

Supplementary figures

New cases of hypochromic microcytic anemia due to mutations in the *SLC11A2* gene and functional characterization of the G75R mutation

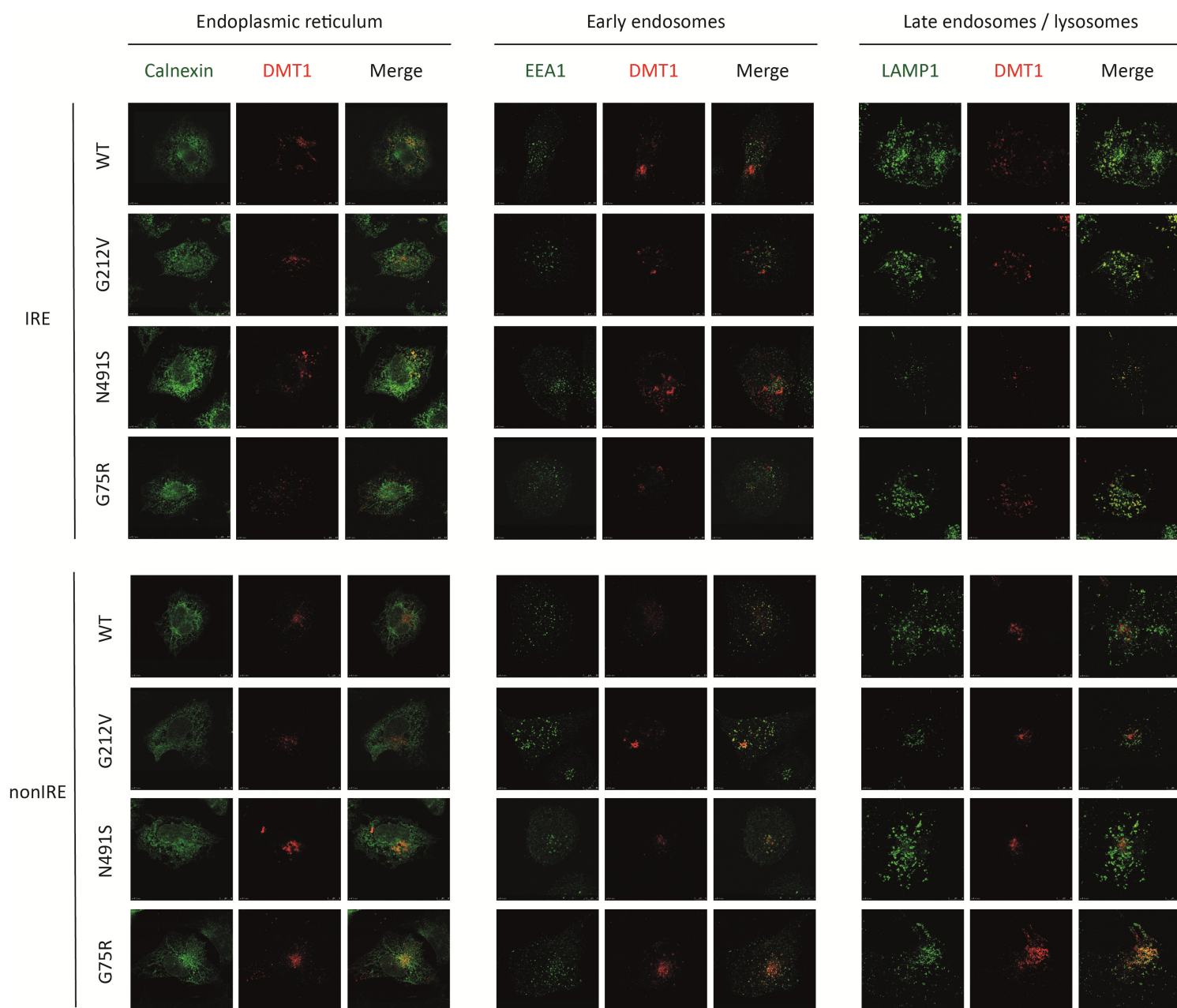
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Supplementary Figure S1. Subcellular localization of wild-type and mutated DMT1 isoforms.

Hutu-80 cells were transiently transfected with a pdsRed2-C1 construct with wild-type (WT), G212V, N491S and G75R mutants of -IRE and -nonIRE isoforms. Cells were incubated with Alexa 488-tagged primary antibodies: anti-calnexin to label endoplasmic reticulum and rabbit anti-LAMP1 to label late endosomes/lysosomes. The figure shows representative split and merged images for each experiment. IRE: iron-responsive element.

Supplementary Figure S2. Comparison of –IRE and –nonIRE DMT1 isoforms’ subcellular localization. Co-localization is expressed as the percentage of DMT1 in the corresponding organelle (n=50). For the WT and each DMT1 mutation (G212V, N491S, G75R), co-localization of the -IRE versus the -nonIRE form in the endoplasmic reticulum (ER; A), early endosomes (B) and late endosomes/lysosomes (C) is compared. $p < 0.05$ was considered significant. **** $p < 0.0001$.

