Materials Design Analysis Reporting (MDAR) Checklist for Authors

The MDAR framework establishes a minimum set of requirements in transparent reporting applicable to studies in the life sciences (see Statement of Task: doi:10.31222/osf.io/9sm4x.). The MDAR checklist is a tool for authors, editors and others seeking to adopt the MDAR framework for transparent reporting in manuscripts and other outputs. Please refer to the MDAR Elaboration Document for additional context for the MDAR framework.

Materials

Antibodies	Yes (indicate where provided: section/paragraph)	n/a
For commercial reagents, provide supplier name, catalogue number and RRID, if available.	Yes. Results/Treatment process /paragraph 1	

Cell materials	Yes (indicate where provided: section/paragraph)	n/a
Cell lines: Provide species information, strain. Provide accession number in repository **OR** supplier name, catalog number, clone number, **OR** RRID	Cell lines were not used in this study.	n/a
Primary cultures: Provide species, strain, sex of origin, genetic modification status.	Primary cultures were not involved in this study.	n/a

Experimental animals	Yes (indicate where provided: section/paragraph)	n/a
Laboratory animals: Provide species, strain, sex, age, genetic modification status. Provide accession number in repository **OR** supplier name, catalog number, clone number, **OR** RRID	Laboratory animals were not involved in this study.	n/a
Animal observed in or captured from the field: Provide species, sex and age where possible	No animal was observed in or captured from the field in this study.	n/a
Model organisms: Provide Accession number in repository (where relevant) **OR** RRID	No model organism was used in this study.	n/a

Plants and microbes	Yes (indicate where provided: section/paragraph)	n/a
Plants: provide species and strain, unique accession number if available, and source (including location for collected wild specimens)	No plant was used in this study.	n/a
Microbes: provide species and strain, unique accession number if available, and source	No microbe was used in this study.	n/a

Human research participants	Yes (indicate where provided: section/paragraph)	n/a
Identify authority granting ethics approval (IRB or equivalent committee(s), provide reference number for approval.	Yes. Methods/paragraph 1	
Provide statement confirming informed consent obtained from study participants.	Yes. Methods/paragraph 1	
Report on age and sex for all study participants.	Yes, in the Figure 6	

Design

Study protocol	Yes (indicate where provided: section/paragraph)	n/a
For clinical trials, provide the trial registration number **OR** cite DOI in manuscript.	This study was not previously registered.	n/a

Laboratory protocol	Yes (indicate where provided: section/paragraph)	n/a
Provide DOI or other citation details if detailed step-by-step protocols are available.	All laboratory procedures were performed using standard commercial kit procedures.	n/a

Experimental study design (statistics details)	Yes (indicate where provided: section/paragraph)	n/a
State whether and how the following have been done**, or** if they were not carried out.		n/a
Sample size determination	We collected all surviving maternal blood relatives of the proband, a total of nine cases.	n/a
Randomisation	Randomization was not involved in this study.	n/a
Blinding	Blinding was not involved in this study.	n/a
Inclusion/exclusion criteria	The proband's maternal and living blood relatives were included in this study.	n/a

Sample definition and in-laboratory replication	Yes (indicate where provided: section/paragraph)	n/a
State number of times the experiment was replicated in laboratory	Because it is a mature commercial test, it is tested only once.	n/a
Define whether data describe technical or biological replicates	During the commercial product validation phase, both data describe technical and biological replicates performed.	n/a

Ethics	Yes (indicate where provided: section/paragraph)	n/a
Studies involving human participants: State details of authority granting ethics approval (IRB or equivalent committee(s), provide reference number for approval.	Yes. Methods/paragraph 1	
Studies involving experimental animals: State details of authority granting ethics approval (IRB or equivalent committee(s), provide reference number for approval.	Laboratory animals were not involved in this study.	n/a
Studies involving specimen and field samples: State if relevant permits obtained, provide details of authority approving study; if none were required, explain why.	The ethics committee approved blood samples for this study. The blood samples were properly disposed of and the remaining samples were destroyed.	n/a

Dual Use Research of Concern (DURC)	Yes (indicate where provided: section/paragraph)	n/a
If study is subject to dual use research of concern, state the authority granting approval and reference number for the regulatory approval	This study does not involve.	n/a

Analysis

Attrition	Yes (indicate where provided: section/paragraph)	n/a
State if sample or data point from the analysis is excluded, and whether the criteria for exclusion were determined and specified in advance.	No sample or data points were excluded in the data processing of this study.	n/a

Statistics	Yes (indicate where provided: section/paragraph)	n/a
Describe statistical tests used and justify choice of tests.		n/a

Data Availability	Yes (indicate where provided: section/paragraph)	n/a
State whether newly created datasets are available, including protocols for access or restriction on access.	Pedigree analysis was used in this study, and statistics were not involved.	n/a
If data are publicly available, provide accession number in repository or DOI or URL.	Study data are available via email to the corresponding author.	n/a
If publicly available data are reused, provide accession number in repository or DOI or URL, where possible.	No publicly available data were reused in this study.	n/a

Code Availability	Yes (indicate where provided: section/paragraph)	n/a
For all newly generated code and software essential for replicating the main findings of the study:		
State whether the code or software is available.	Mature commercial software was used for data analysis, and no new code was used.	n/a
If code is publicly available, provide accession number in repository, or DOI or URL.	Code ownership in commercial companies, contact commercial companies can be obtained.	n/a

Reporting

Adherence to community standards	Yes (indicate where provided: section/paragraph)	n/a
MDAR framework recommends adoption of discipline-specific guidelines, established and endorsed through community initiatives. Journals have their own policy about requiring specific guidelines and recommendations to complement MDAR.		
State if relevant guidelines (eg., ICMJE, MIBBI, ARRIVE) have been followed, and whether a checklist (eg., CONSORT, PRISMA, ARRIVE) is provided with the manuscript.	ICMJE guidelines were followed, as the journal follows ICMJE recommendations for publication.	n/a

Article information: https://dx.doi.org/10.21037/atm-21-6925