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| **Patient ID** | ***SMN1* variants (CNVs)** | ***SMN1* variants (SNVs)** | **ACMG classification of SNVs** |
| ID1 | heterozygous *SMN1* deletion | heterozygous c.821C>T p.(Thr274Ile) | class 5 (PS3, PS4\_MOD, PM3\_STRG, PM2\_SUP, PP3) |
| ID2 | heterozygous *SMN1* deletion | heterozygous c.821C>T p.(Thr274Ile) | class 5 (PS3, PS4\_MOD, PM3\_STRG, PM2\_SUP, PP3) |
| ID9 | none | heterozygous c.469C>T p.(Gln157\*)  heterozygous c.821C>T p.(Thr274Ile) | class 5 (PS3, PS4\_MOD, PM3\_STRG, PM2\_SUP, PP3)  class 5 (PVS1, PM3\_STRG, PM2\_SUP |
| ID10 | none | heterozygous c.549del p.(Lys184Serfs\*29)  heterozygous c.815A>G p.(Tyr272Cys) | Class 5 (PVS1, PM3, PM2\_SUP)  Class 4 (PS4\_MOD, PS3\_SUP, PM3\_STRG, PM2\_SUP, PP3) |