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| Supplementary Table 1. Participants with respiratory dysfunction who have an in-frame mutation and showed a faint and patchy IHC pattern in dystrophin immunostaining.   |  |  |  |  |  |  |  |  |  | | --- | --- | --- | --- | --- | --- | --- | --- | --- | | Age |  | Age at loss of ambulation | Steroid use | Cardiac function | Cardiac medication | FVC (%) | Ventilator use | Genotyping | | 53 | non-ambulant | 53 | never | decreased | yes | NT | part-time NPPV | ex45\_ex47del | | 44 | ambulant | - | never | normal | no | NT | part-time NPPV | ex14\_ex41del | | 38 | ambulant | - | never | normal | no | 78 | never | ex48\_ex49del | |
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| Supplementary Table 2 Genotyping of deletion/duplication mutations.   |  |  |  |  | | --- | --- | --- | --- | |  | Total | BMD | IMD | | ex45\_ex47del | 36 | 36 | 0 | | ex45\_ex48del | 17 | 17 | 0 | | ex45\_ex49del | 9 | 8 | 1 | | ex3\_ex7del | 7 | 2 | 5 | | ex45\_ex55del | 4 | 4 | 0 | | ex48del | 4 | 4 | 0 | | ex14\_ex41del | 3 | 3 | 0 | | ex48\_ex49del | 3 | 3 | 0 | | ex5\_ex9del | 3 | 2 | 1 | | ex13del | 2 | 2 | 0 | | ex19\_ex44del | 2 | 2 | 0 | | ex45\_ex53del | 2 | 2 | 0 | | ex45\_ex54del | 2 | 1 | 1 | | ex51del | 2 | 0 | 2 | | ex6del | 2 | 0 | 2 | | Dp427c-ex1dep | 2 | 0 | 2 | | ex2dup | 2 | 0 | 2 | | ex44dup | 2 | 1 | 1 | | ex10del | 1 | 1 | 0 | | ex10\_ex25del | 1 | 1 | 0 | | ex10\_ex33del | 1 | 1 | 0 | | ex10\_ex34del | 1 | 0 | 1 | | ex10\_ex42del | 1 | 0 | 1 | | ex10\_ex44del | 1 | 0 | 1 | | ex12\_ex43del | 1 | 1 | 0 | | ex13\_ex18del | 1 | 1 | 0 | | ex13\_ex44del | 1 | 1 | 0 | | ex2\_ex3del | 1 | 1 | 0 | | ex24\_ex48del | 1 | 1 | 0 | | ex3\_ex11del | 1 | 0 | 1 | | ex3\_ex4del | 1 | 1 | 0 | | ex3\_ex5del | 1 | 1 | 0 | | ex34\_ex44del | 1 | 1 | 0 | | ex38\_ex43del | 1 | 1 | 0 | | ex4del | 1 | 1 | 0 | | ex42\_ex43del | 1 | 1 | 0 | | ex44del | 1 | 0 | 1 | | ex45del | 1 | 0 | 1 | | ex45\_ex51del | 1 | 1 | 0 | | ex45\_ex56del | 1 | 0 | 1 | | ex46\_ex47del | 1 | 0 | 1 | | ex46\_ex48del | 1 | 0 | 1 | | ex46\_ex50del | 1 | 0 | 1 | | ex46\_ex52del | 1 | 0 | 1 | | ex46\_ex55del | 1 | 0 | 1 | | ex47\_ex48del | 1 | 0 | 1 | | ex48\_ex54del | 1 | 0 | 1 | | ex48\_ex55del | 1 | 1 | 0 | | ex49\_ex52del | 1 | 0 | 1 | | ex50del | 1 | 0 | 1 | | ex51\_ex55del | 1 | 0 | 1 | | ex52del | 1 | 0 | 1 | | ex53del | 1 | 0 | 1 | | ex7\_ex11del | 1 | 0 | 1 | | ex8\_ex9del | 1 | 0 | 1 | | ex3\_ex7dup,ex25dep | 1 | 1 | 0 | | ex17\_ex51dep | 1 | 0 | 1 | | ex2\_ex7dep | 1 | 1 | 0 | | ex3\_ex13dep | 1 | 1 | 0 | | ex3\_ex16dep | 1 | 0 | 1 | | ex3\_ex4dep | 1 | 0 | 1 | | ex3\_ex5dep | 1 | 0 | 1 | | ex34\_ex44dep | 1 | 0 | 1 | | ex38\_ex43dep | 1 | 0 | 1 | | ex52\_ex71dep | 1 | 0 | 1 | | ex56\_ex67dep | 1 | 1 | 0 | | ex8\_ex12dep | 1 | 1 | 0 | |  | 153 | 108 | 45 | |