

Manuscript no. HAEMATOLOGICA/2012/062281 entitled “High frequency of ribosomal protein gene deletions in Italian Diamond Blackfan anemia patients detected by Multiplex Ligation-dependent Probe Amplification (MLPA) assay“

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

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The guarantors of this manuscript confirm that all persons designated as authors qualify for authorship, and that each author has participated sufficiently in the work to take public responsibility for appropriate portions of the content.

2) Authors who participated in the conception of the study: Ugo Ramenghi, Paola Quarello, Emanuela Garelli, Irma Dianzani, Alfredo Brusco

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

- Ugo Ramenghi and Paola Quarello were responsible for following the patients and for patient selection
- Paola Quarello was responsible for clinical data collection
- Emanuela Garelli was responsible for designing the MLPA and real-time PCR study
- Adriana Carando and Patrizia Pappi were responsible for MLPA analysis
- Cecilia Mancini and Alfredo Brusco were responsible for qPCR analysis
- Luciana Vinti and Johanna Svahn were responsible for following patients and for samples and data collection

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

- Paola Quarello was responsible for Table 1.
- Emanuela Garelli was responsible for MLPA data and for Figure 1
- Alfredo Brusco was responsible for qPCR data and for Figure 2
- Emanuela Garelli was responsible for on-line supplementary tables

5) Writing the manuscript. The following authors were responsible for *writing and/or critically revising the manuscript*:

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