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: Dr. Heba Morsy

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

6- Last date of program specification approval: 8/1/2017

B- Professional Information

1- Program aims:

By end of the program, the student should:

1. Have comprehensive knowledge on 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. Know the molecular basis of cancer, the genetics of the immune system and the concepts of pharmacogenetics.
5. Relate to 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. Have comprehensive knowledge on 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. Have comprehensive knowledge on the various methods used for treatment of genetic disorders the concept of gene therapy.
9. Have comprehensive knowledge on the genetic variation in populations, principles of population genetics and Hardy-Weinberg law.
10. Have a comprehensive knowledge on the genetic aspect of development and clinical aspects of human teratology.
11. Develop skills in information technology

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- Discuss 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- Recall the different tools used in molecular genetics, define the different types of human mutations, explain the role of gene mutations in genetic diseases and list the molecular techniques used for diagnosis of genetic disorders.

a4- Recall the metabolic pathways in each of the inborn errors of metabolism, discuss 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, understand phenotype, genotype, and gene frequencies, discuss the Hardy-Weinberg law and understand the underlying principles of disorders with complex inheritance

a6- Recall the role of genetics in cancer, recall 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- Discuss the recent developments in the field of human genetic sciences

a8- Recall the role of cytogenetics in medicine, discuss the different cytogenetic techniques and discuss anomalies of autosomes and sex chromosomes

a9- Describe patterns of human malformations and recall the craniofacial disorders, deafness and syndromes of short stature

a10- Review the genetic aspects of development, limb defects, overgrowth, neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.

a11- Discuss the role of cytogenetics in medicine and recall the various chromosome abnormalities and the different methods used in cytogenetics

a12- Discuss a group of inborn errors of amino acid, carbohydrate, purine and pyrimidine metabolism,

and urea cycle disorders, disorders of protein glycosylation and hyperlipoproteinemias

a13- 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

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

b- Intellectual skills:

b1- Illustrate 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- Demonstrate 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- Illustrate 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- 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.

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

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

b8- Demonstrate 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, explain causes of human malformations and evaluate cases with craniofacial disorders, deafness and short stature

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

b11- Illustrate 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 illustrate urea cycle disorders, disorders of protein glycosylation and hyperlipoproteinemias

b13- Distinguish 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- Illustrate the role of genes in the human gastrointestinal, lymph and blood diseases

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)

Date of Academic Reference standards (ARS) approval by Institute Council: 12/2/2014

3b - Comparison of provision to selected external references

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

ARS / NAQAAE	ARS for M.Sc. in Human Genetics
A1-Basic facts, theories, of the specialty and related subjects/ fields	**a1-** Recognize the role of genetics in medicine, recall the structure and function of genes and chromosomes. **a2-** Discuss genetic counseling and the various methods used for prenatal diagnosis and for treatment of genetic disorders. **a3-** Recognize the molecular basis of inherited disease and the role of proteomics in genetic disorders. **a4-** Recall the metabolic pathways in the inborn errors of metabolism. **a5-** Review genetic diversity in human populations and principles of genetic epidemiology. **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. **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.
A2- Mutual relation between professional practice and effects on	**a7-** Describe patterns of human malformations, review genetic aspects of development, and evaluate cases with craniofacial disorders, deafness. short stature, limb defects, overgrowth,

environment	neural tube defects, fragile X syndrome, muscular dystrophy and congenital myopathies.
A3- Main scientific advances in the field of practice	**Thesis & assignments**
A4-Fundamentals of ethical & legal practice	**a8-** Recognize the fundamentals of ethical and legal practice in Medical Genetics.
A5 -Quality standards of the practice	**Thesis** **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. **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.
A6- Basics and ethics of scientific research	**Thesis**
B1 -Interpret, analyze & evaluate the information to solve problems	**b1-** Interpret, analyze and evaluate basic genetic information to solve problems
B2- Solve some problems that do not conform to classic data (incomplete data)	**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.
B3- Integrate different information to solve professional problems	**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.
B4- Conduct a scientific research &/Or write scientific systematic approach to a research problem (hypothesis)	**Thesis**
B5- Evaluate risks imposed during professional practice.	**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. d6-Develop skills to work safely in a laboratory environment
B6- Plan for professional improvement	**Student questionnaire**
B7- Take professional decisions in wide range of professional situations	**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.
C1- Competent in all basic and some of the advanced professional skills (to be determined according to the specialty board/ department)	**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

C2- Write and appraise reports	**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
C3- Evaluate methods and tools used in specialty	**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.
D1- Communicate effectively using all methods	**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.
D2- Use information technology to improve his/her professional practice	**d3-** Develop skills in information technology
D3- Practice self appraisal and determines his learning needs	**Thesis** **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.
D4- Share in determination of standards for evaluation	**d5-** Develop skills in reading and research

of others (e.g.: subordinates/ trainees etc.)	
D5- Use different sources of information to obtain data	**d3-** Develop skills in information technology **d5-** Develop skills in reading and research
D6- Work in teams as well as a member in larger teams	**d2-** Work as a part of team
D7- Manage scientific meetings and appropriately utilize time	**d2-** Work as a part of team **d3-** Develop skills in information technology **d4-** Develop skills for oral presentation.

4- curriculum structure and contents

4.a program duration: *3-4 years*

4.b program structure:

4.b.i- No. of hours per week in each year/semester:

Semester	Core Courses	Elective Courses
	No. of hours	No. of hours
First semester	9	
Second semester	6	
Third semester	5	
Fourth semester	3	2
Fifth semester	3	2

4.b.ii- No. of credit hours	**Lectures**	**20**	**Practical**	**10**	**Total**	**30**	
	Compulsory	**26**	**Elective**	**4**	**Optional**	**0**	

4.b.iii- No. of credit hours of basic science courses	**No.**	**2**	**%**	**6.67**
4.b.iv- No. of credit hours of courses of social sciences and humanities.	**No.**	**0**	**%**	**0**
4.b.v- No. of credit hours of specialized courses	**No.**	**28**	**%**	**93.33**
4.b.vi- No. of credit hours of other courses	**No.**		**%**	
4.b.vii- Field Training	**Yes**		**No**	**√**

4.b.viii- 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 hours)

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	Selected 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**

5.3- Elective II (Offered by other departments)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1708720	Immunogenetics	**2**	**2**	**-**
1718721	Radio-diagnosis in genetic disorders	**2**	**1**	**2**
1721624	Genetic epidemiology	**2**	**2**	**-**

5.4- Optional – (none)

6- Program admission requirements

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

7- 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+.

2- Submit a thesis validity report by an examination committee approved by the department council and their members include at least two external examiners.

8- 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	Interview	At least 50 %
2- Alumni	Interview	Representative sample
3- Stakeholders (Employers)	Interview	Representative sample
4- External Evaluator(S) or External Examiner (s)	Reports	**Dr. Rabah Shawky**
5- Other		

Dates of Previous editions/revisions:

Editions/Revisions Number	Date
Edition no.1	2009
Edition no. 2	2011
Edition no.3	5/6/2014
Edition no.3, revision no.1	12/2014
Edition no.3, revision no.2	10/2016
Edition no.3, revision no.3	9/2017

Program coordinator:

Name: Dr. Heba Morsy Signature: …….

Department Head:

Name: Prof. Dr. Amal Kotb Signature: …….

Date of Department Council Approval: 6/9/2017

Attached these Matrixes:

****Program Aims vs ILOs matrix***

**** Courses vs Program ILOs matrix***

ILOs / Course title	a1	a2	a3	a4	a5	a6	a7	a8	b1	b2	b3	b4	b5	b6	b7	8b	9b	10 b	c1	c2	c3	C4	c5	c6	d1	d2	d3	d4	d5
Basic Human Genetics	x								x			x							x						x	x	x	x	x
Prevention &Treatment of Gen Disorders		x								x									x						x	x	x	x	x
Molecular Genetics			x								x									x					x	x	x	x	x
Biochemical Genetics				x								x									x				x	x	x	x	x
population Genetics								x										x							x	x	x	x	x
Special Genetics					x								x												x	x	x	x	x
Selected Topics																									x	x	x	x	x
Clinical Cytogenetics						x								x								x			x	x	x	x	x
Clinical Genetics I							x								x	x							x	x	x	x	x	x	x
Clinical Genetics II							x									x	x						x	x	x	x	x	x	x
Cytogenetics	x					x								x								x			x	x	x	x	x
Biochemical Genetics I					x							x									x				x	x	x	x	x
Biochemical Genetics II					x							x									x				x	x	x	x	x
Genes and Diseases	x	x																	x	x	x	x	x	x	x	x	x	x	x

ILOS / Program Aims	a1	a2	a3	a4	a5	a6	a7	a8	a9	a10	a11	a12	a13	a14	b1	b2	b3	b4	b5	b6	b7	b8	b9	b10	b11	b12	b13	b14	c1	c2	c3	c4	c5	c6	d1	d2	d3	d4	d5
1. Have comprehensive knowledge on chromosomal basis of heredity, the patterns of single gene disorders, mitochondrial inheritance and multifactorial inheritance.	X				X										X				X										X						X	X		X	X
2. Recall 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.			X														X													X				X	X	X	X	X
3. Recognize the fundamental concepts the biochemical basis of single gene inheritance and have comprehensive knowledge on inborn errors of metabolism.				X								X	X					X								X	X				X				X	X		X	X
4. Know the molecular basis of cancer, the genetics of the immune system and the concepts of pharmacogenetics.						X																	X							X						X	X		X	X
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.								X			X											X			X							X				X	X		X	X
6. Have comprehensive knowledge on the genetic basis of craniofacial disorders,									X														X										X	X	X	X		X	X

deafness, short stature and muscular dystrophy.																																						
7. Recognize the fundamental concepts and methods in population screening and prenatal diagnosis and the role of genetic counseling in providing information and risk assessment.		X													X											X	X		X			X	X		X	X
8. Have comprehensive knowledge on the various methods used for treatment of genetic disorders the concept of gene therapy.		X					X								X					X												X	X		X	X
9. Have comprehensive knowledge on the genetic variation in populations, principles of population genetics and Hardy-Weinberg law.					X														X														X	X		X	X
10. Have a comprehensive knowledge on the genetic aspect of development and clinical aspects of human teratology.									X	X													X	X							X	X	X	X		X	X
11. Develop skills in information technology																																			X			