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Review report

Final title: Delirious mania in an adolescent with Bardet-Biedl syndrome : A case report

Title at submission: A complex case of Bardet-Biedl syndrome with delirious mania in an adolescent: A rare case report

Reviewer: **MM Jalal Uddin**, ORCID: 0000-0003-0402-7457

1. Comment It is an interesting and rare case report indeed. Though rare case but sometimes may be found such type of case. Overall write up is good but required some amendment. In the line of 43 it is started with Psychiatric symptoms observed in BBS but in line 44 (continuation of the above line) there were written some psychiatric disorders which are not psychiatric symptoms rather psychiatric disorders (psychiatric diagnosis). Please correction these words.

Response We have corrected it accordingly (Line 43-44).

2. Comment In the introduction section it is very short and please mention few lines about delirious mania including features of delirious mania. .

Response We have added it accordingly (Line 44-46).

3. Comment In the lines 52, 53 and 54, there were mentions about some psychiatric symptoms ; Did these symptoms fullfill the criteria of delirious mania?

Response We have elaborated it to fulfill the criteria clinically (Line 57-60 and 68-75).

4. Comment In the line 64, when the patient started talking it was not mentioned.

Response We have revised accordingly (Line 66).

5. Comment In the line 66, it was reported that she had history of RTI and epilepsy, please write in details about history of epilepsy including management and seizure condition at present.

Response We have added the details accordingly (Line 66-68).

6. Comment In the line 71, it was written about the objective evidence of hallucination but what other symptoms were found to diagnose an case of delirious mania, not mentioned specially mood. Please describe symptoms were present to reach a diagnosis of delirious mania.

Response We have elaborated it (Line 57-60 and 68-75).

7. Comment Please elaborate the abbreviated form of MMSE (Line 75).

Response We have elaborated it (Line 76).

8. Comment Please rewrite in the line 76 as the psychiatric diagnosis was made delirious mania based on .....(mention the criteria or symptoms found in the case).

Response We have added it accordingly (Line 79-80).

9. Comment In the line 79, it was seen that escitalopram was prescribed. Please explain the rationality of use escitalopram in this case of delirious mania .

Response We acknowledge our oversight not to document it properly. Previously she got escitalopram for apathy and clonazepam 0.5mg and aripiprazole 10mg for sleep and behavior disturbance. Later on we stopped escitalopram and switch clonazepam to lorazepam. We have corrected the text (Line 83-84).

10. Comment In the discussion section in line 98, it was written that delirious mania .....BBS with signs of confusion or disorientation. Please check it. In the line 114 ethical approval, it was written that informed written consent was obtained (Line 115), please mention the use of photograph also.

Response We have taken consent from caregiver but did not assent due patient's disorientation. We have added it accordingly (Line 124).

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Keywords
Bardet-Biedl syndrome, delirious mania, psychiatric disorder

Ethical approval
Ethical approval was not sought because this is a case report. However, written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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None

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Not applicable

Reviewer: Hurjahan Banu, ORCID: 0000-0002-8115-1761

Overview
The authors highlighted an uncommon feature of Bardet-Biedl syndrome (BBS) which is not rare in our community. I have the following observations regarding the case report.

11. Comment Was there any symptoms present which can mimic delirium like any metabolic derangement or any features of septicemia?

Response The laboratory and diagnostic studies with clinical examination ruled out delirium-like metabolic derangement or any features of septicemia. We have added it (Line 78).

12. Comment Line 51: Other comorbidities should include diabetes mellitus and hypertension.

Response We have included them (Line 54-55).

13. Comment Line 59: Rather than in the middle of the history, family history should be added toward the bottom

Response We have rearranged it (Line 76).

14. Comment At what age did she receive her epilepsy diagnosis? Did she use any medications for epilepsy? Is there a history of convulsions?

Response We have added the details accordingly (Line 66-68).

15. Comment A case summary can be created by including the following information: background, present, past, drug, family, and history that rules out other conditions that could be mistaken for delirious mania. Supportive investigation reports should be added.

Response We have rearranged it accordingly within the limited word count for the case report.

16. Comment The differential diagnosis and the reasons and methods for ruling out certain disorders should be covered in the discussion section.

Response We have revised the discussion (Line 91).

Reviewer: Abed khan, ORCID: 0000-0002-4151-2817

17. Comment This case report is interesting, however I suggest to use the following title:
"Delirious mania in an adolescent with Bardet-Biedl syndrome: A case report"

Response We have changed the title as per the suggestion.

18. Comment Key messages is generic and lacks case-specific insight.
Suggestion: Tailor key messages to this patient—highlight the diagnostic challenge of psychiatric comorbidity in a syndromic adolescent with neurodevelopmental delay.

Response We have revised it (Line 28-32).

19. Comment "Many organs and systems are impacted by the hereditary condition known as Bardet-Biedl syndrome (BBS)."
The sentence is structurally weak and broad. It lacks scholarly tone.

Response We have changed it accordingly (Line 34).

20. Comment Numerical incidence rates or specific regional data should be added.
Suggestion: Include epidemiological data with a citation.

Response We have added it accordingly (Line 35-37).

21. Comment "Together, these five results are referred to as 'the pentad'..."
The term "results" is inappropriate; it should be "features."
Suggestion: Clarify that this pentad historically includes retinal dystrophy, obesity, polydactyly, hypogonadism, and cognitive impairment.

Response We have revised texts to clarify the points suggested (Line 37-40).

22. Comment "Psychomotor slowness, childishness, and reactive emotional liability..."
This grouping is not referenced and blurs symptom categories.
Suggestion: Specify if these are neurodevelopmental vs psychiatric features, and cite accordingly.

Response Reference number 3 was added. We have revised it per suggestion (Line 41-46).

23. Comment "However, delirious mania is a rare manifestation..."
Needs citation. Also, lacks clarity on its definition and clinical importance.
Suggestion: Define delirious mania briefly, then justify its significance in this case.

Response We have revised the text as per the comment (Line 42-48).

24. Comment "A 17-year-old female, 1st issue of consanguineous marriage of his parents..."
Grammatically flawed. "1st issue" is archaic and "his" is incorrect.
Suggested revision: "A 17-year-old girl, the first child of consanguineous parents, presented with..."

Response We have corrected it accordingly (Line 50).

25. Comment "...presented to the Endocrinology Department... with BBS with multiple comorbidities..."
Redundant phrasing ("with BBS with") and lacks diagnostic confirmation method (clinical vs genetic).
Suggestion: Clarify how BBS was diagnosed—phenotypically or molecularly.

Response We have removed the redundant phrase and clarified it accordingly (Line 52-54).

26. Comment "These psychological concerns included mood swings, visual and auditory hallucinations..."
A long list of symptoms is presented without temporal context or hierarchy.
Suggestion: Structure with subsections or a table (e.g., cognitive, affective, behavioral).

Response We have revised it accordingly (Line 57-61).

27. Comment "Her developmental milestones were delayed by 6 months, including head control, sitting at 12 months..."
Confusing-if sitting was achieved at 12 months; the delay is more than 6 months.
Suggestion: Specify normative milestone expectations to contextualize delays.

Response We have specified it accordingly (Line 64-66).
Answer: She had head control at 6 months but didn't sit unassisted until 12 months or walk until 18 months. She spoke her first word at 18 months, indicating delayed developmental milestones.

28. Comment "Words appeared irrelevant."
Colloquial and vague. Suggestion: Use standardized psychiatric terminology such as "speech was disorganized and lacked coherence."

Response We have revised it accordingly (Line 70).

29. Comment "She denied experiencing perceptual problems but did show objective evidence of hallucinations..."
The sentence is contradictory; clarify that insight was lacking despite active symptoms.

Response We have revised it (Line 73-75).

30. Comment "Previous records showed that her IQ was 65. Later on, Mini-Mental State Exam score was 22."

Response We have justified the statement in the text.

31. Comment IQ and MMSE are not directly comparable; consider explaining intellectual disability levels and why MMSE was used in a young adolescent as it is widely used for evaluation of age-related cognitive decline.

Response IQ level 65 was mild intellectual disability. Mini-Mental State Exam test can be administered to individuals who are 18–85 years or older, with some normative data available for children as young as 10 years of age. It is also used to estimate the severity and progression of cognitive impairment and to follow the course of cognitive changes in an individual over time; thus making it an effective way to document an individual's response to treatment.
Reference: Schatz, P. (2011). Mini-Mental State Exam. In: Kreutzer, J.S., DeLuca, J., Caplan, B. (eds) Encyclopedia of Clinical Neuropsychology. Springer, New York, NY. https://doi.org/10.1007/978-0-387-79948-3_199

32. Comment "She was prescribed quetiapine, escitalopram, topiramate, and melatonin..."
No dosages, rationale, or evidence for polypharmacy are provided.
Suggestion: Justify choice and sequencing of medications.

Response There is a lack of clear classification and established treatment guidelines for delirious mania. Dose and rationale are added (Line 82-83 and 102-106).

33. Comment "Excitation, grandiosity, emotional instability, mania's psychosis and insomnia..."
Poor sentence construction and awkward phrasing ("mania's psychosis").
Suggestion: Rewrite as: "Delirious mania presents with features of psychosis, mood lability, heightened arousal, and cognitive disorientation."

Response We have removed that line.

34. Comment "The extremely quick onset of delirious mania is especially common among children and adolescents."
Good point, but should be expanded with references to relevant literature in pediatric psychiatry.

Response We have tried to expand it with reference (Line 97-99)

35. Comment "Although there are no official diagnostic standards for incontinence (as presented here) ..."
Incontinence was never discussed in the case description.
Suggestion: Either introduce the symptom in the case or remove this from the discussion.

Response We have removed it.

36. Comment "Delirious mania is a potential diagnosis to be made if a patient with BBS shows signs of confusion or disorientation."
Redundant phrasing and weak closing.
Suggestion: Discuss whether this condition is under recognised in neurodevelopmental syndromes and suggest screening approaches.

Response We have moved that line and added line 100-102

37. Comment Grammar: Numerous issues with subject-verb agreement, awkward phrasing, redundancy.

Response We have checked and tried our best within limited words range.

38. Comment Scientific tone: Needs to shift from descriptive to analytical in parts of the discussion.

Response We have revised it.

39. Comment This case report presents a compelling and clinically significant co-occurrence of Bardet-Biedl syndrome with delirious mania. The rarity of the psychiatric manifestation and the complexity of comorbid conditions make this a publishable case. However, the manuscript requires substantial revision for clarity, scientific rigor, and narrative structure.
Specific attention should be paid to:
Defining timelines,
Justifying diagnosis and treatment decisions,
Improving grammatical quality, and

Response We have addressed these points.