Genotype-phenotype association and variant characterization in Diamond Blackfan anemia caused by pathogenic variants in *RPL35A*

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Supplemental Table Legends:

Supplemental Table 1: Collaborating Centers with references if available

References
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Supplemental Table 2: Definitions of clinical phenotypes

References:
1. Bessler M, Mason P, Link D, Wilson D. Nathan and Oski's Hematology of Infancy and Childhood E-Book: Expert Consult: Online and Print. In: Orkin SH, ed. Inherited Bone Marrow Failure Syndromes. 7th ed: Elsevier Health Sciences; 2009:351-360.

Supplemental Table 3: Summary of pathogenic mutations in cases

Summary of all pathogenic variants in cohort by mutation type. Pathogenic mutations must have been reported by collaborators or literature as pathogenic and have minor allele frequency <1% in gnomAD. Light gray indicates collaborator cases and dark gray indicates literature cases. Genomic coordinates are either direct coordinates from collaborator data or direct coordinates from literature case. If genomic coordinates were not available, coordinates were estimated based on deleted region reported and then using UCSC genome browser (i.e. 3q27.2), or estimated coordinates based on genes reported to be deleted and then using UCSC genome browser. Any missing coordinates (i.e. chromosomal or protein position) was annotated using SnpEff. Previously reported indicates the exact case was previously reported by colleagues. For literature cases, previously published is the author and PMID where case was identified.

Abbreviations: (-): Method either not mentioned or unclear in article; [1]Variant not found in ClinVar or HGMD; G-DBA=German DBA registry case; F-DBA=French DBA registry case; NCI=National Cancer Institute; DBAR=DBA Registry of North America; aCGH=array comparative genomic hybridization; MLPA= Multiplex ligation-dependent probe amplification; N/A= not applicable

Supplemental Table 4: Summary of available clinical information by case

Abbreviations: 0=absence of phenotype, 1=presence of phenotype; pRBC=packed red blood cell transfusion, HCT =hematopoietic cell transplant; any congenital abnormality includes microcephaly, craniofacial abnormality, skeletal/arm/leg/rib abnormality, cardiac abnormality, renal abnormality, genital abnormality, and other congenital abnormality; specific craniofacial abnormalities included pinna abnormality, wide set eyes, flat nasal bridge, snub nose, DBA face, high arching palate, and cleft palate; ≥3 abnormalities includes congenital abnormalities, intellectual disabilities, and short stature. GI = gastrointestinal; G-DBA=German DBA registry case; F-DBA=French DBA registry case.

Supplemental Table 5: Summary of large deletions with available genomic coordinates by size of deletion

Name of gene indicates a deleted gene and gray indicates gene is not deleted. Estimated deleted genes in cases based upon UCSC Table Browser, input = UCSC known canonical genes hg19. UCSC Table Browser also used to estimate start/stop coordinates for 3q27.2-q29. Direct genomic coordinates from publication was used when available. [a]Coordinates estimated based on 3q27.2-q29 or 3q27.2 qter reported to be deleted in publication. [b]Coordinates estimated based on reported genes deleted in publication. [c]Estimated coordinates and genes deleted in 3q29 Deletion Syndrome based upon Willat, *Am. J. Hum. Genet*, 2005. PMID= PubMed identification number. G-DBA=German DBA registry case

Supplemental Table 6: Pathogenic *RPL35A* non-large deletion variants observed in more than one case

All pathogenic non-large deletion variants identified in more than one case were included. Pathogenic variants were only counted once per family. Percent is based upon a total of 34 unique unrelated families with a non-large deletion variant, made up of 19 collaborator and 15 literature families.