MALTA COLLEGE OF PATHOLOGISTS

GENETICS TRAINING PROGRAMME 2013

(Amended September 2019)

Contents

1 Definition

Genetics (from this point onwards the term Genetics will refer to Clinical/Medical Genetics) is that branch of medicine concerned with the effect of hereditary variation on human development and health and also with the study, diagnosis, management, and prevention of disease in individuals, families and communities.

2 General objectives

The practice of Genetics is based on an in depth knowledge of basic genetic principles, a broad range of knowledge of genetic disease as it affects all body systems and individuals of all ages, including complex disease in which risk is due to genetic causes, and a clear understanding of the principles of genetic counselling. Genetics specialists are expected to work within a multidisciplinary team, in close collaboration with laboratory scientists, genetic counsellors, clinical co-workers and academic colleagues.

The specialty of Genetics is constantly changing and the Geneticist must take account of new knowledge and molecular developments and alter clinical practice accordingly. S/He will be an information resource for other medical specialists. Clinical Geneticists will need a wide range of clinical skills as genetic disorders can affect people of all ages and involve all body systems. Communication skills are particularly important in explaining complex concepts and genetic test results to families enabling them to make informed decisions and choose an appropriate course of action.

The main purpose of this training programme is to provide medical specialist trainees in the field of genetics in Malta within a framework that on successful completion results in the award of the Certificate of Completion of Specialist Training (CCST) in Medical Genetics.

After attainment of the CCST, the medical genetics specialist will have the competencies required to be able to work as a consultant within the Malta National Health Service. Post CCST, a medical geneticist will be in a position to develop further a sub-specialist interest within medical genetics.

3 Training Pathway

Specialty training in Medical Genetics consists of two distinct phases: Basic Specialty Training (BST) and Higher Speciality Training (HST). BST (General Medicine, or Paediatrics) training provides physicians with the ability to investigate, treat and diagnose patients with acute and chronic medical symptoms; and with high quality review skills for managing inpatients and outpatients. Higher speciality training (HST) then builds on the core acute care skills to develop the specific competencies required to practise independently as a Clinical/Medical Genetics specialist.

Core training may be completed in BST General Medicine or General Paediatrics. This training builds upon the competencies acquired during the Foundation Years 1 and 2.

The full curriculum for Basic specialty training in Genetics therefore consists of either (minimum of 24 months):

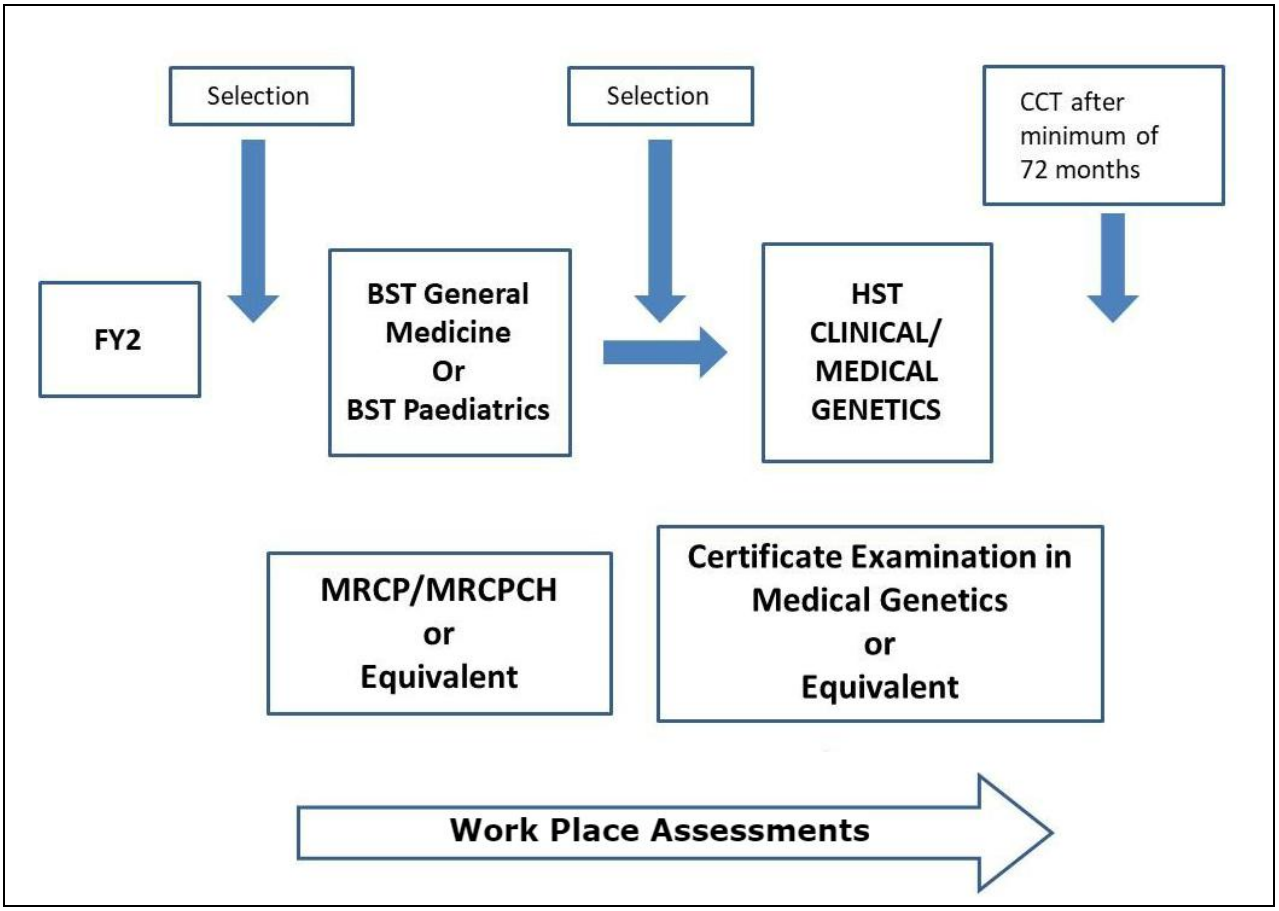
- The Framework for General Professional Training (FGPT) for adult medical specialities (https://deputyprimeminister.gov.mt/en/regcounc/msac/Documents/adult_medical_specialities.pdf)

Or

- The Framework of Competencies for Level 1 Training in Paediatrics (FCL1TPaed) 2008 (https://deputyprimeminister.gov.mt/en/regcounc/msac/Documents/paediatricstrainingprog_.pdf)

And:

Specialty training curriculum for Genetics at Higher Specialist Training level (minimum of 48 months).



BST medical training programmes are designed to deliver core competencies as part of specialty training by acquisition of knowledge, skills and behaviours as assessed by the workplace-based assessments and the MRCP(UK), or equivalent. Programmes are usually for

two years and are broad-based consisting of four to six placements in medical specialties. These placements over the two years must include direct involvement in the acute medical take. Trainees are asked to document their record of workplace-based assessments in an electronic or paper Portfolio (whichever is in place at that moment) which will then be continued to document assessments in specialty training. Trainees completing BST training will have a solid platform of common knowledge and skills from which to continue into HST Training at HST1, where these skills will be further developed and combined with specialty knowledge and skills in order to award the trainee with a certificate of completion of training (CCST).

There are common competencies that should be acquired by all physicians during their training period starting within the undergraduate career and developed throughout the postgraduate career, for example communication, examination and history taking skills. These are initially defined for BST and then developed further in the specialty. This curriculum supports the spiral nature of learning that underpins a trainee's continual development. It recognises that for many of the competences outlined there is a maturation process whereby practitioners become more adept and skilled as their career and experience progresses. It is intended that doctors should recognise that the acquisition of basic competences is often followed by an increasing sophistication and complexity of that competence throughout their career. This is reflected by increasing expertise in their chosen career pathway.

Completion of BST (Medicine or Paediatrics) and acquisition of full MRCP (UK), or MRCPCH or an equivalent examination is a requirement before entry into Specialty training at HST1.

3.1 Duration of Training

Although this curriculum is competency based, the duration of training must meet the European minimum of 3 years for full time specialty training adjusted accordingly for flexible training (EU directive 2005/36/EC). The Malta College of Pathologists has advised that the duration of Genetics specialist training will be 4 years from HST1. Any extensions beyond this 4 year period need to be applied for prospectively, by writing to the Postgraduate Training Coordinator in Pathology who will bring it up at Pathology Training Committee and also the Malta College of Pathologists' Council.

3.2 Higher Genetics Training

Entry Requirements

- Registerable degree in medicine and surgery from the University of Malta or equivalent recognised in the EU
- Full registration with the Medical Council of Malta
- Certificate of Completion of Basic Specialist Training **(Medicine or Paediatrics) and acquisition of full MRCP (UK), or MRCPCH or equivalent examination.**

- **Good command of the Maltese and English language**

Training programme HST

- 12 months (minimum) of approved attachments in genetic laboratory fields, which must include:
    - 3 months (minimum) in a recognised cytogenetics laboratory including molecular cytogenetics techniques
    - 6 months (minimum) in a recognised molecular genetics laboratory.
    - 3 months (minimum) in a recognised biochemical genetics laboratory

- 36 months (minimum) in a Clinical/Medical Genetics unit. Up to 6 months credit may be given for a taught MSc in Clinical Genetics if approval is given in advance. Trainees have to undertake part of their clinical genetic specialty training overseas (minimum of 1 year) at a Centre that is a recognised Genetics Centre with clinical and educational supervision provided, have a personal training programme agreed by the SAC in advance and an agreement from the PGTCPath for training. The latter will then send a recommendation to the Malta College of Pathology who will then send a final recommendation to the SAC. If the SAC recommendation is negative and the trainee still wants to pursue their initiative, the Trainee might have to forfeit his/her training post. This does not affect the Trainee's rights as a doctor within the Malta NHS. A minimum of 2 year clinical training at HST level must be undertaken in the base Centre in Malta. Trainees are expected to take part in all the activities of the units including, attendance and presentation of an assignment based on the annual webcast course in medical genetics; clinical photography; foetal dissections and perinatal autopsies; attendance and presentation of work at genetics meetings/conferences, and training on the various genetic databases used internationally such as ENSEMBL, OMIM, HGMF, Clinvar, LDDB, how to implement them in clinical work and on-call and other out-of-hours commitments.

3.3 Less Than Full Time Training (LTFT)

Trainees who are unable to work full-time are entitled to opt for less than full time training programmes. EC Directive 2005/36/EC requires that:

- LTFT shall meet the same requirements as full-time training, from which it will differ only in the possibility of limiting participation in medical activities.
- The competent authorities shall ensure that the competencies achieved and the quality of part-time training, are not less than those of full-time trainees.

The above provisions must be adhered to. LTFT trainees should undertake a pro rata share of the in- and out-of-hours duties required of their full-time colleagues in the same programme and at the equivalent stage.

EC Directive 2005/36/EC states that there is no longer a minimum time requirement on training for LTFT trainees. In the past, less than full time trainees were required to work a minimum of 50% of full time. With competence-based training, in order to retain competence, in addition to acquiring new skills, less than full time trainees would still normally be expected to work a minimum of 50% of full time. If the trainee is returning or converting to training at less than full time, he/she is required to discuss this with the Postgraduate Training Committee for Pathology (PGTCPath) via the Postgraduate Training Co-ordinators. Permission to enter LTFT shall be assessed and when appropriate, granted by the SAC upon positive recommendation by the PGTCPath and the Malta College of Pathologists

These posts are not supernumerary. Ideally therefore 2 LTFT trainees should share one post to provide appropriate service cover.

Flexible/less than full time trainees should assume that their clinical training will be of a duration pro-rata with the time indicated/recommended, and this should be reviewed during annual appraisal by the PGTCPath.

Trainees entering the Genetics HST programme who later wish to take time off:

1. This could be to pursue a higher research degree eg PhD. In this case the trainee should prospectively apply to the PGTCPath for consideration of a maximum of one (1) year to be counted as part of the HST training. A recommendation is sent by the PGTCPath to the Malta College of Pathologists. The latter will then decide what recommendation to send to the SAC whose decision is final.

   If the Trainee does not wish this period to be counted towards his/her training, he/she still needs to apply for an Out- of-Programme Educational activity to the PGTCPath who will then consider the application. These applications should be sent to PGTCPath at least 6 months prior to leaving the programme.

   If the above rules are not abided to, the Trainee might have to forfeit his/her training post. This does not affect the Trainee's rights as a doctor within the Malta NHS

2. If the trainee wishes to do an Out-of-Programme Experience outside the scope of his/her training, he/she has to apply for consideration to the PGTCPath. The latter will then send a recommendation to the Malta College of Pathologists who will then send a final recommendation to the SAC. If the SAC recommendation is negative and the trainee still wants to pursue their initiative, the Trainee might have to forfeit his/her training post. This does not affect the Trainee's rights as a doctor within the Malta NHS.

3.4 Completion of Training

On completion of the specialist programme, the trainee physician will be competent to function as a specialist in Genetics. At the final assessment, the trainee must demonstrate the knowledge, skills and attitudes relating to gender, culture and ethnicity pertinent to Genetics. In addition, all trainees must demonstrate an ability to incorporate gender, cultural and ethnic perspectives in research methodology, data presentation and analysis. This requires the physician to:

- Perform a complete evaluation of physiological and pathological states relevant to the specific expertise of human Genetics, including, teratology, chromosomal abnormalities, disorders of morphogenesis, inborn errors of metabolism, and monogenic and complex genetic disorders;
- Provide scientifically based, comprehensive and effective diagnosis and management for patients;
- Provide effective genetic counselling for patients and their families;
- Advise the public and health care colleagues on public health aspects of genetics;
- Communicate effectively with medical colleagues, including referring physicians, and other health care professionals;
- Maintain complete and accurate medical records;
- Effectively coordinate or participate in the work of the health care team;
- Be an effective teacher of other physicians (including medical students and specialist trainees), other health care personnel, patients, and general public;
- Demonstrate personal and professional attitudes consistent with the role of Resident specialist and consultant physician;
- Understand the principles and application of practice management and continuing management improvement;
- Be willing and able to appraise accurately his or her own professional performance;
- Be willing and able to keep his or her practice current through various modes of continuing medical education;
- Be able to critically assess the clinical / medical literature;
- Be able to participate in clinical or basic science studies as a leader or member of a research team;
- Understand and apply legal, ethical, and psychosocial dimensions of Genetics; and demonstrate the knowledge, skills and attitudes relating to gender, culture and ethnicity pertinent to Genetics.

The detailed objectives describe minimal standards and in no way exclude the necessity for mastery of additional knowledge, skills or attitudes necessary for the most effective management of patients with genetic disorders.

It is understood that medical trainees successfully completing the Genetics training programme are regarded as fully competent specialists in the clinic with a comprehensive understanding of the work and service provision of the diagnostic genetics laboratories.

3.5 Annual Review

The trainee is to keep a record of training and experience, which must be endorsed annually by the PGTCPath. Trainees can only progress to the next year of the Training Programme

following a successful assessment.

An annual review shall be carried out by a Board nominated by the Pathology Postgraduate Training Committee, composed of three members, including the Chairperson of Pathology (or his/her representative), a Malta College of pathologists representative and a Pathology Postgraduate Training Coordinator, to identify any problems encountered by the trainee and any deficiencies of the training programme.

4 Assessment Process

4.1 *The Assessment System*

The purpose of the assessment system is to:

- Enhance learning by providing formative assessment, enabling trainees to receive immediate feedback, measure their own performance and identify areas for development;
- Drive learning and enhance the training process by making it clear what is required of trainees and motivating them to ensure they receive suitable training and experience;
- Provide robust, summative evidence that trainees are meeting the curriculum standards during the training programme;
- Ensure trainees are acquiring competencies within the domains of Good Medical Practice;
- Assess trainees' actual performance in the workplace;
- Ensure that trainees possess the essential underlying knowledge required for their specialty;
- Inform the Annual Review of Competence Progression (ARCP), identifying any requirements for targeted or additional training where necessary and facilitating decisions regarding progression through the training programme;
- Identify trainees who should be advised to consider changes of career direction.

The integrated assessment system comprises a range of workplace-based assessments and knowledge–based assessments. Individual assessment methods are described in more detail below.

Workplace-based assessments will take place throughout the training programme to allow trainees to continually gather evidence of learning and to provide trainees with formative feedback. They are not individually summative but overall outcomes from a number of such assessments provide evidence for summative decision making. The number and range of these will ensure a reliable assessment of the training relevant to their stage of training and achieve coverage of the curriculum.

4.2 Assessment Blueprint

In the syllabus tables (section 9) the "Assessment Methods" shown are those that are appropriate as possible methods that could be used to assess each competency. It is not expected that all competencies will be assessed and that where they are assessed not every method will be used.

4.3 Certification of Specialist Training

Once the trainee has satisfactorily completed the Training Programme outlined above, and completion of the European Diploma in Medical Genetics and Genomics (EDMGG) or an equivalent qualification as deemed so by the Malta College of Pathologists, he/she will be entitled to obtain the Certificate of Completion of Specialist Training (CCST).

The CCST will be awarded by the Specialist Accreditation Committee, on the recommendation of the PGTCPath and provided the College Council is satisfied of the trainee's proficiency in Medical Genetics.

On fulfilling all the conditions of completion of Higher Genetics Training, the candidate shall inform the Post Graduate Training coordinator in writing and must submit all the necessary documentation in support of the claim, as determined by the Training Programme set by the Malta College of Pathologists. The Pathology Post Graduate Training Committee shall examine and verify the validity of the professional qualifications, work experience and clinical expertise attained by the candidate, and will interview the candidate. The PPGTC shall subsequently send a written recommendation to the College Council. If this recommendation is endorsed by the College council, a positive recommendation will be sent to the Specialist Accreditation Committee, for inclusion of the candidate in the Specialist Register in Medical Genetics.

4.4 Assessment Methods

The methods used to assess progress through training must be valid and reliable. The Curriculum has been re-written, describing the levels of competence which can be recognised. In the tables below, the "Assessment Methods" shown are those that are appropriate as possible methods that could be used to assess each competency. It is not expected that all competencies will be assessed and that where they are assessed not every method will be used. See section 5 for more details.

The assessment grade will be awarded on the basis of direct observation in the workplace by clinical supervisors. Time should be set aside for appraisal following the assessment e.g. of clinical presentations, case management, observation of procedures. As progress is being made, the lower levels of competence will be replaced progressively by those that are higher. Where the grade for an item is judged to be deficient for the stage of training, the assessment should be supported by a detailed note which can later be referred to at the Annual Appraisal Meeting. These methods are described briefly below. More information about these methods including guidance for trainees and assessors is available at the Malta Foundation Programme website and on the JRCPTB website (www.jrcptb.org.uk). Workplace-based assessments should be recorded in the trainee's Portfolio. The workplace-based assessment methods

include feedback opportunities as an integral part of the assessment process; this is explained in the guidance notes provided for the techniques.

They are offered as a means of providing the trainee with attested evidence of achievement in certain areas of the Curriculum e.g. competence in procedural skills, or in generic components. Assessment will also be supported by the trainee's portfolio of achievements and performance at relevant meetings, presentations, audit, in tests of knowledge, attendance at courses and educational events

4.5 Decisions on Progress (ARCP)

The Annual Review of Competence Progression (ARCP) is the formal method by which a trainee's progression through her/his training programme is monitored and recorded. ARCP is not an assessment – it is the review of evidence of training and assessment. The ARCP process is described in *A Reference Guide for Postgraduate Specialty Training in the UK* (the "Gold Guide" – available from www.mmc.nhs.uk).

The PGTCPath is responsible for organising and conducting ARCPs. The evidence to be reviewed by ARCP panels should be collected in the trainee's Portfolio.

The ARCP Decision Aid is included in section 4.6, giving details of the evidence required of trainees for submission to the ARCP panels.

4.6 *Penultimate Year Assessment (PYA)*

The penultimate ARCP prior to the anticipated CCST date will include an external assessor from outside the training programme. The Malta Postgraduate Medical Training Centre (MPMTC) and the PGTCPath will coordinate the appointment of this assessor. This is known as "PYA". The PYA (like the ARCP) will include a face to face component and is intended to be a supportive process for the trainee, where the trainee's progress to date will be reviewed against the curriculum. It will usually be held 12-18 months prior to the expected training completion date. .

4.7 *ARCP Decision Aid: Genetics*

Clinical Genetics ARCP Decision Aid

The table that follows includes a column for each training year which documents the targets that have to be achieved for a satisfactory ARCP outcome at the end of the training year.

Year of Training	HST year 1	HST year 2	HST year 3 (End HST3 = PYA)	HST year 4 (End of HST4 = CCST)
Expected competence	Trainees should be competent in the initial assessment of patients presenting with a common genetic disorder. They should be competent in putting forward a basic plan for genetic investigations and clinical management.	Trainees should be competent in the assessment of patients presenting with the majority of common genetic conditions. Trainees should be competent in their approach to the assessment of patients with some rare genetic disorders.	Trainees should be autonomously competent in the assessment and management of patients presenting with common genetic disorders. Trainees should be competent in the assessment and management of genetic disorders presenting acutely (for example in pregnancy)	Trainees should be autonomously competent in the assessment and management of patients presenting with genetic conditions.
Assessments: European Diploma in Medical Genetics and Genomics (or equivalent)			Attempt/pass EDMGG or equivalent at end of year	Passed EDMGG or equivalent in order to obtain CCST
MSF	Satisfactory		Satisfactory	
Patient Survey		Satisfactory		Satisfactory

mini-CEX*	4 mini-CEX with emphasis on recording family tree, clinical history or clinical examination of patients with genetic conditions. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the mini-CEX)	4 mini-CEX with emphasis on the assessment, management and genetic counselling of patients with genetic conditions (the Educational Supervisor should choose the topic to be assessed for at least 1 of the mini-CEX)	4 mini-CEX with emphasis on the assessment, management and genetic counselling of patients with genetic conditions including those with more complex pedigrees or genetic disorders (the Educational Supervisor should choose the topic to be assessed for at least 1 of the mini-CEX)	4 mini-CEX on the assessment, management and genetic counselling of patients with genetic conditions with the emphasis on complex disorders. (the Educational Supervisor should choose the topic to be assessed for at least 1 of the mini-CEX)
CBD*	4 CBD with emphasis on family tree, clinical history or clinical findings in patients with genetic conditions (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD with emphasis on the assessment, management and genetic counselling of patients with genetic conditions (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD with emphasis on the assessment, management and genetic counselling of patients with genetic conditions including those with more complex pedigrees or genetic disorders (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)	4 CBD on the assessment, management and genetic counselling of patients with genetic conditions with the emphasis on complex disorders (the Educational Supervisor should choose the topic to be assessed for at least 1 of the CbD)
Adult Life Support and Paediatric Life Support	Must have valid ALS and/or PLS	Must have valid ALS and/or PLS	Must have valid ALS and/or PLS	Must have valid ALS and/or PLS

Audit	Evidence of participation in an audit.	Evidence of participation in an audit.	Evidence of completion of an audit with major involvement in design, implementation, analysis and presentation of results and recommendations.	Evidence of completion of an audit with major involvement in design, implementation, analysis and presentation of results and recommendations. Satisfactory portfolio of audit involvement.(AA)
Research	Evidence of critical thinking around relevant clinical questions.	Evidence of critical thinking around relevant clinical questions.	Evidence of developing research awareness and competence through participation in research studies, critical reviews, presentation at relevant research meetings or participation in courses.	Satisfactory academic portfolio with evidence of research awareness and competence. Evidence could include a completed study with presentations or publication, a completed higher degree with a research component or a research degree (MD or PhD).
Teaching	Evidence of participation in teaching of medical students, junior doctors, genetic counsellors or other HPs Assessed by TO.	Evidence of participation in teaching of medical students, junior doctors, genetic counsellors or other HPs	Evidence of participation in teaching with results of students' evaluation. Evidence of understanding of the principles of adult education via training course. Assessed by TO.	Portfolio evidence of ongoing evaluated participation in teaching. Evidence of implementation of the principles of adult education.

Management	Evidence of awareness of and participation in some aspect of management systems: examples might include responsibility for organising rotas, teaching sessions or journal clubs.	Evidence of awareness of and participation in some aspect of management systems: examples might include responsibility for organising rotas, teaching sessions or journal clubs.	Evidence of awareness of managerial structures and functions within the NHS: this could include attendance at relevant courses or participation in relevant local management meetings with defined responsibilities.	Evidence of understanding of managerial structures: for example reflective portfolio entries regarding relevant NHS management activities.
Events giving concern	The following events occurring at any time may trigger review of trainee's progress and possible targeted training: issues of professional behaviour; poor performance in work-place based assessments, poor MSF performance; issues arising from supervisor report or issues of governance including patient safety.			
Educational Supervisor's Report	At least 3 per year. Satisfactory – to include summary of and any actions resulting	At least 3 per year. Satisfactory – to include summary of and any actions resulting	At least 3 per year. Satisfactory – to include summary of and any actions resulting	At least 3 per year. Satisfactory – to include summary of and any actions resulting
Expected competence	Trainees should be competent in the initial assessment of patients presenting with a common genetic disorder. They should be competent in putting forward a basic plan for genetic investigations and clinical management.	Trainees should be competent in the assessment of patients presenting with the majority of common genetic conditions. Trainees should be competent in their approach to the assessment of patients with some rare genetic disorders.	Trainees should be autonomously competent in the assessment and management of patients presenting with common genetic disorders. Trainees should be competent in the assessment and management of genetic disorders presenting acutely (for example in pregnancy).	Trainees should be autonomously competent in the assessment and management of patients presenting with genetic conditions.

*Supervised learning events (SLEs) should be performed proportionately throughout each training year by a number of different assessors across the breadth of the curriculum with structured feedback and action plans to aid the trainee's personal development.

4.8 *Complaints and Appeals*

The trainee is advised to discuss any complaints with his Education Supervisor first; then the Postgraduate Training Coordinator who should discuss the matter with the Training Committee.If the issue is still unresolved he/she needs to write to the Malta College of Pathologists. These complaints/appeals will then be considered in the Malta College Council Meetings according to College, The Malta Postgraduate Training Centre and SAC policies.

The methods used to assess progress through training must be valid and reliable. The Curriculum has been re-written, describing the levels of competence which can be recognised. In the tables below, the "Assessment Methods" shown are those that are appropriate as possible methods that could be used to assess each competency. It is not expected that all competencies will be assessed and that where they are assessed not every method will be used.

The assessment grade will be awarded on the basis of direct observation in the workplace by consultant supervisors. Time should be set aside for appraisal following the assessment e.g. of clinical presentations, case management, observation of procedures. As progress is being made, the lower levels of competence will be replaced progressively by those that are higher. Where the grade for an item is judged to be deficient for the stage of training, the assessment should be supported by a detailed note which can later be referred to at the Annual Evaluation Meeting. The assessment of training shall utilise DOPS (direct observation of procedural skills), Clinical Evaluation Exercise (miniCEX) and Case Based Discussions (CBD) methods adapted for the purpose. They are offered as a means of providing the trainee with attested evidence of achievement in certain areas of the Curriculum e.g. competence in procedural skills, or in generic components. Assessment will also be supported by the trainee's portfolio of achievements and performance at relevant meetings, presentations, audit, in tests of knowledge, attendance at courses and educational events

4.9 Training sites

Training shall take place in a recognised teaching institution or group of institutions recognised by the SAC which offer the trainee practice and knowledge in the full range of the specialty.

5 Supervision and Feedback

5.1 Supervision

All elements of work in training posts must be supervised with the level of supervision varying depending on the experience of the trainee and the clinical exposure and case mix undertaken. Outpatient and referral supervision must routinely include the opportunity to personally discuss all cases if required. As training progresses the trainee should have the opportunity for increasing autonomy, consistent with safe and effective care for the patient.

Trainees will at all times have a named Educational Supervisor and Clinical Supervisor,

responsible for overseeing their education. Depending on local arrangements these roles may be combined into a single role of Educational Supervisor. Supervisors will usually be consultant medical geneticists in NHS practice who have undergone appropriate training in the skills required for such roles. These include understanding the roles of educational and clinical supervisors, ARCP process, educational appraisal, assessment methods, and trainees in difficulty. Examples of such training includes Foundation Programme sessions and Royal College based training development programmes. This may be demonstrated by an educational training portfolio.

The responsibilities of supervisors are reproduced below:

Educational Supervisor

A trainer who is selected and appropriately trained to be responsible for the overall supervision and management of a specified trainee's educational progress during a training placement or series of placements. The Educational Supervisor is responsible for the trainee's Educational Agreement.

Clinical Supervisor

A trainer who is selected and appropriately trained to be responsible for overseeing a specified trainee's clinical work and providing constructive feedback during a training placement. Some training schemes appoint an Educational Supervisor for each placement. The roles of Clinical and Educational Supervisor may then be merged.

The Educational Supervisor, when meeting with the trainee, should discuss issues of clinical governance, risk management and any report of any untoward clinical incidents involving the trainee. The Educational Supervisor should typically be part of the clinical specialty team. Thus if the clinical directorate (clinical chairperson and/or the Clinical Lead in Genetics and/or the Clinical Supervisor) have any concerns about the performance of the trainee, or there were issues of doctor or patient safety, these would be discussed with the Educational Supervisor. These processes, which are integral to trainee development, must not detract from the statutory duty of the hospital to deliver effective clinical governance through its management systems.

Opportunities for feedback to trainees about their performance will arise through the use of the workplace-based assessments, regular appraisal meetings with supervisors, other meetings and discussions with supervisors and colleagues, and feedback from ARCP.

5.2 Appraisal

A formal process of appraisals and reviews underpins training. This process ensures adequate supervision during training provides continuity between posts and different supervisors and is one of the main ways of providing feedback to trainees. All appraisals should be recorded in the Training Portfolio.

Induction Appraisal

The trainee and educational and clinical supervisor should have an appraisal meeting at the

beginning of each post to review the trainee's progress so far, agree learning objectives for the post ahead and identify the learning opportunities presented by the post. Reviewing progress through the curriculum will help trainees to compile an effective Personal Development Plan (PDP) of objectives for the upcoming post. This PDP should be agreed during the Induction Appraisal. The trainee and supervisor should also both sign the educational agreement in the portfolio at this time, recording their commitment to the training process.

Mid-point Review

This meeting between trainee and educational supervisor is mandatory (except when an attachment is shorter than 4 months), but is encouraged particularly if either the trainee or educational or clinical supervisor has training concerns or the trainee has been set specific targeted training objectives at their ARCP. At this meeting trainees should review their PDP with their supervisor using evidence from the portfolio.

Workplace-based assessments and progress through the curriculum can be reviewed to ensure trainees are progressing satisfactorily, and attendance at educational events should also be reviewed. The PDP can be amended at this review.

End of Attachment Appraisal

Trainees should review the PDP and curriculum progress with their educational or clinical supervisor using evidence from the e-portfolio. Specific concerns may be highlighted from this appraisal. The end of attachment appraisal form should record the areas where further work is required to overcome any shortcomings. Further evidence of competence in certain areas may be needed, such as planned workplace-based assessments, and this should be recorded. If there are significant concerns following the end of attachment appraisal then the Postgraduate Training Coordinator for Pathology should be informed.

6 Managing Curriculum Implementation

The PGTC, the Malta College of Pathologists, educational and clinical Supervisors and trainees will together ensure local delivery of the curriculum.

The PGTCPath is responsible to the Malta Postgraduate Training Centre and the Malta College of Pathologists who in turn are responsible to the SAC, and manages the Genetics training programme. The PGTC and the Malta College of Pathologists work with the Malta Postgraduate Training centre to enable trainees to gain the relevant competencies and experience.

The PGTCPath will support educational and clinical supervisors and be part of the ARCP panel. When trainees are running into difficulties the PGTCPath will work with educational supervisors and identify remedial placements in the rotation.

Trainee feedback is through the PGTCPath via trainee surveys.

6.1 Intended Use of Curriculum by Trainers and Trainees

This curriculum and Portfolio are web-based documents which are available from the SAC www.ehealth.gov.mt website.

The educational supervisors and trainers can access the up-to-date curriculum from the www.ehealth.gov.mt website and will be expected to use this as the basis of their discussion with trainees. Both trainers and trainees are expected to have a good knowledge of the curriculum and should use it as a guide for their training programme.

Each trainee will engage with the curriculum by maintaining a portfolio. The trainee will use the curriculum to develop learning objectives and reflect on learning experiences.

6.2 Recording Progress

On enrolling with SAC trainees will be given access to the paper or ePortfolio for Genetics. The Portfolio allows evidence to be built up to inform decisions on a trainee's progress and provides tools to support trainees' education and development.

The trainee's main responsibilities are to ensure the Portfolio is kept up to date, arrange assessments and ensure they are recorded, prepare drafts of appraisal forms, maintain their personal development plan, record their reflections on learning and record their progress through the curriculum.

The supervisor's main responsibilities are to use Portfolio evidence such as outcomes of assessments, reflections and personal development plans to inform appraisal meetings. They are also expected to update the trainee's record of progress through the curriculum, write end-of-attachment appraisals and supervisor's reports.

Educational Supervisors, the PGTCPath and ARCP panels may use the Portfolio to monitor the progress of trainees for whom they are responsible.

The Malta College of Pathologists, the Malta Postgradute Training Centre and the SAC may use summarised, anonymous Portfolio data to support its work in quality assurance.

All appraisal meetings, personal development plans and workplace based assessments should be recorded in the Portfolio. The MSFs will be kept for a period of 15 years by the involved Educational Supervisor. Trainees and supervisors should sign the educational agreement. Trainees are encouraged to reflect on their learning experiences and to record these in the Portfolio. Reflections can be kept private or shared with supervisors.

Reflections, assessments and other Portfolio content should be linked to curriculum competencies in order to provide evidence towards acquisition of these competencies. Trainees can add their own self-assessment ratings to record their view of their progress. The aims of the self-assessment are:

- To provide the means for reflection and evaluation of current practice
- To inform discussions with supervisors to help both gain insight and assists in developing personal development plans.
- To identify shortcomings between experience, competency and areas defined in the curriculum so as to guide future clinical exposure and learning.

Supervisors can sign-off and comment on curriculum competencies to build up a picture of

progression and to inform ARCP panels.

7 Curriculum Review and Updating

The specialty curriculum will be reviewed and updated with minor changes on an annual basis. The curriculum should be regarded as a fluid, living document and the PGTCPath will ensure to respond swiftly to new clinical and service developments. In addition, the curriculum will be subject to three-yearly formal review within the PGTCPath. The PGTCPath will have available:

- The trainees' survey, which will include questions pertaining to their specialty (PGTCPath to provide)
- Specialty-specific questionnaires
- Reports from other sources such as educational supervisors, Malta College of Pathologists, clinical supervisors, Malta Postgraduate Medical Training centre.
- Informal trainee feedback during appraisal.

Evaluation will address:

- The relevance of the learning outcomes to clinical practice
- The balance of work-based and off-the-job learning
- Quality of training in individual posts
- Feasibility and appropriateness of on-the-job assessments in the course of training programmes
- Availability and quality of research opportunities
- Current training affecting the service

Evaluation will be the responsibility of the Genetics trainers, the PGTCPath and Malta College of Pathologists and the SAC. These bodies must approve any significant changes to the curriculum.

Interaction with the NHS will be particularly important to understand the performance of specialists within the NHS and feedback will be required as to the continuing needs for that specialty as defined by the curriculum. It is likely that the NHS will have a view as to the balance between generalist and specialist skills, the development of generic competencies and, looking to the future, the need for additional specialist competencies and curricula. In establishing specialty issues which could have implications for training, the SAC will produce a summary report to discuss with the NHS employers and ensure that conclusions are reflected in curriculum reviews.

Trainee contribution to curriculum review will be facilitated through the involvement of trainees in local faculties of education and through informal feedback during appraisal and College meetings.

The SAC will respond rapidly to changes in service delivery. Regular review will ensure the coming together of all the stakeholders needed to deliver an up-to-date, modern specialty curriculum. The curriculum will indicate the last date of formal review monitoring and document revision.

8 Equality and Diversity

The Malta College of pathologists welcomes applicants from as diverse a population as possible, and actively seeks to recruit people to all its activities regardless of race, religion, ethnic origin, disability, age, gender or sexual orientation.

Appendix 1 - Curriculum & Content of Programme for Higher Genetics Training

Formal lectures, seminars and hands-on training shall form part of the programme for Higher Genetics training. By the end of the educational programme, trainees must have the requisite knowledge, skills and attitudes listed in the curriculum, to diagnose and manage genetic aspects of a wide range of disorders in the following categories, including but not restricted to the conditions specified.

1.1. Good Medical Practice

Good medical practice is core guidance for doctors and sets out the values and principles on which good practice is founded.

Good medical practice is divided into four domains:

1. Knowledge, skills and performance
2. Safety and quality
3. Communication, partnership and teamwork
4. Maintaining trust

The 'GMP' column in the syllabus defines which of the four domains of Good Medical Practice are addressed by each competency.

1.2. Procedures

Objectives

To demonstrate proficiency in clinical procedures related to genetics.

Phlebotomy

Knowledge

Knowledge of technique	GMP 1	mini-CEX,

Skills

Ability to take blood samples from adults and children, including those with special needs	GMP 1, 3	mini-CEX,

Behaviours

Understand the stress of the technique and	GMP 1, 3	mini-CEX,

obtain consent		

Mouth Swab.Mouth Wash

Knowledge

Knowledge of technique and indications for use	GMP 1	mini-CEX,

Skills

Demonstrate ability to obtain samples suitable for analysis	GMP 1, 3	mini-CEX,

Behaviours

Explain procedure appropriately and obtain consent	GMP 1, 3	mini-CEX,

Clinical Photography

Knowledge

Knowledge of technique	GMP 1	mini-CEX MCR
Understand importance and confidentiality of photographic records	GMP 1, 3	mini-CEX MCR

Skills

Demonstrate ability to take photographs of sufficient quality for clinical use		mini-CEX MCR
Use of digital photography and storage of data	GMP 1, 3	mini-CEX MCR

Behaviours

Explain the need for clinical photography and obtain consent	GMP 1, 3	mini-CEX MCR

1.3. Communication Skills and Genetic Counselling

Objectives

Acquire and demonstrate effective communication with patients, relatives and colleagues

Within a Consultation

Knowledge

How to structure a consultation appropriately	GMP 1, 3	CbD, mini-CEX,
The importance of the patient's background, culture, education and preconceptions (beliefs, ideas, concerns, expectations) to the process	GMP: 1,3	CbD, mini-CEX,
Specific techniques and methods that facilitate effective and empathic communication	GMP: 1,3,4	CbD, mini-CEX,
Understand the importance of the developmental stage when communicating with adolescents and young adults	GMP: 1	CbD, mini-CEX,

Skills

Be able to communicate effectively in Maltese and English.	GMP 1, 3	CbD, mini-CEX,
Give clear information and feedback to patients and share information with relatives when appropriate	GMP: 3	CbD, mini-CEX,
Establish a rapport with the patient and relevant others	GMP: 1,3	CbD, mini-CEX,
Utilise open and closed questioning appropriately	GMP: 1,3	CbD, mini-CEX,
Listen actively and question sensitively to guide the patient and to clarify information	GMP: 1,3	mini-CEX,
Identify and manage communication barriers, tailoring language to the individual patient	GMP: 1,3	CbD, mini-CEX,
Deliver information compassionately, being alert to and managing their and your emotional response (anxiety, antipathy etc.)	GMP: 1,3, 4	CbD, mini-CEX,
Use, and refer patients to, appropriate written and other evidence based information sources	GMP: 1,3	CbD, mini-CEX,
Make accurate contemporaneous records of the discussion	GMP: 1,3	CbD, mini-CEX,
Ensure appropriate referral and communications with other healthcare professional resulting from	GMP: 1,3	CbD, mini-CEX,

the consultation are made accurately and in a timely manner		

Behaviours

Approach the situation with courtesy, empathy, compassion and professionalism, especially by appropriate body language and endeavouring to ensure an appropriate physical environment	GMP 1, 3, 4	CbD, mini-CEX, MSF,
Ensure that the approach is inclusive and patient centred and respect the diversity of values in patients, caregivers and colleagues	GMP: 1, 3	CbD, mini-CEX, MSF,
Be willing to provide patients with a second opinion	GMP: 1,3	CbD, mini-CEX, MSF,
Accept uncertainty and use different methods of ethical reasoning to come to a balanced decision where complex and conflicting issues are involved	GMP: 1,3	CbD, mini-CEX, MSF,
Demonstrate:		
• Recognising good advice and continuously promoting values based on non-prejudicial practice (GMP 3)	GMP: 3	MSF,
• Using authority appropriately and assertively; willing to follow when necessary (GMP 3)	GMP: 3	MSF,

Breaking Bad News

Knowledge

Know how to structure the interview and where it should take place	GMP 1	CbD, mini-CEX, MSF,
Be aware of the normal bereavement process and behavior	GMP 1,3	CbD,
How bad news is delivered irretrievably affects the subsequent relationship with the patient	GMP: 1,	CbD, mini-CEX, MSF,
Every patient may desire different levels of explanation and have different responses to bad news	GMP: 1,4	CbD, mini-CEX,
That bad news is confidential but the patient	GMP: 1,	CbD, mini-CEX,

may wish to be accompanied		
Breaking bad news can be extremely stressful for the doctor or professional involved	GMP: 1, 3	CbD, mini-CEX,
“Bad news” may be expected or unexpected and it cannot always be predicted	GMP: 1, 3	CbD, mini-CEX,
Sensitive communication of bad news is an essential part of professional practice	GMP: 1, 3	CbD, mini-CEX,
“Bad news” has different connotations depending on the context, individual, social and cultural circumstances	GMP: 1, 3	CbD, mini-CEX,

Skills

Be able to break bad news in steps appropriate to the understanding of the individual and be able to support distress	GMP 1,3	MSF,
Demonstrate to others good practice in breaking bad news	GMP 1,3	CbD, MSF,
Recognises the impact of the bad news on the patient, caregiver, supporters, staff members and self	GMP 1,3,4	CbD, MSF,
Encourage questioning and ensure comprehension	GMP 1,3	CbD, MSF,
Respond to verbal and visual cues from patients and relatives	GMP 1,3	CbD, MSF,
Act with empathy, honesty and sensitivity avoiding undue optimism or pessimism	GMP 1,3	CbD, MSF,
Structures the interview in an appropriate manner.	GMP 1,3	CbD, MSF,

Behaviours

Show empathy, honesty and sensitivity	GMP 4	MSF,
Show leadership in breaking bad news	GMP 1	CbD, DOPS, MSF ,

Respect the different ways people react to bad news	GMP 1	CbD, DOPS, MSF ,
Ensure appropriate recognition and management of the impact of breaking bad news on the doctor	GMP 2	MSF,

Specific Genetic Issues

Knowledge

Knowledge of ethnic difference in the incidence of genetic disease	GMP 1	CE,
Understanding of cross-cultural issues including consanguinity and arranged marriages	GMP 1	CbD, CE, mini-CEX,
Understanding of religious beliefs and attitudes to prenatal diagnosis and assisted reproduction techniques	GMP 1	CbD, mini-CEX,

Skills

Use of “non-directive” counselling skills	GMP 1,3	CbD, mini-CEX, MSF,
Effective use of co-counsellors	GMP 1,3	CbD, mini-CEX, MSF,
Communication of genetic information and risk to children and adolescents	GMP 1,3	CbD, mini-CEX, MSF,
Communication with adults and children with learning disability	GMP 1,3,4	CbD, mini-CEX, MSF,
Recognising which patients will benefit from referral to psychological services	GMP 1	CbD,

Behaviours

Appreciate patient and family anxieties, both rational and irrational	GMP 1,3,4	CbD, mini-CEX,
Appreciate that every person is influenced by their own culture, ethnicity and beliefs	GMP 1,3,4	CbD, mini-CEX, MSF, , CE
Appreciate the importance of genetic counsellors	GMP 1,3,4	CbD, MSF,
Cultivate habit of reflection and discussion with	GMP 1,3,4	CbD, MSF,

colleagues after counselling sessions		
Readiness to alter practice in light of experience and feed-back	GMP 2, 4	CbD,

Complaints

Knowledge

Be aware of the local complaints procedures	GMP 1,2	MSF, MCR
Be aware of systems of independent review	GMP 1,2	MSF, MCR
Recognise factors likely to lead to complaints (poor communication, dishonesty, clinical errors, adverse clinical outcomes etc.)	GMP 1,2	CbD, MSF, MCR
Recognise the impact of complaints and medical error on staff, patients, and the National Health Service	GMP 1,3	CbD, DOPS, MSF, MCR

Skills

Manage dissatisfied patients / relatives	GMP 1,2	MSF, MCR
Contribute to processes whereby complaints are reviewed and learned from	GMP 1,2,3	CbD, DOPS,MSF, MCR
Explain comprehensibly to the patient the events leading up to a medical error or serious untoward incident, and sources of support for patients and their relatives	GMP 1,2,3,4	CbD, DOPS,MSF, MCR
Deliver an appropriate apology and explanation (either of error or for process of investigation of potential error and reporting of the same)	GMP 1,2,3,4	CbD, DOPS,MSF, MCR
Distinguish between system and individual errors (personal and organisational)	GMP 1,2	CbD, DOPS,MSF, MCR
Show an ability to learn from previous error	GMP 1,2,4	CbD, DOPS,MSF, MCR
Recognise when something has gone wrong and identify appropriate staff to communicate this with	GMP 2	CbD, MCR

Behaviours

Act with honesty and sensitivity and promptly		CbD, MSF, MCR
Be prepared to accept responsibility		CbD, MSF, MCR
Take leadership over complaint issues	GMP 1	CbD, DOPS,MSF, MCR
Recognise the impact of complaints and medical error on staff, patients, and the National Health Service	GMP 1,3	CbD, DOPS,MSF, MCR
Contribute to a fair and transparent culture around complaints and errors	GMP 1, 4	CbD, DOPS,MSF, MCR
Adopt behaviour likely to prevent causes for complaints	GMP 2	CbD, MCR

1.4. History, Examination, Investigations, Management & Note Keeping Skills for Clinical Genetics

Objective

To be able to establish genetic diagnoses by means of clinical history taking, physical examination and use of appropriate investigations and to provide clinical genetic management for patients and families

Knowledge

History		
• Knowledge on how to draw complex pedigrees accurately, including consanguinity loops, recording appropriate information	GMP 1	CbD, mini-CEX,
Investigations		CbD, MSF,
• Surface anatomy	GMP 1	CbD, CE,
• Pitfalls in single gene inheritance including variable expressivity and reduced penetrance, somatic and gonadal mosaicism	GMP 1	CbD, CE,
• Differential diagnoses for genetic disorders	GMP 1	CbD, CE,
• Genetic databases and registers for	GMP 1	CbD, CE,

information retrieval		
Note keeping, letters etc		
• Structure, function and legal implications of medical records & medico-legal reports.	GMP 1,3,4	CbD,
• Know the relevance of data protection legislation pertaining to patient confidentiality	GMP 1	CbD,

Skills

Calculating genetic risk in single gene disorders by hand and by use of a computer programme	GMP 1	CbD, CE,
Present genetic information to a patient in a sensitive and understanding manner	GMP 3,4	CbD, mini-CEX, , MSF
Attention to detail and accuracy in collecting and checking family history and medical data	GMP 2,3	CbD, mini-CEX,
Appreciate the confidentiality and ethical issues arising from family history gathering	GMP 4	CbD,
Clinical history taking, physical examination and use of appropriate investigations	GMP 1	CbD, mini-CEX,
Provide clinical genetic management for patients and families	GMP 1	CbD, CE,

1.5. Formal Genetics and Basic Sciences

Objectives:

- Understand cellular and molecular mechanisms that underpin inheritance in man
- Identify the social and ethical implications of genetic knowledge
- Understand patterns of inheritance and undertake risk assessment
- Have knowledge of emerging genetic technologies and their application (including gene therapy)

Knowledge

The chromosomal basis of heredity (mitosis and meiosis)	GMP 1	CE,
Mechanisms of origin of numerical and structural chromosome abnormalities	GMP 1	CE,

Behaviour of structural chromosome abnormalities at meiosis	GMP 1	CE,
The chemical structure of DNA and replication	GMP 1	CE,
Central dogma of cell biology: transcription and translation	GMP 1	CE,
History of genetics	GMP 1	CE,
Modes of inheritance (Mendelian and non Mendelian)	GMP 1	CbD, CE,
Risk calculations including combinatorial probability and Bayes Theorem	GMP 1	CbD, CE,
The clinical embryology and molecular mechanisms of human malformation syndromes	GMP 1	CbD, CE,
Principles of teratogenesis and pregnancy associated risks	GMP 1	CbD, CE,
Mechanisms of mutagenesis and estimation of mutation rates	GMP 1	CbD, CE,
Identification and critical evaluation of information	GMP 1	CbD, CE,

Skills

Use primary sources of data	GMP 1	CE,
Recognition of different inheritance patterns in pedigrees	GMP 1	CbD, CE,
Pedigree-based calculation of segregation ratios for structural chromosome abnormalities	GMP 1	CbD, CE,
Calculate gene frequencies - the Hardy-Weinberg equilibrium and chi square tests of departure	GMP 1	CE,
Apply knowledge to interpret results of chromosome and molecular genetic analysis	GMP 1	CbD, CE,

Behaviours

Commitment to lifelong self-directed learning	GMP 1	CE,
Appreciation the impact of genetic disorders on individuals and families	GMP 1	CE,
Appreciate potential benefits and harm of new genetic technologies	GMP 1	CbD, CE,
Appreciate public concerns about the application of new genetic technologies	GMP 1	CbD, CE,

1.6. Common Genetic Referrals

Objective

To provide the trainee with the skills and knowledge to be able to carry out specialist diagnosis, assessment and genetic counselling for the conditions previously listed.

Knowledge

The genetic basis and clinical features of common genetic condition including Cystic Fibrosis, Down's syndrome, Fragile X, an x-linked recessive genetic condition	GMP 1	CbD, CE,
The medical and surgical complications of common genetic conditions and indications for referral for specialist opinion	GMP 1	CbD, CE,
Knowledge of long term complications of genetic conditions	GMP 1	CbD, CE,
Molecular/cytogenetic testing and its application to diagnosis, predictive testing, carrier testing and prenatal diagnosis	GMP 1	CbD, CE,
Application and limitations of current tests	GMP 1	CbD, CE,
Knowledge of current clinical treatments for ‘core’ conditions and gene therapy trials	GMP 1	CbD, CE,

Skills

Be able to take a relevant history, perform an appropriate examination and formulate clinical diagnoses	GMP 1	CbD, mini-CEX,
Be able to assess patients and families affected by genetic conditions	GMP 1	CbD,
Judge when it is necessary to sustain supportive relationships with patients with chronic disease	GMP 1,4	CbD,
Be able to discuss reproductive options (AID, ICSI, IVF, pre-implantation diagnosis) with the patient and their partner in a sensitive manner	GMP 1,3	CbD, mini-CEX,
Be able to discuss and formulate management plans with individuals/families	GMP 1,3	CbD, mini-CEX,

Behaviours

Appreciate role of patient education, e.g. in type 1 neurofibromatosis	GMP 1	CbD,
Appreciate the role of the general practitioner in management of chronic disease	GMP 1	CbD,
Appreciate the role of support groups and be willing to provide appropriate information	GMP 1	CbD,
Apply good clinical care and counselling skills	GMP 1	CbD, mini-CEX,

1.7. Neurogenetics

Objective

To provide the trainee with the skills and knowledge to recognise genetic causes of central and peripheral nervous system dysfunction

Knowledge

Classification and molecular basis of common genetic neuromuscular disorders	GMP 1	CbD, CE,
Genetic aspects and clinical presentation of trinucleotide repeat disorders	GMP 1	CbD, CE,

Basic neuropathology and differential diagnosis of hereditary dementias	GMP 1	CbD, CE,
Mitochondrial diseases – clinical, biochemical and genetic features	GMP 1	CbD, CE,
Genetic causes of mental retardation (static and progressive)	GMP 1	CbD, CE,
Genetic contribution to autism and autistic spectrum disorders	GMP 1	CbD, CE,
Genetic contribution to psychiatric disease in adults	GMP 1	CbD, CE,

Skills

Recognise family history data that suggest familial neurological disease	GMP 1	CbD, mini-CEX,
Be able to confirm clinical signs in affected individuals	GMP 1	CbD, mini-CEX,
Be able to draw up a differential diagnosis and institute appropriate genetic testing	GMP 1	CbD, CE, mini-CEX,
Assessment of symptoms and signs in patients at risk of adult-onset neurogenetic disease	GMP 1	CbD, mini-CEX,
Application of protocols for pre-symptomatic diagnosis of Huntington's disease and other neurodegenerative disorders	GMP 1	CbD, mini-CEX,
Make timely, appropriate referrals to other specialists such as neurologists, psychologists, psychiatrists, speech therapists	GMP 1	CbD, MSF,
To perform one audit/review during the rotation	GMP 1,2	AA

Behaviours

Appreciation of family stress caused by risk or eventuality of neurodegeneration	GMP 1,3,4	CbD, MSF,
Appreciate social problems encountered by adults with mild/moderate learning disability	GMP 1,3	CbD, MSF,

Appreciate issues involved in predictive testing	GMP 1,3	CbD, MSF,

1.8. Paediatric Genetics and Dysmorphology

Objectives

To provide the trainee with the skills and knowledge to make syndrome diagnosis in children

Knowledge

Identify normal developmental milestones and diagnose delayed development	GMP 1	CbD, CE, mini-CEX,
Explain morphogenesis in terms of deformation, malformation, disruption and dysplasia	GMP 1	CbD, CE,
Have knowledge of common and rarer dysmorphic syndromes	GMP 1	CbD, CE,

Skills

Be able to take a relevant history, and perform an appropriate examination, obtain illustrative photographs	GMP 1	CbD, mini-CEX,
Have a rational approach to investigation of children with delayed development and/or dysmorphic syndromes.	GMP 1	CbD, mini-CEX,
Formulate differential diagnoses of unknown syndromes	GMP 1	CbD, CE, mini-CEX,
Utilise journals and databases used in syndrome identification	GMP 1	CbD,
Cultivate critical assessment of database information and case reports to identify uncertainty and subjectivity in syndrome diagnosis	GMP 1	CbD,
Be able to provide a diagnostic service within a multidisciplinary clinical team	GMP 1	CbD, mini-CEX, MSF,
Present and discuss cases with colleagues	GMP 1	CbD,

Behaviours

Recognise importance of clinical judgement, timing, and tact when diagnosing and informing parents of an infant with serious malformation or handicap	GMP 1,3,4	CbD, MSF,
Appreciate the emotional reactions of parents following early diagnosis of syndrome or recognition of developmental delay	GMP 1,3,4	CbD, MSF,
Appreciate the adverse reaction families may experience following retraction of a previous diagnosis	GMP 1,3,4	CbD, MSF,
Recognise and explain to families when diagnostic work crosses the boundary into research and the constraints that this imposes	GMP 1,3,4	CbD,

1.9. Cardiac Genetics

Objectives

- Demonstrate the ability to diagnose inherited cardiac conditions (ICC)
- Demonstrate the ability to recommend targeted screening in individuals who are identified as having increased risk of an ICC
- Demonstrate the ability to coordinate appropriate molecular genetic testing

Knowledge

Knowledge of clinical features of ICC syndromes, including Marfan syndrome and related disorders	GMP 1	CbD, CE, mini-CEX,
Knowledge of molecular basis of ICC syndromes	GMP 1	CbD, CE,
Current recommendations concerning cardiac surveillance in ICC families	GMP 1	CE,
Knowledge of genetic causes of sudden adult death	GMP 1	CbD, CE,

Skills

Be able to take a relevant history, perform an appropriate examination	GMP 1	mini-CEX,
Work with bereaved families following sudden adult death	GMP 1,3,4	MSF,
Use of Ghent criteria for diagnosing Marfan syndrome	GMP 1	CE, mini-CEX,
Assessment of screening protocols for at-risk relatives	GMP 1,2	CbD, mini-CEX,
Coordinate diagnostic and predictive genetic testing in ICC families	GMP 1,2	mini-CEX,
Identify at-risk patients and relatives who are eligible to participate in prevention strategies (e.g. therapeutic trials)	GMP 1,2	CbD,
To perform one audit/review during the rotation	GMP 1,2	AA

Behaviours

Demonstrate awareness of the roles of primary care, specialist nurses and genetic counsellors play in assessing families where relatives are at risk of developing ICC	GMP 3	MSF,
Inform patients about lifestyle factors that affect risk	GMP 3,	CbD, mini-CEX,
Support primary and secondary care professionals with the long-term management of selected patients with ICC syndromes	GMP 1,3	CbD, mini-CEX,
Work as a member of a multidisciplinary team	GMP 3,4	MSF,
Understand the impact of ICC risk on individuals and families; and demonstrate awareness of psychological impact of sudden adult death	GMP 2,3	CbD,

1.10. Cancer Genetics

Objectives

- Trainee is able to diagnose rare cancer syndromes and recognise when common cancers are likely to have a single gene basis
- The trainee can recommend targeted screening in individuals who are identified as having increased risk
- Trainee can coordinate appropriate molecular genetic testing

Knowledge

The genetic and environmental factors that affect risk of developing cancer	GMP 1	CE,
Current recommendations concerning tumour surveillance in cancer	GMP 1	CbD, CE,
Knowledge of clinical features of genetic cancer syndromes	GMP 1	CbD, CE,
Knowledge of DNA repair disorders	GMP 1	CE,
Genetic mechanisms in neoplasia: Knudson's two-hit hypothesis, oncogenes	GMP 1	CE,
Knowledge of molecular basis of cancer genetic syndromes	GMP 1	CE,
Knowledge of cancer registers and other sources to verify diagnoses	GMP 1	CE,
Knowledge of disease registers (e.g. von Hippel Lindau disease) to support follow-up of affected and at-risk patients	GMP 1	CE,
Screening protocols for at-risk relatives	GMP 1	CE,
Mechanistic tools for calculating likelihood of cancer being inherited	GMP 1	CbD, CE,

Skills

Identify high risk family from a questionnaire	GMP 1	CE, mini-CEX,
Testing risk prediction algorithms	GMP 1	mini-CEX,
Identify at-risk patients and relatives who are eligible to participate in programmes of cancer	GMP 1	mini-CEX,

prevention strategies		
Interpret Variant of Unknown Significance (VUS)	GMP 1	CE, mini-CEX
To perform one audit/review during the rotation	GMP 1,2	AA

Behaviours

Inform patients about lifestyle factors that affect cancer risk	GMP 1	CbD, CE,
Liaise with other specialists as appropriate e.g. for advice about prophylactic mastectomy and work as a member of a multidisciplinary team	GMP 1	CbD,
Understand the impact of cancer risk on individuals and families	GMP 1	CE, mini-CEX,

1.11. Prenatal Diagnosis and Neonatal Dysmorphology

Objectives

To provide the trainee with the skills and knowledge to undertake genetic assessment of actual and potential problems in the foetus, and provide parents with advice about prognosis and inheritance

Knowledge

Process and limitations of clinical and laboratory diagnostic procedures at neonatal post mortem examination	GMP 1, 2	CbD, CE, mini-CEX,
Knowledge of guidelines on retention and storage of foetal tissues	GMP 1, 2	CbD, CE, mini-CEX,
Know the natural history of prenatally diagnosed conditions including autosomal and sex chromosome aneuploidy syndromes	GMP 1, 2	CbD, CE, mini-CEX,
Knowledge of the Maltese legal framework pertaining to termination of pregnancy	GMP 1, 2	CbD, CE, mini-CEX,
Knowledge of Council of Europe Guidelines on Tissue storage	GMP 1, 2	CbD, CE, mini-CEX,
Knowledge of Non-invasive Pre-natal Testing (NIPT) and Non-Invasive Pre-natal Diagnosis (NIPD)	GMP 1, 2	CbD, CE, mini-CEX,

Knowledge of the role of NIPD/NIPT in terms of antenatal testing	GMP 1, 2	CbD, CE, mini-CEX,
Knowledge of the differences between NIPD and NIPT	GMP 1, 2	CbD, CE, mini-CEX,

Skills

Interpret family history data and trace old medical records	GMP 1, 2	CbD,
Perform post-mortem clinical analysis of the neonate (examination, measurements, photography, radiology, tissue sampling and storage for diagnostic studies)	GMP 1,2,3	CbD,
Use syndrome databases in syndrome diagnosis	GMP 1 , 2	CbD,
Provide genetic advice for women who may be at risk of an affected foetus	GMP 3,4	CbD,
Assess clinical significance of chromosome, DNA, and foetal imaging studies in the context of foetal abnormality or risk thereof	GMP 1,2	CbD,
Formulate differential diagnoses and assess prognosis	GMP 1,2,3	CbD, CE,
Sensitive disclosure of abnormal test results or diagnoses in the antenatal period	GMP 3,4	CbD,CE,
Ability to organise an NIPD for an at risk couple	GMP 3,4	CbD, CE,
Understand the sensitivity & specificity of test	GMP 1,2	CbD,

Behaviours

Appreciate the different perspectives on advantages and disadvantages of prenatal diagnosis in each situation	GMP 1,2	CbD, MCR
Non-judgmental appreciation of the ethical and religious dimensions to prenatal diagnosis	GMP 1,2	CbD, MCR
Awareness of the adverse psychological effects of miscarriages and foetal loss	GMP 1,2	CbD, MCR

Appreciate the role of relevant patient support groups and other counselling services	GMP 1,2,4	CbD, MCR

1.12. Biochemical Genetics & Metabolic Diseases (These skills have to be acquired during the years working in a foreign centre where such a service is available)

Objectives

- To become competent with the diagnosis, treatment and follow up of patients with common Hereditary Metabolic Diseases (HMDs).
- To become familiar with the management of patients in acute metabolic crisis and also with the multidisciplinary care required for patient with chronic diseases, including psychosocial care.

Knowledge

Understand basic physiology & biochemistry including fluid and electrolyte balance	GMP 1	CbD, CE,
Understand metabolic response to fasting, lactate, ammonia, amino, organic & fatty acids	GMP 1	CbD, CE,
Understand oxidative phosphorylation, lysosomal and peroxisomal metabolism	GMP 1	CbD, CE,
Galactose & pathophysiology in Galactosemia	GMP 1	CbD, CE,
Glucose lactate profile and lactate/pyruvate ratios	GMP 1	CbD, CE,
Understand cholesterol and steroid metabolism	GMP 1	CbD, CE,
Metabolic functional studies: including lactate/pyruvate profiling, fasting studies, & investigation of hyperammonaemia	GMP 1,2	CbD, CE,
Metabolic tests required in the investigation of developmental delay	GMP 1	CbD, CE,
Drug management & experience of drugs used in the treatment of metabolic intoxication	GMP 1,2	CbD, CE,
The principles of dialysis for metabolic intoxication	GMP 1,2	CbD, CE,
The applications of liver, HSCT and stem cell	GMP 1	CbD, CE,

transplantation		
The principles of gene therapy	GMP 1,2	CbD, CE,
The general nutritional parameters & the use of nutritional unwell & unstable diet regimes	GMP 1	CbD, CE,
The parameters used to measure normal intellectual and psychological development, the assessment of IQ, behaviour and neuro psychological function	GMP 1	CbD, CE,
The applications of clinical research	GMP 1,2	CbD, CE,

Skills

To be familiar with Enzyme Replacement Therapy protocols for Lysosomal Storage Diseases & other therapies e.g. chaperones, substrate inhibition	GMP 1,2	CbD, mini-CEX,
To be familiar with the principles of gene therapy	GMP 1,2	CbD, mini-CEX,
To be familiar with the interpretation of specialist biochemical testing, including plasma amino & urine organic acid analysis, acylcarnitine profiles, mitochondrial respiratory chain enzymology & lysosomal screening enzymology tests	GMP 1,2	CbD, mini-CEX,
To be familiar with the post mortem metabolic genetic autopsy	GMP 1,2	CbD, mini-CEX,
To become familiar with the principles of Newborn Screening, the local and European practice and understand the different opportunities of genetic screening	GMP 1,2	CbD, mini-CEX,
To perform one audit/review during the rotation	GMP 1,2	AA

1.13. Laboratory Genetics

Objective

To acquire skills and knowledge to interpret genetic laboratory results within a clinical setting, by completing an attachment in the genetic laboratories

Knowledge

Techniques for conventional chromosome analysis in different tissues	GMP 1,2	CbD, CE,
Laboratory techniques and application of cytogenetic tests e.g. Array CGH, FISH	GMP 1,2	CbD, CE,
Apply array-CGH in different clinical settings and interpretation of CNV's (including use of databases such as DECIPHER and ECARUCA)	GMP 1,2	CbD, CE,
Use of ISCN nomenclature	GMP 1,2	CbD, CE,
Molecular genetic techniques in common usage – (DNA extraction, Southern Blotting, PCR, DNA sequencing)	GMP 1,2	CbD, CE,
Exome and genome sequencing: an understanding of the principles and practice of exome and genome sequencing	GMP 1,2	CbD, CE,
Next generation sequencing (exomic & genomic) sequencing	GMP 1,2	CbD, CE,
Application of DNA-based testing for gene mapping, linkage and mutation detection	GMP 1,2	CbD, CE,
Potential application of new DNA technologies	GMP 1,2	CbD, CE,
Sensitivity and specificity of laboratory tests	GMP 1,2	CbD, CE,
Use of DNA and molecular cytogenetic methods in pre implantation diagnosis	GMP 1,2	CbD, CE,
The operation of national newborn screening programme	GMP 1,2	CbD, CE,
Pre implantation genetics diagnosis	GMP 1,2	CbD, CE,
Interpretation of clinical consequences of abnormal karyotypes and molecular test results	GMP 1,2	CbD, CE,
Incidentals and awareness of how and where these arise and how to manage them	GMP 1,2	CbD, CE,
Awareness of ESHG document on incidental findings	GMP 1,2	CbD, CE,

Non-invasive prenatal testing and non-invasive prenatal screening; cell-free fetal DNA (cffDNA): an understanding of the sensitivity, specificity and positive predictive value of cffDNA for aneuploidy screening	GMP 1,2	CbD, CE,
Understand techniques for conventional cytogenetic analysis in different tissues	GMP 1,2	CbD, CE,
Interpret clinical consequences of chromosomes rearrangements	GMP 1,2	CbD, CE,
Understand the principles and application of next generation sequencing (NGS) technologies including targeted panels, clinical exome sequencing, whole exome sequencing, whole genome sequencing	GMP 1,2	CbD, CE,
Interpret the large data set created from NGS using basic bioinformatics, filtering techniques, clinical and functional data	GMP 1,2	CbD, CE,
Know OMIC technologies and their current and future applications	GMP 1,2	CbD, CE,
Be aware of the Human Genome Variation (HGVS) nomenclature for single gene variants	GMP 1,2	CbD, CE,
Understand the sensitivity and specificity of laboratory tests	GMP 1,2	CbD, CE,

Skills

Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines	GMP 1,2	CBD,
Willingness to liaise with colleagues to interpret laboratory results	GMP 1,2	MSF
Liaise with molecular and cytogenetics scientists in analysis of test results	GMP 2,3	MSF
Provide advice to laboratory on the wording of reports to referring clinicians	GMP 2,3,4	CbD, mini-CEX, , MSF
Genetic risk calculation based on laboratory test	GMP 1,2	CbD, mini-CEX,

results (e.g. MLINK, Bayesian analysis)		
Be aware of importance of Bioinformatics & be able to do database searches (Decipher & Ensembl)	GMP 1,2	CbD,
Awareness of possibility of incidental findings and the methods required to interpret variants of unknown significance	GMP 1,2	CbD,
Awareness of the need for confirmation of diagnosis where screening techniques are used	GMP 1,2	CbD,
Undertake genetic risk calculation based on laboratory test results (incorporation of genetic test results into Bayesian calculations)	GMP 1,2	CbD, mini-CEX,
Interpret results of cytogenetic, molecular cytogenetic, molecular genetic and biochemical tests	GMP 1,2	CbD, mini-CEX,
Use databases including ENSEMBL, USCS and locus-specific databases for interpretation of results	GMP 1,2	CbD, mini-CEX,
Liaise with laboratory scientists and bioinformaticians in the analysis of test results	GMP 3,4	MSF
Provide advice to genetic laboratory colleagues on the wording of reports to referring clinicians	GMP 2,3,4	MSF
Familiarity with the use of proteomics/ metabolomics to identify the underlying pathology of a disease/syndrome	GMP 1,2	CbD, mini-CEX,
To perform one audit/review during the rotation	GMP 1,2	AA

Behaviours

Develop awareness of the importance of informed consent in relation to storage of DNA and cell lines	GMP 2,3	CbD, mini-CEX,
Be able to take informed consent when undertaking genomic analyses	GMP 1,2,3	CbD, mini-CEX,
Demonstrate awareness of the potential for	GMP 1,2	CbD, mini-CEX,

incidental findings in genomic analyses and the complexity of these from the patient perspective		
Recognise the importance and impact of genetic test results for families and communicate implications of results clearly to them	GMP 1,2,3	CbD, mini-CEX,
Show willingness to liaise with colleagues to interpret laboratory results	GMP 3,4	MSF
Be able to adapt to new techniques and tests as they arise and incorporate them into clinical practice appropriately	GMP 1,2	CbD, mini-CEX,

1.14. Organisation and Provision of Genetics Services for Populations

Objectives

To identify practical, legal and ethical issues arising from operation of genetic registers. To know the criteria against which screening programmes for genetic diseases and susceptibilities are judged.

Knowledge

The genetic characteristics in different populations, common gene frequencies and disease prevalence	GMP 1,2	CbD, CE,
The factors that influence decisions to instigate programmes of population screening for genetic diseases	GMP 1,2	CbD, CE,
Sensitivity, specificity and predictive values of screening tests	GMP 1,2	CbD, CE,
Knowledge of current screening programmes	GMP 1,2	CbD, CE,
Knowledge of appropriate population-based registers	GMP 1,2	CbD, CE,

Skills

Encourage patients to adopt a healthier lifestyle with specific emphasis on risk factor avoidance and promotion of behaviours that reduce risk of developing disease	GMP 1,3	CbD, mini-CEX,
Team-working with database managers, genetic		

associates and nurse specialists in: • 'cascade screening' and provision of genetic services for extended families with common single gene disorders (e.g. thalassaemia, cystic fibrosis, gangliosidosis, familial Mediterranean fever, fragile X syndrome, Huntington's disease) • family based screening for individuals at high risk of developing cancer • contribute to the maintenance of any national or hospital based genetic register systems		
Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)	GMP 1,2	
Be able to explain the benefits and consequences of screening programmes	GMP 1,3	CbD, mini-CEX,
Be aware of neonatal screening programmes in EU	GMP 1	CbD, mini-CEX

Behaviours

Appreciate ethical and social dimensions to population screening	GMP 1,2	
Understand the central role of patient education	GMP 1,2	CbD, mini-CEX,
Appreciate the value of specialised clinics (breast clinics, lipid and cardiovascular risk factor clinics)	GMP 1,2	

1.15. Joint Specialist Clinics

Objectives

To equip the trainee with skills and knowledge to provide genetic advice within multidisciplinary clinic settings

Knowledge

Genetic contribution with other specialists including: • Child development • Vision	GMP 3,4	CbD, mini-CEX, , MSF

• Hearing • Endocrine • Skeletal dysplasia • Neurological • Cranio-facial malformation • Tumour surveillance • Cardiac		

Skills

Team working skills	GMP 3,4	MSF
Develop skills and liaisons needed to nurture new services, even in settings such as health centres or child development centres, outside of the genetics department	GMP 1,3	MSF

1.16. Patient Education and Disease Prevention

Objective

To ensure that the trainee has the knowledge, skills and attitudes to be able to educate patients effectively about genetic disease.

Knowledge

Educating patients about: • disease • investigations • management	GMP 1,2	CbD, CE,
Know disease course and manifestations	GMP 1,2	CbD, CE,
Know investigation procedures including possible alternatives / choices	GMP 1,2	CbD, CE,
Management strategies for genetic disease	GMP 1,2	CbD, CE,
Awareness of the EU recommendation on rare disease and how it impacts on patients with rare disease .	GMP 1,2	CbD, mini-CEX,

Awareness articles 12 & 13 of the EU cross border directive for travel for patients with rare diseases and the application of European Reference Networks	GMP 1,2	CbD, mini-CEX,
Knowledge of the role of the Orphanet website and its use as a resource for patients with rare diseases and professionals who care for such patients	GMP 1,2	CbD, mini-CEX,
Understand the risk factors that may influence certain genetic diseases, including: • Life style • Smoking • Alcohol • Medication	GMP 1,2	CbD, CE,
Knowledge of teratogenic potential of medication	GMP 1,2	CbD, CE,
Know the methods of data collection and their limitations	GMP 1,2	CbD, mini-CEX,
Know principles of 1^{o} & 2^{o} prevention & screening	GMP 1,2	CbD, mini-CEX,

Skills

Assess an individual patient's risk factors	GMP 1,2	CbD, CE, , mini-CEX
Consider the positive & negative aspects of prevention and importance of patient confidentiality	GMP 2,4	CbD, mini-CEX,
Give information to patients clearly in a manner that they can understand including written information	GMP 2,3	CbD, mini-CEX,
Respect patient choice	GMP 3, 4	CBD, mini-CEX
Consider involving patients in developing mutually acceptable investigation plans	GMP 3	CBD, mini-CEX
Use Orphanet effectively to source information	GMP 1,2	Mini-CEX

Behaviours

Encourage participation in appropriate disease prevention or screening programmes	GMP 1,3	CbD, CE,
Encourage patients to access further information and patient support groups	GMP 3	CbD, mini-CEX,

1.17. Use of Databases

Objective:

To ensure that the trainee becomes proficient in the use of Decipher and Ensembl web based browser platforms (databases) for comparative genomics (human genome variants.)

Knowledge

Be able to upload a case on to Decipher	GMP 1,2	CbD, mini-CEX,
Be able to navigate through Ensembl and identify genes of interest within patients Copy Number Variation (CNV)	GMP 1,2	CbD, mini-CEX,
Be able to investigate a variant and determine its pathogenicity or otherwise using ClinVar and Exac databases	GMP 1,2	CbD, mini-CEX,
Knowledge of NHS processes used to determine pathogenicity of a variant	GMP 1,2	CbD, mini-CEX,

Skills

Identifying genes of interest and CNV's on databases	GMP 1,2	CbD, mini-CEX,
Determination of the pathogenicity of a variant using appropriate methodologies and platforms	GMP 1,2	CbD, mini-CEX,
Presentation of cases at clinical meetings	GMP 1	MSF