|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Table 1**. **Patients with Platelet Disorders Associated With Platelet Membrane Glycoproteins Abnormalities** | | | | | | | | | | | | |
| **Laboratory values** | | | | | **Molecular data** | | | | | | | |
| **Probands** | **Gender/age**  **(M/F year)** | **BS** | **Plat. (x109/L)** | **MPV (fL)** | ***Gene*** | **Exon** | **Nucleotide**  **change** | **Amino acid**  **change** | **Gen.** | **ACMG** | **Diagnosis** | **Previously reported** |
| 1 | 59/F | 13 | 18 | 19,5 | *GP9* | 3 | c.212T>C | p.Phe71Ser | Hmz | P | BSS | Yes |
| **2** | **70/M** | **11** | **15** | **nd** | ***GP1BA*** | **2** | **c.578T>C** | **p.Leu193Pro** | **Hmz** | **VUS/L P** | **BSS** | **No** |
| 3 | 49/F | 8 | 13 | 15 | *GP9* | 3 | c.259delT | p.Trp87Glyfs\*41 | Hmz | L P | BSS | Yes |
| 4 | 95/F | 14 | 174 | 11 | *ITGB3* | 5 | c.665T>C | p.Leu222Pro | Hmz | P | GT | Yes |
| 5 | 55/F | 9 | 164 | 11 | *ITGB3*  *ITGB3* | 10  11 | c.1658\_1660delCCT  c.1702T>A | p.Ser553del  p.Cys568Ser | Htz  Htz | P  P | GT | Yes  Yes |
| 6 | 4/F | 11 | 243 | 8 | *ITGB3*  *ITGB3* | 5  12 | c.665T>C  c.1924G>T | p.Leu222Pro  p.Glu642\* | Htz  Htz | P  P | GT | Yes  Yes |
| **7** | **45/F** | **10** | **172** | **11** | ***ITGB3*** | **8** | **c.1036-2A>G** |  | **Hmz** | **P** | **GT** | **No** |
| 8 | 52/F | 10 | 164 | 12 | *ITGA2B* | 12 | c.1063G>A | p.Glu355Lys | Hmz | P | GT | Yes |
| 9 | 29/F | 2 | 100 | 10 | *GP1BA* | 2 | c.1282T>C | p.Ser428Pro | Htz | VUS/LP | MABSS | Yes |
| **10** | **11/F** | **0** | **80** | **12** | ***GP1BA*** | **2** | **c.97T>C** | **p.Cys33Arg** | **Htz** | **L P** | **MABSS** | **No** |
| 11 | 8/F | 9 | 220 | 10 | *ITGA2B*  *ITGB3* | 13  3 | c.1358G>A  c.176T>C | p.Arg453Gln  p.Leu59Pro | Htz  Htz | VUS/LP  B | GTlike | **No**  Yes |
| **12** | **64/F** | **10** | **270** | **12,7** | ***ITGA2B*** | **28** | **c.2906C>T** | **p.Ala969Val** | **Htz** | **VUS/LP** | **GTlike** | **No** |
| 13 | 41/F | 11 | 246 | 11,1 | *ITGA2B* | 4 | c.439C>G | p.Leu147Val | Htz | B | GTlike | Yes |
| **14** | **25/F** | **nd** | **267** | **7** | ***ITGA2B*** | **10** | **c.892-2A>T** |  | **Htz** | **P** | **GTlike** | **No** |
| **15** | **37/M** | **2** | **173** | **11** | ***ITGA2B*** | **19** | **c.1912G>A** | **p.Val638Ile** | **Htz** | **VUS** | **GTlike** | **No** |
| 16 | 37/F | 6 | 168 | 12 | *ITGB3* | 13 | c.2131C>T | p.Pro711Ser | Htz | VUS/LP | GTlike | Yes |
| 17 | 21/M | 2 | 339 | 9 | *ITGA2B* | 29 | c.2965G>A | p.Ala989Thr | Htz | VUS/LB | GTlike | Yes |
| 18 | 49/F | 3 | 60 | 14,9 | *ITGB3*  *ITGA2B* | 11  4 | c.1807G>A  c.439C>G | p.Gly603Ser  p.Leu147Val | Htz  Htz | L P  B | ADMT | Yes  Yes |
| **19** | **34/F** | **0** | **167** | **9,4** | ***ITGA2B*** | **20** | **c.2033C>T** | **p.Ala678Val** | **Htz** | **VUS/LP** | **ADMT** | **No** |
| 20 | 16/M | 1 | 109 | 13 | *GP9* | 3 | c.182A>G | p.Asn61Ser | Htz | P | ADMT | Yes |
| 21 | 58/F | 1 | 116 | 12,7 | *ITGB3* | 10 | c.1658\_1660delCCT | p.Ser553del | Htz | P | ADMT | Yes |
| 22 | 7/M | 2 | 87 | 12 | *ITGB3*  *ITGB3* | 4  14 | c.505C>T  c.2278C>T | p.Arg169\*  p.Arg760Cys | Htz  Htz | P  VUS/LP | ADMT | Yes  Yes |
| *Abbreviations: BS, Bleeding Score (ISTH Bleeding Assessment Tool); Plat., Platelet count; MPV, Mean platelet volume; Gen., Genotype; Hmz, homozygous; Htz, Heterozygous; LP, Likely Pathogenic; P, Pathogenic; VUS, Variant of Uncertain Significance; BSS, Bernard-Soulier Syndrome; MABSS, monoallelic BSS; GT, Glanzmann thrombasthenia; GT-like, GT-like syndrome; ADMT, autosomal dominant macrothrombocytopenia* | | | | | | | | | | | | |