Case reports are the primary form of summative assessment in clinical genetics advanced training and an alternative to an exit examination. They are assessed centrally by members of the Advanced Training Committee (ATC) and ensure that there is uniformity in assessment of trainees across Australia and New Zealand.

Twelve case reports must be submitted and assessed as satisfactory to complete Advanced Training in Clinical Genetics. The minimum requirements for submission of case reports are three in the first year, four in the second year and five in the third year of training. Case Reports are due by 15 September each year. Completion of case reports is required to progress to the next year of training; if the required number of case reports are not submitted annually by the provided deadline in the handbook, the training period will become ineligible for certification. A case report should be written on each one of the case-based discussions submitted.

For clinical genetics and cancer genetics trainees, over the course of three years, the case reports should address clinical problems in the following areas:

- Cytogenetics
- Dysmorphology/clinical diagnosis
- Molecular genetic testing/clinical correlation
- Genomics
- Biochemical genetics
- Prenatal diagnosis
- Mendelian genetic disorders e.g. cystic fibrosis, Huntington’s disease, myotonic dystrophy

For metabolic medicine trainees, at least five of the total 12 case reports should focus on:

- Management/treatment issues relevant to the case with a list of relevant counselling/psychosocial issues*
- Counselling/psychosocial issues with a list of relevant management/treatment issues

In addition, two general clinical genetic case reports should be submitted. These should be written as outlined for trainees in clinical genetics.

*Note the option for some of the metabolic cases to include a management section relevant to the case, instead of a detailed counselling section. It is suggested that metabolic trainees submit a combination of cases covering metabolic management and/or more detailed counselling issues. The management section will be marked rigorously by a metabolic specialist. The metabolic management case reports should include a list of relevant counseling issues and a pedigree. If there are important errors or omissions in the management described the case will be marked as unsatisfactory. The metabolic cases focusing on the counseling issues should include a list of management issues relevant to the case and a pedigree.
Publications may be submitted as case reports (with the trainee as the first author). A counselling issues section should be appended to publications submitted as case reports. No more than two publications per year should be submitted as case reports where it is not possible to include counselling issues (e.g. review articles).

**Acceptable standards for case reports**

**Format**

The case report should include the following details and information:

- Summary
- Referral details
- Clinical section including:
  - History
  - Pedigree
    - Standard of pedigree - Please use standardised nomenclature as described in J Genet Counsel (2008) 17:424–433
    - If a case is submitted without a pedigree the EO will return the case to the trainee without submitting for review by the markers
  - Clinical examination
  - Investigations
  - Discussion
  - Differential diagnosis
  - Literature review

- Counselling issues should not just address diagnostic problems or recurrence risk. Topics may include:
  - Reasons for seeking assessment
  - Dilemmas faced by consult
  - Emotions: fear, grieving, guilt, anger, psychological defence mechanisms, consultant’s understanding of discussion of testing, penetrance/occurrence risk, recurrence risk, natural history, variability, prenatal and/or diagnostic testing
  - Benefits/limitations of testing & uncertainty
  - Offer of plans for counselling to relatives at risk

The counselling section is important and should account for at least 10% of the word count. The most common reason for cases being marked unsatisfactory is poor attention to this section. It should focus on the case under discussion.

Prior to choosing cases to write up, trainees should consider what issues they will focus on. Discuss suitable cases with your supervisors or genetic counsellors in your service so you have a clear idea about what you will discuss in the counselling section. It is much easier to write a good report if the case was a good example of a common counselling issue faced in a clinical genetics consultation or if there were particularly difficult issues faced by the patient/family or clinicians/genetic counsellors. At least 2-3 references from relevant counselling literature should be cited in this section.

- Outcome
- Follow-up & management
Written by Dr Juliet Taylor and Associate Professor Carolyn Ellaway

Modified from Guidelines for writing and marking case reports - Associate Professor Edwin Kirk

- Bibliography/reference list – choose one style of referencing and use this consistently within the report

**Word limit**

All trainees have to comply with a word limit of between **1500 - 2000 words** for each case report, not including references. This does NOT apply if the trainee is submitting a publication (either already accepted/published or submitted to a journal) with appended counselling discussion. If the case report exceeds 2000 words, it will be immediately returned by the EO to the trainee and will not be submitted for review by the markers.

**General guidance**

Individuals should be referred to as patients. If it is necessary to identify an individual, it is preferable to use a numeric designation, e.g. Patient 1 or their numerical reference as per the pedigree, rather than using any other identifying notations such as initials. Terms should not be abbreviated unless they are used repeatedly, and the abbreviation is helpful to the reader.

Sequence variants should be described in the text and tables using both DNA and protein designations whenever appropriate. Sequence variant nomenclature should follow the current HGVS guidelines (see varnomen.hgvs.org for examples of acceptable nomenclature). Human gene nomenclature should follow the standards of the HUGO Gene Nomenclature Committee (HGNC), see [https://www.genenames.org/](https://www.genenames.org/).

Clarity of expression is important so try to write the report in a concise and focused way. Part of the purpose of writing case reports is for assessment of your written communication, which is an important skill for a clinical geneticist. The description of the case should be succinct. Think about what is important: important to the diagnosis, important to the decisions that were made, important to the patient and relevant to counselling issues. Discussion of the disorder likewise should be concise and focused on what is clinically important. The emphasis will of course vary from case to case. The differential diagnosis may be very important in a dysmorphology case and completely irrelevant in a case of predictive testing for Huntington disease. Cases will be marked as unsatisfactory if there are important omissions but may also be sent back for revision if there is irrelevant material included, which suggests a lack of understanding of what is important to the case.

It is important to use grammatically correct English and minimise typographical errors in the report. Ensure that your case report is proof-read by your supervisor and other colleagues. Aim for the report to be at the standard of a paper submitted to a medical journal for publication.

Avoid “cut and paste” of diagrams or tables directly from publications. Consider whether any diagram/table included is necessary and adds to the quality of your report. Including a table containing the results of every investigation performed on a patient is never appropriate. Only include the pertinent positive and negative results.

Reference to the relevant literature is important but does not need to be exhaustive. Aim for 10-20 references for the entire case. Reference to primary literature is preferable to reference to GeneReviews and the like; we want you to read and understand where the information comes from.
Examples of representative reports will be provided on request. Please contact the EO.

**Marking of case reports**

Two reviewers are assigned to each case and the trainee will receive either a satisfactory or unsatisfactory mark. A case must be marked satisfactory by both reviewers to pass. If a case is assessed as unsatisfactory, the reviewers will try to make it as clear as possible what the deficiencies are and how the case could be improved. The trainee will then have three months to resubmit this case report addressing the reviewer’s comments.

Case reports will be marked and returned to trainees after eight weeks. Where delays occur, the EO will be in contact with you with further advice.

If you have questions about a submitted or marked case report, please contact the EO directly at ClinicalGenetics@racp.edu.au.

**Sources:**
- Clinical genetics advanced training handbook