**Table 3. Genes associated with DBA syndrome and genetic phenocopies**

| Gene symbol | Inheritance | Chromosome location | New protein symbol | Approximate frequency | References (see manuscript) |
|---|---|---|---|---|---|
| **DBA SYNDROME: RIBOSOMOPATHY[1]** | | | | | |
| **Small ribosomal subunit (11 genes)** | | | | | |
| *RPS7* | AD | 2p | eS7 | < 1% | 107 |
| *RPS10* | AD | 6p | eS10 | 3% | 54 |
| *RPS15A* | AD | 16p | uS8 | <1% | 108 |
| *RPS17* | AD | 15q | eS17 | 1% | 109 |
| *RPS19* | AD | 19q | eS19 | 25% | 110 |
| *RPS20* | AD | 8q | uS10 | < 1% | 57,111 |
| *RPS24* | AD | 10q | eS24 | 2.4% | 112 |
| *RPS26* | AD | 12q | eS26 | 6.6% | 54 |
| *RPS27* | AD | 1q | eS27 | < 1% | 113 |
| *RPS28* | AD | 19p | eS28 | < 1% | 114 |
| *RPS29* | AD | 14q | uS14 | < 1% | 115 |
| **Large ribosomal subunit (13 genes)** | | | | | |
| *RPL4* | AD | 15q | uL4 | < 1% | 116 |
| *RPL5* | AD | 1p | uL18 | 7% | 55 |
| *RPL8* | AD | 8q | uL2 | < 1% | 117 |
| *RPL9* | AD | 4p | uL6 | < 1% | 13,54 |
| *RPL11* | AD | 1p | uL5 | 5% | 55 |
| *RPL15* | AD | 3p | eL15 | < 1% | 32,118 |
| *RPL17* | AD | 18q | uL22 | < 1% | 15 |
| *RPL18* | AD | 19q | eL18 | < 1% | 119 |
| *RPL26* | AD | 17P | uL24 | < 1% | 120 |
| *RPL27* | AD | 17q | eL27 | < 1% | 113 |
| *RPL31* | AD | 12q | el31 | < 1% | 42 |
| *RPL35* | AD | 3q | uL29 | < 1% | 119 |
| *RPL35A* | AD | 9q | eL33 | 3% | 121 |
| **Ribosomal protein chaperones (2 genes)** | | | | | |
| *TSR2* | X | X | | < 1% | 114 |
| *HEATR3* | AR | 16q | | < 1% | 59 |
| **DBA SYNDROME OTHER[2]** | | | | | |
| *GATA1* | X | X | | < 1% | 23,122-124 |
| *TP53 (GOF)* | AD | AD | | < 1% | 24,25 |
| CANDIDATE GENES[3] | | | | | |
| *RPS11* | AD | 19q | uS17 | < 1% | 47 |
| *RPL3* | AD | 22q | uL3 | < 1% | |
| *RPL10* | AD | X | uL16 | < 1% | |
| *RPL10A* | AD | 6p | uL11 | < 1 % | |
| *RPL19* | AD | 17q | eL19 | < 1% | |
| *RPL34* | AD | 4q | eL34 | < 1% | |
| *RPL0* | AD | 12q | uL10 | < 1% | |
| GENETIC PHENOCOPIES[4] | | | | | |
| *ADA2* | AR | 22q11.1 | | | 27,29,43 |
| *EPO* | AR | 7q22.1 | | | 26 |

[1] Bona fide ribosomopathy genes validated functionally (ribosomal biogenesis defect or presence of somatic genetic rescue).

[2] Genes affecting pathways implicated in DBA syndrome and associated with hyporegenerative anemia.

[3] Considered putative due to lack of studies demonstrating impaired ribosomal biogenesis.

[4] Diseases with different pathomechanisms that can manifest with pure red cell aplasia.

**Abbreviations**: AD, autosomal dominant; X, X-linked recessive; AR, autosomal recessive; GOF, gain-of-function