### Supplementary Table 3. Twenty-four pathogenic variants in *ARR3* that identified in our cohort (including three previously reported)

| Position (at chrX) | Change | Family Effect | ACMG/AMP Evidence | gnomAD AF | RE VEL | CA DD | SIFT Polyphen2 | PRO VLEAN | BD | NetG HSF |
|------------------|--------|---------------|--------------------|-----------|--------|------|---------------|-----------|----|---------|
| No. | (GRCh37/hg19) | | | | | | | | | | |
| 1 | 69489238 | c.3G>A | p.M1I? | 1 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 0.01 | NA | NA | NA | NSSC | NSSC | NSSC |
| 2 | 6949511 | c.9-1G>A | SA | 1 | PVS1,PS4,PM2,PP1 | P | none | NA | 22.7 | NA | NA | SSC | SSC | SSC |
| 3 | 69489952 | c.103G>A | p.G35S | 1 | PS2,PS4,PM2,PP3 | P | none | 0.54 | 28.6 | D | D | D | NSSC | NSSC | SSC |
| 4 | 69498988 | c.139C>T | p.R47* | 2 | PS1,PS4,PM2,PP4 | P | none | 35.0 | 125.4 | D | D | D | NSSC | NSSC | SSC |
| 5 | 69495932 | c.146T>G | p.L49W | 1 | PS4,PM2,PP1,PP3,PP4 | LP | none | 0.31 | 24.1 | D | Pr | D | NSSC | NSSC | SSC |
| 6 | 69495935 | c.149T>C | p.F50S | 1 | PS1,PS4,PM2,PP1,PP3,PP4 | LP | 5.5E-06 | 0.43 | 23.9 | D | D | D | NSSC | NSSC | SSC |
| 7 | 69496018 | c.232C>T | p.Q78* | 1 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 32.0 | NA | NA | NA | SSC | NSSC | SSC |
| 8 | 69496025 | c.239T>C | p.L80P | 1 | PS1,PS4,PM2,PP1,PP4 | P | none | 0.08 | 9.2 | T | B | D | NSSC | SSC | SSC |
| 9 | 69496084 | c.298C>T | p.R100* | 3 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 34.0 | NA | NA | NA | SSC | SSC | SSC |
| 10 | 69496131 | c.345G>C | p.Q115H | 1 | PVS1,PS4,PM2,PM6,PP3 | P | none | NA | 37.0 | NA | NA | NA | SSC | SSC | SSC |
| 11 | 69496281 | c.361C>A | p.P121T | 1 | PS4,PM2,PM6,PP3 | P | none | 0.63 | 25.3 | D | D | D | NSSC | SSC | SSC |
| 12 | 69496298 | c.389G>T | p.K167* | 1 | PS1,PS4,PM2,PP4 | P | none | 0.37 | 30.0 | NA | NA | NA | SSC | SSC | SSC |
| 13 | 69496323 | c.499A>T | p.K167* | 1 | PS1,PS4,PM2,PP4 | P | none | 37.0 | 30.0 | NA | NA | NA | SSC | SSC | SSC |
| 14 | 69497269 | c.520G>T | p.E174* | 1 | PS1,PS4,PM2,PP4 | P | none | 35.0 | 30.0 | NA | NA | NA | SSC | SSC | SSC |
| 15 | 69497290 | c.520delG | p.E174Rfs*7 | 0 | PVS1,PS4,PM2,PM4,PP4 | P | none | NA | 50.0 | NA | NA | NA | SSC | SSC | SSC |
| 16 | 69497926 | c.707C>G | p.T236R | 1 | PS4,PM2,PM6,PP3 | P | none | 0.35 | 24.7 | D | D | D | SSC | SSC | SSC |
| 17 | 69497928 | c.707C>G | p.T236R | 1 | PS4,PM2,PM6,PP3 | P | none | 0.35 | 24.7 | D | D | D | SSC | SSC | SSC |
| 18 | 69497978 | c.757delC | p.Q253Rfs*7 | 1 | PS1,PS4,PM2,PP4 | P | none | NA | 35.0 | NA | NA | NA | SSC | SSC | SSC |
| 19 | 69498430 | c.844_845insT | p.R282Lfs*10 | 1 | PS1,PS4,PM2,PP1,PP4 | P | none | NA | 35.0 | NA | NA | NA | SSC | SSC | SSC |
| 20 | 69498479 | c.893C>A | p.A298D | 1 | PS4,PM2,PM5,PP1,PP4 | P | none | 0.63 | 26.2 | D | D | D | SSC | SSC | SSC |
| 21 | 69500067 | c.928G>T | p.E310* | 1 | PS1,PS4,PM2,PP1,PP4 | P | none | NA | 35.0 | NA | NA | NA | SSC | SSC | SSC |
| 22 | 69500068 | c.929_930del | p.E310Afs*4 | 6 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 35.0 | NA | NA | NA | SSC | SSC | SSC |
| 23 | 69500102 | c.963_964del | p.R321Sfs*3 | 5 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 35.0 | NA | NA | NA | SSC | SSC | SSC |
| 24 | 69500614 | c.1014-2A>G | SA | 1 | PVS1,PS4,PM2,PP1,PP4 | P | none | NA | 32.0 | NA | NA | NA | SSC | SSC | SSC |

**Abbreviations:** AF = allele Frequency; B = benign; CA = classification; D = damage; N = neutral; NA = not applicable; NSSC = no splicing site change; P = pathogenic; Pr = probably damage; T = tolerant; Un = unknown splicing effect; SA = splicing acceptor; SSC = splicing site change. The p.L80P, p.R100*, and p.A298D were reported before.