

Program SPECIFICATION FOR Master Degree in Human Genetics

Code: 1713700

University: Alexandria

Faculty: Medical Research Institute

Program Specification

A- Basic information

1- Program title: M.Sc. Human Genetics

2- Program type: single double multiple

3- Department(s): Human Genetics

4- Coordinator: Assistant professor Dr. Lubna Mohamed

5- External evaluator(s): Dr. Rabah Shawky

6- Last date of program specification approval: 5/6/2014

B- Professional Information

1- Program aims:

By end of the program, the student should:

1. Differentiate chromosomal basis of heredity and the patterns of single gene disorders, non classical modes of inheritance, mitochondrial inheritance and multifactorial inheritance
2. Recognize the fundamental concepts in molecular genetics, know the different types of mutations and polymorphism and have comprehensive knowledge of the tools and techniques of molecular genetics.
3. Recall the fundamental concepts the biochemical basis of single gene inheritance and have comprehensive knowledge on inborn errors of metabolism.
4. Distinguish the molecular basis of cancer, the genetics of the immune system and the concepts of pharmacogenetics.
5. Recall the role of cytogenetics in medicine and have comprehensive knowledge on human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies and sex chromosomes abnormalities.
6. Explore the genetic basis of craniofacial disorders, deafness, short stature and muscular dystrophy.
7. Recall the fundamental concepts and methods in population screening and prenatal diagnosis and the role of genetic counseling in providing information and risk assessment.
8. Differentiate the various methods used for treatment of genetic disorders the concept of gene therapy.
9. Examine the genetic variation in populations, principles of population genetics and Hardy-Weinberg law.

10. Distinguish the genetic aspect of development and clinical aspects of human teratology.
11. Explore skills in information technology
12. Use systematic approaches to design and conduct scientific research.

2- Intended learning outcomes (ILOs)

a- knowledge and understanding:

- a1-** Recall the role of genetics in medicine, recall the structure and function of genes and chromosomes, understand the different modes of inheritance of single gene disorders and chromosomal basis of heredity.
- a2-** Recognize genetic counseling and recurrence risk assessment and recall population screening for genetic diseases and the various methods used for prenatal diagnosis and for treatment of genetic disorders.
- a3-** Differentiate the different tools used in molecular genetics, define the different types of human mutations, recognize the role of gene mutations in genetic diseases and list the molecular techniques used for diagnosis of genetic disorders.
- a4-** list the metabolic pathways in each of the inborn errors of metabolism, recall the relationship between the biochemical abnormalities and the phenotypic expression of the disease, and discuss the different types of treatment of those diseases.
- a5-** Discuss genetic diversity in human populations, distinguish phenotype, genotype, and gene frequencies, discuss the Hardy-Weinberg law and distinguish the underlying principles of disorders with complex inheritance
- a6-** Recall the role of genetics in cancer, list the types of genes that have been implicated in initiating cancer, describe the genetic basis of the immune response and discuss diseases in which immune-related genes contribute to disease.
- a7-** Explore the recent developments in the field of human genetic sciences
- a8-** Recall the role of cytogenetics in medicine, discuss the different cytogenetic techniques and compare anomalies of autosomes and sex chromosomes
- a9-** Analyze patterns of human malformations and recall the craniofacial disorders, deafness and syndromes of short stature
- a10-** Differentiate the genetic aspects of development, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.
- a11-** Recognize the role of cytogenetics in medicine and recall the various chromosome abnormalities and the different methods used in cytogenetics
- a12-** Recall a group of inborn errors of amino acid, carbohydrate, purine and pyrimidine metabolism,

and urea cycle disorders, disorders of protein glycosylation and hyperlipoproteinemias

a13- Differentiate the genetic basis and pathophysiology of a group of inborn errors of metabolism including sphingolipidoses, oligosaccharidosis, porphyrias and disorders of peroxisomes, lipid, lipoprotein, fatty acids, copper, iron and vitamin D metabolism

a14- Explore the role of genes in the human gastrointestinal, lymph and blood diseases

b- Intellectual skills:

b1- Recognize the role of genetics in medicine, relate the structure and function of genes and chromosomes, differentiate between the various modes of inheritance, and apply basic genetic knowledge to clinical problem solving.

b2- Recall the indications of prenatal diagnosis, differentiate between the various methods used for treatment of genetic disorders, and relate population screening for genetic disease to maximize the benefits and minimize any harm.

b3- Differentiate between various types of mutations and polymorphism, evaluate the principles of molecular diseases and disorders of hemoglobin and apply basic molecular knowledge to solve the problems of genetic diseases.

b4- Recognize the biochemical basis of inborn errors of metabolism, relate the precise biochemical abnormalities and the disease phenotype and relate the treatment of the inborn error to the biochemical defect.

b5- Distinguish the genetic diversity in human populations, demonstrate the factors that disturb Hardy-Weinberg equilibrium and evaluate cases with complex inheritance and differentiate between disorders with complex inheritance and other modes of inheritance.

b6- Recognize the role of genetics in cancer, the application of pharmacogenetics, and differentiate between the various gene systems that govern immune function.

b7- Explore the recent developments in the field of human genetic sciences

b8- Recall the role of cytogenetics in medicine, differentiate between the various methods used in cytogenetics and evaluate cases with anomalies of autosomes and sex chromosomes

b9- Differentiate cases with proportionate and disproportionate short stature, causes of human malformations and evaluate cases with craniofacial disorders, deafness and short stature

b10- Recall the genetic aspects of development and evaluate cases with limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.

b11- Recognize the role of cytogenetics in medicine and distinguish the various chromosome abnormalities and the different methods used in cytogenetics

b12- Distinguish the genetics and metabolic features of inborn errors of amino acid, carbohydrate, purine and pyrimidine metabolism, and compare urea cycle disorders, disorders of protein glycosylation and hyperlipoproteinemias

b13- Recall the genetic basis and pathophysiology of a group of inborn errors of metabolism including sphingolipidoses, oligosaccharidosis, porphyrias and disorders of peroxisomes, lipid, lipoprotein, fatty acids, copper, iron and vitamin D metabolism

b14- Distinguish the role of genes in the human gastrointestinal, lymph and blood diseases

b15- Write a thesis protocol using a scientific systematic approach to a research problem.

c- professional and practical skills:

c1- Construct and examine a pedigree

c2- Develop skills to differentiate between the tools used in molecular genetics

c3- Develop skills to diagnose the inborn errors of metabolism

c4- Construct and examine a karyotype

c5- Assess cases with different malformations

c6- Develop skills in clinical genetic examination and evaluation

d- General and transferable skills:

d1- Communicate through group discussion

d2- Work as a part of team

d3- Develop skills in information technology

d4- Develop skills for oral presentation

d5- Develop skills in reading and research

3- Academic standards

3a External references for standards (Benchmarks)

Generic Academic Reference Standards of the National Authority for Quality Assurance and Accreditation of Education (NAQAAE)

Adopted at MRI council 12/2/2014 and re-adopted at 15/1/2023

Last date of Academic Reference standards (ARS) approval by Institute

Council: 15/1/2023

3b Comparison of provision to selected external references

Comparison between Generic Academic Standards of NAQAAE and ARS of Master degree in Human Genetics

<div style="text-align: right;">ARS</div> <div style="text-align: left;">NAQAAE</div>	ARS for M.Sc. in Human Genetics
A1-Basic facts, theories, of the specialty and related subjects/ fields	<p>a1- Recognize the role of genetics in medicine, recall the structure and function of genes and chromosomes.</p> <p>a2- Discuss genetic counseling and the various methods used for prenatal diagnosis and for treatment of genetic disorders.</p> <p>a3- Recognize the molecular basis of inherited disease and the role of proteomics in genetic disorders.</p> <p>a4- Recall the metabolic pathways in the inborn errors of metabolism.</p> <p>a5- Review genetic diversity in human populations and principles of genetic epidemiology.</p> <p>a6- Recognize the role of cytogenetics in medicine, recall the various chromosome abnormalities and the different methods used in cytogenetics and discuss anomalies of autosomes and sex chromosomes, and recognize the role of genetics in cancer.</p> <p>a7- Describe patterns of human malformations, review genetic aspects of development, and evaluate cases with craniofacial disorders, deafness. short stature, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.</p>
A2- Mutual relation between professional practice and effects on environment	<p>a7- Describe patterns of human malformations, review genetic aspects of development, and evaluate cases with craniofacial disorders, deafness. short stature, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.</p>
A3- Main scientific advances in the field of practice	<p>a7- Describe patterns of human malformations, review genetic aspects of development, and evaluate cases with craniofacial disorders, deafness. short stature, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.</p>
A4-Fundamentals of ethical & legal practice	<p>a8- Recognize the fundamentals of ethical and legal practice in Medical Genetics.</p>
A5 -Quality standards of the practice	<p>a6- Recognize the role of cytogenetics in medicine, recall the various chromosome abnormalities and the different methods used in cytogenetics and discuss anomalies of autosomes and sex chromosomes, and recognize the role of genetics in cancer.</p> <p>a7- Describe patterns of human malformations, review genetic aspects of development, and evaluate cases with craniofacial disorders, deafness. short stature, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.</p>
A6- Basics and ethics of scientific research	<p>a2- Discuss genetic counseling and the various methods used for prenatal diagnosis and for treatment of genetic disorders.</p>
B1 -Interpret, analyze & evaluate the information to solve problems	<p>b1- Interpret, analyze and evaluate basic genetic information to solve problems</p>

<p>B2- Solve some problems that do not conform to classic data (incomplete data)</p>	<p>b2- Relate the structure and function of genes and chromosomes. b4- Review molecular basis of genetic diseases and proteomics in relation to genetic diseases b5- Relate the precise biochemical abnormalities and the disease phenotype and demonstrate the application of pharmacogenetics b7- Differentiate between the various methods used in cytogenetics, demonstrate the syndromes with autosomal anomalies, and evaluate cases with sex chromosome anomalies, review role of genetics in cancer. b8- Evaluate cases with craniofacial disorders, deafness, neural tube defects, fragile X syndrome, overgrowth syndromes, and reproductive disorders, differentiate cases with proportionate and disproportionate short stature and cases with muscle dystrophy, congenital myopathies or spinal muscle atrophy. Discuss clinical aspects of human teratology and evaluate cases exposed to different teratogens.</p>
<p>B3- Integrate different information to solve professional problems</p>	<p>b3- Demonstrate the value of prenatal diagnosis and the various methods used for treatment of genetic disorders. b5- Relate the precise biochemical abnormalities and the disease phenotype and demonstrate the application of pharmacogenetics. b6- Explain the genetic diversity in human populations, demonstrate the factors that disturb Hardy-Weinberg equilibrium and evaluate cases with complex inheritance and differentiate between disorders with complex inheritance and other modes of inheritance. b7- Differentiate between the various methods used in cytogenetics, demonstrate the syndromes with autosomal anomalies, and evaluate cases with sex chromosome anomalies, review role of genetics in cancer. b8- Evaluate cases with craniofacial disorders, deafness, neural tube defects, fragile X syndrome, overgrowth syndromes, and reproductive disorders, differentiate cases with proportionate and disproportionate short stature and cases with muscle dystrophy, congenital myopathies or spinal muscle atrophy. Discuss clinical aspects of human teratology and evaluate cases exposed to different teratogens.</p>
<p>B4- Conduct a scientific research &/Or write scientific systematic approach to a research problem (hypothesis)</p>	<p>b9- Write a thesis protocol using a scientific systematic approach to a research problem.</p>
<p>B5- Evaluate risks imposed during professional practice.</p>	<p>b7- Differentiate between the various methods used in cytogenetics, demonstrate the syndromes with autosomal anomalies, and evaluate cases with sex chromosome anomalies, review role of genetics in cancer. b8- Evaluate cases with craniofacial disorders, deafness, neural tube defects, fragile X syndrome, overgrowth syndromes, and reproductive disorders, differentiate cases with proportionate and disproportionate short stature and cases with muscle dystrophy, congenital myopathies or spinal muscle atrophy. Discuss clinical aspects of human teratology and evaluate cases exposed to different teratogens.</p>

<p>B6- Plan for professional improvement</p>	<p>b7- Explore the recent developments in the field of human genetic sciences</p>
<p>B7- Take professional decisions in wide range of professional situations</p>	<p>b1- Interpret, analyze and evaluate basic genetic information to solve problems b2- Relate the structure and function of genes and chromosomes. b4- Review molecular basis of genetic diseases and proteomics in relation to genetic diseases b5- Relate the precise biochemical abnormalities and the disease phenotype and demonstrate the application of pharmacogenetics. b6- Explain the genetic diversity in human populations, demonstrate the factors that disturb Hardy-Weinberg equilibrium and evaluate cases with complex inheritance and differentiate between disorders with complex inheritance and other modes of inheritance. b7- Differentiate between the various methods used in cytogenetics, demonstrate the syndromes with autosomal anomalies, and evaluate cases with sex chromosome anomalies, review role of genetics in cancer. b8- Evaluate cases with craniofacial disorders, deafness, neural tube defects, fragile X syndrome, overgrowth syndromes, and reproductive disorders, differentiate cases with proportionate and disproportionate short stature and cases with muscle dystrophy, congenital myopathies or spinal muscle atrophy. Discuss clinical aspects of human teratology and evaluate cases exposed to different teratogens.</p>
<p>C1- Competent in all basic and some of the advanced professional skills (to be determined according to the specialty board/ department)</p>	<p>c1- Differentiate between the tools used in molecular genetics c2- Diagnose inborn errors of metabolism. c3- Construct and examine a karyotype. c4- Construct and examine a pedigree and gain skills in clinical genetic examination and evaluation</p>
<p>C2- Write and appraise reports</p>	<p>c1- Differentiate between the tools used in molecular genetics c2- Diagnose inborn errors of metabolism. c3- Construct and examine a karyotype. c4- Construct and examine a pedigree and gain skills in clinical genetic examination and evaluation</p>
<p>C3- Evaluate methods and tools used in specialty</p>	<p>c1- Differentiate between the tools used in molecular genetics c2- Diagnose inborn errors of metabolism. c3- Construct and examine a karyotype. c4- Construct and examine a pedigree and gain skills in clinical genetic examination and evaluation.</p>
<p>D1- Communicate effectively using all methods</p>	<p>d1- Communicate through group discussion d2- Work as a part of team d4- Develop skills for oral presentation d5- Develop skills in reading and research.</p>
<p>D2- Use information technology to improve his/her professional practice</p>	<p>d3- Develop skills in information technology</p>
<p>D3- Practice self appraisal and determines his learning needs</p>	<p>d5- Develop skills in reading and research. d3- Develop skills in information technology</p>

4.b.v- Program levels (in credit-hours system)

Student is required to pass at least 12 credit hours with CGPA not less than C+ before submitting a thesis proposal.

5- Program Courses : 5.1- Compulsory (26 CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713701	Basic Human Genetics	3	3	-
1713702	Prevention & Treatment of Genetic Disorders	3	3	-
1713703	Molecular Genetics	3	2	2
1713704	Biochemical Genetics	3	2	2
1713705	population genetics	2	2	-
1713706	Special Genetics	2	2	-
1713707	Recent Topics	1	1	-
1713708*	Clinical Cytogenetics	3	2	2
1713709*	Clinical Genetics I	3	2	2
1713710*	Clinical Genetics II	3	2	2
1713711**	Cytogenetics	2	1	2
1713712**	Biochemical Genetics I	3	2	2
1713713**	Biochemical Genetics II	3	2	2
1713714**	Genes and Diseases	1	1	-

*compulsory for medical students

** compulsory for non medical students

5.2- Elective I (offered by the Human Genetic Dept)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713715	Embryology	1	1	-
1713716	Genetics of reproductive disorders	2	2	-
1713717	Genomics I	1	1	-
1713718	Special biochemical genetics	2	1	2
1713719	Special clinical genetics	2	1	2
1713721	Proteomica & Bioinformatics	2	2	-
1713722	Pharmacogenetics	2	2	-

1713723	Blood genetic Disorders	2	1	2
1708720	Immunology	2	1	2
1718721	Radio-diagnosis	2	1	2
1713724	Genetic epidemiology	2	2	-

5.4- Optional – (none)

6- Program admission requirements

Graduate students with a M.B.Ch.B. of Medicine, B.Sc. of Science or Pharmacy

7- Teaching methods used :

- Lecture
- Practical/Clinical
- Brainstorming
- Discussion Groups
- Problem Solving
- Self-Directed Learning

8- Regulations for progression and program completion

For the progression and completion of the program to obtain the degree of M.Sc, the student must:

- 1- Complete 30 credit hours with CGPA of at least C+ through courses
- 2- Complete 8 credit hours with through thesis.
- 3- Submit a thesis validity report by an examination committee approved by the department council and their members include at least two external examiners.

9- Evaluation of Students enrolled in the program.

Tool evaluation	Intended learning outcomes being assessed
Written	ILOs a & b
Practical	ILOs c
Oral	ILOs a ,b & d
Semester Work	ILOs b & d

Evaluation of the Program

Evaluator	Tool	Sample
1- Senior students	questionnaire	At least 50 %
2- Alumni	questionnaire	Representative sample
3- Stakeholders (Employers)	meeting	Representative sample
4- External Evaluator(S) or External Examiner (s)	Report	Dr. Rabah Shawky
5- Other		

Program coordinator:

Name: Dr Lubna Mohamed

Signature:

Lubna Mohamed

Department Head:

Prof. Dr. Ebtessam Abdalla

Signature *Ebtessam Abdalla*

Date of Department Council Approval: 30/ 8 / 2023

Attributes Vs Aim

Generic Graduate Attributes of NAQAAE	Graduate Attributes By the end of this program, the Graduate <i>should be able to</i>	Program aims
Apply the basics and methodologies of scientific research and using its various tools proficiently.	Integrate knowledge in all areas of 'Medical Genetics' to be able to understand inherited diseases and the diagnostic and screening methodologies which may be employed to benefit individuals, families or populations.	<ul style="list-style-type: none"> -Differentiate chromosomal basis of heredity and the patterns of single gene disorders, non classical modes of inheritance, mitochondrial inheritance and multifactorial inheritance -Recognize the fundamental concepts in molecular genetics, know the different types of mutations and polymorphism and have comprehensive knowledge of the tools and techniques of molecular genetics. -Recall the fundamental concepts the biochemical basis of single gene inheritance and have comprehensive knowledge on inborn errors of metabolism. -Distinguish the molecular basis of cancer, the genetics of the immune system and the concepts of pharmacogenetics
Use the analytical methods in the field of specialty	Run a genetics clinic proficiently, manage cases with genetic diseases in a multidisciplinary approach and conduct comprehensive sessions of genetic counseling.	<ul style="list-style-type: none"> -Recall the fundamental concepts and methods in population screening and prenatal diagnosis and the role of genetic counseling in providing information and risk assessment. -Explore the genetic basis of craniofacial disorders, deafness, short stature and muscular dystrophy. -Distinguish the genetic aspect of development and clinical aspects of human teratology.
Apply specialized knowledge in the field of specialty and integrate it with relevant knowledge in his professional practice.	Upgrade skills to keep with pace of advances in the field and be aware of limitations and challenge	-Differentiate the various methods used for treatment of genetic disorders the concept of gene therapy.
Demonstrate awareness of current problems and modern visions in the field of specialty	. Identify the ethical, legal and socio-cultural issues related to genetic research and genetic testing.	-Recall the fundamental concepts and methods in population screening and prenatal diagnosis and the role of genetic counseling in providing information and risk assessment.
Identify professional problems in the field of specialty and propose solutions to them.	Construct and examine a karyotype, differentiate between the tools used in molecular genetics, diagnose inborn errors of metabolism, construct and examine a pedigree and implement new technologies for diagnosis of rare syndromes and genetic disorders.	.-Recall the role of cytogenetics in medicine and have comprehensive knowledge on human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies and sex chromosomes abnormalities.



Master an appropriate of professional skills in the field of including use of technology.	-Apply principles of genetic research and academic writing and utilize different genetic/genomic databases and data-mining tools. -Evaluate data from different genetic analyses in the context of the clinical situation and respond effectively to limitations of genetic testing.	-Differentiate the various methods used for treatment of genetic disorders the concept of gene therapy.
Communicate efficiently and lead work teams.	Utilize genetic testing resources effectively to balance costs with potential utility of results.	-Examine the genetic variation in populations, principles of population genetics and Hardy-Weinberg law.
Take Decision in different professional contexts.	Incorporate genetic testing and results of genetic research into health-care practice in order to achieve health improvements	-Explore skills in information technology -Use systematic approaches to design and conduct scientific research.
Employ the available resources to achieve the highest benefit and maintain them.	Train and educate junior peers in clinical practice of medical genetics.	
Show awareness of his/her role in community development and environmental preservation in light of global and regional changes.	Develop skills of presenting cases in conferences and meetings and communicate effectively with colleagues within a team work environment.	-Explore skills in information technology -Use systematic approaches to design and conduct scientific research.

