| 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" | Page 1/Line 1-3 | Title page/Paragraph 1 |
| Key Words | 2       | 2 to 5 key words that identify diagnoses or interventions in this case report, including "case report" | Page 1/Line 24-25 | Title page/Paragraph 14 |
| Abstract (no references) | 3a | Introduction: What is unique about this case and what does it add to the scientific literature? | Page 2/Lines 17-19 | Abstract/Paragraph 1 |
|  | 3b | Main symptoms and/or important clinical findings | Page 2/Lines 7-12 | Abstract/Paragraph 1 |
|  | 3c | The main diagnoses, therapeutic interventions, and outcomes | Page 2/Lines 12-15 | Abstract/Paragraph 1 |
|  | 3d | Conclusion—What is the main “take-away” lesson(s) from this case? | Page 2/Lines 15-17 | Abstract/Paragraph 1 |
| Introduction | 4 | One or two paragraphs summarizing why this case is unique (may include references) | Page 3/Lines 2-21 | Introduction/Paragraphs |
| Patient Information | 5a | De-identified patient specific information | Page 4/Lines 2 - Page 5 | Case |
|  | 5b | Primary concerns and symptoms of the patient | Page 4/Lines 13-19 | Case |
|  | 5c | Medical, family, and psycho-social history including relevant genetic information | Page 4/Lines 10-Pages | Case |
|  | 5d | Relevant past interventions with outcomes | Page 4/Lines 20-Pages | Case |
| Clinical Findings | 6 | Describe significant physical examination (PE) and important clinical findings | Page 5/Lines 9-Pages 6/Lines | Case |
| Timeline | 7 | Historical and current information from this episode of care organized as a timeline | Page 5/Lines 9-Pages 6/Lines | Case |
| Diagnostic Assessment | 8a | Diagnostic testing (such as PE, laboratory testing, imaging, surveys). | Page 4/Lines 4-6 | Case |
|  | 8b | Diagnostic challenges (such as access to testing, financial, or cultural) | N/A. The genetic | N/A |
|  | 8c | Diagnosis (including other diagnoses considered) | Page 4/Lines 10-12 | Case |
|  | 8d | Prognosis (such as staging in oncology) where applicable | N/A - Not applicable to | N/A |
| Therapeutic Intervention | 9a | Types of therapeutic intervention (such as pharmacologic, surgical, preventive, self-care) | Page 5/Lines 9-14 | Case |
|  | 9b | Administration of therapeutic intervention (such as dosage, strength, duration) | Page 5/Lines 9-Pages 6/Lines | Case |
|  | 9c | Changes in therapeutic intervention (with rationale) | Page 5/Lines 9-Pages 6/Lines | Case |
| Follow-up and Outcomes | 10a | Clinician and patient-assessed outcomes (if available) | Page 5/Line 9-Page 6/Line | Case |
|------------------------|-----|------------------------------------------------------|---------------------------|------|
|                        | 10b | Important follow-up diagnostic and other test results | Page 6/Lines 14-18        | Case |
|                        | 10c | Intervention adherence and tolerability *(How was this assessed?)* | Page 5/Line 12; Page      | Case |
|                        | 10d | Adverse and unanticipated events                     | Page 5/Line 16; Page      | Case |
| Discussion             | 11a | A scientific discussion of the strengths AND limitations associated with this case report | Page 10/Lines 3-11        | Discussion/Paragraph |
|                        | 11b | Discussion of the relevant medical literature *with references* | Page 7/Line 1 - Page      | Discussion/Paragraphs |
|                        | 11c | The scientific rationale for any conclusions *(including assessment of possible causes)* | Page 7/Lines 1 - Page     | Discussion/Paragraphs |
|                        | 11d | The primary “take-away” lessons of this case report *(without references)* in a one paragraph conclusion | Page 10/Lines 6-11        | Discussion/Paragraph 12 |
| Patient Perspective    | 12  | The patient should share their perspective in one to two paragraphs on the treatment(s) they received | Page 10/Lines 14-20       | Patient |
| Informed Consent       | 13  | Did the patient give informed consent? Please provide if requested | Yes ✅                      | No   |

Article information: [http://dx.doi.org/10.21037/atm-20-5540](http://dx.doi.org/10.21037/atm-20-5540)

*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.*