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. |
| Primary cultures: Provide species, strain, sex of origin, genetic modification status. | Primary cultures were not involved in this study. |

| 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. |
| 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. |
| Model organisms: Provide Accession number in repository (where relevant) OR RRID | No model organism was used in this study. |

| 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. |
| Microbes: provide species and strain, unique accession number if available, and source | No microbe was used in this study. |

| 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 |

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