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 : Prof. Dr. Nahla Nazmy

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

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

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. Acquire and add knowledge through research and reasoning on chromosomal inheritance, mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases.
3. Describe metabolic and molecular basis of different genetic disorders.
4. Recognize patterns of human malformations and deformations.
5. Acquire knowledge 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.
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 programme.
9. Proficient in conducting research .
10. Use information technology to increase the genetic knowledge.

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- Explain 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- Discuss 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- Recall human craniofacial, skeletal and connective tissue disorders including craniofacial syndromes, craniosynostosis, anomalies of bone structure, bone density, chondrodysplasias, Marfan syndrome, Ehhler 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, icthyosis and epidermolysis bullosa.
a13- State the mechanisms of development, stem cells and cell signaling.
a14- Recall 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.

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 dyplasias, 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.

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 solve 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)

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

3b Comparison of provision to selected external references

Table of comparison between NAQAAE and ARS

ARS / NAQAAE	ARS for Ph.D. in Human Genetics
A1-Basic facts , theories, of the specialty and related subjects/ fields	**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 **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. **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. **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. **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.
A2- Mutual relation between professional practice and effects on environment	**A6-** Describe mitochondrial inheritance, multifactorial inheritance, complex disease, and pathogenetics of diseases. **A7-** Recognize the fundamental concepts and methods in genetic epidemiology, the molecular basis of cancer, the biological basis of aging, pharmacogenetics and pharmacogenomics. **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.
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	**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. **A10-** Know details of ethical and legal considerations in medical genetics .
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	Design, conduction & publishing of scientific research through thesis
A7- Ethical considerations in different types of scientific research	Recognize Ethical considerations in different types of scientific research **t**hrough thesis
B1- Analyze, deduce, extrapolate & evaluation of information	**B1-** Differentiate between disease-causing DNA mutations and polymorphisms, the different modes of inheritance and chromosomal disorders **B2-** Apply basic genetic knowledge to clinical problem solving **,** calculate simple statistics in epidemiological researches on genetic disorders **B3-** Illustrate the different genetic errors responsible for human cancers 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-** Apply basic principles of clinical genetics to the 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. **B7-** Illustrate current genetic pathophysiological mechanisms of cardiovascular diseases, respiratory diseases and renal disorders. **B8-** Illustrate the molecular basis of hematological, endocrinological and immunological disorders
B2- Solve the majority of problems in the specialty according to the available	**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 . **B9-** Illustrate the genetic causes and inheritance patterns of inborn errors of metabolism, clinical presentations of various metabolic

data (complete or incomplete)	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 dyplasias, connective tissue disorders and patterns of human malformations.
B3- Conduct research studies that add to the existing specialty knowledge	Conduct research studies that add to the existing specialty knowledge through thesis and assignments
B4- Publish scientific articles/papers (in indexed journals)	Publish scientific articles/papers (in indexed journals**)** through thesis
B5- Plan and implement (or supervise implementation of) enhancement & Improvement approaches to practice	Plan and implement (or supervise implementation) of enhancement & Improvement approaches to practice through student questionnare **B11-** Argue future considerations in medical genetics.
B6- Take decisions in various professional situations (including dilemmas & controversial issues)	**B10**- Use critical judgment to assess different craniofacial syndromes**,** skeletal dyplasias, connective tissue disorders and patterns of human malformations. **B12-** Evaluate the genetic basis of genetic ophthalmologic disorders**,** 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	Add to the specialty field through creativity & innovation through thesis
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	**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 .

according to the specialty board/ department)	**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 *and improve* 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 *and improve* 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 practice and enhance performance of juniors	Evaluate and improve methods and tools used in specialty through student questionnaire
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	**D4-** Develop skills in reading, critical and self appraisal

continuous learning	
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 credit hours	**Lectures**	**17**	**Practical**	**7**	**Total**	**24**
	Compulsory	**22**	**Elective**	**2**	**Optional**	**0**

4.b.iii- No. of credit hours of basic science courses	**No.**	**2**	**%**	**8.3**
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.**	**22**	**%**	**91.7**
4.b.vi- No. of credit hours of other courses	**No.**	**-**	**%**	**-**
4.b.vii- Field Training	**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

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**

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

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	-

5.3- Elective II (2 CH)

Code No.	Course Title	No. of credit hours	No. of hours /week	
			Lecture	Practical
1718821	Radio-diagnosis in Genetic Disorders	2	1	2

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**

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- 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+.

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) External Examiner (s)	Reports	**Prof. Dr. Samia El Temtamy**
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: Prof. Dr. Nahla Nazmy Signature

Department Head:

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

Date of Department Council Approval: 6/9/2017

Attached these Matrices:

1*Program Aims vs ILOs matrix

2 * Courses vs Program ILOs matrix

3*ARS vs ILOs matrix

4*Teaching methods vs Course matrix