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Information about the contributions of each person named as having participated in the study

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2) Authors who participated in the conception of the study:
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3) Design & Methods. The following authors were responsible for specific investigations:

- Roberta Bottega and Daniela De Rocco were responsible for molecular analysis
- Alessandro Pecci was responsible for immunofluorescence analysis
- Nuria Pujol-Moix was responsible for TEM analysis
- Alessandro Pecci, Erica De Candia, Nuria Pujol-Moix, Paula G. Heller, Patrizia Noris, Gian Marco Podda, Ana Glembotsky, Marco Cattaneo and Carlo L. Balduini were responsible for enrolling patients and acquiring clinical data

4) Results. The following authors were responsible for specific portions of the results, including figures and tables:

- Roberta Bottega, Anna Savoia and Daniela De Rocco were responsible for the identification of NBEAL2 mutations, splicing mutation analysis and missense pathogenicity analysis. They were also responsible for Tab.1, Fig.1(A,C and D) and Fig.2
- Alessandro Pecci was responsible for Fig.3
- Nuria Pujol-Moix was responsible for Fig.1B
- Alessandro Pecci, Erica De Candia, Nuria Pujol-Moix, Paula G. Heller, Patrizia Noris, Gian Marco Podda, Ana Glembotsky, Marco Cattaneo and Carlo L. Balduini were responsible for characterization of patients with biallelic, monoallelic and no mutations of NBEAL2. They were also responsible for Tab.2, Tab.3, Tab.1S, Tab.2S and Tab.3S

5) Writing the manuscript. The following authors were responsible for writing the manuscript:

- Roberta Bottega, Alessandro Pecci, annd Anna Savoia
- All the other authors read, commented, and approved the manuscripts

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