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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**: Paula Aranaz, Cristina Hurtado, Ignacio Erquiaga, Itziar Miguéliz, Francisco Javier Novo, José Luis Vizmanos

3) **Design & Methods**. The following authors were responsible for specific investigations:

- Paula Aranaz was responsible for the design and performance of the mutational analysis and functional assays
- Cristina Hurtado collaborated with the mutational analysis of the patients and controls and with the functional assays
- Ignacio Erquiaga contributed to the design and development of the functional assays
- Itziar Miguéliz collaborated with the mutational analysis of the patients and controls
- Cristina Ormazábal contributed with the design of the mutational study
- Ion Cristóbal contributed to the design of the functional assays
- José Luis Vizmanos was responsible for the design of all studies of the work

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

- Paula Aranaz was responsible for all results of the study
- Cristina Hurtado collaborated with the mutational analysis of the patients and controls and contributed to the results obtained in the functional assays
- Itziar Miguéliz collaborated with the mutational analysis of the patients and controls
- Ignacio Erquiaga collaborated with the results obtained in the functional assays
- José Luis Vizmanos collaborated with the analysis of all the results obtained in the study
- Paula Aranaz and José Luis Vizmanos made together all figures and tables of the manuscript

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

- Paula Aranaz wrote the manuscript
- Ignacio Erquiaga carefully read and comments the manuscript
- Marina García Delgado carefully read and comments the manuscript
- Francisco Javier Novo carefully read and comments the manuscript
- José Luis Vizmanos wrote the manuscript

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