

Program SPECIFICATION FOR PhD Degree in Human Genetics

Code: 1713800

University: Alexandria

Faculty: Medical Research Institute

Program Specification

A- Basic information

1- Program title : PhD in Human Genetics

2- Program type: single ☒ double ☐ multiple ☐

3- Department(s) : Human Genetics

4- Coordinator : Pro. Dr. Nahla Nazmy

5- External evaluator(s): Prof. Dr. Samia El Temtamy

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

B- Professional Information

1- Program aims:

Provide the students with knowledge, skills and critical awareness to make has significant contribution to research and services provided by the department

By the end of this program the student should:

1. Recognize the principles of human genetics and diseases associated and demonstrate their ability to solve difficult professional problems and think critically.
2. Differentiate and list chromosomal inheritance, mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.
3. Describe metabolic and molecular basis of different genetic disorders.
4. Discuss patterns of human malformations and deformations.
5. List the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine.
6. Acquire clinical and laboratory genetic skills to diagnose various genetic disorders.
7. Improve basic and advanced procedural/ practical skills using technology and innovation.
8. Integrate concepts and relate ideas covered in different parts of the degree program.
9. Use systematic approaches to design and conduct scientific research.
10. Conduct research studies that add to the existing specialty knowledge.
11. Use information technology to increase the genetic knowledge. Develop skills for oral presentation, and work as a part of team.

2- Intended learning outcomes (ILOS)

a- knowledge and understanding:

- a1- Recall basic facts of the genome structure and gene expression, role of mutations in human disease, the principles of population genetics, segregation and genetic linkage analyses.
- a2- Discuss mitochondrial genes in degenerative diseases, cancer and ageing, multifactorial inheritance, complex disease, and pathogenetics of diseases.
- a3- Recognize the fundamental concepts and methods in genetic epidemiology, the molecular basis of cancer, the biological basis of aging, pharmacogenetics and pharmacogenomics.
- a4- Describe the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine.
- a5- Recall metabolic disorders in addition to the pathogenesis, clinical manifestations, and management of human inherited biochemical disorders.
- a6- Recognize the genetics of cardiovascular, respiratory, renal as well as gastrointestinal diseases as congenital heart diseases, inherited cardiomyopathies, molecular basis of hypertension, cystic fibrosis, asthma, cystic diseases of the kidneys, nephrotic syndrome and cancer colon.
- a7- Define the genetics of endocrinological disorders, genetic basis of common human hematological and immunological disorders, with special emphasis in the hemoglobinopathies and thalassemias, hemophiliac, leukemias and lymphomas, immunodeficiency disorders and autoimmune diseases.
- a8- Recall the genetics of neurologic as well as neuromuscular disorders including genetics of epilepsy, hereditary ataxias, neural tube defects, Duchenne as well as other muscle dystrophies and congenital myopathies.
- a9- Discuss the genetic basis of human male infertility, female infertility, fetal loss, abnormal body size and proportion and human malformation including hand malformations.
- a10- Review human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies, sex chromosomes abnormalities, deletions and other structural abnormalities of the autosomes.
- a11- Identify human craniofacial, skeletal and connective tissue disorders including craniofacial syndromes, craniosynostosis, anomalies of bone structure, bone density, chondrodysplasias, Marfan syndrome, Ehler Danlos syndrome and heritable diseases affecting elastic tissue.
- a12- Discuss the genetics of ophthalmologic disorders, hereditary hearing loss and dermatologic disorders including optic atrophy, defects of cornea, retinoblastoma, anomalies of skin pigmentation, ichthyosis and epidermolysis bullosa.
- a13- State the mechanisms of development, stem cells and cell signaling.
- a14- Recognize recent trends in biochemical genetics.
- a15- Discuss the genomic basis of disease.
- a16- Define the metabolic and molecular basis of deafness, eye disorders, and skin disease .
- a17- Recall the effects of genomics on health and genomic alterations on cardiology and obesity
- a18- Define various genetic syndromes in which hand malformation is a feature.
- a19- Design, conduction, publishing and recognize ethical considerations in different types of scientific research through thesis .

b- Intellectual skills:

- b1-** Differentiate between disease-causing DNA mutations and polymorphisms and illustrate the importance of epigenetics and the use of segregation analysis.
- b2-** Interpret the role of mitochondria in health and disease and illustrate pathogenetics of diseases.
- b3-** Assess the different genetic errors responsible for human cancers, the relation between genomics, health and disease, and the basic principles of pharmacogenetics.
- b4-** Integrate all phases of the genetic counseling process, the various genetic testing methods, the different genetic tests used in carrier and prenatal screening with clinical genetic applications.
- b5-** Analyze the genetic causes and inheritance patterns of inborn errors of metabolism, clinical presentations of various metabolic disorder and how to appropriately work up a patient suspected of having a metabolic disorder.
- b6-** Apply current genetic pathophysiological mechanisms of cardiovascular diseases, respiratory diseases and renal disorders.
- b7-** Illustrate the molecular basis of hematological and immunological and endocrine disorders.
- b8-** Interpret the role of genetics in neurological disorders as those of basal ganglia, cerebral cortical development, neural tube defects, epilepsy, tics, ataxias and phakomatoses, and neuromuscular disorders including muscle dystrophies, neuropathies, myopathies, spinal muscle atrophy, motor neuron disease.
- b9-** Apply basic principles of clinical genetics to the dysmorphic cases, human infertility, abnormal growth and different types of malformations.
- b10-** Evaluate the different genetic syndromes caused by chromosomal abnormalities, sex chromosomes abnormalities and the genetics of deletion syndromes.
- b11-** Use critical judgment to assess different craniofacial syndromes, skeletal dysplasias, connective tissue disorders and patterns of human malformations.
- b12-** Evaluate the genetic basis of genetic ophthalmologic disorders, deafness and dermatologic disorders.
- b13-** Evaluate the mechanisms of development, stem cells and cell signaling.
- b14-** Evaluate recent trends in biochemical genetics.
- b15-** Assess the genetic basis of genomic diseases and the effect of genome rearrangements on the phenotype.
- b16-** Interpret the metabolic and molecular basis of eye disorders, deafness and skin disorders.

b17- Assess the effect of genome structure on health and demonstrate the effect of genome alterations on disorders of heart, obesity and infectious diseases.

b18- Interpret the genetic syndromes in which hand malformation is a cardinal feature.

b19- Write a thesis protocol using a scientific systematic approach to a research problem.

b20- Prepare scientific articles/papers to be published in indexed journals.

c- professional and practical skills:

c1- Diagnose patients with mitochondrial disorders and patients with complex disorders, interpret results of investigations performed to patients with mitochondrial disorders , write and appraise reports

c2- Choose the appropriate tests for suspected genetic metabolic diseases; including the appropriate material to be tested and interpret the results of biochemical tests, their clinical significance and limitations.

c3- Choose the appropriate tests for diagnosing immunologic, hematologic and endocrinologic genetic diseases and interpret the results of molecular tests, their clinical significance and limitations.

c4- Examine, diagnose and counsel cases with genetic neurologic and neuromuscular diseases, chose the appropriate test and interpret the result of these tests.

c5- - Examine, diagnose and counsel cases with chromosome and sex anomalies, perform a blood culture for chromosome analysis and examine a karyotype and differentiate between various types of chromosomal abnormalities , chose the appropriate cytogenetic test and interpret the result of these tests.

c6- Gain competency for diagnosing craniofacial, skeletal and connective tissue genetic disorders, including the recognition of the physical features, clinical variability and natural history, all of which will help to formulate a differential diagnosis.

c7- Examine, diagnose and interpret the test results of patients with genetic ophthalmologic disorders, hereditary hearing loss and patients with genetic skin disorders.

c8- Develop counseling skills and recognize the ethical dilemmas in genetic disorders and utilize computerized genetic database resources.

c9- Use recent technologies to diagnose patients with inborn errors of metabolism

c10- Diagnose genomic disorders through high technology laboratory methodologies

c11- Interpret the results and limitations of tests done to diagnose eye, ear and skin genetic disorders .

c12- Examine, diagnose, interpret the results of tests of patients with hand malformations.

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, teach and evaluate others

d5- Develop skills in reading , research and self appraisal

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

Table of comparison between NAQAAE and ARS

Generic Academic Standards	ARS for Ph.D. in Human Genetics
A1-Basic facts , theories, of the specialty and related subjects/ fields	<p>A1- Recall facts of the genome structure and gene expression, role of mutations in human disease, Mendelian inheritance patterns the principles of population genetics, segregation and genetic linkage analyses</p> <p>A2- Discuss the genetic basis of human female & male infertility, a clinical approach to the dysmorphic child, clinical teratology, human malformations, abnormal mental development and abnormal body size and proportions.</p> <p>A3- Recall metabolic disorders, hematological and immunological disorders, with special emphasis in the hemoglobinopathies and thalassemias, hemophiliac, leukemias and lymphomas, immunodeficiency disorders and autoimmune diseases.</p> <p>A4- Identify human craniofacial, skeletal and connective tissue disorders, human hereditary hearing impairment, congenital blindness and other ophthalmologic disorders and genetic disorders of the skin, fundamental genetic basis of the cardiovascular, respiratory, renal, gastrointestinal and endocrinological disorders, genetic basis of the human mental, behavioral, neurologic and neuromuscular disorders.</p> <p>A5- Review human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies, sex chromosomes abnormalities, deletions and other structural abnormalities of the autosomes.</p>

A2- Mutual relation between professional practice and effects on environment	<p>A6- Describe mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.</p> <p>A7- Recognize the fundamental concepts and methods in genetic epidemiology, the molecular basis of cancer, the biological basis of aging, pharmacogenetics and pharmacogenomics.</p> <p>A8- Explain the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine.</p>
A3- Recent advances in the field of practice	A9- Identify future considerations in medical genetics and quality standards of the practice
A4-Details of ethical & legal practice	<p>A8- Explain the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine.</p> <p>A10- Know details of ethical and legal considerations in medical genetics .</p>
A5 -Quality standards of the practice	A9- Identify future considerations in medical genetics and quality standards of the practice
A6- Design, conduction & publishing of scientific research	A11- Design, conduction & explore publishing of scientific research.
A7- Ethical considerations in different types of scientific research	A12- Recognize ethical considerations in different types of scientific research through thesis
B1- Analyze, deduce, extrapolate & evaluation of information	<p>B1- Differentiate between disease-causing DNA mutations and polymorphisms, the different modes of inheritance and chromosomal disorders</p> <p>B2- Apply basic genetic knowledge to clinical problem solving , calculate simple statistics in epidemiological researches on genetic disorders</p> <p>B3- Illustrate the different genetic errors responsible for human cancers and the basic principles of pharmacogenetics.</p> <p>B4- Integrate all phases of the genetic counseling process, the various genetic testing methods, the different genetic tests used in carrier and prenatal screening with clinical genetic applications.</p>

	<p>B5- Apply basic principles of clinical genetics to the dysmorphic cases, human infertility, abnormal growth and different types of malformations.</p> <p>B6- Evaluate the different genetic syndromes caused by chromosomal abnormalities, sex chromosomes abnormalities and the genetics of deletion syndromes.</p> <p>B7- Illustrate current genetic pathophysiological mechanisms of cardiovascular diseases, respiratory diseases and renal disorders.</p> <p>B8- Illustrate the molecular basis of hematological, endocrinological and immunological disorders</p>
B2- Solve the majority of problems in the specialty according to the available data (complete or incomplete)	<p>B4- Integrate all phases of the genetic counseling process, the various genetic testing methods, the different genetic tests used in carrier and prenatal screening to solve problems in clinical genetic .</p> <p>B9- Illustrate the genetic causes and inheritance patterns of inborn errors of metabolism, clinical presentations of various metabolic disorder and how to appropriately work up a patient suspected of having a metabolic disorder.</p> <p>B10- Use critical judgment to assess different craniofacial syndromes, skeletal dysplasias, connective tissue disorders and patterns of human malformations.</p>
B3- Conduct research studies that add to the existing specialty knowledge	B14- Conduct research studies that add to the existing specialty knowledge.
B4- Publish scientific articles/papers (in indexed journals)	B15- Prepare scientific articles/papers to be published in indexed journals
B5- Plan and implement (or supervise implementation of) enhancement & Improvement approaches to practice	<p>Plan and implement (or supervise implementation) of enhancement & Improvement approaches to practice through student questionnaire</p> <p>B11- Argue future considerations in medical genetics.</p>
B6- Take decisions in various professional situations (<p>B10- Use critical judgment to assess different craniofacial syndromes, skeletal dysplasias, connective tissue disorders and patterns of human malformations.</p> <p>B12- Evaluate the genetic basis of genetic ophthalmologic disorders,</p>

including dilemmas & controversial issues)	deafness and dermatologic disorders B13- Assess the genetic basis of mental and behavioral and neuromuscular disorders
B7- Add to the specialty field through creativity & innovation	B14- Write a thesis protocol using a scientific systematic approach to a research problem.
B8- Manage discussions on basis of evidence and proofs	B4- Integrate all phases of the genetic counseling process, the various genetic testing methods, the different genetic tests used in carrier and prenatal screening to solve problems in clinical genetic .
C1- Competent in all basic and all required advanced professional skills (to be determined according to the specialty board/ department)	C1- Develop the skills to apply the molecular knowledge in clinical practice C2- Perform a blood culture for chromosome analysis and examine a karyotype and to differentiate between different types of chromosomal abnormalities . C3- Develop counseling skills and recognize the ethical dilemmas in genetic disorders C4- Interpret pedigree data and calculate risk C5- Develop a working knowledge of the various genetic testing methods C6- Examine and diagnose patients with various genetic disorders and to accurately record the findings and write the reports .
C2- Write and appraise reports	C6- Examine and diagnose patients with various genetic disorders and to accurately record the findings and write the reports.
C3- Evaluate <u>and improve</u> methods and tools used in specialty	C1- Develop the skills to apply the molecular knowledge in clinical practice C2- Perform a blood culture for chromosome analysis and examine a karyotype and to differentiate between different types of chromosomal abnormalities . C5- Develop a working knowledge of the various genetic testing methods. Evaluate <u>and improve</u> methods and tools used in specialty through student questionnaire .
C4- Use technology to advance practice	C1- Develop the skills to apply the molecular knowledge in clinical practice C2- Perform a blood culture for chromosome analysis and examine a karyotype and to differentiate between different types of chromosomal abnormalities .
C5- Plan professional development courses to improve	Evaluate and improve methods and tools used in specialty through student questionnaire

practice and enhance performance of juniors	
D1- Communicate effectively using all methods	D1- Communicate through group discussion
D2- Use information technology to improve his/her professional practice	D2- Develop skills in information technology
D3- Teach and evaluate others	D3- Develop skills for oral presentation, teach and evaluate others
D4- Perform self appraisal & seek continuous learning	D4- Develop skills in reading, critical and self appraisal
D5- Use different sources of information to obtain data	D2- Develop skills in information technology D5- Develop skills in reading and research
D6- Work in teams as well as a member in larger teams	D6- Work as a part of team
D7- Manage scientific meetings and appropriately utilize time	D7- Manage scientific meetings and appropriately utilize time through monthly scientific meeting in the department

4- curriculum structure and contents

4.a program duration: minimal 4 academic years including thesis

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	4	
Second semester	4	
Third semester	4	
Fourth semester	4	
Fifth semester	2	2
Sixth semester	4	

4.b.ii- No. of lectures Practical Thesis Total

Compulsory Elective Optional

4.b.v- No. of credit hours of specialized courses No. %

4.b.vi- No. of credit hours of other courses No. %

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 (24CH).

5- Program Courses

5.1.1- Compulsory (15 CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713801	Advanced Genetics I	2	2	-
1713802	Advanced Genetics II	2	1	2
1713803	Advanced Genetics III	2	2	-
1713804	Clinical Genetic Applications	1	1	-
1713807	Approach to Specific Disorders II	2	1	2
1713808	Approach to Specific Disorders III	2	2	-

1713810	Approach to Specific Disorders V	2	1	2
1713812	Approach to Specific Disorders VII	2	1	2
	Total	15	11	8

5.1.2- Compulsory for medical students (7CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713805	Approach to Clinical Problems	1	1	-
1713806	Approach to Specific Disorders I	2	1	2
1713809	Approach to Specific Disorders IV	2	1	2
1713811	Approach to Specific Disorders VI	2	1	2
	Total	7	4	6

5.1.3- Compulsory for non medical students (7CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713813	Advanced biochemical genetics I	1	1	-
1713814	Advanced biochemical genetics II	2	1	2
1713815	Genomic basis of disease	2	1	2
1713816	Approach to Specific Disorders IX	2	1	2
	Total	7	4	6

5.2- Elective I (2CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713817	Genomics II	2	2	-
1713818	Special clinical genetic	2	1	2
1713827	Embryology	2	2	-
1718821	Radio-diagnosis	2	1	2

5.3- Elective II (2 CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical

5.4- Optional – (none)

6- Program admission requirements

Postgraduate students with a M.Sc. in Human Genetics.

All students with an MSc in a field relevant to Human Genetics, the student should sit for a supplementary course (13 CH) and pass a qualifying exam to be eligible to register to PhD in Human Genetics.

Pre-requisite for PhD

Core courses (6CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713821	Basic Human Genetics	3	3	-
1713822	Basic Molecular Genetics	3	2	2
	Total	6	5	2

Elective courses (7CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1713823	Basic biochemical genetics	3	2	2
1713824	Clinical Genetics	4	3	2
1713825	Special Biochemical Genetics	4	3	2
1713826	Cytogenetics	3	2	2

7- Teaching and Learning Methods

- Lecture
- Practical/Clinical
- Brainstorming

- Discussion Groups
- Problem Solving
- Case Study
- Self-Directed Learning
- Critical Appraisal

Regulations for progression and program completion

For the progression and completion of the program to obtain the degree of PhD in Human Genetics, the student must :

- 1- Complete 24 credit hours with CGPA of at least C+ through courses.
- 2- Complete 24 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 one 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	questionnaire	At least 50 %
2- Alumni	questionnaire	Representative sample

3- Stakeholders (Employers)	meeting	Representative sample
4- External Evaluator(S) External Examiner (s)	Reports	Prof. Dr. Samia El Temtamy
5- Other	-	

Program coordinator :

Name: Prof. Dr. Nahla Nazmy

Signature *Nahla Nazmy*

Department Head:

Name: Prof. Dr. Ebtesam Abdalla

Signature *Ebtesam Abdalla*

Date of Department Council Approval: 30/8/2023

Attached these Matrices:

****Generic Attributes * Program Attributes * Program Aims***

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

**** Program ILOs vs Courses + thesis matrix***

****ARS vs ILOs matrix***

****Teaching methods vs Course matrix***

ARS Vs AIM Matrix
Degree:PhD **Code: 1713800**

AIM \ ARS	1	2	3	4	5	6	7	8	9	10	11
A1- Recall facts of the genome structure and gene expression, role of mutations in human disease, Mendelian inheritance patterns the principles of population genetics, segregation and genetic linkage analyses	x										
A2- Discuss the genetic basis of human female & male infertility, a clinical approach to the dysmorphic child, clinical teratology, human malformations,				x							

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

abnormal mental development and abnormal body size and proportions.										
A3- Recall metabolic disorders, hematological and immunological disorders, with special emphasis in the hemoglobinopathies and thalassemias, hemophiliac, leukemias and lymphomas, immunodeficiency disorders and autoimmune diseases.			x							
A4- Identify human craniofacial, skeletal and connective tissue disorders, human hereditary hearing impairment, congenital				x						

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

blindness and other ophthalmologic disorders and genetic disorders of the skin, fundamental genetic basis of the cardiovascular, respiratory, renal, gastrointestinal and endocrinological disorders, genetic basis of the human mental, behavioral, neurologic and neuromuscular disorders										
A5- Review human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies, sex chromosomes abnormalities, deletions and other structural abnormalities of the autosomes.		x								

ARS Vs AIM Matrix

Degree:PhD

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A6- Describe mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases		x									
A7- Recognize the fundamental concepts and methods in genetic epidemiology, the molecular basis of cancer, the biological basis of aging, pharmacogenetics and pharmacogenomics.	x										
A8- Explain the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques,					x						

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine.											
A9- Identify future considerations in medical genetics and quality standards of the practice						X	X				
A10- Know details of ethical and legal considerations in medical genetics.	X				X						
A11- Design, conduction & explore publishing of scientific research									X		
A12- Recognize ethical considerations in different types of scientific research through thesis										X	
B1- Differentiate between disease-causing DNA mutations and polymorphisms, the different modes of		X	X								

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

inheritance and chromosomal disorders											
B2- Apply basic genetic knowledge to clinical problem solving , calculate simple statistics in epidemiological researches on genetic disorders	x										
B3- Illustrate the different genetic errors responsible for human cancers and the basic principles of pharmacogenetics.			x								
B4- Integrate all phases of the genetic counseling process, the various genetic testing methods, the different genetic tests used in carrier and prenatal screening with clinical genetic applications							x				
B5- Apply basic principles of clinical genetics to the				x							

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

dysmorphic cases, human infertility, abnormal growth and different types of malformations.										
B6- Evaluate the different genetic syndromes caused by chromosomal abnormalities, sex chromosomes abnormalities and the genetics of deletion syndromes.		x								
B7- Illustrate current genetic pathophysiological mechanisms of cardiovascular diseases, respiratory diseases and renal disorders.					x					
B8- Illustrate the molecular basis of hematological, endocrinological and immunological disorders			x							
B9- Illustrate the			x							

ARS Vs AIM Matrix

Degree:PhD

Code: 1713800

genetic causes and inheritance patterns of inborn errors of metabolism, clinical presentations of various metabolic disorder and how to appropriately work up a patient suspected of having a metabolic disorder.											
B10- Use critical judgment to assess different craniofacial syndromes, skeletal dysplasias, connective tissue disorders and patterns of human malformations				X							
B11- Argue future considerations in medical genetics.							X				
B12- Evaluate the genetic basis of genetic ophthalmologic disorders, deafness and dermatologic disorders.				X							
B13- Assess the genetic basis of mental and behavioral and neuromuscular				X							

ARS Vs AIM Matrix

Degree:PhD

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disorders .											
B14- Conduct research studies that add to the existing specialty knowledge.									X		
B15- Prepare scientific articles/papers to be published in indexed journals										X	
C1- Develop the skills to apply the molecular knowledge in clinical practice						X					
C2- Perform a blood culture for chromosome analysis and examine a karyotype and to differentiate between different types of chromosomal abnormalities						X					
C3- Develop counseling skills and recognize the ethical dilemmas in genetic disorders.								X			
C4- Interpret pedigree data and calculate risk								X			
C5- Develop a						X	X				

ARS Vs AIM Matrix

Degree:PhD

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working knowledge of the various genetic testing methods											
C6- Examine and diagnose patients with various genetic disorders and to accurately record the findings and write the reports .						X		X			
D1- Communicate through group discussion											X
D2- Develop skills in information technology											X
D3- Develop skills for oral presentation, teach and evaluate others											X
D4- Develop skills in reading, critical and self appraisal.									X		
D5- Develop skills in reading and research.									X	X	
D6- Work as a part of team.											X

ARS Vs ILOs Matrix

Degree:PhD Code: 1713800

<div>ILOS</div> <div>ARS</div>	A1	A2	A3	A4	A5	A6	A7	A8	A9	A10	A11	A12	A13	A14	A15	A16	A17	A18	A19	B1	B2	B3	B4	B5	B6	B7	B8	B9	B10	B11	B12	B13	B14	B15	B16	B17	B18	B19	B20	C1	C2	C3	C4	C5	C6	C7	C8	C9	C10	C11	C12	D1	D2
A1- Recall facts of the genome structure and gene expression, role of mutations in human disease, Mendelian inheritance patterns the principles of population genetics, segregation and genetic linkage analyses	X														X		X																																				
A2- Discuss the genetic basis of human female & male infertility, a clinical approach to the dysmorphic child, clinical teratology, human malformations,								X																																													

Degree:PhD

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[illegible]

ARS Vs ILOs Matrix

Degree:PhD

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blindness and other ophthalmologic disorders and genetic disorders of the skin, fundamental genetic basis of the cardiovascular, respiratory, renal, gastrointestinal and endocrinological disorders, genetic basis of the human mental, behavioral, neurologic and neuromuscular disorders

A5- Review human syndromes caused by chromosomal abnormalities and the clinical genetics of common autosomal trisomies, sex chromosomes abnormalities, deletions and other structural abnormalities of the autosomes.

x

Degree:PhD

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[illegible]

Degree:PhD

Code: 1713800

[illegible]

Degree:PhD

Code: 1713800

[illegible]

ARS Vs ILOs Matrix

[illegible]

Degree:PhD

Code: 1713800

[illegible]

Degree:PhD

Code: 1713800

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Degree:PhD

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[illegible]

Graduate Attributes of Ph.D. Program in Human Genetics

Generic Graduate Attributes of NAQAAE	Graduate Attributes of Doctor of Philosophy in Human Genetics
	By the end of this program, Graduate of Doctor of Philosophy in Human Genetics should be able to
Master the basics and methodologies of scientific research.	Conduct scientific research with proficiency.
Work continuously to add to his/her knowledge in the field of specialty.	Differentiate and list chromosomal inheritance, mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.
Apply the analytical and critical approach to knowledge in the field of specialty and related fields.	Solve difficult professional problems and think critically through recognizing the metabolic and molecular basis of different genetic disorders and patterns of human malformations and deformations.
Integrate knowledge in the field of specialty with related knowledge, deduce and develop relationships between them.	Integrate the fundamental principles of human genetics and diseases associated and relate ideas covered in different parts of the degree programme.
Demonstrate a deep awareness of current problems and modern theories in the field of specialty.	Demonstrate a deep awareness on the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine .
Identify professional problems and find innovative solutions to solve	Solve difficult professional problems with innovation and think critically.

them.	
Master a wide range of professional skills in the field of specialty.	Acquire clinical and laboratory genetic skills to diagnose various genetic disorders.
Develop new methods, tools and methods for professional practice.	Improve basic and advanced procedural/ practical skills using technology and innovation to diagnose various genetic disorders.
Use appropriate technological means to serve his professional practice.	Use information technology to increase the genetic knowledge. Integrate concepts and relate ideas covered in different parts of the degree program.
Communicate efficiently and lead work teams in various professional scenarios.	Communicate efficiently, lead work teams and develop skills for oral presentation,
Take Decision in light of available data.	Integrate concepts and relate ideas covered in different parts of the degree program
Employ and develop available resources efficiently and work to find new resources.	Improve basic and advanced procedural/ practical skills using technology and innovation.
Show awareness of his/her role in community development and environmental preservation	Conduct research studies that add to the existing specialty knowledge
Act in a manner that reflects a commitment to integrity, credibility, and professionalism.	Act in a manner that reflects a commitment to integrity, credibility, and professionalism through continuous training and self education.
Commit to continuous self-development and transfer his/her knowledge and experiences to others.	Commit to continuous self-development and transfer his/her knowledge and experiences to others through seminars and self directed learning.

Signature *Nahla Nazmy*

Signature Ebtessam Abdallah

Date of Approval **30/8/2023**

Attributes Vs AIM Matrix
Degree:PhD **Code: 1713800**

AIM \ Attributes	1	2	3	4	5	6	7	8	9	10	11
Conduct scientific research with proficiency.									X	X	
Differentiate and list chromosomal inheritance, mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.		X									
Solve difficult professional problems and think critically through recognizing the metabolic and molecular basis of different genetic disorders and			X	X							

Attributes Vs AIM Matrix

Degree:PhD

Code: 1713800

patterns of human malformations and deformations.											
Integrate the fundamental principles of human genetics and diseases associated and relate ideas covered in different parts of the degree programme.	X							X			
Demonstrate a deep awareness on the process of genetic counseling, carrier and prenatal screening, cytogenetic and molecular diagnostic techniques, strategies for treatment of genetic disorders, bioinformatics, and the ethical, social and legal issues in genetic medicine .					X						
Solve difficult professional problems with innovation and think critically.						X		X			
Acquire clinical and laboratory genetic skills to diagnose various genetic	X					X					

Attributes Vs AIM Matrix

Degree:PhD

Code: 1713800

disorders.											
Improve basic and advanced procedural/ practical skills using technology and innovation to diagnose various genetic disorders.						X	X				
Use information technology to increase the genetic knowledge. Integrate concepts and relate ideas covered in different parts of the degree program.								X			X
Communicate efficiently, lead work teams and develop skills for oral presentation,											X
Integrate concepts and relate ideas covered in different parts of the degree program								X			
Improve basic and advanced procedural/ practical skills using technology and innovation.							X	X			
Conduct research studies that add to									X	X	

Attributes Vs AIM Matrix

Degree:PhD

Code: 1713800

the existing specialty knowledge											
Act in a manner that reflects a commitment to integrity, credibility, and professionalism through continuous training and self education.											X
Commit to continuous self-development and transfer his/her knowledge and experiences to others through seminars and self directed learning.											X

Matrix for ILOs of PhD of Human Genetics and its aims

ILOS	A 1	A 2	A 3	A 4	A 5	A 6	A 7	A 8	A 9	A 10	A 11	A 12	A 13	A 14	A 15	A 16	A 17	A 18	A 19	B 1	B 2	B 3	B 4	B 5	B 6	B 7	B 8	B 9	B 10	B 11	B 12	B 13	B 14	B 15	B 16	B 17	B 18	B 19	B 20	C 1	C 2	C 3	C 4	C 5	C 6	C 7	C 8	C 9	C 10	C 11	C 12
Aims																																																			
1. Know and recognize the principles of human genetics and diseases associated and demonstrate their ability to solve difficult professional problems and think critically.	X	X	X	X		X	X	X	X	X	X	X			X					X						X	X				X	X					X	X	X			X									
2. Acquire and add knowledge through research and reasoning on chromosomal inheritance, mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.		X								X										X	X	X							X							X															
3. Describe metabolic and molecular basis of different genetic disorders.			X		X								X	X		X	X							X		X									X	X	X			X											
4. Recognize patterns of									X									X										X							X		X			X											

[illegible]

[illegible]

Courses vs Program ILOs matrix for PhD in Human Genetics

Course title	a1	a2	a3	a4	a5	a6	a7	a8	a9	a10	a11	a12	a13	a14	a15	a16	a17	a18	a19	b1	b2	b3	b4	b5	b6	b7	b8	b9	b10	b11	b12	b13	b14	b15	b16	b17	b18	b19	b20	c1	c2	c3	c4	c5	c6	c7	c8	c9	c10	c11	c12	d1	d2	d3	d4	d5
Advanced Genetics I	x																			x																																x	x	x	x	x
Advanced genetics II		x																			x																															x	x	x	x	x
Advanced genetics III			x																			x																														x	x	x	x	x
Clinical genetic application				x																			x																													x	x	x	x	x
Approach to specific disorders II					x																			x																												x	x	x	x	x
Approach to specific disorders III						x											x								x																											x	x	x	x	x
Approach to specific disorders V							x																			x																										x	x	x	x	x
Approach to specific disorders VII								x																			x																									x	x	x	x	x
Approach to clinical problems									x																			x																								x	x	x	x	x
Approach to specific disorders I										x																				x																						x	x	x	x	x
Approach to specific disorders IV											x																																									x	x	x	x	x
Approach to specific disorders VI												x																																								x	x	x	x	x

Teaching and Learning Methods Vs Courses Matrix

Degree: PhD Code: 1713800

	Course code: 801	Course code:802	Course code:803	Course code:804	Course code:805	Course code:806	Course code:807	Course code:808	Course code:809	Course code:810	Course code:811
Lecture	●	●	●	●	●	●	●	●	●	●	●
Practical/Clinical						●	●		●	●	●
Brainstorming	●		●	●	●	●	●	●			
Discussion Groups	●	●	●	●	●	●	●	●	●	●	●
Problem Solving		●				●		●	●		
Case Study		●		●		●		●	●	●	●
Training Workshops											
Self-Directed Learning	●	●	●	●	●	●	●	●	●	●	●
e-learning											
Project											
Critical Appraisal		●								●	●

Teaching and Learning Methods Vs Courses Matrix
Degree: PhD **Code: 1713800**

	Course code: 812	Course code:813	Course code:814	Course code:815	Course code:816	Course code:817	Course code:818	Course code:820
Lecture	●	●	●	●	●	●	●	●
Practical/Clinical	●		●	●	●		●	●
Brainstorming	●	●	●	●	●	●	●	●
Discussion Groups	●	●	●	●	●	●	●	●
Problem Solving	●							
Case Study	●						●	
Field Training								
Role playing								
Training Workshops								
Self-Directed Learning	●	●	●	●	●	●	●	●
e-learning								
Project								
Critical Appraisal								

Teaching and Learning Methods Vs Courses Matrix
Degree: PhD **Code: 1713800**

	Course code: 821	Course code:822	Course code:823	Course code:824	Course code:825	Course code:826
Lecture	●	●	●	●	●	●
Practical/Clinical		●	●	●	●	●
Brainstorming	●	●	●	●	●	
Discussion Groups	●	●	●	●	●	●
Problem Solving	●			●		●
Case Study				●		
Field Training						
Role playing						
Training Workshops						
Self-Directed Learning	●	●	●	●	●	●
e-learning						
Project						
Critical Appraisal						