

Table S1. Identification of high-confidence heterozygous and homozygous variants

| Type | Total Variants | Total High Confidence | Heterozygous High Confidence | Homozygous High Confidence |
|-----------------------------|-----------------------|------------------------------|-------------------------------------|-----------------------------------|
| Total SNVs | 51,875 | 45,058 | 25,570 | 19,488 |
| Total gene-associated SNVs | 51,829 | 45,020 | 25,555 | 19,465 |
| Total coding | 23,100 | 21,587 | 12,772 | 8,815 |
| Missense: benign | 9,049 | 8,476 | 4,741 | 3,735 |
| Missense: deleterious (LoF) | 2,436 | 2,124 | 1,632 | 492 |
| Nonsense (LoF) | 118 | 105 | 75 | 30 |
| Synonymous | 11,497 | 10,882 | 6,324 | 4,558 |
| 5'UTR | 1,403 | 1,169 | 593 | 576 |
| 3'UTR | 2,130 | 1,882 | 1,039 | 843 |
| Intron | 23,539 | 19,324 | 10,585 | 8,739 |
| Ts/Tv | — | 2.61 | — | — |
| Rare SNV | 5,246 | 2,767 | 2,227 | 540 |