Apparent recessive inheritance of sideroblastic anemia type 2 due to uniparental isodisomy at the *SLC25A38* locus

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Keywords: uniparental isodisomy, SLC25A38, sideroblastic anemia type II.

Running title: uniparental isodisomy causes sideroblastic anemia type II.

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Methods

Patients, genomic DNA preparation, and NGS analysis

Patient diagnosis was based on medical history, clinical findings, laboratory data, morphological analysis of peripheral blood and aspirated bone marrow. DNA samples were obtained after signed informed consent of the parents and according to the Declaration of Helsinki. The local University Ethical Committees approved the DNA sampling and the collection of patient samples ('Federico II' University of Naples). Genomic DNA preparation, genetic testing by targeted next-generation sequencing for hereditary anemias, and validation of the variant were performed as previously described.[4]

Oligo-SNP array

The oligo-SNP array analysis followed the protocol provided by the manufacturer, with the analysis on the proband and the parents performed using CytoGenomics v5.0.2 (Agilent Technologies, USA).

Statistical analysis

Statistical significances were determined using Student's t-tests. Two-sided $p < 0.05$ was considered as statistically significant.