score |
|---|---|---|---|---|
| **Content** | Presentation includes **all** information **relevant** to the topic. | Presentation does **not** have all information relevant to the topic. | The Presentation is **lacking** in elements required. | |
| | Information arranged in an **organized** fashion. | Information is presented in an **unorganized** fashion. | There are many **gaps** in information presented. | |
| **Presentation mechanics and layout** | The Presentation has an element of **creativity** and **style**. | Presentation is **not creative**. | The Presentation **lacks style**. | |
| | **No** error: grammatical, spelling punctuation | **Few** errors: grammatical, spelling punctuation | **Many** errors: grammatical, spelling punctuation | |
| **Presentation** | The presenter speaks clearly and puts information with complete **understanding**. The presenter has **prepared** what he/she will say. | There are several **confusing** moments among and the information presented is not completed. The presenter has **not practiced** to prepare. | The information that is presented **lacks** the **essential** elements of what is expected. **No preparation** was done for the presentation. | |
| | **Effective** body language. **Maintained** eye contact with audience. | **Puzzling** body language. **Interrupted** eye contact with audience. | **Negative** body language: (Staring, crossed arms, overuse of hands, poor posture, frowning). **Avoidance** of eye contact with audience. | |
| | **Presentation topic/ID:** | | | |
| | **Please put your total score / 10** | | | |

## Course quality evaluation

After the end of the course, please give your **FEEDBACK** through the following link:

https://docs.google.com/forms/d/1u7KxjAURMIwH4HjWtVVjwO5CUjKZLs1ATyO6uTU61yM/edit