**CARE Checklist of information to include when writing a case report**

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| **Topic** | **Item No** | **Checklist item description** | **Reported on Page Number/Line Number** | **Reported on Section/Paragraph** |
| Title | 1 | The diagnosis or intervention of primary focus followed by the words “case report” | P1/L1 | title |
| Key Words | 2 | 2 to 5 key words that identify diagnoses or interventions in this case report, including "case report" | P2/L41 | Key words |
| Abstract  (Structured summary) | 3a | Background: state what is known and unknown; why the case report is unique and what it adds to existing literature. | P2/L22-L24 | Abstract |
| 3b | Case Description: describe the patient’s demographic details, main symptoms, history, important clinical findings, the main diagnosis, interventions, outcomes and follow-ups. | P2/L24-L32 | Abstract |
| 3c | Conclusions: summarize the main take-away lesson, clinical impact and potential implications. | P2/L36-L39 | Abstract |
| Introduction | 4 | One or two paragraphs summarizing why this case is unique **(may include references)** | P3/L46 | Clinical features |
| Patient Information | 5a | De-identified patient specific information | P3/L47-L73 | Clinical features |
| 5b | Primary concerns and symptoms of the patient | P3/L47-L73 | Clinical features |
| 5c | Medical, family, and psycho-social history including relevant genetic information | P5/L106-L112 | Family history |
| 5d | Relevant past interventions with outcomes | P4/L90-P5/L105 | Treatment and outcome |
| Clinical Findings | 6 | Describe significant physical examination (PE) and important clinical findings | P3/L47-L62 | Clinical features |
| Timeline | 7 | Historical and current information from this episode of care organized as a timeline | P3/L47-L62 | Clinical features |
| Diagnostic Assessment | 8a | Diagnostic testing (such as PE, laboratory testing, imaging, surveys). | P3/L63-L73 | Clinical features  Gene sequencing results |
| 8b | Diagnostic challenges (such as access to testing, financial, or cultural) |  | NA |
| 8c | Diagnosis (including other diagnoses considered) | P3/L63-L73 | Clinical features |
| 8d | Prognosis (such as staging in oncology) where applicable | P4/L90-P5/L105 | NA |
| Therapeutic Intervention | 9a | Types of therapeutic intervention (such as pharmacologic, surgical, preventive, self-care) | P4/L90-P5/L105 | Treatment and outcome |
| 9b | Administration of therapeutic intervention (such as dosage, strength, duration) | P4/L90-P5/L105 | Treatment and outcome |
| 9c | Changes in therapeutic intervention (with rationale) |  | NA |

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| Follow-up and Outcomes | 10a | Clinician and patient-assessed outcomes (if available) |  | NA |
| 10b | Important follow-up diagnostic and other test results | P4/L90-P5/L105 | Treatment and outcome |
| 10c | Intervention adherence and tolerability (How was this assessed?) |  | NA |
| 10d | Adverse and unanticipated events |  | NA |
| Discussion | 11a | A scientific discussion of the strengths AND limitations associated with this case report | P7/L148-P8/L186 | Discussion |
| 11b | Discussion of the relevant medical literature **with references** | P7/L148-P8/L186 | Discussion |
| 11c | The scientific rationale for any conclusions (including assessment of possible causes) | P8/L187-L198 | Conclusion |
| 11d | The primary “take-away” lessons of this case report (without references) in a one paragraph conclusion |  | NA |
| Patient Perspective | 12 | The patient should share their perspective in one to two paragraphs on the treatment(s) they received |  | NA |
| Informed Consent | 13 | Did the patient give informed consent? Please provide if requested | **Yes √** | **No** |

\*As the checklist was provided upon initial submission, the page number/line number reported may be changed due to copyediting and may not be referable in the published version. In this case, the section/paragraph may be used as an alternative reference.