

**Table S1:** Classification of variants associated with arthrogryposis in our cohort, according to the recommendations of the American College of Medical Genetics (ACMG) [28].

| Family<br>(Patients #) | Gene          | Variant  | Classification (class) | ACMG Criteria                              |
|------------------------|---------------|--|------------------------|--|
| Fy1 (1-3)              | <i>TPM2</i>   | NM_003289.4:c.[463G>A];[=]<br>NP_003280.2:p.[(A155T)];[=]                      | likely pathogenic (4)  | PM2, PM5, PP1, PP2, PP3, PP5               |
| Fy2 (4-6)              | <i>TNNT3</i>  | NM_006757.3:c.[187C>T];[=]<br>NP_006748.1:p.[(R63C)];[=]                       | likely pathogenic (4)  | PM5, PM2, PP1, PP3, PP5 (used as moderate) |
| Fy3 (7-8)              | <i>TNNI2</i>  | NM_001145829.2:c.[499_501del];[=]<br>NP_001139301.1:p.[(E167del)];[=]          | likely pathogenic (4)  | PM1, PM2, PM4, PP1, PP3, PP5               |
| Fy4 (9)                | <i>ECEL1</i>  | NM_004826.4:c.[1630C>T];[1700C>G]<br>NP_004817.2:p.[(R544C)];[(P567R)]         | likely pathogenic (4)  | PM2, PM3, PP3, PP5, PP4                    |
| Fy5 (10)               | <i>CHRNA1</i> | NM_005199.4:c.[459dup];[459dup]<br>NP_005190.4:p.[(V154Sfs*24)];[(V154Sfs*24)] | pathogenic (5)         | PVS1, PM2, PM3, PP4, PP5                   |
| Fy6 (11)               | <i>TGFBR2</i> | NM_001024847.2:c.[1595dup];[=]<br>NP_001020018.1:p.[(H532Qfs*9)];[=]           | likely pathogenic (4)  | PM2, PM4, PP3, PP1                         |

## References

[28]. Richards, S.; Aziz, N.; Bale, S.; Bick, D.; Das, S.; Gastier-Foster, J.; Grody, W.W.; Hegde, M.; Lyon, E.; Spector, E.; et al. Standards and guidelines for the interpretation of sequence variants: A joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet. Med.* 2015, 17, 405–424. <https://doi.org/10.1038/gim.2015.30>.