**Table e-1** Clinical data and characteristics of ANXA11 gene mutations in ten Chinese ALS and ALS-FTD patients identified in the current study

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Case number | 534 | 395 | 474 | 506 | 545 | 718 | 660 | 595 | 588 | 479 |
| Sex | Male | Female | Male | Female | Male | Male | Female | Male | Male | Male |
| Age of onset(years) | 71 | 71 | 70 | 59 | 41 | 40 | 61 | 60 | 37 | 55 |
| Site of onset | bulbar | bulbar | bulbar | Lift limb | Right limb | Lift limb | Lift limb | Left limb | bulbar | Right limb |
| Survival time(months)a | 28 | 24 | 39(alive) | 40(alive) | 95(alive) | 21(alive) | 11(alive) | 23(alive) | 18(alive) | 67(alive) |
| Cognitive impairment | No | Yes | No | No | No | No | No | No | No | Yes |
| Sarcoidosis | No | No | No | No | No | No | No | No | No | No |
| Diagnosis | FALS | SALS | ALS-FTD | SALS | SALS | SALS | SALS | SALS | SALS | SALS |
| cDNA Changeb | c.107C>G | c.107C>G | c.107C>G | c.119A>G | c.174-2A>G | c.382G>A | c.687T>A | c.904C>T | c.904C>T | c.1471G>A |
| Protein change | p.P36R | p.P36R | p.P36R | p.D40G | p.A58\_Q187del | p.V128M | p.S229R | p.R302C | p.R302C | p.G491R |
| Mutation in controls | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 | 0/384 |
| ExAC((MAF %) | 0.00090% | 0.00090% | 0.00090% | - | - | - | - | 0.0041% | 0.0041% | 0.0033% |
| dbSNP | rs199988035 | rs199988035 | rs199988035 | - | - | - | - | rs142183550 | rs142183550 | rs777207980 |
| PolyPhen2 | Probably damaging | Probably damaging | Probably damaging | Probably damaging | - | Benign | Benign | Probably damaging | Probably damaging | Probably damaging |
| SIFT | Deleterious | Deleterious | Deleterious | Tolerated | - | Tolerated | Tolerated | Deleterious | Deleterious | Deleterious |
| Mutation Taster | Disease causing | Disease causing | Disease causing | Disease causing | Diseasecausing | Polymorphism | Disease causing | Disease causing | Disease causing | Disease causing |

Abbreviations: cDNA, complementary deoxyribonucleic acid; FALS, familial amyotrophic lateral sclerosis; SALS, sporadic amyotrophic lateral sclerosis; FTD, frontotemporal dementia; ExAC, Exome Aggregation Consortium; MAF, minor allele frequency; dbSNP, database of single Nucleotide Polymorphism; PolyPhen2, Polymorphism Phenotyping v2. SIFT, sorting intolerant from tolerant amino acid substitutions.

a Disease onset to September 2017

b Mutation position based on NM\_145869.