

**Clinical Genetics Case Report Marking Criteria**

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| **Trainees Name** |   |
| **Case Study Title** |  |
|  | Submission 1 [ ]  | Resubmission 1 [ ]  | Resubmission 2 [ ]  |
| **OUTCOME** |
| **[ ]  Satisfactory - Meets satisfactory standard. Minor revision required in no more than 1 criterion.** **[ ]  Resubmit ≥ 2 areas require minor revision or ≥ 1 area requires major revision****[ ]  Fail – Does not meet any of the criteria.****General Comments and Feedback****Reviewers are encouraged to provide a balance of positive feedback as well as indicate areas for improvement. For reports graded unsatisfactory, reviewer's comments should point out inadequacies in the report and promote the trainee’s learning.** |
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 **Purpose**

The aim of this Case Report is to guide the trainee’s learning through structured feedback and help the supervisor evaluate the expertise and judgement exercised in clinical cases. This is a summative assessment

**Requirement**

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 appointed delegates to 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 of the case-based discussions submitted.

|  | **Major revisions** | **Minor revisions** | **Satisfactory** | **Comments** |
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| 1. **Summary**
 | Summary is irrelevant to the case | Some aspects of the summary omitted | Summary includes an overview of the problem or problems that the case addresses, a brief description of the case and the issues to be discussed |  |
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| 1. **Referral details**
 | Source of and reason for referral not stated.Major aspects. eg: Reasons for referral omitted. | Source of and reason for referral unclear.Some aspects omitted.  | Source of and reason for referral clear.Referral provides information on who referred the patient (type of doctor) and reason for referral. |  |
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| 1. **Clinical section**
 | Major omissions in the case presentation eg key aspects of the history or examination findings omitted.Clinical section is irrelevant to caseClinical section omits key points in the issues coveredClinical section is not linked back to the caseClinical section is of inadequate length (It should form 50 – 60% of the case study)Trainee has misunderstood key aspects of the caseCase does not include clinical genetics/metabolic/cancer genetic issuesClinical section does not include:* History
* Pedigree – use of standardised nomenclature as described in J Genet Counsel (2008) 17:424-433
* Clinical examination
* Investigations
* Discussion
* Differential diagnosis (if relevant)
* Literature review
* Outcome
* Management and follow-up
 | Some aspects of the clinical section not linked to the caseSome statements in the Clinical section not appropriately supported by citations Clinical section does not include some of:* History
* Pedigree – use of standardised nomenclature as described in J Genet Counsel (2008) 17:424-433
* Clinical examination
* Investigations
* Discussion
* Differential diagnosis (if relevant)
* Literature review
* Outcome
* Management and follow-up
 | Good discussion of the issues relevant to the case described.Clinical section linked appropriately to the case.Clinical section includes:* History
* Pedigree – use of standardised nomenclature as described in J Genet Counsel (2008) 17:424-433
* Clinical examination
* Investigations
* Discussion
* Differential diagnosis (if relevant)
* Literature review
* Outcome
* Management and follow-up
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| 1. **Management section (for Metabolic reports)**
 | Metabolic management issues are not well covered and / or do not relate specifically to the caseManagement issues not linked back to the caseManagement issues lack key pointsErrors and omissions in the management described in the case.Inadequate supporting references  | A few aspects of the metabolic management issues are incomplete but generally well doneSome aspects of the management issues are not linked to the caseManagement issues are borderline and further attention should be included | Metabolic management issues have been adequately addressed  |  |
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| 1. **Counselling section** 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
 | Discussion inadequate as limited to diagnostic problem and/or recurrence riskCounselling issues does not cover issues relevant to the practice of clinical genetics careCounselling issues omits key points in the issues coveredCounselling issues is not linked back to the casePoor attention to counselling issues, and does not account for at least 10% of the word count. No mention of at least 2-3 references have been included from relevant counselling literature have been sitedErrors and omissions in the management described in the case. (only for metabolic medicine reports) | A few aspects of the Counselling issues incomplete but generally well done. Eg: omissions in the medical or psychosocial historySome aspects of the counselling section not linked to the caseCounselling issue is borderline and further attention should be includedLimited reference to counselling literature (< 2-3 references have been included) | Good discussion of counselling  issues that are relevant to the case, discussion included appropriate examples of the counselling techniques employed during the consult with the patient and/or family members.Accounts for at least 10% of the word count. At least 2-3 references have been included from relevant counselling literature have been sited |  |
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| 1. **Reflection**
* Reflections on the case; trainee emotions and responses
* Brief paragraph summarizing the case, drawing a few conclusions, and reflecting on the case or mentioning the lessons learned
 | No reflection or thoughts on the case included. | Insufficient reflection either in scope or detail included. | Appropriate reflections on the case including (if relevant) areas where case management and/or counselling could have been improved . |  |
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| 1. **Bibliography/Reference List**
 | Uses different style of referencing and is not consistent throughout the entire report. Many statements in the Clinical and/or counselling section(s) **not** supported by citations and/or citations used do not support statements made. | References have been provided, in majority of aspects.Use of one reference style, however inconsistency can be found in some areas of the report.Some statements in the clinical or counselling section not appropriately supported by citations. | References have been provided. One style of referencing has been chosen and is consistent throughout the report (eg. Vancouver Guidelines).Key statements supported by relevant and contemporaneous citations.10-20 references have been cited |  |
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| 1. **Academic Writing**
 | Presentation, clinical report and/or counselling section disjointed and/or difficult to follow.Poor spelling and/or grammar with a significant impact on the readability of the assignment.Poor quality references used e.g. PowerPoint presentations, low quality websites, low quality journals, outdated sources.Generally inconsistent or incorrect referencing style.Inadequate number of references used.Important omissions made, which suggests a lack of understanding of what is important to the case.Plagiarised. | Minor issues with flow and structure of case study.Some spelling and grammatical errors.Not written in the third person.A few poor-quality references used.Scattered mistakes in referencing. | Individuals have been referred to as patients, or have been de-identified accordingly (e.g.: Patient 1). Sequence variants should be described in the text and tables using both DNA and protein designations whenever appropriate. Current HGVS guidelines have been followed (see varnomen.hgvs.org for examples of acceptable nomenclature).Written in a concise and focused way. Written communication is clear. Clear, logical structure.No/very few spelling or grammatical errors.Written in third person, except the personal reflection.Relevant, up to date, high quality references used.Consistent, correct referencing style (Vancouver Guidelines) used.Discussion is focused on what is clinically important.Has been proofread by supervisor. |  |
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| 1. **Word limit**
 | More than 2000 words (Case Reports will automatically be returned to trainees if they have gone over the word limit)Less than 1500 words. | Not applicable. | Word limit is between 1500-2000 and is appropriate.(This does NOT apply if the trainee is submitting a publication (either already accepted/published or submitted to a journal) with appended counselling discussion.)  |  |
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