



Topic	Item	Checklist item description	Reported on Line
<b>Title</b>	1	The diagnosis or intervention of primary focus followed by the words "case report" .....	✓
<b>Key Words</b>	2	2 to 5 key words that identify diagnoses or interventions in this case report, including "case report" .....	✓
<b>Abstract (no references)</b>	3a	Introduction: What is unique about this case and what does it add to the scientific literature? <i>we identified two new features</i>	
	3b	Main symptoms and/or important clinical findings .....	<i>small left atrium, growth abnormality, and urogenital/anal abnormality</i>
	3c	The main diagnoses, therapeutic interventions, and outcomes .....	<i>Termination of pregnancy</i>
	3d	Conclusion—What is the main "take-away" lesson(s) from this case? <i>The growth abnormalities are more severe than cardiac abnormality present</i>	
<b>Introduction</b>	4	One or two paragraphs summarizing why this case is unique (may include references) <i>This case identified two new features, namely a small left atrium and anal atresia</i>	
<b>Patient Information</b>	5a	De-identified patient specific information. ....	
	5b	Primary concerns and symptoms of the patient. ....	<i>whether to continue the pregnancy</i>
	5c	Medical, family, and psycho-social history including relevant genetic information .....	<i>not include</i>
	5d	Relevant past interventions with outcomes .....	NO
<b>Clinical Findings</b>	6	Describe significant physical examination (PE) and important clinical findings. <i>thickened NT, TGR, small left atrium, anal atresia</i>	
<b>Timeline</b>	7	Historical and current information from this episode of care organized as a timeline <i>12<sup>th</sup> w → 25<sup>th</sup> w → 30<sup>th</sup> w</i>	
<b>Diagnostic Assessment</b>	8a	Diagnostic testing (such as PE, laboratory testing, imaging, surveys) <i>a 1.12 Mb de novo deletion on chromosome 6q24.3q25.1</i>	
	8b	Diagnostic challenges (such as access to testing, financial, or cultural) <i>whether the prognosis of the fetus is good</i>	
	8c	Diagnosis (including other diagnoses considered) <i>6q25.1 (TAB2) microdeletion</i>	
	8d	Prognosis (such as staging in oncology) where applicable <i>not good</i>	
<b>Therapeutic Intervention</b>	9a	Types of therapeutic intervention (such as pharmacologic, surgical, preventive, self-care) .....	<i>surgical treatment</i>
	9b	Administration of therapeutic intervention (such as dosage, strength, duration) .....	NO
	9c	Changes in therapeutic intervention (with rationale) .....	NO
<b>Follow-up and Outcomes</b>	10a	Clinician and patient-assessed outcomes (if available) .....	<i>not available</i>
	10b	Important follow-up diagnostic and other test results .....	<i>refused to consent to a fetal autopsy</i>
	10c	Intervention adherence and tolerability (How was this assessed?) .....	NO
	10d	Adverse and unanticipated events .....	NO
<b>Discussion</b>	11a	A scientific discussion of the strengths AND limitations associated with this case report .....	Yes
	11b	Discussion of the relevant medical literature <b>with references</b> .....	Yes
	11c	The scientific rationale for any conclusions (including assessment of possible causes) .....	Yes
	11d	The primary "take-away" lessons of this case report (without references) in a one paragraph conclusion .....	✓
<b>Patient Perspective</b>	12	The patient should share their perspective in one to two paragraphs on the treatment(s) they received .....	
<b>Informed Consent</b>	13	Did the patient give informed consent? Please provide if requested .....	Yes <input checked="" type="checkbox"/> No <input type="checkbox"/>