

February 2005

REGULATIONS

FOR ADMISSION TO THE FELLOWSHIP OF THE COLLEGE OF MEDICAL GENETICISTS OF SOUTH AFRICA

1.0 OBJECTIVES

- 1.1 The candidate who passes these examinations must be able to fulfil the role of a specialist Medical Geneticist in the medical and academic communities, and in society at large
- 1.2 Central to these examinations is their licensing function: persons awarded the FCMG(SA) who, in addition, fulfil the other requirements of the Medical, Dental and Supplementary Health Services Act may register and practise as specialist Medical Geneticists in terms of the Act
- 1.3 The following paragraphs indicate briefly the range of competencies that can be expected of the specialist Medical Geneticist. The specialist Medical Geneticist:
 - 1.3.1 should be able to offer care for all conditions commonly occurring in Medical Genetics;
 - 1.3.2 should possess particular competence in caring for patients with medical genetic conditions and birth defects;
 - 1.3.3 must be able to judge when to seek the help of other specialists and sub specialists; and
 - 1.3.4 must be able to act as the patient's advocate, advisor and guide within the discipline of Medical Genetics

The examination comprises Part I and Part II: Part II must be passed within six years of passing Part I.

PART I

2.0. ADMISSION TO THE PART I EXAMINATION

(to be read in conjunction with the Instructions)

- 2.1 For admission to Part I of the examination, the candidate must hold a qualification to practise medicine which is registered with the Health Professions Council of South Africa.
- 2.2 Completed the period of community service.
- 2.3 Preference will given to applicants who have at least twelve months experience in Paediatrics and/or Obstetrics and Gynaecology and/or

Internal Medicine. This experience should be obtained in a level 2 or higher facility.

- 2.4 The examination is written prior to completing 30 months as a fulltime registrar in a teaching department of Medical Genetics (registered with HPCSA).
- 2.5 The Senate of the CMSA, through its Examinations and Credentials Committee, will review all applications for admission to the examination and may also review the professional and ethical standing of candidates.

3.0 SYLLABUS FOR THE PART I EXAMINATION

- 3.1 The subjects covered by the examination will include the basic sciences of Medical Genetics (including molecular and cell biology; laboratory techniques and interpretation of laboratory results, mechanisms of commonly occurring genetic disorders and birth defects; elementary statistics; public health genetics, and applied anatomy, physiology and embryology), applicable ethical aspects and principles of genetic counselling. Knowledge will be required of all those aspects of the subjects which should form part of the general education of any specialist and particularly of those aspects applicable to Medical Genetics.

- 3.1.1 A syllabus of subjects and notes forming a general guide to Part I of the examination is attached (Appendix A). This is not exhaustive and the candidate is expected to have knowledge of basic sciences as related to Medical Genetics as well as allied areas of new developments.

- 3.1.2 Basic sciences must be regarded as an applied science to the principles and practice of medical genetics. Basic science questions can and will be asked in Part I, but may also be asked (with clinical application) in Part II of the examination.

- 3.2 Candidates will be expected to have completed a minimum 80 hour genetic counselling course (40 hours basic counselling skills course and 40 hours experiential training, over an 18 month period).

- 3.2.1 The standard of counselling will be assessed by the course providers in 4 staged assessments.

- 3.2.2 Candidates would be expected to manage routine clinical and genetic counselling cases e.g. counselling for women of advanced maternal age, antenatal diagnosis, Down syndrome and other common chromosome abnormalities, single gene disorders, and common multifactorial conditions.

4.0 CONDUCT OF THE PART I EXAMINATION

Part I examination, with an overall pass mark of 50%, comprises:

Three written papers (no oral examination) as follows:

- 4.1 Paper 1: Essay questions (3 hours)
Medical Genetics
- 4.2 Paper 2: Short questions (3 hours)
Medical Genetics
- 4.3 Paper 3: Short questions (3 hours)
Genetic Counselling and Medical Ethics
- 4.4 Each of the 3 papers contributes 1/3 to the final mark.
- 4.5 Candidates who obtain less than 45% in any paper will fail the examination.

5.0 TRAINING FOR THE PART I EXAMINATION

The candidate must submit evidence that he/she has completed the following training in posts approved for the purposes by the HPCSA before admission to Part I of the examination.

5.1 General

5.1.1 Twelve months in resident hospital posts recognised for pre-registration purposes by the Health Professions Council of South Africa (i.e. 12 months internship).

5.1.2 Candidates must fulfil the requirements of the HPCSA other than the registrar training for specialist registration (i.e. Community Service posts).

5.1.3 Fulfilled the clinical requirements as specified in 2.3

5.2 Medical Genetics:

Completed at least 12 months in a full-time post as a registrar in a HPCSA registered Medical Genetics unit, such posts having been recognised by the HPCSA for the purpose of the Fellowship examination.

PART II

6.0 ADMISSION TO THE PART II EXAMINATION
(to be read in conjunction with the Instructions)

6.1 For admission to Part II the candidate must present evidence of

6.1.1 having passed Part I (or the Part I Fellowship examination of one of the Colleges with which there is an agreement of reciprocity for the primary examinations)

- 6.1.2 having been qualified for not less than 5 years, including the year of internship
 - 6.1.3 having completed the training set out in 9.0 of the regulations
 - 6.1.4 having at least 3 years fulltime training as a registrar in a teaching unit of Medical Genetics registered with the HPCSA at the time of initial entry. Consideration may be given in special circumstances.
 - 6.1.5 a certificate of proficiency from the Head of Department and the HPCSA registered clinical head of the teaching unit (Appendix B).
 - 6.1.6 having fulfilled the requirements for a logbook set out in Appendix C (Logbook) of the regulations.
 - 6.1.7 The CMSA Senate, through its Examinations and Credentials Committee, will review every application for admission to the examination, and may also consider the professional and ethical standing of the candidate
- 6.2 Regulations for completion of the logbook and case report/Medical Genetics paper are as follows:
- 6.2.1 Requirements are set out in Appendix C.
 - 6.2.2 Before being allowed to enter for the examination, a candidate shall submit a logbook fully completed, and one case report (or Medical Genetics manuscript) complying with the regulations in force at the time of entry into the examination.
 - 6.2.3 Candidates must present themselves for the examination within two years of having completed their logbook and case report (or Medical Genetics report). Failing this, a new logbook and new case report (or Medical Genetics manuscript) will be required.
 - 6.2.4 Candidates who fail the examination may again enter the examination on the basis of their original logbook and commentaries, within a period of 2 years.
 - 6.2.5 The logbook and commentaries must reach the Academic Registrar in Johannesburg as detailed in Appendix C.

7.0 SYLLABUS FOR THE PART II EXAMINATION

- 7.1 The subjects covered by the examination will include clinical genetics as well as the basic sciences of Medical Genetics, laboratory techniques and interpretation of laboratory results, public health

genetics, ethical aspects and genetic counselling (as applied from Part I of the examination).

7.1.1 A syllabus of subjects and notes forming a general guide to Part II of the examination is attached (Appendix D). This is not exhaustive and the candidate is expected to have knowledge of the full spectrum of clinical genetics as well as of areas of new advances.

7.1.2 Basic science questions can and will be asked in Part II of the examination, but with a clinical application.

7.1.3 Candidates will be expected to have completed a logbook, reflecting one case report and a minimum of 400 cases covering a broad spectrum of genetic disorders and/or birth defects (See Appendix C).

7.1.4 Candidates would be expected to be able to fully assess clinical cases for appropriate diagnosis, investigation, counselling and holistic management.

7.1.5 Genetic disorders and birth defects important in the South African context should be emphasised. Cost-effective investigation and management should be stressed.

8.0 CONDUCT OF THE PART II EXAMINATION

CONDUCT OF THE EXAMINATION

The Part II examination, with an overall pass mark of 50%, comprises:

8.1 Written examination (40% of total):

Two written papers, of 3 hours duration each, on:

8.1.1 Medical Genetics and such aspects of medicine, paediatrics, obstetrics and gynaecology, neurology, public health and basic medical sciences as are relevant to the practice of Medical Genetics. Paper 1 will be long essay type questions examining broad categories and approaches. Paper 2 will be shorter questions examining a specific topic or aspect.

8.2 Objective Structured Clinical Examination (OSCE) (10% of total mark):

8.2.1 The Convenor will, with the other 2 examiners, determine the content and set up the OSCE examination

8.2.2 This will include 10-20 stations with questions on pedigree

analysis, chromosomal analysis (including FISH), biochemical analysis, molecular genetic tests and their interpretation, fetal ultrasound, fetal pathology, radiology, dysmorphological photographs and any other topics relevant to the clinical practice of Medical Genetics.

8.2.3 Candidates will provide written answers.

8.3 The clinical examination (40% of total mark):

8.3.1 The Convenor is responsible for organising the clinical examinations. This entails identifying and selecting appropriate patients for examination as well as locating an appropriate examination venue

8.3.2 The long case (20% of total mark)

This will usually comprise a complex disorder of genetic origin, with multiple organ system involvement. Sixty minutes will be allocated during which time the candidate should take a thorough history, conduct a relevant physical evaluation (with examiners not present) and prepare for a clinical evaluation. The evaluation by two examiners (plus an observer) will be allocated 30 minutes.

8.3.3 Short cases (20% of total mark)

Two short cases will be evaluated for a period of 20-30 minutes each by the candidate. A short history and examination of one organ system (at least) will be required. Thereafter the candidate will be evaluated by two examiners (plus an observer). Examiners will be present throughout the allocated time.

8.4 Oral examination (10% of total mark)

8.4.1 The content of the oral examination will be decided upon by the Convenor and examiners on the day of the clinical examination

8.4.2 An oral examination of 20-30 minutes duration will be undertaken by two examiners (plus an observer).

8.4.3 In cases where the oral examination will not make a significant differences to the candidates overall mark, the oral examination may be waived at the discretion of the convenor and examiners.

8.5 The examiners will submit all their assessments in percentages.

8.6 Candidates who obtain less than 50% for the written part of the examination will fail the examination and not be eligible to participate in the clinical component of the examination.

8.7 Candidates who obtain less than 60% for the practical part of this examination (OSCE and clinical examination combined) will fail the examination.

8.8 Candidates who obtain less than 50% overall will fail the examination.

8.9 The overall examination will be overseen by a Convenor.

9.0 **TRAINING FOR THE PART II EXAMINATION**

The candidate must submit evidence that he/she has completed the following training in posts approved for the purposes by the CMSA before admission to Part II of the examination.

9.1 **General:**

Twelve months in resident hospital posts recognised for pre-registration purposes by the Health Professions Council of South Africa (i.e. 12 months internship).

9.2 Candidates must fulfil the requirements of the HPCSA for community service.

9.3 Candidates must have completed a minimum of 36 month in a full-time post as registrar or clinical assistant, or a full-time post providing equivalent experience in a Medical Genetics unit, such posts having been recognised for the purpose of the Fellowship examination.

9.4 The CMSA Senate, through its Examinations and Credentials Committee, will review every application for admission to the examination, and may also consider the professional and ethical standing of the candidate

9.5 Successful completion of the examination for FCMG does not guarantee registration as a specialist with the HPCSA and it is the candidates responsibility to ensure the he/she meets the HPCSA requirements for registration.

10.0 ADMISSION AS A FELLOW

10.1 The candidate having passed the examination and having been admitted as a Fellow of the College of Medical Geneticists of South Africa, will be asked to sign a declaration, as under:

I, the undersigned,
do solemnly and sincerely declare

that while a member of the CMSA I will at all times do all within my power to promote the objects of the CMSA and uphold the dignity of the CMSA and its members

that I will observe the provisions of the Memorandum and Articles of Association, By-laws, Regulations and Code of Ethics of the CMSA as in force from time to time

that I will obey every lawful summons issued by order of the Senate of the said CMSA, having no reasonable excuse to the contrary

and I make this solemn declaration faithfully promising to adhere to its terms

Signed at this
..... day

of
.....

Signature

.....

Witness

.....

(who must be a Founder, Associate Founder, Fellow, Member, Diplomate or Commissioner of Oaths)

10.2 A two-thirds majority of members of the CMSA Senate present at the relevant meeting shall be necessary for the award to any candidate of a Fellowship.

10.3 A Fellow shall be entitled to the appropriate form of certificate under the seal of the CMSA.

10.4 In the event of a candidate not being awarded the Fellowship (after having passed the examination), the examination fee shall be refunded in full.

10.5 The first annual subscription is due one year after registration (statements are rendered annually).

APPENDIX A

1.0 SYLLABUS FOR PART I OF THE FCMG EXAMINATION

This examination requires a wide applied knowledge of the basic sciences. It is impossible to define this level of knowledge precisely, but candidates may expect to be examined on information which should form part of the general education of any specialist. A more detailed knowledge of medical and public health genetics, together with the principles and practices of genetic counselling, medical ethics and of laboratory genetic tests, will be required. A broad overview of anatomy, physiology, normal and abnormal embryology, virology, pharmacology and pathology as applied to Medical Genetics, is also required.

The following list of subjects and notes forms a general guide to the information upon which questions will be based. It is not meant to be comprehensive and questions may be asked on topics which are not covered directly in this guide.

1.0 MEDICAL GENETICS

Cellular and Molecular Basis of inheritance

- The cell
- Structure and function of DNA, RNA and proteins
- Transcription
- Translation
- Gene regulation
- Mutations and mutagenesis
- DNA repair mechanisms
- Mitosis and meiosis
- Cell cycle

Pedigree construction and analysis

Cytogenetics and chromosome abnormalities

- Chromosome nomenclature
- Chromosome structure
- Chromosome number
- Sex chromosomes
- Methods of chromosome analysis
 - Karyotyping
 - FISH
 - Molecular cytogenetics
- Chromosome abnormalities
 - Numerical abnormalities
 - Structural abnormalities
 - Microdeletion syndromes

Anatomy, Physiology And Embryology

- Principles of embryology and application to dysmorphology
- Fertilisation and gastrulation

Physiological changes of pregnancy and adaptation of the neonate/infant to extra-uterine life

Developmental genetics

Gender determination

Developmental gene families

Twinning

Candidates are expected to have a broad knowledge of the anatomy, physiology and embryology of the major organ systems and their application to Medical Genetics.

Cardiovascular system

Respiratory system

Gastro-intestinal system

Reticulo-endothelial system

Urogenital system

Musculoskeletal system

Central and peripheral nervous system

Sensory systems

Ophthalmic system

Auditory system

Olfactory system

Tactile system

Endocrine system

Reproductive system and pregnancy

Patterns of inheritance

Mendelian inheritance

Autosomal dominant inheritance

Autosomal recessive inheritance

X-linked dominant inheritance

X-linked recessive inheritance

Y-linked inheritance

Polygenic and multifactorial inheritance

Non-Mendelian Inheritance

X-chromosome inactivation

Mitochondrial inheritance

Dynamic repeat disorders

Genomic imprinting

Mosaicism

Complex disorders

Population genetics

Hardy Weinberg equilibrium principle

Founder effect

Gene flow

Genetic drift

Genetic polymorphism

Segregation analysis

Genetic linkage

Consanguinity

Twins and twinning

Molecular genetics

Principles of recombinant DNA technology
 Techniques for DNA analysis and genetic testing
 Linkage analysis
 Mutation analysis
 DNA polymorphisms
 DNA sequencing
 PCR
 Southern blotting
 RNA analysis
 FISH
 Microarray technology
 Flow cytometry
 Monoclonal antibodies
 New technology
 Indications for genetic testing
 Identifying disease genes
 Gene therapy

Developmental genetics

Gender determination
 Developmental gene families

Biochemical genetics

Principles of biochemical analysis
 Inborn errors of metabolism
 Urea cycle disorders
 Disorders of amino-acid metabolism
 Organic acid disorders
 Disorders of carbohydrate metabolism
 Disorders of steroid metabolism
 Disorders of lipid metabolism
 Lysosomal storage disorders
 Disorders of purine/pyrimidine metabolism
 Disorders of porphyrin biosynthesis
 Peroxisomal disorders

Parentage Testing

Forensic testing

Bioinformatics

Knowledge and use of electronic databases

Biostatistics

Statistical and epidemiological techniques
 Mendelian disorders including risk calculations
 Bayes theorem
 Empiric risks
 Consanguinity
 Evidence based medicine

Pharmacogenetics

Immunogenetics

Cancer Genetics

- Oncogenes
- Tumour suppressor genes
- Repair genes
- Familial cancers
 - Breast cancer
 - Bowel Cancers
 - Multiple endocrine neoplasias

Clinical Genetics

- Congenital Abnormalities
- Teratogens
- Common single gene disorders
- Common multifactorial disorders
- Common chromosome abnormalities
- Clinical features, genetic principles and molecular basis of common diseases including:
 - Haemoglobinopathies
 - Fragile X syndrome
 - Cystic fibrosis
 - Albinism
 - Haemophilia
 - Duchenne muscular dystrophy
 - Huntington disease
 - Myotonic dystrophy
 - Neurofibromatosis
 - Spinal muscular atrophy
- Carrier detection and predictive testing
- Genetic factors in common diseases such as:
 - Diabetes
 - Hypertension
 - Cardiovascular disease
 - Psychiatric disorders

Prenatal diagnosis

- Techniques used
 - Amniocentesis
 - CVS
 - Cordocentesis
 - Ultrasound
 - Biochemical screening
 - Preimplantation genetic diagnosis
 - New techniques
- Termination of Pregnancy Act
- Reproductive options

Public Health Genetics

- Genetic Screening
- Birth defect surveillance
- Genetic Registers

- Epidemiology of birth defects
- Care and prevention of genetic disorders
- Community Genetics
- Parent support groups and NGOs
- HIV/AIDS and other STDs

Medical ethics

- Informed consent
- Testing of minors
- Genetic screening
- Eugenics
- Ethics and the law
- Social responsibilities of medical geneticists
- Ethnicity and health

Medico-legal issues

Paediatrics in Genetics

- Examination of the newborn
- Examination of child
 - Growth
 - Child development

Future genetics

- Genomics
- Proteomics
- Phenomics
- Gene therapy
- Pharmacogenetics

2.0 GENETIC COUNSELLING

Basic counselling skills course – minimum 40 hours and minimum 40 hours experiential time supervised by qualified Medical Geneticists and genetic counsellors

- Pedigree Drawing
- Family history taking
- Confidentiality and autonomy
- Adult onset disease
 - Predictive/pre-symptomatic) testing
 - Susceptibility testing
- Testing of children
- Termination of pregnancy
- Multiple miscarriages
- Prenatal diagnosis
- Screening tests
- Diagnostic tests
- Support groups and resources
- Genetics and public health
- Grief counselling
- Family development and dynamics

Child development

RECOMMENDED READING FOR PART I

1. Medical Genetics

BONTHRON D, FITZPATRICK D, PORTEOUS M and TRAINER A (1998) Clinical Genetics A Case-based Approach. London, WB Saunders Company Limited.

BURKE W et al Public Health Genetics in the 21st Century (AC)

HARPER PS (2004) Practical Genetic Counselling. 6th ed. Oxford, Butterworth Heineman.

JONES KL (2006) Smith's Recognisable Patterns of Human Malformations. 6th ed. Philadelphia, WB Saunders Company.

KORF BR (2000) Human Genetics, A problem-based approach. 2nd edition Boston, Blackwell Science.

MARIEB, A Essentials of Human Anatomy and Physiology, 7th edition, Benjamin Cummings, 2003

MOORE KL and PERSUAD TVN Medical Embryology. 8th ed. Baltimore: The Williams & Wilkins Company, 2000.

MUELLER RF and YOUNG ID (2004) Emery's Elements of Medical Genetics. 12th ed. Edinburgh, Churchill Livingstone.

STRACHAN T and READ AP (1999) Human Molecular Genetics. 2nd ed. New York, BIOS Scientific Publishers Ltd.

2. Genetic Counselling

EGAN G (1986) The Skilled Helper. 3rd ed. New York, Brooks Cole.

HARPER PS (1998) Practical Genetic Counselling. 5th ed. Oxford, Butterworth Heineman.

KESSLER S (1979) Genetic Counselling, Psychological Dimensions. New York, Academic Press.

NUSSBAUM RL, MCINNES RR AND WILLARD HF (2004) Thompson & Thompson Genetics in Medicine. 6th edition. Philadelphia, Saunders.

APPENDIX B

1.0 CERTIFICATE OF PROFICIENCY

1.1 Ongoing evaluation for medical geneticists in training

1.1.1 This form must be completed by the head of the department and the HPCSA registered clinical head of the Medical Genetics training unit in which the candidate receives training.

Please complete in writing (not typed) and initial each item. The completed form must be submitted to the CMSA.

Name of candidate:

Date of completion of Part 1 exam:.....

Date of HPCSA registration as a registrar in Medical Genetics:

.....

Academic training unit:

.....

Commencement of Medical Genetics training (day/month/year):

.....

Supervising Medical Geneticist:

.....

1.1.2 Six monthly reviews

Candidates require six monthly reviews throughout the 4 years of registrar training. Please indicate the dates for every review period. Rate the candidate's ability for the following categories as inadequate, adequate or excellent.

Theoretical knowledge:

- Medical Genetics
- Clinical Genetics
- Laboratory Tests
- Counselling

Clinical skills in:

- Medical Genetics
- Clinical Genetics
- Laboratory Tests
- Counselling

Attitude

- Motivation
- Commitment
- Follow-up
- Communication
- Team-work
- Maintenance of good ethical standards
- Client relationship

1.1.3 Research experience

1.1.3.1 State number of congress presentations delivered:

- Local
- National
- International

1.1.3.2 Publications

- Local
- National
- International

1.1.3.3 Research – awards

1.1.3.4 Projects – in progress

Provide a brief outline (one paragraph) as an annexure. Indicate clearly the candidate’s role in the project/s

1.1.3.5 Projects – completed

Provide a brief outline (one paragraph) as an annexure. Indicate clearly the candidate’s role in the project/s

1.2 HEAD OF TRAINING UNIT:

The above details correctly reflect the assessment of this candidate:

NAME:.....

SIGNATURE:.....

DATE:.....

1.3. HEAD OF DEPARTMENT:

I endorse that the above details correctly reflect the assessment of this candidate:

NAME:.....

SIGNATURE:.....

DATE:.....

APPENDIX C

1.0 REGULATIONS FOR THE LOGBOOK FOR THE FCMG(SA) PART II

- 1.1 Before being admitted to Part II of the examination for the FCMG(SA), the logbook and a minimum of ONE case report (or other Medical Genetic manuscript) accepted for publication in a peer-reviewed journal must be accepted by the CMSA. The logbook and manuscript with the current assessment fee must reach the Academic Registrar of the CMSA at least three months before the published date of the commencement of the written examination for the Part II FCMG(SA).
- 1.2 The logbook should demonstrate exposure to a minimum of 100 cases per year, during the 4 years of training. These cases will be documented, covering a broad spectrum of genetic disorders and/or birth defects. For each case, the date and place of evaluation, name and/or hospital number of the client, diagnosis, management summary, role of the trainee, and signature of the supervisor needs to be specified. The logbook also needs to include a record of the trainee's departmental presentations of topics and cases, as well as evidence of at least 2 presentations at a national SASHG or international Human Genetics congress.
- 1.3. The publication should have been submitted or published in a Pubmed/Index Medicus indexed journal.
- 1.4 Candidates will be notified when their logbooks and publication(s) have been accepted. In the event of the logbook and publication(s) being rejected, the candidate will be notified what additional work is required.
- 1.5 The rule as to the time of presentation shall apply to new or modified logbooks and/or publication(s) and the appropriate fee shall be payable on each occasion when these are resubmitted.

APPENDIX D

1.0 SYLLABUS FOR PART II OF THE FCMG(SA) EXAMINATION

This examination requires a wide specialist knowledge of medical and clinical genetics. Candidates would be expected to have a good and broad general knowledge of clinical genetics, including the latest developments in the speciality and a clear understanding of evidence based principles of practice. In addition knowledge of the basic sciences of Medical Genetics, laboratory techniques and interpretation of laboratory results, public health genetics, medical ethics and genetic counselling, as applied from Part I of the examination are required.

The syllabus below forms a general guide to the knowledge which the candidates will be expected to have acquired. It is not exhaustive and questions may well be asked on topics which are not covered directly in this syllabus, or conditions which have not been mentioned. The examination aims to test specialist rather than subspecialist knowledge.

2.0 CLINICAL GENETICS

Genetic history-taking

Pedigree construction and analysis

Dysmorphology

Principles of normal and abnormal embryogenesis

Malformations, dysplasias, disruptions and deformations

Approach to the fetus with birth defects

Examination of the fetus

Fetal management

Approach to the child with birth defects

Dysmorphology history

Dysmorphology examination

Dysmorphology diagnosis

Use of databases

Common chromosomal abnormalities

Trisomy 13, 18 and 21

Sex chromosome abnormalities – Turner syndrome, XXY, XYY, XXX

Triploidy

Deletion/Duplication syndromes

Wolf-Hirschorn syndrome

Cri du chat syndrome

Velocardiofacial syndrome

Prader Willi syndrome

Angelman syndrome

Miller-Dieker syndrome

Williams syndrome

Tricho-rhino-phalangeal syndrome

Smith-Magenis syndrome
Pallister-Killian syndrome

Multifactorial disorders

Neural tube defects
Cleft-lip and –palate
Talipes equinovarus
Congenital heart disease
Common complex disorders eg diabetes, hypertension, psychiatric disorders

Teratogens

Principles of teratogenesis

Alcohol

Drugs:

Warfarin

Anti-epileptics

Retinoic Acid

Thalidomide

Infections:

Rubella

Toxoplasmosis

CMV

Syphilis

HIV

Varicella

Parvovirus

Maternal Phenylketonuria

Hyperthermia

Maternal disorders

Thyroid

Diabetes

Hypertension

Cardiac disease

SLE

Malformations and disruptions

ADAM sequence

Amniotic band sequence

Limb-body wall sequence

Connective Tissue Disorders

Osteogenesis imperfecta

Ehlers-Danlos syndrome

Marfan syndrome

Pseudoxanthoma elasticum

Beals syndrome

Homocystinuria

Stickler syndrome

Cutis laxa

Skeletal dysplasias

Including classification, terminology, clinical and radiological assessment

Osteochondrodysplasias

- Achondroplasia
- Hypochondroplasia
- Thanatophoric dysplasia
- Achondrogenesis syndromes
- Short-rib polydactyly syndromes
- Spondyloepiphyseal dysplasia syndromes
- Metaphyseal dysplasia syndromes
- Chondrodysplasia punctata syndromes
- Osteopetrosis
- Sclerosteosis
- Pyknodystosis
- Cleido-cranial dysostosis
- Campomelic dysplasia

Craniosynostosis syndromes

- Apert syndrome
- Pfeiffer syndrome
- Crouzon syndrome
- Saethre-Chotzen syndrome
- Frontonasal dysplasia
- Greig syndrome
- Antley-Bixler syndrome

Multiple exostoses syndrome

Nail-patella syndrome

Langer mesomelic dysplasia

Acrodysostosis

Albright hereditary osteodystrophy

Paget's disease

Kyphomelic dysplasia

Klippel-Feil syndrome

Jarcho-Levine syndrome

Limb defects

Polydactyly syndromes

Syndactyly syndromes

Ectrodactyly syndromes

- Ectrodactyly-ectodermal dysplasia-clefting syndrome

- Adams-Oliver syndrome

Radial ray abnormality syndromes

- Thrombocytopaenia absent radius syndrome

- Fanconi anaemia syndromes

Ulna ray abnormality syndromes

- Holt-Oram syndrome

Phocomelia

- Robert syndrome

- Grebe syndrome

Poland sequence

Pterygium syndromes

Femoral hypoplasia-unusual facies syndrome

Sirenomelia

Caudal dysplasia sequence

Short stature syndromes

Russel-Silver syndrome
 Rubinstein-Taybi syndrome
 Dubowitz syndrome
 Brachmann de Lange syndrome
 Johanssen-Blizzard syndrome
 Seckel syndrome
 Hallermann-Streiff syndrome
 Ellis van Creveld syndrome

Other common syndromes

Noonan syndrome
 Costello syndrome
 Cardio-facio-cutaneous (CFC) syndrome
 Aarskog syndrome
 Robinow syndrome
 Opitz syndromes
 Floating harbour syndrome
 Kabuki syndrome

Facial and limb defects

Miller syndrome
 Nager syndrome
 Townes-Brocks syndrome
 Mohr syndrome
 Oculo-dento-digital syndromes
 Oto-palato-digital syndromes
 FG syndrome
 Larsen syndrome

Cranio -facial defects

Moebius sequence
 Blepharophimosis syndrome
 Robin sequence
 Van der Woude syndrome
 Fronto-nasal dysplasia
 Fraser syndrome
 Branchio-oculo-facial syndrome
 Branchio-oto-renal syndrome

Inborn errors of metabolism and storage diseases

Urea cycle disorders
 Ornithine trans-carbamylase deficiency
 Disorders of amino-acid metabolism
 Phenylketonuria
 Alkaptonuria
 Homocystinuria
 Maple syrup urine disease
 Disorders of carbohydrate metabolism
 Galactosaemia
 Hereditary fructose intolerance

- Glycogen storage diseases
 - Pompe's disease (GSD II)
 - McArdle's disease (GSDV)
 - Von Gierke's disease (GSDI)
- Disorders of steroid metabolism
 - Congenital adrenal hyperplasia
 - Androgen insensitivity
- Disorders of lipid and lipoprotein metabolism
 - Familial hypercholesterolaemia
 - Smith-Lemli-Opitz syndrome
- Lysosomal storage disorders
 - Mucopolysaccharidoses
 - Hurler/Scheie syndrome (MPSI)
 - Hunter syndrome (MPS II)
 - Sanfilippo syndrome (MPSIII)
 - Morquio syndrome (MPSIV)
 - Maroteaux-Lamy syndrome (MPSVI)
 - Sly syndrome (MPSVII)
- Sphingolipidoses
 - Tay-Sachs disease
 - Gaucher disease
 - Niemann Pick disease
 - Canavan disease
- Disorders of purine/pyrimidine metabolism
 - Lesch-Nyhan syndrome
 - Adenosine deaminase deficiency
- Organic acid disorders
 - Methylmalonic acidaemia
 - Propionic acidaemia
- Disorders of porphyrin biosynthesis
- Peroxisomal disorders
 - Zellweger syndrome
 - Adrenoleukodystrophy
- Fatty Acid Transport and Oxidation disorders
- Copper metabolism
 - Wilson disease
 - Menkes disease
- Iron metabolism
 - Haemochromatosis

Hamartoses

- Neurofibromatosis
- Tuberous sclerosis
- Incontinentia pigmenti
- Hypomelanosis of Ito
- Sturge-Weber syndrome
- Von Hippel Lindau disease
- Linear sebaceous nevus syndrome
- Goltz syndrome
- Aicardi syndrome

Ectodermal dysplasias

- Rapp-Hodgkin EDS

Hypohidrotic EDS

Overgrowth syndromes

Beckwith-Wiedemann syndrome
 Weaver syndrome
 Sotos syndrome
 Proteus syndrome
 Klippel-Trenaunay-Weber syndrome
 Marshall Smith syndrome

Cancer syndromes

Inherited breast cancers
 Inherited bowel cancers
 Familial adenomatous polyposis coli
 Hereditary non-polyposis cancer
 Peutz-Jeughers syndrome
 Multiple endocrine neoplasias
 Von Hippel Lindau
 Retinoblastoma
 Wilms tumour
 Gorlin syndrome

Chromosome breakage syndromes

Ataxia telangiectasia
 Fanconi anaemia syndromes
 Cockayne syndrome
 Progeria
 Bloom syndrome
 Xeroderma pigmentosa

Brain anomalies

Neural tube defects
 Lissencephaly
 Holoprosencephaly
 Dandy Walker syndromes
 Agenesis of corpus callosum syndromes
 Microcephaly
 Macrocephaly
 Hydrocephaly

Mental retardation syndromes

X-linked mental retardation
 Fragile X mental retardation syndrome
 Alpha-thalassaemia mental retardation syndromes
 Coffin Lowry syndrome
 MASA syndrome
 Other syndromes
 Fryns syndrome
 Cohen syndrome
 Pallister-Hall syndrome
 Bardet-Biedl syndromes
 Coffin-Siris syndrome

Brain and neuro-muscular syndromes

Arthrogyposis syndromes
 Walker-Warburg syndrome
 Maarden-Waker syndrome
 Acro-callosal syndrome
 Freeman-Sheldon syndrome
 Pena-Shokier syndrome
 Meckel-Gruber syndrome
 Hydrolethalus syndrome
 Neu-Laxova syndrome
 Cerebro-oculo-facial-skeletal syndrome
 Schwartz-Jampel syndrome

Dementias

Huntington disease
 Alzheimer disease
 Parkinson disease

Epilepsies**Associations**

VATER
 VACTERL
 MURCS
 CHARGE
 OEIS
 Oculo-auriculo-vertebral spectrum

Deafness syndromes

Waardenburg syndrome
 Treacher Collins syndrome
 Oculo-auriculo-vertebral spectrum
 Pendred syndrome
 Non-syndromic deafness

Visual impairment syndromes

Colour blindness
 Albinism
 Hereditary retinal and choroidal degeneration syndromes
 Macular degeneration syndromes
 Cataract syndromes
 Retinoblastoma
 Lenz microphthalmia syndromes
 Peters anomaly
 Congenital blindness

Dermatological disorders

Ectodermal dysplasias
 Albinism
 Pigmentary abnormalities
 Piebaldism
 Ichthyoses
 Keratoderma

Keratolytic winter erythema
 Epidermolysis bullosa syndromes
 Xeroderma pigmentosum
 Porphyria
 Pigmentation disorders – piebaldism
 Lipoid proteinosis
 Skin cancers

Respiratory system

Cystic fibrosis
 Kartagener syndrome
 Asthma

Dynamic repeat disorders

Fragile X
 Myotonic dystrophy
 Huntington disease
 Spinocerebellar ataxia
 Friedreich's ataxia

Neuromuscular disorders

Muscular dystrophies and myopathies

- Duchenne and Becker muscular dystrophy
- Emery-Dreifuss muscular dystrophy
- Autosomal recessive muscular dystrophy
- Limb-girdle dystrophy
- Facio-scapulo-humeral dystrophy
- Congenital myopathies

 Spinal muscular atrophy
 Hereditary motor and sensory neuropathies
 Myotonic dystrophy
 Non-dystrophic myotonias

- Periodic paralysis
- Myotonia congenital
- Motor neurone disease

 Movement disorders

- Huntington disease
- Spino-cerebellar ataxias

Immunological disorders

Immuno-deficiency disorders
 Disorders of granulocyte function
 Autoimmunity

Haematological disorders

Inherited bleeding/clotting disorders

- Haemophilias
- Von Willebrands disease
- Factor V Leiden
- Antithrombin 3 deficiency
- Thrombocytopaenia absent radius

 Inherited anaemias

- Haemoglobinopathies and thalassaemias
- Red cell membrane disorders
- Red cell disorders
- Fanconi anaemia
- Blood groups
 - Rhesus feto-maternal incompatibility
- Leukaemias and lymphomas

Urogenital disease

- Congenital renal or urinary tract disorders
- Renal cystic disease
- Nephrotic syndrome
- Renal tubular acidosis
- Renal and urogenital tumours
- True hermaphroditism
- Androgen insensitivity syndrome
- Infertility

Cardiovascular system

- Congenital heart disease
 - Velo-cardiofacial syndrome
- Cardiomyopathies
- Familial dysrhythmias and conduction disorders
- Disorders of venous and lymphatic system

Pregnancy

- Teratogenic and harmful drugs
- Maternal medical problems
 - Diabetes
 - Cardiac
 - Endocrine
 - Epilepsy
 - SLE
- Prevention of neural tube defects
- HIV/AIDS
- Pregnancy monitoring
 - Identification of high risk pregnancy

Prenatal diagnosis

- Chorionic villus sampling
- Ultrasound
- Amniocentesis
- Cordocentesis
- Preimplantation genetic diagnosis

3.0 RECOMMENDED READING FOR PART II

3.1 Textbooks

AASE JM Diagnostic dysmorphology Plenum Medical book company
1990

BONTHRON D, FITZPATRICK D, PORTEOUS M and TRAINER A (1998) *Clinical Genetics A Case-based Approach*. London, WB Saunders Company Limited.

CONNOR M and FERGUSON-SMITH M (1997) *Essential Medical Genetics*. 5th ed. Oxford, Blackwell Science Ltd.

GORLIN RJ, COHEN MM and HENNEKAM RCM (2001) *Oxford Monographs on Medical Genetics No.42. Syndromes of the Head and Neck*. 3rd ed. Oxford, Oxford University Press.

JONES KL (2006) *Smith's Recognisable Patterns of Human Malformations*. 6th ed. Philadelphia, WB Saunders Company.

KORF BR (2000) *Human Genetics, A problem-based approach*. 2nd edition Boston, Blackwell Science.

MOORE KL and PERSUAD TVN (1993) *The Developing Human. Clinically Orientated Embryology*. 5th ed. Philadelphia, WB Saunders Company.

NUSSBAUM RL, MCINNES RR AND WILLARD HF (2004) *Thompson & Thompson Genetics in Medicine*. 6th edition. Philadelphia, Saunders

RIMOIN DL, CONNOR JM, PYERITZ RE, KORF B (eds) (2002) *Emery and Rimoin's Principles and Practice of Medical Genetics*. Fourth Edition. Churchill Livingstone

SCRIVER CR, BEAIDET AL, SLY WS and VALLE D (eds) (1995) *The Metabolic and Molecular Basis of Inherited Disease*. New York, McGraw-Hill Inc.

TRENT RJ (1997) *Molecular Medicine An Introductory Text*. 2nd ed. New York, Churchill Livingstone.

STRACHAN T and READ AP (1999) *Human Molecular Genetics*. 2nd ed. New York, BIOS Scientific Publishers Ltd.

3.2 Databases

London Medical Databases
 Online Mendelian Inheritance in Man
 POSSUM
 Radiological Electronic Atlas of Malformation Syndromes
 Gene Reviews

3.3 Important Reference Periodicals and Journals including:

Am J Hum Genet
 Am J Med Genet

BMJ
 Clin Genet
 Hum Genet
 Human Molecular Genetics
 JAMA
 J Med Genet
 Lancet
 Nature Genetics
 New Engl J Med
 S Afr Med J

APPENDIX E: GUIDELINES FOR CANDIDATES AND EXAMINERS

1.0 CANDIDATES

- 1.1 Recognised training centres should have a supervisor for registrars in training. The supervisor should be on the panel of examiners and be familiar with the examination and the CMSA regulations
- 1.2 The role of the supervisor should include discussion of the regulations for the FCMG(SA) examination with prospective candidates; indication of the breadth and depth required for different aspects of the examination; discussion of the methods of assessments used in the examination, informing the candidate of the limitations of his or her hospital as a training institution – comment)
- 1.3 On written request written reports on their performance will be made available to unsuccessful candidates after the examinations from the CMSA convener. These must be such as to allow unsuccessful candidates to learn where they have made mistakes and correct their deficiencies in specific areas

2.0 EXAMINERS

- 2.1 Question papers will be carefully reviewed by the convener and other examiners before the examinations, and all care will be taken to ensure that the questions are appropriate and free from ambiguities, grammatical errors, errors of vocabulary and spelling errors
- 2.2 At least two examiners will examine each candidate in the clinical parts of the examination. Examiners should play a minor role in the examination of candidates with whom they have worked closely in the recent past
- 2.3 In the clinical parts of the examinations, each examiner should submit his or her own independent assessment of each candidate. Discrepancies between the assessments will be discussed at the examiners meeting. The consistency of the examinations as a whole will be assessed
- 2.4 Examiners should familiarise themselves with the basic theoretical considerations involved in examinations, in medical examinations in particular

- 2.5 All new examiners should undergo a period of familiarisation during which they act as observers of the clinical and oral parts of the examination. During this period they will not submit assessments of candidates.

3.0 THE CLINICAL EXAMINATION

3.1. THE LONG CASE

3.1.1 Candidates

You will be asked to take a comprehensive history and perform a complete physical examination on a patient. You will have one hour to do this. The patient will usually have multiple problems which may be related or unrelated. When you have completed the history and examination you will be required to present your findings, a diagnosis, differential diagnosis and management plan to two examiners (an observer may be present). You will be assessed on the **accuracy** and **completeness** of your evaluation and **management plan** which should demonstrate your ability to prioritise problems, plan cost-effective and safe investigation and rational management. You will be expected to demonstrate insight into preventative strategies and prognosis. After the presentation of your findings and discussion, your examiners may ask you to interpret and integrate the results of relevant special investigations into your differential diagnosis and management plan. Your examiners may choose to expand the discussion to aspects of management, pathophysiology, pharmacology, genetics or other relevant areas (e.g. screening) in which a competent specialist Medical Geneticist would be expected to be knowledgeable. The standard expected is that of a specialist Medical Geneticist.

3.1.2 Examiners

The standard is that which is expected of a competent specialist Medical Geneticist.

NOTE FOR EXAMINERS

- i) The purpose of the long case is to ensure that a candidate is able to elicit a history, perform a competent physical examination and formulate a reasonable plan of further investigation and management.
- ii) Ideally, examiners should have an opportunity to take a brief history and perform a physical examination on patients beforehand.

3.2. THE SHORT CASE

3.2.1 **Candidates**

You will be asked to examine two short cases. Usually there will only be one organ/system involved and you will be informed which this is. If there are related problems in other systems you will be expected to identify and examine these systems if necessary. You will have 20 - 30 minutes for the brief history and physical examination of each patient and after this will be expected to present your finding to two examiners (an observer may be present). You will be assessed on the **accuracy** of your elicitation of the **symptoms** and **signs** and your **final diagnosis**. You may be asked to discuss your interpretation of the severity of the problem and a plan for cost-effective and safe management.

3.2.2 **Examiners**

The standard is that which is expected of a competent specialist Medical Geneticist

NOTE FOR EXAMINERS:

The purpose of the short cases is to evaluate the candidates' clinical skills and emphasis should be placed on accuracy of description and interpretation of clinical features.