Introduction

Clinical genetics is defined as the science and arts on the practice of diagnosis, management and prevention of genetic disorders. Genetic disorders and inborn errors of metabolism (IEM) are individually rare disorders but collectively, they form an important component of the health burden and a main cause of chronic illnesses in childhood. IEMs occur in out of 2,000 - 3,000 births. Three percent of newborns have major birth defects. With an annual delivery of 600,000 births, about 300 and 20,000 newborns will be affected with IEMs and birth defects respectively in Malaysia every year. In addition, about 200-300 individuals with p-thalassaemia major are diagnosed a year. Many other conditions with strong genetic susceptibility such as cancers, neurodegenerative disorders and psychiatric illnesses collectively known as adult onset genetics has become increasingly prominent. Other multifactorial conditions involve the interplay between genetic and environmental factors such as diabetes, asthma, hypertension and osteoarthritis are now recognised as the main cause of chronic ill health in the general population.

Biotechnology has also improved the diagnostic armamentarium of the clinical geneticists. Treatment for many genetic diseases is available. Genetic counselling is recommended for all patients with genetic diseases and before any genetic testing are done. This is important in situations where presymptomatic and predictive testing is done. Clinical genetics is recognised as a medical specialty in most developed countries in the 1990s. To provide optimal genetic services to this group of population, it was recommended that at least 1-2 clinical geneticists are available per one million population. On the basis of this recommendation, Malaysia requires the service of at least 25-50 clinical geneticists. A formal programme of training and accreditation of clinical geneticist is needed to ensure proper genetic care is provided by competent personnel.

1. Entry Criteria
   i) A recognised postgraduate degree in paediatrics. Ministry of Health candidates may need to be gazetted as a paediatric specialist before application for training can be approved.
   ii) Interest in paediatric genetics and metabolic medicine as shown by involvement in CME and research activities viz presentation at conferences/ seminars, writing and publication of journal articles and conducting studies in medical genetics and metabolic medicine.
   iii) Submission of application form for training
   iv) Approval by the clinical genetics sub-specialty committee
In the event that there are more applicants than training posts, additional criteria for selection will be:
   a) Research output, publications and presentation at scientific meetings
   b) Seniority in service
   c) Having worked in a district hospital after passing his/her postgraduate examinations
   d) Participation in professional bodies relevant to the sub speciality.

2. Duration of Training Programme

Duration of training is 3 years, with at least one year of training overseas in a recognised centre for clinical genetics or metabolic medicine.

3. Training Programme

*Topics and core knowledge to be covered include:*
- Human embryology and fetal physiology
- Basic genetics, molecular and cell biology
- Dysmorphology and birth defects
- Biochemical genetics (Inherited Metabolic Disorders)
- Mendelian and non-mendelian genetic disorders
- Prenatal genetics and teratology
- Adult onset genetics, including cancer genetics
- Genetic counselling for all aspects mentioned and risk calculation
- Basic laboratory understanding & skills in genetic testing
- Bioinformatics and population genetics
- Ethical, legal and social issues
- Epidemiology, Audit, Quality Assurance and Research

*Methods of Training*
Training will be done in the form of service provision (including clinics and call duties), continuing medical education activities e.g. ward rounds, case discussion, case conferences, seminars, journal sessions and teaching and supervision of junior doctors, nurses and other allied staff, and involvement in research. Training is scheduled over the 3-year period during which acquisition of laboratory knowledge and skills must be done concurrently. Teaching and on-call duties will be carried out throughout the 3 years.

\textit{i) Communication and clinical genetics training}
- Able to obtain family history and interpret pedigree accurately
- Able to perform a clinical examination and relevant assessment of the client to arrive at a provisional diagnosis and differential diagnoses
- Able to perform recurrence risk calculation
- Able to communicate with all clients and family regarding genetic concepts, risk, disease burden in the context of a multi-cultural society
• Able to provide genetic counselling in all genetic conditions, encompassing breaking bad news, dealing with uncertainty, counselling for people of all ages in non-directive manner and helping clients make informed choice.
• Able to develop strong interpersonal skills and able to provide grief and bereavement counselling
• Able to identify and provide relevant and evidence-based therapeutic and management strategies.
• Capable of using databases specifically genetic databases on the World Wide Web to gain information about genetic disorders (e.g. OMIM, Gene Test) and commercial dysmorphology databases.
• Able to initiate genetic tests and understand the scientific, ethical, social and legal issues relating to genetic testing. Specific issues include:
  o Genetic tests: how to order them and their interpretation
  o Population screening and interpretation
  o Targeted group screening (e.g. cascade testing)
  o Carrier testing, predictive/presymptomatic testing
  o Prenatal diagnosis, pre-implantation diagnosis
  o Medical ethics
  o Principles of medical law relating to privacy and right to know

Log book: 100 cases per year, with a minimum of 10 cases for each core areas listed in the Log Book.

ii) Performance of procedures / skills to be acquired

For the first 2 year of training, in addition to the usual clinical training, a candidate must concurrently spend at least a 3-month rotation/year in the following laboratories or clinical area:

A. Cytogenetics laboratory.
   • Cell and tissue culture
   • Routine karyotyping
   • Other techniques e.g. fluorescence in situ hybridization (FISH)
   • Log book: total 20 cases (10 observed; 10 self-performed and interpreted)

B. Molecular genetics laboratory
   • DNA extraction
   • Polymerase chain reaction (PCR)
   • Gel electrophoresis
   • Genomic sequencing
   • Mutation screening methods
   • Primer designs and various other techniques in molecular genetics
   • Log book: total 20 PCR reactions (10 observed; 10 self-performed & interpreted)
C. Biochemical genetic laboratory
   • Amino acid analysis Organic acid analysis Acylcarnitine profiling Urine mucopolysaccharides test Enzyme assays Newborn screening laboratory • Log book: total 20 analyses (10 observed; 10 self-performed and interpreted)

D. The fourth 3-month rotation in the same year may be in one of the following disciplines:
   I). Fetal-medicine Unit
      • Prenatal counselling clinic
      • Opportunity to familiarise with fetal ultrasonography, maternal screening and diagnostic procedures
      • Multi-disciplinary approach to management of fetal abnormalities
      • Log book: 10 cases
   II) Skeletal dysplasia Unit / craniomaxillofacial Unit
      • Interpretation of skeletal imaging techniques
      • Multi-disciplinary approach to management of genetic orthopaedic and craniomaxillofacial diseases
      • Log book: 10 cases
   III). Cancer genetics Unit
      • Interpretation of haematological and solid tumors findings
      • Predictive counselling and testing
      • Log book: 10 cases
   IV). Neurogenetics Unit
      • Interpretation of various neurological testing techniques
      • Predictive counseling and testing
      • Log book: 10 cases

   iii) Presentation at conferences and publications
      • Attendance and presentation (oral or poster) at conferences (minimum once a year)
      • Publication of peer-reviewed articles in journal (minimum once a year)
      • Case report or case review, 2 per year, each 2000 words with 10 references. Case reports may be published in peer-reviewed journals.

   iv) Attendance at professional meetings & courses (certification)
      • Journal clubs and structured laboratory meetings (monthly)
      • Genetic workshops and genetic counselling courses (once a year)
At the end of the training, the candidate must have the knowledge, skills and attitude to be able to perform the following functions independently:

1) Dysmorphology diagnosis
2) Assessment of recurrence risk
3) Determination of burden of care of the conditions
4) Initiate appropriate investigations of genetic and metabolic disorders
5) Diagnosis and management of all birth defects, genetic disorders and IEMs
6) Stabilisation of acute crises and unexpected problems
7) Perform genetic counselling appropriately
8) Perform clinical procedures e.g skin biopsy, hair analysis
9) Liaison with genetic and metabolic laboratories
10) Patient and community education
11) Data collection, participation in registry and evaluation
12) Research and outcome surveillance
13) Involvement in graduate, postgraduate and allied health personnel education
14) Administration of a genetics & metabolic services
15) Involvement in publication of articles in clinical genetics
16) Involvement in lay and professional bodies in clinical genetics

3. Review and Evaluation of Progress of Training

i) Approval of course of training for individual candidates prior commencement of training

ii) A log-book on training and professional development as provided by the board which will include information on hospital census, credentialing of procedures performed, service provided at clinics, out-of-office hours call duties, CME and teaching responsibilities, research and publications.

v) Quarterly assessment and yearly documentation of candidate's performance by the supervisors in the format provided by the subspecialist committee. Supervisor's rating of performance and attitudes will also be incorporated in the above document.

vi) An exit interview conducted by the Clinical Genetics Sub-Specialty Committee when deemed necessary.

4. Training Centre and Trainers/Supervisors

- All hospitals with a genetic & metabolic service (See appendix A) managing more than 100 new cases and 200 follow-up cases a year, respectively can be accredited as training centres at least for part of the training. Centres managing less than the above stipulated cases may be considered as training centres on a case-by-case basis by the Clinical Genetics Sub-Specialty Committee.
• Training must be done under the supervision of an accredited clinical geneticist who has been appointed as a trainer by the clinical genetics sub-specialty committee.
• The ratio of trainer to trainees is suggested to be not more than two (2). However, this will be determined by service conditions and workload of the unit.
• No trainee will spend more than two years under the supervision of one specific trainer.

5. Clinical Genetics Sub-specialty Committee.

This Committee shall consist of the following members who will decide on the training program, the criteria for accreditation, and suitability of candidates to be accredited as paediatric geneticists and metabolic medicine specialists. The Committee shall consist of representation of clinical geneticists from Ministry of Health Malaysia, Universities and relevant medical genetics professional bodies.

Membership to the Committee will be by appointment of the Chairperson of the National Credentialling Committee upon the recommendation of the College of Paediatrics of the Academy of Medicine of Malaysia. The recommended term of office is 2 years, at the end of which committee members are eligible for re-appointment.
6. **Criteria for Accreditation of Clinical Geneticists in Malaysia**

1. Any doctor can request to be registered if he/she fulfils **ALL** the following requirements:

   **A) A recognised basic medical degree**
   A basic medical degree recognized by the Malaysian Medical Council

   **B) A recognised postgraduate qualification**
   One of the following paediatric postgraduate degrees recognized by the Malaysian Paediatric Specialty Board:
   - Master of Paediatrics awarded by Universiti Malaya, M.Med (Paed) awarded by Universiti Kebangsaan Malaysia or Universiti Sains Malaysia
   - MRCP (UK) up to year 2000
   - MRCPCH by Royal College of Child Health UK
   - MRCPI (Ireland)
   - FRACP
   - M.Med in Paediatrics (Singapore)
   - Any other equivalent paediatric or specialty postgraduate degrees recognised by the Malaysian Paediatric Specialty Board on a case by case basis

2. Completed postgraduate training in Clinical Genetics in recognised training centres

   - Completion of a minimum of 3 years of clinical genetics training in centres which fulfilled the criteria stipulated by the Malaysian Clinical Genetics Subspecialty Committee (refer to Appendix A), under the supervision of clinical genetics trainers who fulfilled the criteria stipulated by the Malaysian Clinical Genetics Sub-specialty Committee.

   - This period of training does not include the time the applicant spent during his/her housemanship nor the period when undergoing training for the basic paediatric postgraduate degrees

   - The candidate must furnish evidence of satisfactory clinical genetics subspecialty training such as:
     - Log book of core procedures and patients or clients seen
     - Portfolio with supporting document where relevant, e.g. a valid certificate of completion of training in a genetics theory course, published research papers or abstracts, certificates of attendance at conferences, courses or workshops
     - Satisfactory supervisors' reports on Clinical Genetics Clinical Core Competency

The above criteria will be reviewed from time to time.
Appendix A
Checklist and criteria for accreditation of centre for Clinical Genetics

Name of Candidate

A. Clinical Genetics work load and services

1. Number of new cases consultations a year
   (Minimum 100 per year) _____ peryr

2. Total number of follow-up cases seen in genetics clinic per year
   (Minimum 200 per year) _____ peryr

3. Provides care and laboratory support services
   Yes [ ] No [ ]

4. Provides follow-up care
   Yes [ ] No [ ]

B. Training Facilities for Clinical Genetics

1. Total number of accredited clinical geneticists in the hospital (At least one)

2. Meeting room with audio-visual aids
   Yes [ ] No [ ]

3. A Medical Library on site
   Yes [ ] No [ ]

4. Access to Medline and literature search
   Yes [ ] No [ ]

C. Educational activities in clinical genetics
   (Please furnish a copy of weekly or monthly teaching activities)

1. Number of teaching ward round with clinical geneticist /week
   (Minimum 1 per week) ___________________

2. Number of hours/week of rostered clinical genetics education
   (Minimum 1 hour per week)

3. Applicant was able to attend at least 70% of the educational opportunities
   Yes [ ] No [ ]

4. Number of conference and workshops attended per year
   (Minimum 1 per/year)

AND

D. Clinical Cases (see Log Book)

E. Laboratory activities in clinical genetics
   (Please furnish details in Log Book)
Appendix B
Report on Level of Achievement of Clinical Genetics Key Competencies by Supervisors/Referees

(Each supervisor/referee must submit a separate report)

Candidate's Name: ________________________________

Candidate's Identification Number or Passport number: ________________________

Period of training: from _____________ to ________________

Name of supervisor/referees: ________________________________

Report

I hereby certified that the above information is true and accurate.

Signature of supervisor: ________________________________

Date of Report: _____________________________

Prepared by:
Members in Clinical Genetics Sub-specialty Committee:

1) Professor Dr Thong Meow Keong (University of Malaya)
2) Dr Keng Wee Teik (Paediatric Institute Kuala Lumpur / Hospital Pulau Pinang)
3) Assoc Professor Dr Zarina Abdul Latif (UKM)
4) Assoc Professor Dr Zilfalil Alwi (USM)
5) Dr Rowani Mohd Rawi (USM)
6) Dr Ngu Lock Hock (Paediatric Institute Kuala Lumpur)
7) Dr Choy Yew Sing (Private practice)
**SUMMARY OF POSTS/CLINICAL ATTACHMENTS**
**SINCE ENTERING TRAINING PROGRAMME**

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<th>Hospital</th>
<th>Designated specialty</th>
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**Name of trainee**
### PRESENTATIONS (JOURNAL & DYSMORPHOLOGY CLUBS / NATIONAL / INTERNATIONAL MEETINGS)

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<th>Date</th>
<th>Authors(s)</th>
<th>Title</th>
<th>Conference/meeting/oral/poster</th>
<th>Abstract reference (if any)</th>
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Name of trainee

Signature of trainee
## RESEARCH AND PUBLICATIONS

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<th>Date(s)</th>
<th>Project / Paper title</th>
<th>Supervisor</th>
<th>Grant support</th>
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Name of trainee

Signature of trainee
# ADDITIONAL ACTIVITIES DURING HIGHER MEDICAL TRAINING

*Cumulative record entered serially THROUGHOUT course of training*

## COURSES / CONFERENCES ATTENDED

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<th>Subject</th>
<th>Degree/diploma/certificate obtained (if any)</th>
<th>Evaluation of course</th>
</tr>
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Name of trainee: ____________________________

Signature of trainee: ____________________________
CLINICAL GENETICS - DATABASE & COMPUTING EXPERIENCE

Date

Give details of experience of computer databases in relation to clinical genetics

I confirm that the experience above has been demonstrated and that a satisfactory level of competence has been demonstrated.

Educational supervisor's signature

Trainee's signature

YEAR OF TRAINING (please circle) ONE / TWO / THREE
CLINICAL GENETICS: CLINICAL EXPERIENCE

_up to 10 consultations may be recorded in any year - please note date and case/family record number_

**CHROMOSOMAL DISORDERS**  

\[NB F=fetus/stillbirth & L=livingJ\]

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I confirm that the experience _above_ has been acquired & that a satisfactory level of competence has been demonstrated.  

Educational supervisor's signature & date

Trainee's signature: _lie_  

YEAR OF TRAINING (please circle) ONE / TWO / THREE
CLINICAL GENETICS: CLINICAL EXPERIENCE

up to 10 consultations may be recorded in any year - please note date and case/family record number

CANCER GENETICS

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I confirm that the experience above has been acquired & that a satisfactory level of competence has been demonstrated. I Educational supervisor’s signature & date

Trainee’s signature: [signature] Year of training (please circle) One / Two / Three
CLINICAL GENETICS: CLINICAL EXPERIENCE

up to 10 consultations may be recorded in any year - please note date and case/family record number

CRANIOMAXILLOFACIAL UNIT

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I confirm that the experience above has been acquired & that a satisfactory level of competence has been demonstrated. 

Educational supervisor's signature & date

Trainee's signature: 

YEAR OF TRAINING (please circle) ONE / TWO / THREE
**CLINICAL GENETICS: CLINICAL EXPERIENCE**

*up to 10 consultations may be recorded in any year - please note date and case/family record number*

**DYSMORPHOLOGY (MULTIPLE CONGENITAL ANOMALIES - SYNDROMES NAME IF KNOWN)** Please note whether diagnosis (D) or counselling (C) or both (D/C) [NB F=fetus/stillbirth & L=living]

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I confirm that the experience *above* has been acquired & that a satisfactory level of competence has been demonstrated. I

**Educational supervisor’s signature & date**

**Trainee’s signature**

**YEAR OF TRAINING (please circle) ONE / TWO / THREE**
CLINICAL GENETICS: CLINICAL EXPERIENCE
up to 10 consultations may be recorded in any year - please note date and case/family record number

INBORN ERRORS OF METABOLISM

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I confirm that the experience above has been acquired & that a satisfactory level of competence has been demonstrated. [ ] Educational supervisor's signature & date

Trainee's signature [ ] YEAR OF TRAINING (please circle) ONE / TWO / THREE
CLINICAL GENETICS: CLINICAL EXPERIENCE
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GENERAL GENETIC COUNSELLING/COMMON CONDITIONS WITH GENETIC COMPONENT

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<td>Asthma</td>
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<td>Epilepsy</td>
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<td>Psychiatric illness</td>
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<td>Pregnancy loss</td>
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I confirm that the experience above has been acquired & that a satisfactory level of competence has been demonstrated. Educational supervisor’s signature & date

Trainee’s signature: [Signature]  YEAR OF TRAINING *(please circle)* ONE / TWO / THREE
### CLINICAL GENETICS: CLINICAL EXPERIENCE
*up to 10 consultations may be recorded in any year - please note date and case/family record number*

#### NEUROLOGICAL/NEUROMUSCULAR/NEUROPSYCHIATRIC

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<th>DISORDER</th>
<th>CONSULT 1</th>
<th>CONSULT 2</th>
<th>CONSULT 3</th>
<th>CONSULT 4</th>
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<td>Spinocerebellar ataxia</td>
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<tr>
<td>Mitochondrial abnormalities</td>
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</table>

Other *(please specify)*

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Trainee's signature: ___________  YEAR OF TRAINING (please circle) ONE / TWO / THREE
CLINICAL GENETICS: CLINICAL EXPERIENCE
up to 10 consultations may be recorded in any year - please note date and case/family record number

DYSMORPHOLOGY (SINGLE CONGENITAL ANOMALIES)

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<td>Abdominal wall defects</td>
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<td>Other <em>(please specify)</em></td>
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Trainee's signature __________________________ |
YEAR OF TRAINING *(please circle)* ONE / TWO / THREE
COMMON SINGLE GENE DISORDERS

<table>
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<td>Visual loss</td>
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<td>Deafness</td>
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<td>Haemophilia</td>
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<td>Thalassaemia</td>
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<tr>
<td>Other haematological</td>
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<tr>
<td>Other connective tissue</td>
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</tbody>
</table>

I confirm that the experience described above has been acquired and that a satisfactory level of competence has been demonstrated. 

Educational supervisor’s signature & date

Trainee’s signature

YEAR OF TRAINING (please circle) ONE / TWO / THREE
<table>
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<th>Date(s)</th>
<th>Details</th>
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Educational supervisor's signature

Trainee's signature

| YEAR OF TRAINING (please circle) ONE / TWO / THREE |
CLINICAL GENETICS: CLINICAL EXPERIENCE

*up to 10 consultations may be recorded in any year - please note date and case/family record number*

Prenatal and Teratology Counselling

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<thead>
<tr>
<th>Condition</th>
<th>Record # &amp; date</th>
<th>Condition</th>
<th>Record # &amp; date</th>
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</table>

I confirm that the experience *above* has been acquired & that a satisfactory level of competence has been demonstrated. | Educational supervisor's signature & date

Trainee's signature | [ ] YEAR OF TRAINING (*please circle*) ONE / TWO / THREE
I confirm that the experience above has been acquired & that a satisfactory level of competence has been demonstrated. Educational supervisor's signature & date

Trainee's signature: [Signature]

YEAR OF TRAINING (please circle) ONE / TWO / THREE
<table>
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<tr>
<th>FROM</th>
<th>TO</th>
<th>Type of experience</th>
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Educational supervisor's signature

Trainee's signature

YEAR OF TRAINING (please circle) ONE / TWO / THREE
CLINICAL GENETICS - LABORATORY EXPERIENCE
BIOCHEMICAL GENETICS

FROM    TO    Type of experience

I confirm that the experience above has been demonstrated and that a satisfactory level of competence has been demonstrated.
Educational supervisor's signature

Trainee's signature

YEAR OF TRAINING (please circle) ONE / TWO / THREE
<table>
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<tr>
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<th>Type of experience</th>
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Educational supervisor's signature

Trainee's signature

YEAR OF TRAINING *(please circle)* ONE / TWO / THREE
## PRACTICAL EXPERIENCE IN COUNSELLING SITUATIONS

<table>
<thead>
<tr>
<th>Situation</th>
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<tbody>
<tr>
<td>Breaking bad news:</td>
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<td>Predictive test counselling:</td>
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<tr>
<td>Communicating complex risks:</td>
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<tr>
<td>Communicating uncertain risks:</td>
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<tr>
<td>Bereavement issues (post TOP, NND etc):</td>
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<tr>
<td>Counselling in context of family conflict eg paternity queries, confidentiality etc:</td>
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<tr>
<td>Other counselling situations:</td>
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I confirm that the experience above has been acquired and that a satisfactory level of competence has been demonstrated.

Educational supervisor's signature

Name of trainee

IIYear of training (please circle) ONE / TWO / THREE