**Table S1** Frequency in the general population (minor allele frequency, MAF) of the genetic variants identified in infertile men and women which are present in the dbSNP database (https://www.ncbi.nlm.nih.gov/snp/). The reported allele frequencies were retrieved from the following public databases: GnomAD\_exome and TOPMED. CADD is a tool for scoring the deleteriousness of single nucleotide variants as well as insertion/deletions variants in the human genome

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Variant (protein)** | **dbSNP ID** | **GnomAD global v3.1.2 (exomes)** | **TOPMED global** | **CADD score** |
| ***PLCZ1*** | **p.D46N** | rs202206940 | 0.00002635 | 0.000026 | 23.0 |
|  | **p.Q94\*** | rs138801851 | 0.0001447 | 0.000144 | 31.0 |
|  | **p.I120M** | rs79487790 | 0.007517 | – | 19.6 |
|  | **p.R141H** | rs202034240 | 0.00001974 | 0.000015 | 18.1 |
|  | **p.C196\*** | rs535719220 | 0.000006580 | 0.000004 | 36.0 |
|  | **p.R197H**  | rs781075636 | 0.000006574 | 0.000015 | 29.6 |
|  | **p.L224P** | rs144902254 | 0.0004730 | 0.000638 | 21.4 |
|  | **p.H233L**  | rs200061726 | – | 0.000589 | – |
|  | **p.V326Kfs\*25** | rs777169092 | 0.00006586 | 0.000053 | 19.5 |
|  | **p.N377del** | rs758499880 | 0.000006573 | 0.000026 | 18.6 |
|  | **p.A384V** | rs1334320857 | – | – | – |
|  | **p.H398P** | rs1359777791 | 0.000006917 | 0.000011 | 29.2 |
|  | **p.R412Efs\*15** | rs1165227004 | 0.00001325 | 0.000004 | 32.0 |
|  | **p.I489F** | rs757326350 | – | – | – |
|  | **p.S500L** | rs10505830 | 0.03120 | 0.031195 | 23.5 |
|  | **p.M578T** | rs758723831 | 0.00004603 | 0.000060 | 27.1 |
| ***WBP2NL*** | **p.Q5E** | rs17002790 | 0.02773 | 0.031255 | 24.7 |
|  | **p.D121G** | rs133335 | 0.4836 | 0.475413 | 21.3 |
|  | **p.C170F** | rs17002802 | 0.02995 | 0.034799 | 23.0 |
|  | **p.Q285H** | rs2301521 | 0.7431 | 0.238634 | 0.00100 |
| ***WEE2*** | **p.E75Vfs\*6** | rs768323979 | 0.00003944 | 0.000042 | 17.5 |
|  | **p.K195N** | rs1448207084 | – | 0.000011 | – |
|  | **p.R200\*** | rs754062320 | 0.00001315 | 0.000019 | 36.0 |
|  | **p.R207C** | rs200326581 | 0.0001053 | 0.000042 | 29.0 |
|  | **p.D234H** | rs1554415096 | – | – | – |
|  | **p.Q288H** | rs973812791 | 0.00001314 | 0.000008 | 24.7 |
|  | **p.L316F** | rs371571990 | 0.000006569 | 0.000008 | 24.9 |
|  | **p.H337Yfs\*24** | rs1370452353 | – | 0.000004 | – |
|  | **p.R410W** | rs905047580 | 0.000006573 | 0.000004 | 23.9 |
|  | **p.T493Nfs\*39** | rs1554416415 | – | – | – |
|  | **p.Y526D** | rs35683659 | 0.005769 | – | 9.43 |
| ***TUBB8*** | **p.I4L** | rs199817418 | 0.00001315 | 0.000019 | 10.0 |
|  | **p.P70L** | rs782247878 | – | 0.000004 | – |
|  | **p.G98R** | rs1339474843 | 0.000006568 | 0.000004 | 19.7 |
|  | **p.E108K** | rs541770879 | 0.00001970 | – | 22.4 |
|  | **p.T143Dfs\*12** | rs782683305 | – | – | – |
|  | **p.A196T** | rs1554738381 | – | 0.000004 | – |
|  | **p.R241P** | rs782631950 | – | – | – |
|  | **p.R262Q** | rs869025610 | – | 0.000004 | – |
|  | **p.G308S** | rs782575307 | 0.00001314 | – | 21.7 |
|  | **p.R380H** | rs1554738022 | – | – | – |
|  | **p.Q424\*** | rs1356357815 | 0.000006809 | – | 32.0 |
| ***TLE6*** | **p.R409Q** | rs1310808966 | – | – | – |
|  | **p.S510Y** | rs767222404 | – | – | – |
|  | **p.E541K** | rs1176718387 | – | – | – |
| ***PADI6*** | **p.R457\*** | rs766389713 | 0.000006574 | 0.000011 | 38.0 |
| ***TRIP13*** | **p.V247M** | rs1203102465 | – | 0.000004 | – |
| ***BTG4*** | **p.M1?** | rs755402000 | – | – | – |
| ***ZAR1*** | **p.P172=** | rs755756974 | – | 0.000004 | – |
| ***NLRP5*** | **p.R143P** | rs746147069 | 0.000006576 | 0.000004 | 16.3 |
|  | **p.R462C** | rs199475775 | 0.00007884 | 0.000072 | 15.6 |
|  | **p.R533P** | rs752560793 | 0.00003942 | 0.000043 | 8.04 |
|  | **p.S893T** | rs769920247 | 0.00005925 | 0.000110 | 12.3 |
|  | **p.L1116W** | rs773877703 | – | 0.000004 | – |
| ***NLRP7*** | **p.K619Nfs\*6** | rs749049980 | – | 0.000004 | – |
| ***CDC20*** | **p.Y228C** | rs1309457938 | – | – | – |
| ***DNAH17*** | **p.R350\*** | rs371400048 | 0.00001314 | – | 34.0 |
|  | **p.M1130I** | rs189737946 | 0.000006572 | – | 21.1 |
| ***ACTL9*** | **p.S345L** | rs1478948010 | – | 0.000008 | – |
|  | **p.V380L** | rs532021673 | 0.00006574 | – | 24.3 |
|  | **p.Y403\*** | rs34687433 | 0.00006574 | – | 24.3 |
| ***ACTL7A*** | **p.R155\*** | rs755704105 | – | 0.000008 | – |
|  | **p.G362R** | rs779515458 | 0.00001315 | 0.000008 | 27.4 |