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# Supplementary Materials

**Supplementary Table S1**. Missense Variants Identified in the Cohort

| SNP ID | HGVSC | HGVSP | Consequence | Suggested ACMG classification | ACMG criteria | ALFA | | PAGE | Number of individuals | Variant frequency in the cohort |
|---|---|---|---|---|---|---|---|---|---|---|
| rs1042034 | c.13013G>A | p.(Ser4338Asn) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,7893<br>0,73226 | SA 0,7255 | 54 | 0,9000 |
| rs679899 | c.1853C>T | p.(Ala618Val) | missense variant | Benign | BA1, BS2, BP6 | LA1<br>LA2 | 0,3342<br>0,41981 | SA 0,3794 | 41 | 0,6833 |
| rs676210 | c.8216C>T | p.(Pro2739Leu) | missense variant | Benign | BA1, BS2, BP6 | LA1<br>LA2 | 0,2018<br>0,25946 | SA 0,2714 | 35 | 0,5833 |
| rs1367117 | c.293C>T | p.(Thr98Ile) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,2535<br>0,27 | SA 0,2714 | 26 | 0,4333 |
| rs1801702 | c.12809G>C | p.(Arg4270Thr) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0<br>0 | SA Not Reported | 21 | 0,3500 |
| rs17240441 | c.35_43del-TGGCGCTGC | p.(Leu12_Leu14del) | missense variant | Benign | BA1, BS2, BP3, BP6 | LA1<br>LA2 | 0<br>0 | SA Not Reported | 15 | 0,2500 |
| rs1801701 | c.10913G>A | p.(Arg3638Gln) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,0805<br>0,09295 | SA 0,0933 | 6 | 0,1000 |
| rs1042031 | c.12541G>A | p.(Glu4181Lys) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,1590<br>0,1401 | SA 0,11 | 4 | 0,0667 |
| rs1801699 | c.5741A>G | p.(Asn1914Ser) | missense variant | Benign | BA1,BS2, BP6 | LA1<br>LA2 | 0,0203<br>0,0489 | SA 0,9506 | 3 | 0,0500 |
| rs12691202 | c.2188G>A | p.(Val730Ile) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,0285<br>0,0253 | SA Not Reported | 2 | 0,0333 |
| rs6752026 | c.433C>T | p.(Pro145Ser) | missense variant | Benign | BA1, BS2, BP6 | LA1<br>LA2 | 0,0382<br>0,0076 | SA 0,0106 | 2 | 0,0333 |
| rs539614975 | c.3379C>T | p.(Pro1127Ser) | missense variant | VUS | PM2 | LA1<br>LA2 | Not Reported<br>Not Reported | SA Not Reported | 2 | 0,0333 |
| rs2163204 | c.8353A>C | p.(Asn2785His) | missense variant | Benign | BA1,BS2, BP4, BP6 | LA1<br>LA2 | 0,9952<br>0,9717 | SA 0,9818 | 2 | 0,0333 |
| rs769491475 | c.9871C>T | p.(Arg3291Cys) | missense variant | VUS | PM2, BP6 | LA1<br>LA2 | 0<br>0 | SA Not Reported | 1 | 0,0167 |
| rs533617 | c.5768A>G | p.(His1923Arg) | missense variant | Benign | BA1, BS2, BP6 | LA1<br>LA2 | 0,0172<br>0,01221 | SA 0,0096 | 1 | 0,0167 |
| rs61736761 | c.3634C>A | p.(Leu1212Met) | missense variant | Benign | BA1, BS2, BP6 | LA1<br>LA2 | 0,0268<br>0,0058 | SA 0,0056 | 1 | 0,0167 |
| rs12713450 | c.13451C>T | p.(Thr4484Met) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,0397<br>0,0088 | SA 0,0151 | 1 | 0,0167 |
| rs72653077 | c.3427C>T | p.(Pro1143Ser) | missense variant | Benign | BA1, BS2, BP4, BP6 | LA1<br>LA2 | 0,0009<br>0,0012 | SA 0,0005 | 1 | 0,0167 |
| rs12713843 | c.3383G>A | p.(Arg1128His) | missense variant | Benign | BS1, BS2, BP6 | LA1<br>LA2 | 0,0037<br>0,00379 | SA 0,004 | 1 | 0,0167 |
| rs141225768 | c.4663A>G | p.(Ile1555Val) | missense variant | Likely Benign | BS1, BP4, BP6 | LA1<br>LA2 | 0,0024<br>0 | SA 0 | 1 | 0,0167 |
| rs72653098 | c.8912A>C | p.(Asn2971Thr) | missense variant | Likely Benign | PM2, BP4, BP6 | LA1<br>LA2 | 0,9986<br>0,9979 | SA 0,9995 | 1 | 0,0167 |
| rs142638069 | c.5110G>A | p.(Ala1704Thr) | missense variant | Likely benign | BP4, BP6 | LA1<br>LA2 | 0<br>0,0004 | SA 0 | 1 | 0,0167 |
| rs531341535 | c.3443T>A | p.(Leu1148His) | missense variant | VUS | PM2, BP6 | LA1<br>LA2 | 0<br>0 | SA Not Reported | 1 | 0,0167 |

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