

High frequency of ribosomal protein gene deletions in Italian Diamond-Blackfan anemia patients detected by multiplex ligation-dependent probe amplification assay

Paola Quarello,^{1*} Emanuela Garelli,^{2*} Alfredo Brusco,^{3,4} Adriana Carando,² Cecilia Mancini,³ Patrizia Pappi,⁴ Luciana Vinti,⁵ Johanna Svahn,⁶ Irma Dianzani,^{7,8} and Ugo Ramenghi^{2,8}

¹Pediatric Onco-Hematology, Regina Margherita Children's Hospital, Turin; ²Hematology Unit, Department of Pediatrics, University of Turin; ³Department of Genetics, Biology and Biochemistry, University of Turin; ⁴Medical Genetics Unit, San Giovanni Battista Hospital, Turin; ⁵Department of Hematology-Oncology, Paediatric Hospital Bambino Gesù, Rome; ⁶Hematology Unit, G. Gaslini Children's Hospital, Genoa; ⁷Department of Health Sciences, University of Eastern Piedmont, Novara; ⁸IRCAD Interdisciplinary Research Center of Autoimmune Diseases, Novara, Italy

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Online Supplementary Table S1. Probe set used for MLPA.

Gene	Exon	Sequences of 5' half-probe/ Sequences of 3' half-probe	Amplicon size
<i>RPS17</i>	1	GTGAGCTGGGCCCTTCACGTCAATGTTAGAGGTTGTTACGTGCT AGGCGCTGTTCAAGCACTGGGAATTAGCTAAACAAAATGACCATTACTGTCATGGAGCTC	146bp
<i>RPS17</i>	4	GGTAAGTGTGTTGCTGGATTCTAAAGTGGTATTTTCCTG GTCAAAAACCATCAGTAGGCTTATTATCCAAGGTCACC	120bp
<i>RPS19</i>	1	GTGAAGGACGGAAGATGATAGCCACATTTCTTCTCGCCCTTCCCCTAG GTTCCCTGTCACAGTTCCGCCCTTACTACTCCCACTTCCGCCAGGGAACAGCCACTTC	150bp
<i>RPS19</i>	2	http://www.mrc-holland.com	115bp
<i>RPS19</i>	6	http://www.mrc-holland.com	110bp
<i>RPS26</i>	1	CCATGGATAAATAAACAACACTAGGAACGCATTTCCACCCTAGATTTTCAG CAGAAATGCTGAATGTAAAGGAATATTTGAGTAAAGTGAGTTGCCGTTCC	138bp
<i>RPS26</i>	4	GTGTTCTCCCTCCACCTCTATCAGCTTCCCATGCATTTGTAGCCTGAA TACATCAGTTTGTGAGAGGATGGTGGTCAAGTTCTTTGG	129bp
<i>RPL5</i>	1	GGATGGCGTTAGATTGCTAGCTCTGA CTTGGTTCGAGGTGCAGTTCCAGCGTGGCCTTAAATG	105bp
<i>RPL5</i>	6	GATACCATCTGGGAGGAAAGCTCTTCATAGCATATGTGT GCTATATTCTGGACTTTCCATTATCTCAGACATGTACACAGGA	125bp
<i>RPL11</i>	4	CTTGCACTCGAGGATCTGTAAAGCACCACATGGCTTAAAGGTGGATGA AGGAAACTGTTCCCTCCAGCCAGTGATTTGAAACTTTGGAGAGAGGCAGTA	142bp
<i>RPL35A</i>	1	GGCATTGTTGTCCTCCGAACCTCC TACATGAAACTCCCATCCAAAAGGAATTTGCTTTAGG	101bp
<i>RPL35A</i>	5	GCCACCACACCTGCCAAGTCTTTGTGATACTATGCATTTGTTCAATG CAGATTGGGAAACTTAAAATTTGAATGGAGATTATGTTGATGG	133bp

Online Supplementary Table S2. Real-time PCR probes.

Gene	Probe	Primer Forward	Primer Reverse	Amplicon size
<i>RPS17</i>	55	CACTCTTCCCAGAAGGTTGC	CCTTCTTCACGGTTTTGGTG	88bp
<i>RPS17</i>	51	TGCAGTTGTGTGGGTAGAG	GACAAGTGCCAGCTGATCTGT	65bp
<i>RPS19</i>	7	GCTAAGGTCCCGCCTCTG	GAGCGAAAAGGAATGAGG	76bp
<i>RPS19</i>	79	CCGACCTCTCTGGCTGACTA	GAGCAGGCTAATGAAAGCAGA	73bp
<i>RPS19</i>	23	ACAGGTAAGGCCTGCGTTT	TATGGGCTTTGAAATGCTT	70bp
<i>RPS19</i>	87	GCAGAGACCCCTTGACTAA	ACCCAGCATGGTTTGTCTAA	91bp
<i>RPS26</i>	14	TCCAGAATTGCTCCGAGTC	GCGGAAGAAAATCGAGGTTA	70bp
<i>RPL5</i>	61	TGCAGCAGGTAGGCACAC	CTGAACTTGGGTACGGAGGA	82bp
<i>RPL5</i>	43	GAAAACAGGTTGGGAATGGTT	CGAGTTTGCACAATTACACCA	61bp
<i>RPL11</i>	85	CCATCATGGCCGGTGAGTAG	ATGGATGGCGACGGATAA	60bp
<i>RPL11</i>	20	AGCCCCCTTCTCAGATGAT	GGCTCTGAATCCCATCCTTT	91bp
<i>RPL35A</i>	58	GCCAGCCATTGATTCTACG	AAACACCATCAGCCCAACAT	73bp
<i>RPL35A</i>	57	GGCCAGGACACACTCC	GGGAAATGCCATGAAGTGAT	70bp
<i>CDK2</i>	17	TCCCCTCATCAAGGTAATGC	CACCAGTCCCCAAGACAT	66bp
<i>RNF41</i>	47	TCTCTGGCAGGTTACACAC	CCAATTTCCCAAAGTCCTGA	66bp