Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 89801

Title: A 3M Syndrome Patient with a Novel Mutation: A case report from China

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06366507
Position: Peer Reviewer
Academic degree: MD
Professional title: Professor, Researcher
Reviewer's Country/Territory: China
Author's Country/Territory: China

Manuscript submission date: 2023-11-13

Reviewer chosen by: Yu-Lu Chen

Reviewer accepted review: 2023-12-07 08:26

Reviewer performed review: 2023-12-08 02:09

Review time: 17 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
Creativity or innovation of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation

Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

Scientific significance of the conclusion in this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No scientific significance
Language quality	[ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [Y] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The article found that a younger girl had a rare recessive genetic disorder, and did a detailed clinical examination and whole exon sequencing, and found that she had inherited the disease-causing mutation from both parents. Additionally, the identification of the c.5683+1G>C variant in the OBSL1 gene is noteworthy, as it has not been previously reported in public databases. In addition to her own genetic disease, she also complicated with spinal deformity is also the first report. Although this study shows a novel locus in 3MS, there are some problems listed as follow: 1. What is the basis for dividing 3MS into 3 types? 2. Does having two mutation sites at the same time lead to more severe disease manifestations? 3. How to take effective treatment measures against gene mutation? 4. Is there a link between scoliosis and mutated genes? 5. The use of punctuation in some parts of the article is not standard, and it is recommended to modify it carefully. 6. What is the full name of MVV in the resulting paragraph? This case report is novel and has sufficient sequencing and pedigree verification, so I recommend publishing this article.

Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 89801

Title: A 3M Syndrome Patient with a Novel Mutation: A case report from China

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 04032648
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Mexico

Author's Country/Territory: China

Manuscript submission date: 2023-11-13

Reviewer chosen by: Yu-Lu Chen

Reviewer accepted review: 2023-12-10 22:14

Reviewer performed review: 2023-12-28 05:21

Review time: 17 Days and 7 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
Creativity or innovation of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation

Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

Scientific significance of the conclusion in this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No scientific significance
Language quality	[ ] Grade A: Priority publishing [ ] Grade B: Minor language polishing [Y] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Manuscript 89801 A Case Report of China 3M Syndrome Patient with a Novel Mutation Review comments In general, English should be improved along the manuscript to correct grammar issues and spelling mistakes and improve the use of English. Along the manuscript, authors should comply with the proper nomenclature for genes and proteins, e.g., gene symbols should be italicized, with all letters in uppercase, and proteins should be designated as the gene symbol but not italicized, with all letters in uppercase. For instance, the authors say, “In this case, we employed whole exon sequencing to sequence the CUL7… genes”. 1 Title. The title can be improved to be grammatically correct; instead of …China 3M Syndrome.. should say … Chinese 3M Syndrome … 4 Background The background section is missing information describing the function of the 3M complex under physiological conditions together with the molecular basis for the malfunction of the mutations in OBSL1, Cullin 7 and CCDC8 in disease. A description of the predicted domain(s) or protein fold of OBSL1 could be helpful to understand the effect of the mutations. A brief description of the already-known mutations in OBSL1, Cullin 7, and CCDC8 is desired. A small attempt to

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

include this information was presented in the Discussion section, but it is scant and limited mainly to what can be found in the corresponding Uniprot section. 5 Methods. In the methodology section regarding "Basic information," some details are missing along the text. For example, it mentions that family medical records and consanguinity were extracted from hospital records, but no information is mentioned in the text. 6 Results. The head of the table and the figure legends should be improved, keeping in mind these should be auto-consistent and should be understandable on their own without reading the manuscript. For example, the head of Table 1 says, "Clinical characteristics and molecular findings of the patient"; however, out of context of the manuscript, this title does not inform the type of clinical patient. The symbology used along the table could be improved, for instance, "long slender tubular bones, +." Authors could expand this information somewhere in the table. How much is the + symbol referring to in order to be considered +? Does the "–" symbol mean that the feature is typical? If so, why should all the typical features be included in the table, making reading long and tedious? A phrase in the text mentioning that other features were within the normal standard would suffice. Inconsistencies referring to the patient as 15- or 16-year-old are along the text and should be corrected. The authors' main contribution is a novel mutation in the OBSL1 gene corresponding to c.5683+1G>C together with a second already known mutation c.3341G>A. The authors mention that the c.5683+1G>C variant potentially affects mRNA splicing, leading to decreased OBSL1 protein expression and loss of protein function, which in turn contributes to disease occurrence. Since the authors performed whole exome sequencing, it is assumed that this mutation is present in an exome. Which one? At the coding level, authors should state the outcome of the mutation. Is it an insertion, a deletion, or a frameshift that introduces of a stop codon that truncates the protein? Exactly what does "potentially affects mRNA splicing" mean? The phrase seems ambiguous as it stands. Can the authors perform a northern blot or an

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

immune blot to sustain their claim? At the coding level, the second mutation corresponds to p.Trp1114Ter which truncates the protein in the IGc2 domain. What does IGc2 domain refer to? Authors never mentioned in the text that OBSL1 consists of a protein with 4 tandem N-terminal immunoglobulin (Ig)-like domains, a central fibronectin domain, and 13 C-terminal Ig domains. Hence, this type of information needs to be mentioned in the manuscript. What is the function of each domain of the protein? 7 Discussion Authors mention that disruption of this complex results in microtubule damage, abnormal chromosome separation, and cell death. What does microtubule damage mean? By "chromosome separation," do the authors mean chromosome segregation? The narrative in the discussion section is messy, disorganised, and difficult to follow. For example, the authors say, "As orthopedists, it is important for us to provide symptomatic treatment and follow-up care, considering the high likelihood of encountering patients with this syndrome." 3MS is a rare disease, with approximately 200 cases reported worldwide. The purpose of the paragraph about the growth hormone (GH) therapy section is unclear, as they mention that their patient does not show growth abnormalities. Finally, in the concluding remarks, the authors state that sequencing of CUL7, OBSL1, and CCDC8 genes is necessary to confirm the clinical diagnosis and provide appropriate genetic counseling. Although true, the authors should be more specific to aid the audience, e.g. it is advisable to sequence the whole gene(s) or simply the exons as presented in this manuscript.

Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

RE-REVIEW REPORT OF REVISED MANUSCRIPT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 89801

Title: A 3M Syndrome Patient with a Novel Mutation: A case report from China

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 04032648
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Mexico

Author's Country/Territory: China

Manuscript submission date: 2023-11-13

Reviewer chosen by: Ji-Hong Liu

Reviewer accepted review: 2024-01-15 22:51

Reviewer performed review: 2024-01-17 20:55

Review time: 1 Day and 22 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [ ] Grade C: Good [Y] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[ ] Grade A: Priority publishing [ ] Grade B: Minor language polishing [Y] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

Baishideng Publishing Group

7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: office@baishideng.com
https://www.wjgnet.com

SPECIFIC COMMENTS TO AUTHORS

The manuscript is very different to that initially revised so I prefer not to comment on this revised version