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Compound heterozygosity in PKLR gene for a previously unrecognized intronic polymorphism and a rare missense mutation as a novel cause of severe pyruvate kinase deficiency

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Contributions: Shruti Bagla planned and executed the PCR and Sanger confirmation of the mutations and the allele-specific cDNA experiments. Kanta Bhambhani provided the clinical care from age 8 weeks and provided critical case details. Manisha Gadgeel and Steve Buck are research assistants and performed the ektacytometry/flow cytometry studies. Prof. Jian-Ping Jin provide critical advice on the allele-specific sequencing studies and reviewed the manuscript. Dr Ravindranath supervised the exome sequencing analysis, identified the potential importance of the intronic mutations. Drs Bagla and Ravindranath equally shared in the writing of the manuscript.