**Table S1. Cohort of internal patients with nonsense mutations in *DMD* gene.**

| **Exon** | **DNAchange** | **Protein change** | **Disease** | **Reference** |
| --- | --- | --- | --- | --- |
| 5 | c.313A>T | p.(Lys105\*) | BMD | [1] |
| 6 | c.433C>T | p.(Arg145\*) | DMD | [1] |
| 6 | c.433C>T | p.(Arg145\*) | DMD | [2] |
| 6 | c.433C>T | p.(Arg145\*) | DMD | [2] |
| 6 | c.433C>T | p.(Arg145\*) | DMD | [2] |
| 7 | c.583C>T | p.(Arg195\*) | DMD | [2] |
| 8 | c.724C>T | p.(Gln242\*) | DMD | [2] |
| 8 | c.701C>A | p.(Ser234\*) | DMD | [2] |
| 8 | c.701C>A | p.(Ser234\*) | DMD | [2] |
| 8 | c.724C>T | p.(Gln242\*) | DMD | [1, 3] |
| 10 | c.1093C>T | p.(Gln365\*) | DMD | [2] |
| 10 | c.1062G>A | p.(Trp354\*) | DMD | [1] |
| 10 | c.1062G>A | p.(Trp354\*) | DMD | [2] |
| 10 | c.1062G>A | p.(Trp354\*) | DMD | [2] |
| 10 | c.1132C>T | p.(Gln378\*) | DMD | Unpublished |
| 11 | c.1292G>A | p.(Trp431\*) | DMD | [2] |
| 14 | c.1652G>A | p.(Trp551\*) | DMD | [2] |
| 16 | c.1865C>G | p.(Ser622\*) | DMD | [1] |
| 16 | c.1873C>T | p.(Gln625\*) | DMD | Unpublished |
| 17 | c.2125C>T | p.(Gln709\*) | DMD | [2] |
| 17 | c.2077T>C | p.(Gln693\*) | DMD | [2] |
| 18 | c.2201G>A | p.(Trp734\*) | DMD | Unpublished |
| 20 | c.2414C>G | p.(Ser805\*) | DMD | [2] |
| 23 | c.3151C>T | p.(Arg1051\*) | DMD | [2] |
| 23 | c.3151C>T | p.(Arg1051\*) | DMD | Unpublished |
| 24 | C.3242C>A | p.(Ser1081\*) | DMD | [2] |
| 24 | c.3259C>T | p.(Gln1087\*) | DMD | [1] |
| 26 | c.3580C>T | p.(Gln1194\*) | DMD | Unpublished |
| 28 | c.3843G>A | p.(Trp1281\*) | DMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | BMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | DMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | DMD | [2] |
| 29 | c.4027G>T | p.(Glu1343\*) | DMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | DMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | DMD | [2] |
| 29 | c.3940C>T | p.(Arg1340\*) | BMD | [2] |
| 29 | c.3940C>T | p.(Arg1314\*) | BMD | [2] |
| 30 | c.4117C>T | p.(Gln1373\*) | DMD | [2] |
| 34 | c.4729C>T | p.(Arg1577\*) | DMD | [2] |
| 35 | c.4996C>T | p.(Arg1666\*) | DMD | [2] |
| 35 | c.4979G>A | p.(Trp1660\*) | BMD | [2] |
| 37 | c.5209C>T | p.(Gln1737\*) | DMD | [2] |
| 39 | c.5530C>T | p.(Arg1844\*) | DMD | [2] |
| 41 | c.5773G>T | p.(Glu1925\*) | BMD | [2] |
| 41 | c.5773G>T | p.(Glu1925\*) | BMD | Unpublished |
| 41 | c.5899C>T | p.(Arg1967\*) | BMD | [1] |
| 41 | c.5899C>T | p.(Arg1967\*) | DMD | [2] |
| 41 | c.5899C>T | p.(Arg1967\*) | DMD | [2] |
| 41 | c.5899C>T | p.(Arg1967\*) | DMD | [2] |
| 42 | c.6023C>A | p.(Ser2008\*) | DMD | [2] |
| 42 | c.6103G>T | p.(Glu2035\*) | DMD | Unpublished |
| 48 | c.7006C>T | p.(Gln2336\*) | DMD | [2] |
| 59 | c.8713C>T | p.(Arg2905\*) | DMD | [2] |
| 59 | c.8713C>T | p.(Arg2905\*) | DMD | Unpublished |
| 60 | c.8944C>T | p.(Arg2982\*) | DMD | [2] |
| 65 | c.9461T>A | p.(Leu3154\*) | DMD | [2] |
| 65 | c.9474T>G | p.(Tyr3158\*) | DMD | [2] |
| 65 | c.9558T>G | p.(Tyr3186\*) | DMD | [2] |
| 70 | c.10108C>T | p.(Arg3370\*) | DMD | [2] |
| 70 | c.10141C>T | p.(Arg3381\*) | DMD | [2] |
| 76 | c.10801C>T | p.(Gln3601\*) | BMD | Unpublished |

**References**

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2. Torella A, Trimarco A, Blanco Fdel V, Cuomo A, Aurino S, Piluso G, et al. One hundred twenty-one dystrophin point mutations detected from stored DNA samples by combinatorial denaturing high-performance liquid chromatography. J Mol Diagn. 2010;12(1):65-73. Epub 2009/12/05. doi: 10.2353/jmoldx.2010.090074. PubMed PMID: 19959795; PubMed Central PMCID: PMCPMC2797720.

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