CMSA
The Colleges of Medicine of South Africa NPC
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JOHANNESBURG OFFICE
EXAMINATIONS & CREDENTIALS

JUNE 2023

R E G U L A T I O N S

FOR ADMISSION TO THE FELLOWSHIP OF THE

COLLEGE OF MEDICAL GENETICISTS OF SOUTH AFRICA

FCMG(SA)

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 A broad outline of what is expected of a specialist Medical Geneticist is as follows:

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 congenital disorders;

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

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

1.5 It is the candidate's responsibility to become aware of regulations of their training unit and ensure compliance with those regulations in addition to these regulations.

These regulations should be read in conjunction with the guidelines for candidates and examiners (Appendix E)

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 The candidate must be currently appointed in a post as a registrar in a HPCSA registered Medical Genetics unit, such posts having been recognised by the HPCSA for the purpose of the Medical Genetics training. Application for exemption will be considered in special circumstances by the CMG.

2.3 The examination must be successfully completed prior to completing 30 months in a full time registrar post in an HPCSA registered Medical Genetics unit. Application for exemption will be considered in special circumstances by the CMG provided that the application is fully supported by the relevant head of the training unit.

2.4 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 COMPETENCIES AND SYLLABUS FOR THE PART I EXAMINATION

3.1 The focus of the FCMG Part I examination is the application of basic sciences and applicable ethical aspects and principles of genetic counselling relevant to Medical Genetics.

3.1.1 The competencies required and a guide to learning is included as Appendix A. This is not exhaustive and the candidate is expected to have a broad knowledge of these topics including areas of new advances.

4.0 CONDUCT OF THE PART I EXAMINATION

The examination will consist of 2 written papers of equal weighting

4.1 **Paper 1** will comprise a combination of Single best answer (SBA) questions, very short and short answer questions that may include short scenarios with a total of **125 marks**

4.2 **Paper 2** will comprise short answer questions and may include scenarios with a total of **125 marks**

4.3 Each paper will be of at least 3 hours' duration.

4.4 The examination will require electronic answering. Time for typing has been factored in.

4.5 The pass mark will be 50% for the 2 papers combined with no subminima scores.

4.6 The examination (questions and answers) will be conducted in English only

4.7 If more answers are given than are asked for, only the requested number of answers will be marked starting from the first answer.

4.8 The mark allocation should be used as a guide to what is expected.

5.0 TRAINING FOR THE PART I EXAMINATION

The candidate must submit evidence that they occupy an HPCSA accredited training post in a registered training unit approved for the purposes by the HPCSA before admission to Part I of the 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 within the preceding 6 years.

6.1.2 having completed the training set out in 9.0 of the regulations

6.1.3 having completed at least 36 months fulltime training as a registrar in a teaching unit of Medical Genetics registered with the HPCSA at the time of writing the examination. Special consideration may be given to candidates who have been in their training post for less than 36 months who are supported by the head of the training unit to enter the examination if agreed upon by the CMG.

6.1.4 a certificate of proficiency from the Head of Department and the HPCSA registered clinical head of the teaching unit (Appendix B)

6.1.5 having fulfilled the requirements for a Portfolio set out in Appendix C of the regulations

6.1.6 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 Portfolio are as follows:**

6.2.1 Requirements are set out in Appendix D.

6.2.2 Before being allowed to enter for the examination, a candidate shall submit a Portfolio summary as indicated in appendix D that has been assessed and approved by the head of the training unit. Any unsatisfactory or outstanding requirements should be addressed in the certificate of proficiency (appendix C).

6.2.3 Candidates must present themselves for the examination within two years of having their Portfolio completed and approved. Failing this, a new Portfolio will be required.

6.2.4 Candidates who fail the examination may again enter the examination on the basis of their original Portfolio, within a period of 2 years.

6.2.5 The Portfolio must reach the Academic Registrar in Johannesburg as detailed in 9.0 of the regulations.

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7.0 COMPETENCIES AND SYLLABUS FOR THE PART I EXAMINATION

7.1 The focus of the FCMG Part II examination will be on integrated knowledge of the aetiology, epidemiology, presentation, clinical features, selecting and interpreting appropriate investigations, management, and prognosis in medical genetic disorders as well as the context of genetics and genomics in health care including the interpretation, utility and implementation of genetic and genomic data.

7.1.1 The competencies required and a guide to learning is included as Appendix B. This is not exhaustive and the candidate is expected to have knowledge of the full spectrum of medical genetic practice 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 clinical application.

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 and congenital disorders that are common and those that are 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

Part II Examinations for College of Medical Genetics

The examination will consist of 2 written papers and an online clinical/oral/practical assessment.
Clinical focus: Includes integration of clinical genetics, basic science, medical genetics, genetic counselling and ethical principles

8.1 **Written examination**

8.1.1 The examination will consist of two papers of equal weighting
8.1.2 Each paper will be 3 hours in duration
8.1.3 Each paper will be worth 125 marks
8.1.4 Paper 1 will comprise MCQ questions and very short and short answer questions.
8.1.5 Paper 2 will comprise very short and short answer questions of an integrative nature.
8.1.6 There will be no sub-minima criteria, and an average mark of 50% over the two papers will be required for invitation to the oral component of the examination
8.1.7 The examination (questions and answers) will be conducted in English only.
8.1.8 If more answers are given than are asked for, only the requested number of answers will be marked starting from the first answer given.

8.2 Candidates who achieve the required marks in the written component but who fail the oral component of the assessment will be exempt from the written component of the examination in the next semester. Such exemption applies to one sitting only and must be exercised in the following semester except under very exceptional circumstances.

8.3 **Clinical/ Oral/ Practical assessment**

The current format of the examination is described below. The format of the examination is subject to change depending on circumstances. Candidates will be informed timeously of any changes to the format.

8.3.1 A modified clinical/practical /or al examination will be conducted in the form of an electronic written Objective Structured Clinical Examination and an online platform based Structured Oral Examination (SOE). There will be a minimum of 2 examiners p er station and each candidate will be exposed to at least 3 different examiners during the examination.

8.3.2 Objective Structured Clinical Examination (OSCE)

Number of stations: 1

Duration of station: 30 minutes (30 marks) (with an additional 10 minutes allowed for typing)

Electronic written examination

PowerPoint slides or word documents will be used and may include case histories and charts, test results, still images, photos and diagrams. Use of electronic databases may be included.

8.3.3 Structured Oral Examination (SOE)

Number of stations: 4 of 15 minutes (20 marks each), and 1 of 30 minutes (40 marks)

Duration of stations: 90 minutes in total

Online platform based examination

PowerPoint slides will be used and may include case histories and charts, test results, still images, photos, diagrams and digital imaging. In addition to verbal responses, candidates may also be asked to provide written or demonstrated responses. Use of electronic databases may be included.

Candidates are encouraged to consider their answers carefully as, if more than one answer is given, the first answer will be that which is assessed.

8.4 **WEIGHTING OF THE EXAMINATION FOR FCMG(SA) PART II**

Written Paper I	(25%)
Written Paper II	(25%)
Clinical/Practical/Oral exam	(50%)

A score of 50% or more is required in the Clinical/Practical/Oral exam to achieve an overall pass score.

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 or appropriate equivalence in the case of international trainees.

9.1 General:

The mandatory period in resident hospital posts recognised for pre-registration purposes by the Health Professions Council of South Africa

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

9.3 Candidates must have completed a minimum of 36 months 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 the FCMG(SA) does not guarantee registration as a specialist with the HPCSA and it is the candidates responsibility to ensure they meet all the HPCSA requirements for registration

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10.0 ADMISSION AS A FELLOW

10.1 Only candidates who have completed training in a CMSA recognised registrar post may be awarded a fellowship if successful in the examination.

10.2 **Candidates who have written the examination as a prerequisite from the HPCSA for inclusion on the specialist register are not eligible to be awarded a Fellowship but will be sent a letter confirming their success in the examinations**

All other candidates will be asked to sing a declaration as below:

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 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).

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APPENDIX A

COMPETENCIES EXPECTED FOR THE FCMG PART 1 EXAMINATION:

Understand the structure, function, replication, and epigenetic control of human genetic/genomic material.
Understand the embryological basis of congenital disorders, with an emphasis on dysmorphology and teratology.
Understand the role of genetics in embryology, physiology, and pathophysiology (causation of diseases that have a genetic component).
Apply this understanding particularly to common chromosomal, Mendelian, mitochondrial, multifactorial, and teratogenic disorders.
Apply principles of genetics and genomics to therapeutics.
Understand the terminology and nomenclature used in Medical Genetics.
Understand the principles and basic practicalities of laboratory genetics, with an emphasis on cytogenetics/genomics, and molecular genetics/genomics.
Understand clinical genetic/genomic testing in the context of screening, diagnosis, preimplantation and prenatal diagnosis, presymptomatic, carrier and cascade testing.
Understand principles of variant classification and interpretation.
Apply this understanding to laboratory diagnostic tests and their interpretation.
Recognise patterns and apply clinical principles to diagnose common genetic conditions.
Apply the basic principles of public health, epidemiology, and community health as well as interpretation of family history to identify individuals at risk of a genetic disorder (or their children) to offer appropriate screening and/or preventative strategies.
Apply these principles to the understanding of inheritance patterns.
Understand communication skills and counselling techniques and apply this to taking a personal, psychosocial, medical, and family history as well as to explaining genetic disease to patients and the family members at risk of genetic disease.
Know the symbols used in pedigree drawing and be able to draw and interpret a pedigree and family history.
Understand the principles of clinical examination and be able to distinguish normal from abnormal clinical features.
Understand basic statistics and apply this to the interpretation of growth charts and laboratory investigations, as well as in the research setting.
Develop computer skills and literacy to enable the use of various genetic databases in clinical practice.
Understand the broad principles of bioethics in the context of medical genetics.
These competencies will form the underpinning for achieving the competencies required of a specialist Medical Geneticist.

To guide learning some of the required knowledge to achieve these competencies is outlined in the below. The majority of the FCMG part 1 examination will interrogate basic genetic science and the principles of medical genetics, genetic counselling, and bioethics. This knowledge may be tested through examples and therefore a basic knowledge of commonly encountered genetic syndromes is required. Some common conditions are included in the list of topics to assist the candidate as examples of what might be expected. Likewise, the domains and topics found below are not exhaustive because medical genetics is a rapidly developing field. As a result, this does not exclude other topics being included in the examination and the importance and relevance of certain topics may change with time. Candidates are encouraged to engage with their training unit consultants for further guidance.

FCMG Part 1	
Domain	**Topic**
Cell structure and cycle	Describe the cell structure, its organelles and functions
	Describe the cell cycle including apoptosis
	Describe the stages of cell division and medical relevance of mitosis and meiosis
	Describe human gametogenesis and fertilization and the transmission of genomic material
Human Genome structure and function	Describe the organization of the human genome, including coding and non-coding regions
	Describe the chromosomal structure of the human genome
	Describe the organization and structure of genes (exons, introns, promoter regions, enhancers, silencers, etc.)
	Describe the structure of DNA, how it is replicated and maintained (DNA repair mechanisms)
	Explain the types of nucleic acids, their functions and differences
	Explain basic gene expression: transcription through to translation, gene regulation (including splicing, tissue specificity, non-coding RNAs), post-translational modifications
	Understand broad concepts of epigenetic control of gene expression.
	Describe the organisation and structure of the mitochondrial genome
Genetic variation	Understand normal versus pathogenic variation
	Differentiate single nucleotide, copy number and chromosomal variation
	Understand the basic use of HGVS nomenclature
	Describe different types of gene mutations/variants (missense, nonsense, frameshift, splicing, intronic etc)
	Understand the functional significance of different types of gene variants (e.g. gain vs loss of function, haploinsufficiency, dominant-negative, nonsense mediated decay, etc.)
	Understand the concept of hypomorphic versus pseudo-deficient alleles, and approaches to determine their clinical significance
	Discuss the origins and effects of mosaicism, including effect on recurrence risk and phenotype
Cytogenetics	Describe the general concepts of numerical autosomal and sex chromosomal abnormalities (e.g. aneuploidy, supernumerary marker, polyploidy)
	Describe general concepts of structural chromosome abnormalities (Robertsonian translocation, reciprocal translocation, inversions, ring chromosomes)
	Describe the aetiology of chromosome abnormalities (e.g. non-disjunction, breakage and repair, non-homologous recombination, mitotic recombination, sister chromatid exchange, uniparental disomy, copy-neutral loss of heterozygosity)
	Understand mosaicism as it relates to cytogenetics
	Understand the basic use of ISCN nomenclature
	Describe various cytogenetic techniques, their indications and limitations: i. Chromosome analysis ii. FISH

	iii. Copy number analysis, e.g. MLPA, array iv. Aneuploidy QF-PCR v. CNV and structural rearrangement calling from WES/WGS
Molecular genetics methods	Understand the basic principles of the polymerase chain reaction
	Understand the basic concepts of nucleic acid sequencing, including Sanger and massively parallel sequencing, RNA sequencing, long range sequencing
	Understand the basic principles of molecular techniques , including qPCR, QF-PCR, MLPA, TP-PCR, WES, whole genome sequencing and their applications/limitations (allele drop-out, primer binding site polymorphisms, large deletions, etc.)
	Understand the basic principles of nucleic acid hybridization assays (e.g. Southern blot)
	Understand the basic principles and applications of linkage analysis
	Describe the concept of identity testing and its use as an adjunct method to establish the relationship between samples (i.e. maternal cell contamination, sample identity matching)
	Susceptibility and predictive tests
	Discuss the use of Direct to Consumer (DTC) or over the counter testing with reference to genetic testing
Principles of clinical/diagnostic laboratory genetic testing	Discuss collecting specimens for laboratory and pre-analytical handling
	Understand concepts of test validity and utility
	Discuss accreditation and quality assurance in the context of a genetic laboratory
	Understand the elements of a genetic report
	Understand the implications and reporting of pathogenic, benign and variants of unknown significance
	Discuss trouble-shooting where results are out of keeping with clinical data
Interpretation of results and Clinical Bioinformatics	Describe sequence variants using appropriate nomenclature (i.e. HGVS)
	Explain the classification of sequence variants and the criteria used (e.g. ACMG-AMP)
	Understand the principles of bio-informatics with emphasis on clinical applications
	Use of genome browsers and ‘omic databases e.g. Decipher, Clinvar, Gnomad etc
	Understand tools to interrogate the genome e.g. Clingen, Varsome
Basic statistics and research methods	Interpret basic statistical analysis, e.g. average, mean, median, SD
	Understand statistical assessment of test validity e.g. test accuracy, sensitivity, specificity, predictive values
	Basic understanding of methods for analysis of data:; chi square, Fisher two-tailed, t-test, odds ratio
	Broad concepts in different types of research methodology (Quantitative, qualitative, genetic and genomic)
Population genetics	Understand the concepts of screening for genetic conditions on at levels (individual, population etc.)

	Population based registries
	Describe the key concepts of human genetic variation in populations, including of founder mutations
	Explain consanguinity and its implications for genetic inheritance and disease
	Describe and apply the Hardy-Weinberg equilibrium
Pedigree drawing and analysis	Be able to draw an accurate pedigree using standard nomenclature
	Demonstrate the ability to analyse pedigrees for inheritance patterns
Multifactorial inheritance	Explain liability to multifactorial trait based on the normal distribution curve (Bell curve)
	Describe the role of twin studies and 'heritability' of multifactorial disorders
	Understand GWAS studies
	Understand the principles of the polygenic risk score
	Describe the contribution of the environment and epigenetics to multifactorial inheritance
Chromosomal inheritance	Understand recurrence risks related to aneuploidies and chromosomal non-disjunction
	Understand recurrence risks of genomic rearrangements including inversions, reciprocal translocations and Robertsonian translocations.
	Understand the meiotic segregation of rearranged chromosomes and the effects of recombination events
Single gene / Mendelian inheritance	Describe the principles of autosomal dominant, autosomal recessive, X-linked and Y-linked inheritance
	Understand the concepts of penetrance, expressivity and anticipation
	Explain the relationship between mutations/variants and inheritance patterns (i.e. dominant negative effect etc.)
Non-Mendelian inheritance	Describe common non-Mendelian inheritance and their aetiologies, including uniparental disomy, imprinting, mosaicism, unstable triplet repeats, pseudoautosomal inheritance, etc
	Describe genetic diagnostic testing for these conditions
Mitochondrial inheritance	Understand matrilineal inheritance as it pertains to population genetics and mitochondrial disorders
	Understand heteroplasmy as a basis for clinical heterogeneity of mitochondrial disorders
	Differentiate the role of nuclear and mitochondrial genes in mitochondrial disease
	Describe general features of mitochondrial disorders
Risk calculation	Calculate recurrence risk in Mendelian, non-Mendelian, chromosomal and multifactorial and complex disorders
	Demonstrate the ability to modify a priori risk by one conditional factor (basic Bayesian analysis)
Ethical and medicolegal principles	Describe the basic principles of medical ethics and how they may relate to genetic conditions
	Informed consent in clinical and research context

	Privacy and confidentiality
	Legal aspects of termination of pregnancy
	Ethical issues of genetic tests (predictive testing, sex selection, incidental findings
	Ethical issues of genomic test (e.g. incidental findings)
	Understand the history of clinical genetics in terms of prior eugenics/dysgenics
	Patient advocacy
	Patient records and note keeping, including POPIA
	Recognising and reporting abuse
Immunology	Basics of immunoglobulins (e.g. genetic changes in clonal development of cells producing IgG)
	Basics of different types of immunity (inate vs adaptive) and cellular immunity
	Principles of inborn errors of immunity
Pharmacogenetics/genomics	Describe how genotype may be used to improve assessment of drug responsiveness, and risk of drug reactions
	Medication use and adverse reactions in genetic disease (e.g. PV, G6PDdef)
Public Health	Epidemiology in relation to genetic disorders
	Screening vs diagnostic testing
	Biostatistics
	Use of "big data"
	Patient support groups
	Prevention of congenital disorders
Genetic counselling	Pre-test counselling; antenatal, postnatal, predictive, screening
	Understand the principles of genetic counselling and communication
	Describe appropriate indications for referral for genetic counselling
	Demonstrate the basic techniques of genetic counselling , e.g. breaking bad news
	Understanding non-verbal communication
	Reactions to genetic counselling
	Understand the grieving process
	Identify abnormal responses and need for psychotherapy
	Understand the difference in counselling techniques across different age groups, intellectual ability and cross cultural counselling
Clinical - general	Take a relevant genetic history and draw the pedigree, interpret the history and pedigree
	Basic examination techniques including use of growth charts.
	Understand the principles of general and organ specific examination

	Measurements as they relate to dysmorphological examination
	Demonstrate correct use of HPO and dysmorphological terms
	Principles of clinical photography
	Describe the methods of assessment of phenotypic variations, syndrome identification and diagnosis, including generally accessible computer diagnostic aids (e.g. OMIM)
Common disorders	Describe the molecular basis, pathogenesis and clinical features of common chromosomal, microdeletion syndromes. Examples: - Aneuploidy (13, 18, 21, X, Y) - 22q11.2 deletion
	Describe the molecular basis, pathogenesis and clinical features of common single gene conditions: Examples: - Autosomal dominant: achondroplasia, neurofibromatosis type 1, tuberous sclerosis - Familial cancers: HBOC, Lynch, retinoblastoma - Autosomal recessive: common haemoglobinopathies, spinal muscular atrophy, cystic fibrosis - X-linked disorders: Dystrophinopathies, Haemophilia A/B
	Describe the molecular basis, pathogenesis and clinical features of common triplet repeat conditions. Examples: - Fragile X, myotonic dystrophy, Huntington disease
	Describe the molecular basis, pathogenesis and clinical features of common imprinting disorders. Examples: - PWS/AS, BWS/RSS
	Describe the pathogenesis and common clinical features of other common birth defects. Examples: - Teratogens: Fetal alcohol spectrum disorder, diabetic embryopathy, congenital CMV - Multifactorial birth defects: neural tube defect, cleft lip and/or palate - Sequences: oligohydramnios sequence, holoprosencephaly sequence
Applied embryology	Describe key embryological processes
	Morphogenesis in terms of deformation, malformation, disruption, dysplasia, sequence, syndrome, association
	Teratogens, types and mechanisms
	Understand and describe the embryology processes underlying common congenital disorders
Applied anatomy and physiology	Basic anatomy and physiology of the human body: i. Skeletal system ii. Cardiovascular system iii. Gastro-intestinal system iv. Neurological system v. Genito-urinary system vi. Respiratory system vii. Haematology and immunity
	Physiology and pathophysiology related to common genetic disorders (CAH, PV, SMA etc.)

Normal growth and development	Normal growth parameters
	Normal anatomic variation
	Describe normal childhood developmental domains and red flags i. Gross motor ii. Fine motor/vision iii. Hearing and speech iv. Personal and social
Prenatal genetics	Describe the principles of and approaches to prenatal screening (invasive and non-invasive)
	Compare approaches to prenatal diagnosis versus postnatal diagnosis
	Describe the general principles of biochemical, molecular and cytogenetic testing on prenatal samples
	Describe the different techniques used in prenatal diagnosis, include limitations, risks and benefits
Cancer genetics	Define the concepts of inherited predisposition to cancer, oncogene activation, tumour suppressor inactivation, alteration of cell cycle control and DNA repair genes
	Explain and contrast inherited versus somatic mutations
	Describe the relevance of cytogenetic and molecular analysis to cancer diagnosis, prognosis and monitoring (e.g. gene expression, recurrent rearrangement, recurrent pathogenic variants)
	Knowledge of common genetic cancers; mechanisms (including micro-satellite instability, loss of heterozygosity, Knudson hypothesis), inheritance, main features of cancer syndromes in particular but not limited to HBOC, CRC
Inborn errors of metabolism	Describe the different categories of proteins in a cell (structural, enzymes, transport, receptor proteins etc.), their modes of action and means of regulation
	Describe the biochemical consequences of a primary enzyme block in a metabolic pathway and the way clinical and pathological signs may be produced
	Understand the principles of newborn screening
	Understand the deleterious effects of toxic metabolites on the fetus (e.g. maternal PKU)
	Biochemical classification of IEM
	Mechanism of IEM and metabolic pathways
	Principles of treatment for IEM
	Storage diseases, principles and mechanisms
Treating genetic disorders	Gene therapy principles and applications
	Principles and applications of gene based therapies
	Chaperones, principles of
	Enzyme Replacement Therapy
	Gene editing
	Dietary and conventional medication use

With a field that is developing so rapidly, it is recognised that many sources of study will be online but a short list of texts to consider follows**:**

- Aase JM. *Diagnostic dysmorphology.* Plenum Medical book company
- Moore KL and Persuad TVN. *The Developing Human. Clinically Orientated Embryology.* Philadelphia, WB Saunders Company
- Nussbaum R, McInnes RR and Willard HF. *Thompson & Thompson Genetics in Medicine.* Philadelphia, Saunders
- Rimoin DL, Connor JM, Pyeritz RE, Korf B. (eds) *Emery and Rimoin's Principles and Practice of Medical Genetics.* Churchill Livingstone
- Scriver CR, Beaidet AL, SLY WS and Valle D. (eds) *The Metabolic and Molecular Basis of Inherited Disease.* New York, McGraw-Hill Inc
- Trent RJ. *Molecular Medicine An Introductory Text.* New York, Churchill Livingstone
- Strachan T and Read AP. *Human Molecular Genetics.* New York, BIOS Scientific Publishers Ltd

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APPENDIX B

COMPETENCIES EXPECTED OF A SOUTH AFRICAN MEDICAL GENETICIST PREPARING FOR FCMG PART II EXAMINATION.

Be able to take and interpret a focused history and family history for children and adults presenting with possible genetic disorders.

Be competent in examination of children and adults with possible genetic disorders with a focus on identifying dysmorphology and recognizing patterns of common genetic disorders,

Be able to use adjuncts to dysmorphology assessment including dysmorphology databases and AI tools.

Be able to provide an accurate risk assessment and genetic counselling in a genetic consultation, appropriate to the context e.g., prenatal, cancer.

Be able to accurately estimate recurrence risk of a disorder based on available information, and to communicate to the patient/family.

Be able to investigate common presentation encountered in medical genetic practice, including use of relevant non-genetic tests.

Be able to manage common presentation encountered in medical genetic practice, including consideration of referrals and non-medical management (e.g. grants)

Be aware of the aetiology and natural history of common congenital structural disorders.

Be able to diagnose and manage common paediatric dysmorphic syndromes.

Be able to diagnose and manage common inherited inborn errors of metabolism.

Be able to diagnose and manage common adult-onset genetic disorders.

Be able to diagnose and manage common neurogenetic and cardiogenetic disorders.

Be able to gather appropriate information and provide risk assessment to individuals and families with cancer.

Be able to recognise and manage common cancer genetic syndromes.

Be able to offer appropriate screening and invasive genetic testing in the prenatal setting to pregnant women with or at risk for congenital disorders in their child.

Be able to counsel on the cause, differential diagnosis, investigation and prognosis of common congenital abnormalities in the prenatal setting.

Be able to recognise indicators suggesting a rare genetic disorder at any age and have an approach to investigating further in this setting.

Be able to decide on the most appropriate genetic and genomic investigations in common scenarios and be aware of the limitations of these.

Be able to assess significance of a possible teratogen exposure, and to counsel appropriately.

Be able to interpret laboratory reports reflecting genetic/genomic testing results and to provide the laboratory with clinical support in interpreting results.

Be able to differentiate the role of clinical versus non-clinical genetic/genomic tests.

Be familiar with bioinformatic tools commonly used by clinicians in assessing variants.

Be able to apply sound ethical principles in medical genetic practice.

Be aware of current legislation related to the practice of medical genetics.

Be able to apply common public health principles to management of genetic disorders in a community.

Be able to initiate and conduct clinically relevant research ethically.

Be able to stay up to date with developments in medical genetics and genomics, using journal articles and other resources.

To guide learning, some of the required knowledge and skills to achieve these competencies is outlined in the below. The majority of the FCMG part 2 examination will be focused on integrated knowledge of the aetiology, epidemiology, presentation, clinical features, selecting and interpreting appropriate investigations, management, and prognosis in more common medical genetic disorders that the trainee has been exposed to, conditions that underlie important principles in medical genetics and genomics and approaches to clinical presentations relevant to the medical geneticist. The candidate will also require a working knowledge of the context of genetics and genomics in health care; the interpretation, utility and implementation of genetic and genomic data and an understanding of how to apply this in both research and different clinical environments. The list provided below is not exhaustive as this is a rapidly developing field, and the importance and relevance of certain topics may change with time. As a result, this does not exclude other topics being included in the examination. The standard expected is that of a competent and safe medical genetics practitioner. Candidates are encouraged to engage with their training unit consultants for further guidance.

FCMG Part 2		
Domain	**Approaches**	**Specific topics**
General Genetics	Developmental delay Intellectual disability Autism Spectrum disorder	Disorders caused by chromosome abnormalities and copy number variation: Down syndrome Edward syndrome Patau syndrome Triploidy Turner syndrome Klinefelter syndrome 22q11.2del syndrome Williams syndrome Prader Willi and Angelman syndrome Wolf Hirschhorn syndrome Cri du chat 1p36 deletion Smith-Magenis syndrome Pallister-Killian syndrome 15q11-13 duplication syndrome 16p11.2 deletion Important single gene disorders: Noonan syndrome and common Ras'opathies CHARGE syndrome Fragile X syndrome Rett syndrome Cornelia De Lange syndrome Bardet Biedl syndrome and common ciliopathies Rubinstein Taybi syndrome Kabuki syndrome Mabry syndrome DDX3X syndrome

		Coffin Siris syndrome Coffin Lowry Opitz G syndrome Aarskog syndrome Robinow syndrome ATRX syndrome
	Visual impairment	Oculocutaneous albinism Retinitis pigmentosa Retinoblastoma Cataract Aniridia Ectopia lentis Stargardt Disease Leber congenital amaurosis
	Approach to Hearing loss Types of hearing loss Syndromic vs non syndromic hearing loss	Waardenburg syndrome Branchio-oto-renal syndrome Usher syndrome Jervell and Lange-Nielsen syndrome Pendred syndrome Hermansky Pudlak
Neurogenetics	Infantile hypotonia	Spinal muscular atrophy Myotonic dystrophy
	Epileptic encephalopathy	Dravet syndrome
	Structural CNS abnormalities	Holoprosencephaly Ventriculomegaly Dandy Walker malformation Agenesis of corpus callosum Neural tube defects Schizencephaly
	Neuronal migration abnormalities	Lissencephaly
	Leukodystrophy	X- linked adrenoleukodystrophy Leigh syndrome
	Microcephaly	Primary microcephaly
	Macrocephaly	Greig cephalopolysyndactyly
	Neuroregression	Mitochondrial hepatopathy Neuronal ceroid lipofuscinoses
	Movement disorders	Huntington Disease Familial Parkinson disease
	Myopathies and muscular dystrophies	Duchenne and Becker muscular dystrophy Facioscapulohumeral dystrophy RYR1 congenital myopathy STAC3 myopathy Myotubular myopathy Centronuclear myopathy
	Hereditary neuropathies	HMSN type 1 Hereditary neuropathy with liability to pressure palsies
	Ataxia	Spinocerebellar ataxia type 1, type 2 and type 7 Friedreich's ataxia
	Dementia	Alzheimer disease
	Hereditary Spastic paraplegia	
Cardiogenetics	Connective tissue disorders	Marfan syndrome Loeys Dietz syndrome Ehlers Danlos syndrome – classical, hypermobile and

		vascular subtypes Homocystinuria Beals syndrome
	Aortic rupture/ dissection	
	Sudden cardiac death	
	Familial cardiomyopathy	Hypertrophic cardiomyopathy Dilated Cardiomyopathy Arrhythmogenic RV
	Familial arrythmia	Brugada syndrome Long QT syndrome
	Infantile cardiomyopathy	Pompes disease Barth syndrome
	Congenital heart disease	
Dysmorphology	Principles of normal and abnormal embryogenesis Approach to the fetus with *multiple congenital abnormalities* Approach to the child with congenital abnormalities Assessment of facial dysmorphism Growth proportions	Malformation, dysplasia, disruption and deformation Syndromes, sequences and associations Use of dysmorphology databases or AI-based adjuncts to assessment
	Overgrowth	Beckwith Wiedemann syndrome Sotos syndrome *PIC3CA* related overgrowth Proteus syndrome
	Hemihypertrophy	
	Obesity	
	Short stature	Albright's hereditary osteodystrophy
	Growth retardation	Russel Silver syndrome Primordial dwarfism
	Skeletal dysplasia	Achondroplasia Thanatophoric dysplasia Osteogenesis imperfecta type 1,2,3 and 4 Campomelic dysplasia Chondrodysplasia punctata Diastrophic dysplasia Common Collagen II disorders Cleidocranial dysostosis Pycnodysostosis Osteopetrosis
	Multiple congenital abnormalities	VACTERL association Oculoauriculovertebral spectrum disorder Moebius sequence OEIS MURCS
	Cleft lip and palate	Pierre Robin sequence Van de Woude syndrome Otopalatodigital syndrome
	Craniosynostosis	FGFR2 related craniosynostosis Saethre-Chotzen syndrome
	Disorders of sexual development Sex reversal	Congenital adrenal hyperplasia X-linked adrenal hypoplasia Kallman syndrome Abnormalities of androgen synthesis Androgen insensitivity syndrome Swyer syndrome Ovotesticular DSD

	Pigmentary abnormalities	Tuberous sclerosis Neurofibromatosis type 1 Incontinentia Pigmenti Hypomelanosis of Ito
	Dermatological disorders	Epidermolysis Bullosa Hypohydrotic ectodermal dysplasia
	Arthrogryposis	Amyoplasia congenita
	Structural limb abnormalities Radial ray abnormalities Ectrodactyly Limb reduction Phocomelia	Poland sequence Holt Oram syndrome Thrombocytopaenia absent radius syndrome Townes Brock syndrome EEC syndrome Adams Oliver syndrome Amniotic band sequence Robert syndrome
Teratogens	Assess clinical relevance of teratogen exposures: Alcohol / recreational drugs Infections Medications Maternal illness	Fetal alcohol spectrum disorders Congenital cytomegalovirus infection Congenital rubella syndrome Warfarin embryopathy Retinoic Acid embryopathy Antiepileptic embryopathy Diabetic embryopathy Maternal phenylketonuria Maternal hypothyroidism
Multisystem disorders		Cystic fibrosis and CFTR related disorders
Haematological disorders	Haemoglobinopathies Bleeding disorders Hypercoagulability	Sickle cell disease alpha and beta thalassaemia G6PD deficiency Haemophilia A and B Von Willebrand disease Hereditary haemorrhagic telangiectasia Factor V Leiden Protein C and S deficiency
Renal disorders		AD and AR Polycystic Kidney Disease Alport syndrome
Hepatic disorders		Haemachromatosis Alpha 1 antitrypsin deficiency Alagille syndrome
Prenatal genetics	Aneuploidy risk assessment	Techniques, and implications
	Techniques, limitations	
	Preimplantation genetic diagnosis	Techniques, requirements
	Antenatal ultrasound abnormalities	Omphalocoele Congenital diaphragmatic hernia Gastroschisis Lower urinary tract obstruction “Soft markers” Echogenic bowel Raised nuchal translucency Hydrops fetalis Cardiac lesions Talipes CNS abnormalities

	Foetal loss/ miscarriage	
	Infertility	
Cancer Genetics	Genetics of common cancers Breast Colon Prostate	BRCA1/2 HNPCC and related conditions Familial adenomatosis polyposis Use of Panels
	Risk assessment in cancer genetics	
	Tumour/ cancer syndromes	Von Hippel Lindau syndrome Li Fraumeni syndrome Multiple endocrine neoplasia type 1 Neurofibromatosis type 2 Fanconi anaemia Wilms tumour PTEN hamartoma syndrome Congenital mismatch repair deficiency Gorlin syndrome Xeroderma pigmentosum
	Somatic evaluation of germline familial cancer risk	
	Principles of gene based targeted therapy	PARP inhibitors
	Genomic Prognostication in cancer	Expression arrays Liquid biopsy
Mitochondrial genetics	Inheritance and expression determinants in mitochondrial disorders Heteroplasmy and genetic counselling	MELAS Lebers hereditary optic neuroretinopathy MERRF NARP Kearne Sayers Syndrome CPEO Alpers Mitochondrial depletion syndrome
	Common presentations of mitochondrial disorders	
Metabolic genetics	Investigating suspected metabolic disease	
	Treatment of metabolic conditions	
	Common presentations of metabolic disease	Hypoglycaemia Hyperammonaemia Lactic acidosis Metabolic acidosis high anion gap
	Common Metabolic disorders (including those with founder effects in South Africa	Urea cycle defects OTC deficiency Organic acidurias Glutaric aciduria Type1 Amino acidaemias Fatty acid oxidation disorders MCAD deficiency Galactosaemia Glycogen storage disorders Lysosomal storage disorders Mucopolysaccharidoses Type 1,2 , 3, 4 and 6 Gaucher disease Peroxisomal disorders Nieman Pick disease Zellweger syndrome Smith Lemli Opitz syndrome

		Menkes disease Wilson disease Congenital disorders of glycosylation Familial hypercholesterolaemia Variegate Porphyria
Immune related conditions	Common single gene conditions affecting the innate and adaptive immune system	Wiscott Aldrich syndrome SCID Bruton's Agammaglobulinemia IPEX Congenital Disorders of Glycosylation
Premature ageing	Conditions causing premature ageing	Cockayne syndrome Hutchinson-Gilford progeria
Principles of therapies	Therapeutic approaches for genetic disease	Therapy approaches in lysosomal storage disorders, SMA, DMD, sickle cell disease and cystic fibrosis, metabolic disorders
Genetic testing and interpretation	Interpreting chromosomal karyotype and chromosomal microarray results	Translocations Supernumerary markers Inversions Reduced penetrance CNVs
	Interpreting molecular genetic results	Common techniques, advantages and disadvantages, and limitations Variant calling Expansion mutations Understanding WES ad WGS advantages and challenges
	Applying ACMG criteria for variant interpretation	Criteria used Limitations and cautions
	Use of common databases to analyse significance of results	Clinvar GnomAD Varsome Decipher OMIM ClinGen
	Selection and interpretation test according to context	Screening, diagnosis, prenatal diagnosis, pre-symptomatic, family and carrier testing
	Differentiate clinical and non-clinical genetic test contexts	Clinical testing vs research vs direct to consumer testing
Non-genetic investigations	Appropriately order and interpret common non-genetic investigations	Identify common abnormalities on: - Imaging (e.g. MRI, CT, sonar, Xray) - Common laboratory investigations
Genomic medicine	Pharmacogenomic principles	Warfarin pharmacogenomics
	Genetic contribution to common complex disorders	Diabetes Hypertension
	Polygenic risk scores	Implementation Pitfalls
	Direct to consumer testing / SNV associations	
Psychiatric genetics	Genetic understanding of schizophrenia and bipolar disorder	
Genetic counselling	Principles Risk assessment Facilitating decision making	Breaking difficult news Estimate and communicate recurrence risk Cascade screening Have a genetic counselling approach suitable to each main context: prenatal, paediatric, cancer, adult, predictive
Public health genetics	Screening for genetic disorders Evaluating incidence of congenital disorders	Newborn screening Population screening Carrier screening

	Structuring genetic services	
ELSI	Ethical principles Ethical dilemmas	Predictive testing Testing in children TOP act and TOP practice
Research	es and principles of methodology earch principles of research ethics	Basis statistics including incidence, prevalence, sensitivity, specificity, positive and negative predictive values Basic principles of quantitative clinical, qualitative, genetic and genomic research relevant to medical genetics Informed consent Broad consent Research in minors and vulnerable populations
Professional development	Foundations for professional development	Be able to critique journal articles in the field Other resources and approaches to track developments in the field

With a field that is developing so rapidly, it is recognised that many sources of study will be online but a short list of texts to consider follows**:**

- Bonthron D, Fitzpatrick D, Porteous M and Trainer A. *Clinical Genetics A Case-based Approach.* London, WB Saunders Company Limited
- Battaglia A, Viskochil D, Carey JC,. Cassidy SB. *Cassidy and Allanson's Management of Genetic syndromes* John Wiley and Son
- Connor M and Ferguson-Smith M. *Essential Medical Genetics.* Oxford, Blackwell Science Ltd
- Firth H and Hurst J *Oxford Desk Reference Clinical Genetics and Genomics* Oxford University Press
- Gorlin RJ, Cohen MM and Hennekam RCM. *Oxford Monographs on Medical Genetics No.42. Syndromes of the Head and Neck.* Oxford University Press
- Jones KL. *Smith's Recognisable Patterns of Human Malformations*. Philadelphia, WB Saunders Company
- Korf BR. *Human Genetics, A problem-based approach.* Boston, Blackwell Science

APPENDIX C

1.0 CERTIFICATE OF PROFICIENCY TO ENTER FCMG PART II EXAMINATION

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

Please complete and initial or sign each item. The completed form must be submitted to the CMSA.

Name of candidate: …….....…….……

Date of completion of Part 1 examination: .....…….……..

Date of HPCSA registration as a registrar in Medical Genetics and relevant training number:

…….....……..

Academic training unit: .....……

Confirmation that all 6 monthly reviews have been completed and found to be satisfactory. If unsatisfactory, please provide details:

.....…….....

.....

Confirmation that the summary of portfolio (Appendix 3) to be submitted is complete and accurate. If incomplete, please provide details including remedial action planned:

.....…….....

.....

Confirmation that the logbook accurately reflects the case exposure and that the required 75% of the final minimum required cases in each category has been met. If not, please provide details and remedial action planned.

.....…….....

.....

Head of training unit:

The above details correctly reflect the assessment of this candidate:

NAME…….

SIGNATURE…….……

DATE:…..…….……

APPENDIX D

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

1.1 Before being admitted to Part II of the examination for the FCMG(SA), the Portfolio summary and required documents as detailed below 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 FCMG(SA) Part II and be sent within 2 weeks to the CMG portfolio examiner.

1.2 The convenor of the FCMG Part II examination will be responsible for notifying the CMSA of the portfolio examiner/s and providing contact details.

1.3 Candidates will be notified when their Portfolio has been accepted. In the event of the Portfolio being rejected, the candidate will be notified what additional work is required.

FCMG portfolio review and assessment:
The overall purpose of the portfolio review is to obtain an independent assessment of a candidate's training and readiness to write the FCMG part 2 examination and as an extension of that to shortly be competent to practice independently as a specialist Medical Geneticist.
The aspects to be assessed are:

1. Review of activities to confirm a broad exposure to activities to achieve a solid academic base for practicing in the field of Medical Genetics is required.
2. Review of clinical exposure to ensure adequate and broad experience of the common genetic disorders and reasons for referral to Medical Genetics services as well as some exposure to rare genetic disorders.
3. Assessment of presentation/s to judge the suitability and content of presentations for teaching purposes.
4. Assessment of written communication that confirms the ability to prepare reports for the referring healthcare practitioner and treatment/care plans.
5. Evidence of ability to develop and carry out a research project

The following documentation is required to be submitted to the CMSA as the portfolio in a **single pdf** document.
Of

- Name, HPCSA registration and training number, academic qualifications and University affiliation.
- A summary of number and type of academic meetings attended (both internal and external) and of lectures and presentations given by the candidate.
- A legible pdf copy of two such academic presentations, journal clubs or lectures and 1 presentation at a national dysmorphology meeting or equivalent. Each submission should include a brief reflective statement of what was learnt.
- A record of attendance at relevant courses and congresses.
- The abstract submitted to at least one national or international congress.
- A summary of the logbook reflecting clinical exposure listing the number of adult, cancer, paediatric and fetal medicine cases seen.
- A copy of the MMed research protocol and ethics approval.
- Confirmation of each 6 monthly assessment as satisfactory or if unsatisfactory the remedial action recommended and signed by the head of training unit. (See appendices D1, D2 and D3)
- A cover letter from the head of the training unit confirming satisfactory progress of the candidate, that all requirements have been met for entry into the examination and that the portfolio has been checked and is complete. If any requirements have not been met these should be specifically addressed in the letter spelling out remedial action. In addition, if the case exposure required for training has not been met, the letter should include the written commitment of the head of the training unit that they will ensure this training requirement is met by the end of their training time.

In addition, an extract from the record of case exposure (logbook) should be submitted documenting cases seen. Each individual may only be reflected once at the first contact with the trainee, and each entry should include the following information:

1. Patient Identifier – ideally hospital numbers should be used but where none are available the DOB and initials may be used (no names to be included)
2. The diagnosis or presenting problem.
3. The site of the consultation
4. Whether it was an initial or follow up consultation with the genetic service
5. The category of clinical case
6. If the consultation was observed or directly or indirectly supervised
7. The name and / or signature of the supervising health care professional

The **minimum** case exposure expected by the completion of training period is reflected in the table below. At least 75% of the final number in each category is required for entry into the FCMG part 2. Exceptions will only be considered in unusual circumstances and will require a written request from the candidate supported by their head of training unit.

Case category **Minimum** total cases reflected. (The balance of the total minimum number of cases may be made up of either directly or indirectly supervised cases)	**Observed/Directly supervised.** (Patient seen in person with the supervising HCP – no more than 30% may be observed cases) **Minimum** cases reflected	**Indirectly supervised.** (Managed independently by trainee with supervision as required after consultation) **Minimum** cases reflected
Paediatric (150 total)	40	90
Adult (50)	15	25
Cancer (50)	15	25
Fetal medicine (50)	15	25
TOTAL (300)	95	175

After submission of the portfolio, the portfolio examiner will request 8 to 10 copies of written communication or a detailed summary regarding cases chosen at random. A minimum of 80% of these should be submitted within 2 weeks of receiving the request, in a single pdf document.

Trainees must keep their own comprehensive portfolio of learning as suggested in the portfolio document on the CMG website. This will reflect more detail than will be required for submission to the CMG portfolio examiner.

APPENDIX D1

ASSESSMENTS REQUIRED FOR MEDICAL GENETIC TRAINEES:

(To be overseen by the head of the training unit)

Every 6 months:

Multi-observer reports using modified CanMed scale by a minimum of 2 observers of which at least one must be a consultant medical geneticist (appendix D2)
1 reflective case report
2 x mini clinical evaluation exercises - 1 counselling and 1 clinical case.
Review of the record of learning including patient case exposure
Self-reflection on progress, strengths and weaknesses including identifying gaps in training.

Annually:

1 formal case-based assessment including at least 2 examiners.

APPENDIX D2

MULTI-OBSERVER REPORT (ADAPTED WITH PERMISSION FROM CANMED)

	EXPECTATIONS				
	Rarely Meets	**Inconsistently meets**	**Generally meets**	**Sometimes exceeds**	**Consistently exceeds**
MEDICAL EXPERT					
Demonstrates the basic scientific and clinical knowledge relevant to medical genetics					
Obtains individual medical and family histories that are appropriate, accurate and well organised					
Performs physical examinations that are appropriate , accurate and well organised					
Recognises, describes and interprets laboratory and imaging findings relevant to genetic disease					
Understands and makes effective use of information technology medical databases					
Integrates pertinent information to arrive at complete and accurate clinical decisions					
Provides continuity of care and periodically assesses the appropriateness of care					
Recognises urgent clinical situations, prioritises correctly and effects prompt and appropriate management					
Overall is proficient in technical and procedure skills. Minimises risks and discomfort to the patient					
Please comment on strengths and weaknesses of the candidate and provide a rationale for your ratings					

	EXPECTATIONS				
	Rarely Meets	**Inconsistently meets**	**Generally meets**	**Sometimes exceeds**	**Consistently exceeds**
COMMUNICATOR					
Establishes a therapeutic relationship with patients and families					
Demonstrates the practice of genetic counselling by communicating well with patients and families, listening effectively, providing clear and thorough explanations of investigations, diagnosis and management and helping the individual and family choose an appropriate course of action.					
Recognises biases and cultural differences and their impact on communication and patient care					
Communicates well with peers and other health professionals. Effectively provides and receives information. Presents cases with accuracy and insight					
Provides accurate, understandable and timely documentation to referring physicians, care givers and families					
Handles conflict.					
Please comment on strengths and weaknesses of the candidate and provide a rationale for your ratings					

	EXPECTATIONS				
	Rarely Meets	**Inconsistently meets**	**Generally meets**	**Sometimes exceeds**	**Consistently exceeds**
COLLABORATOR					
Interacts effectively with other members of all the interdisciplinary health care team and respects their roles and expertise					
Consults and delegates effectively					
Collaborates with research scientists to advance human knowledge					
Please comment on strengths and weaknesses of the candidate and provide a rationale for your ratings					

	EXPECTATIONS				
	Rarely Meets	**Inconsistently meets**	**Generally meets**	**Sometimes exceeds**	**Consistently exceeds**
MANAGER					
Utilises resources effectively to balance patient care, own learning needs and outside activities					
Allocates finite health care resources widely					
Works effectively and efficiently in a health care team. Adheres to policies and procedures,					
Demonstrates and understanding of the principles of practice management relevant to medical genetics					
Demonstrates an understanding of the importance of quality assurance relating to clinical care Appropriate follow up of cases					
Please comment on strengths and weaknesses of the candidate and provide a rationale for your ratings					

	EXPECTATIONS				
	Rarely Meets	**Inconsistently meets**	**Generally meets**	**Sometimes exceeds**	**Consistently exceeds**
HEALTH ADVOCATE					
Recognises and responds appropriately in advocacy situations					
Understands the role of the medical geneticist in intervening on behalf of patients with regard to ethical, social, economic and biologic factors that may impact on health					
Understands the role of the medical geneticist in intervening on behalf of the community with regard to ethical, social, economic and biologic factors that may impact on health					
Understands the importance of participating in public policy discussions and decision making					
Please comment on strengths and weaknesses of the candidate and provide a rationale for your ratings					

APPENDIX D3

REGISTRAR ASSESSMENT SUMMARY

Registrar name:

Year of training:

Assessment period:

Documents submitted by candidate and reviewed	Unsatisfactory	Satisfactory
Summary of cases		
Case report		
Self-reflection of progress		
Summary of record of learning		

Objective assessments	Unsatisfactory	Satisfactory
Clinical mini- CEX		
Counselling mini-CEX		
Annual case based examination		

Multi observer reports	Unsatisfactory	Satisfactory
Observer 1		
Observer 2		
Observer 3 (optional)		
Observer 4 (optional)		
Observer 5 (optional)		

Learning Goals

Remedial action if required

Signed: **Date:**

Name:

Designation:

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

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, moderator 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/oral/practical part of the examination. Examiners should ideally play a minor role in the examination of candidates with whom they have worked closely in the recent past

2.3 In the clinical/oral/practical 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/ORAL/PRACTICAL EXAMINATION (150 marks)

3.1. **The counselling case (30 marks):**

3.1.1 ***Candidates:***

You will be asked to make an assessment of a clinical scenario and provide appropriate, comprehensive genetic counselling in the form of a written typed response, simulated interaction or verbal response. This should include communication of relevant clinical, genetic and recurrence information in a manner that is accessible to the reader. You will have 40 minutes to complete all aspects of this station. The scenario chosen should be that a competent medical geneticist would be expected to be familiar with and 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 counselling case is to ensure that a candidate is able to synthesise the important aspects of the clinical scenario and communicate these in in a clear, understandable and empathetic way.

3.1.2…/

3.2 The short cases (20 marks each):

3.2.1 *Candidates:*

You will be taken through a short series of slides pertaining to clinical cases that you will be required to answer questions on. These may include slides pertaining to history, examination findings, which may include photographs, and investigations, which may include laboratory results, X-rays or imaging. The scope of the case may include all aspects of clinical practice including detection and interpretation of findings, diagnosis and differential diagnosis, and management. Each case is 15 minutes long and you are encouraged to keep your answers short.

3.2.2 ***Examiners:***

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

3.3 The long case (40 marks):

3.3.1 *Candidates:*

This case is 30 minutes long and although similar to the short cases in format, it will require more detailed and comprehensive assessment of the scenario and may include all aspects of medical genetic practice including interpretation of pedigree or molecular genetic information.

NOTE FOR EXAMINERS:

The purpose of the long case is to evaluate the candidates' clinical reasoning and interpretation of additional sources of information and emphasis should be placed on the holistic interpretation of the scenario.

JOHANNESBURG
June 2023