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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 |

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