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.