Supplementary Table S1. Summary of the clinical and hematological parameters of the patients and mutation carriers.

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Parameters | II-3 | II-5 | II-7 | II-11 | II-15a | III-9 | III-15 | III-14 | IV-5 |
| Sex-age (at diagnosis) | F-60 | F-57 | F-55 | F-51 | M-43 | M-29 | F-18 | M-14 | M-5 |
| Year of presentation | 2017 | 2017 | 2016 | 2017 | 2010 | 2016 | 2016 | 2016 | 2017 |
| WBC (109/L) |  5.06 |  5.31 |  5.02 |  6.34 |  5.26 |  3.77 |  5.53 |  5.49 |  9.16 |
| Hb (g/dL) |  14.8 |  14.6 |  16.9 |  14.2 |  22.5 |  19.8 |  16.5 |  18.9 |  16.0 |
| PCV (L/L) |  0.45 |  0.44 |  0.51 |  0.43 |  0.64 |  0.53 |  0.48 |  0.56 |  0.48 |
| RBC (1012/L) |  4.89 |  4.73 |  5.52 |  4.49 |  7.00 |  5.01 |  5.48 |  6.12 |  5.87 |
| MCV (fL) |  93.2 |  92.2 |  92.6 |  94.9 |  91.3 | 106.0 |  87.3 |  90.7 |  81.9 |
| MCH (pg) |  30.4 |  30.9 |  30.6 |  31.7 |  32.1 |  39.5 |  30.1 |  30.8 |  27.2 |
| Platelets (109/L) | 215.0 | 183.0 | 210.0 | 154.0 | 180.0 | 154.0 | 289.0 | 170.0 | 293.0 |
|  Genotype | codon 109/A | codon 109/A | codon 109/A | codon 109/A | codon 109/A | codon 109/A | codon 109/A | codon 109/A | codon 109/A |

WBC: white blood cell count; Hb: hemoglobin; PCV: packed cell volume; RBC: red blood cell count; MCV: mean corpuscular volume; MCH: mean corpuscular Hb; codon 109/A: Hb San Diego [109(G11)ValMet or *HBB*: c.328G>A].

a Proband.