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Exome sequencing identified RPS15A as a novel causative gene for Diamond-Blackfan anemia

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Contributions: YO, AS-O, YShiraishi, HT, KC and SM performed bioinformatics analyses of the resequencing data. FI, KY and TT processed and analyzed genetic material, prepared the library and performed sequencing. FI, TT, RK and AK performed the Northern blot analyses and RT-PCR analyses. FI, TT, TF and RK performed genome editing using CRISPR/Cas9 system. TU, SI, YN, and NK performed zebrafish experiments. YSasahara, KT, TK, HM, HK, SO, AOhga, SK, and EI collected specimens and were involved in planning the project. EI and SOgawa led the entire project. FI, NK and EI wrote the manuscript. All author participated in discussions and interpretation of the data and results.