**Supplementary Material**

**Matrilineal analysis of mutations in *DMD* in a multigenerational South Indian cohort using *DMD* gene panel sequencing**

Arun Shastry1#\*, Sankaramoorthy Aravind 1,3\*, Meeta Sunil2\*, Keerthi Ramesh1, Berty Ashley1, Nithyanandan T.2, Ramprasad VL2, Ravi Gupta2, Somasekar Seshagiri4, Upendra Nongthomba3, Sameer Phalke2#

1. Dystrophy Annihilation Research Trust (DART), Bangalore, India
2. MedGenome Labs, Bangalore, India
3. Indian Institute of Science (IISc), Bangalore, India
4. SciGenom Research Foundation, Cochin, India

\* Equal Contributions

# Corresponding Authors:

Sameer Phalke: sameer.p@medgenome.com

 Arun Shastry: arunshastry@dartindia.in

**Figure Legend**

**Supp. Fig. S1:** QC metrics of the sequenced data: (a) Percentage of raw data above Q30, (b) read alignment percentage across the cohort (c) % *DMD* gene coverage the probands and the other unaffected family members and (d) average depth of *DMD* gene in the proband and the other family members.

**Supp. Fig. S2:** Pedigree diagrams of all the 22 families

**Table Legend:**

**Supp. Table S1**: List of family, sample, relationship with proband and age of ambulation loss

**Supp. Table S2:** List of additional 80 genes, along with associated OMIM phenotype, included in the panel

**Supp. Table S3**: Data quality and processing summary

**Supp. Table S4**: Comparison of *DMD* mutation analysis using Illumina True Sight (TRUPNL) and *DMD* gene panel (MGM *DMD* Panel)

**Supp. Table S5**: List of inherited/*de-novo* *DMD* mutation

Supp. Fig. S1

C

a

c

b

d

Supp. Fig. S2



**Supp. Table S1**

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Family ID** | **Sample ID** | **Sample Info** | **Sample Name** | **Gender** | **Age** | **Age at loss of ambulation** |
| **FM-1** | M-1 | Mother | DMD2-M | F |   |   |
| **FM-1** | S-1 | Sister | DMD2-S | F |   |   |
| **FM-1** | P-1 | Proband | DMD2 | M | 13 | 12 |
| **FM-2** | GM-2 | GrandMother | DMD22-GM | F |   |   |
| **FM-2** | M-2 | Mother | DMD22-M | F |   |   |
| **FM-2** | P-2 | Proband | DMD22 | M | 16 | 10 |
| **FM-3** | GM-3 | GrandMother | DMD12-GM | F |   |   |
| **FM-3** | B-3 | Brother | DMD12-B | M |   |   |
| **FM-3** | M-3 | Mother | DMD12-M | F |   |   |
| **FM-3** | P-3 | Proband | DMD12 | M | 17 | 14 |
| **FM-4** | GGM-4 | Great GrandMother | DMD21-GGM | F |   |   |
| **FM-4** | GM-4 | GrandMother | DMD21-GM | F |   |   |
| **FM-4** | M-4 | Mother | DMD21-G | F |   |   |
| **FM-4** | P-4 | Proband | DMD21 | M | 10 | 7 |
| **FM-5** | GM-5 | GrandMother | DMD3-GM | F |   |   |
| **FM-5** | M-5 | Mother | DMD3-M | F |   |   |
| **FM-5** | P-5 | Proband | DMD3 | M | 8 | 8 |
| **FM-6** | GM-6 | GrandMother | DMD1-GM | F |   |   |
| **FM-6** | M-6 | Mother | DMD1-M | F |   |   |
| **FM-6** | B-6 | Brother | DMD1-B | M |   |   |
| **FM-6** | P-6 | Proband | DMD1 | M | 9 | Still Walking |
| **FM-7** | GM-7 | GrandMother | DMD17-GM | F |   |   |
| **FM-7** | M-7 | Mother | DMD17-M | F |   |   |
| **FM-7** | P-7 | Proband | DMD17 | M | 11 | 8 |
| **FM-8** | GGM-8 | Great GrandMother | DMD15-GGM | F |   |   |
| **FM-8** | GM-8 | GrandMother | DMD15-GM | F |   |   |
| **FM-8** | M-8 | Mother | DMD15-M | F |   |   |
| **FM-8** | P-8a | Proband | DMD15 | M | 15 | 9 |
| **FM-8** | P-8b | Proband | DMD16 | M | 17 | 9 |
| **FM-10** | M-10 | Mother | DMD36-M | F |   |   |
| **FM-10** | P-10 | Proband | DMD36 | M | 11 | Walking |
| **FM-11** | M-11 | Mother | DMD38-M | F |   |   |
| **FM-11** | P-11 | Proband | DMD38 | M | 11 | Walking |
| **FM-12** | M-12 | Mother | DMD40-M | F |   |   |
| **FM-12** | P12 | Proband | DMD40 | M | 13 | Walking |
| **FM-13** | GGM-13 | Great GrandMother | DMD8-GGM | F |   |   |
| **FM-13** | GM-13 | GrandMother | DMD8-GM | F |   |   |
| **FM-13** | M-13 | Mother | DMD8-M | F |   |   |
| **FM-13** | B-13 | Brother | DMD8-B | M |   |   |
| **FM-13** | P-13 | Proband | DMD8 | M | 12 | 8 |
| **FM-14** | GGM-14 | Great GrandMother | DMD20-GGM | F |   |   |
| **FM-14** | GM-14 | GrandMother | DMD20-GM | F |   |   |
| **FM-14** | M-14 | Mother | DMD20-M | F |   |   |
| **FM-14** | P-14 | Proband | DMD20 | M | 17 | 13 |
| **FM-15** | GM-15 | GrandMother | DMD6-GM | F |   |   |
| **FM-15** | M-15 | Mother | DMD6-M | F |   |   |
| **FM-15** | S-15 | Sister | DMD6-S | F |   |   |
| **FM-15** | P-15 | Proband | DMD6 | M | 10 | walking |
| **FM-16** | GGM-16 | Great GrandMother | DMD5-GGM | F |   |   |
| **FM-16** | GM-16 | GrandMother | DMD5-GM | F |   |   |
| **FM-16** | M-16 | Mother | DMD5-M | F |   |   |
| **FM-16** | B-16 | Brother | DMD5-B | M |   |   |
| **FM-16** | P-16 | Proband | DMD5 | M | 10 | 7 |
| **FM-17** | M-17 | Mother | DMD4-M | F |   |   |
| **FM-17** | S17 | Sister | DMD4-S | F |   |   |
| **FM-17** | P-17a | Proband | DMD4 | M | 11 | walking |
| **FM-17** | P-17b | Proband | DMD7 | M | 16 | 14 |
| **FM-18** | GM-18 | GrandMother | DMD14-GM | F |   |   |
| **FM-18** | M-18 | Mother | DMD14-M | F |   |   |
| **FM-18** | S-18 | Sister | DMD14-S | F |   |   |
| **FM-18** | P-18 | Proband | DMD14 | M | 10 | walking |
| **FM-20** | GM-20 | GrandMother | DMD11-GM | F |   |   |
| **FM-20** | M-20 | Mother | DMD11-M | F |   |   |
| **FM-20** | P-20 | Proband | DMD11 | M | 6 | walking |
| **FM-21** | GM-21 | GrandMother | DMD9-GM | F |   |   |
| **FM-21** | M-21 | Mother | DMD9-M | F |   |   |
| **FM-21** | B-21 | Brother | DMD9-B | M |   |   |
| **FM-21** | P-21 | Proband | DMD9 | M | 8 | 8 |
| **FM-22** | GM-22 | GrandMother | DMD10-GM | F |   |   |
| **FM-22** | M-22 | Mother | DMD10-M | F |   |   |
| **FM-22** | P-22 | Proband | DMD10 | M | 7 | walking |
| **FM-23** | M-23 | Mother | DMD82-M | F |   |   |
| **FM-23** | P-23 | Proband | DMD82 | M | 14 | 11 |
| **FM-24** | GM-24 | GrandMother | DMD18-GM | F |   |   |
| **FM-24** | M-24 | Mother | DMD18-M | F |   |   |
| **FM-24** | S-24 | Sister | DMD18-S | F |   |   |
| **FM-24** | P-24 | Proband | DMD18 | M | 7 | NA |

**Supp. Table S2.**

|  |  |  |
| --- | --- | --- |
| # |  **Gene**  | **OMIM phenotype** |
| 1 |  ***ACTA1*** | **congenital actin myopathy with cores, congenital actin myopathy with excess of thin myofilaments, autosomal domiant or recessive nemaline myopathy 3, congenital myopathy with fiber-type disproportion 1** |
| 2 |  ***ANO5*** | **Gnathodiaphyseal dysplasia, Miyoshi muscular dystrophy 3, limb-girdle muscular dystrophy-2L** |
| 3 |  ***B3GALNT2*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 11** |
| 4 |  ***B3GNT1*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 13** |
| 5 |  ***BAG3*** | **dilated cardiomyopathy-1HH, myofibrillar myopathy-6** |
| 6 |  ***BIN1*** | **Autosomal recessive centronuclear myopathy-2** |
| 7 |  ***BVES*** | **Limb-girdle muscular dystrophy type 2X** |
| 8 |  ***CAPN3*** | **limb-girdle muscular dystrophy-2A** |
| 9 |  ***CAV3*** | **Familial hypertrophic cardiomyopathy, elevated serum creatine phosphokinase, long QT syndrome 9, limb-girdle muscular dystrophy type 1C, Tateyama type distal myopathy, rippling muscle disease** |
| 10 |  ***CCDC78*** | **centronuclear myopathy 4** |
| 11 |  ***CFL2*** | **autosomal recessive nemaline myopathy-7** |
| 12 |  ***CHKB*** | **Megaconial type congenital muscular dystrophy** |
| 13 |  ***CNTN1*** | **Compton-North congenital myopathy** |
| 14 |  ***COL12A1*** | **Ullrich congenital muscular dystrophy 2, Bethlem myopathy 2** |
| 15 |  ***COL6A1*** | **Bethlem myopathy, Ullrich congenital muscular dystrophy** |
| 16 |  ***COL6A2*** | **Bethlem myopathy 1, Ullrich congenital muscular dystrophy 1, congenital myosclerosis**  |
| 17 |  ***COL6A3*** | **Bethlem myopathy 1, Ullrich congenital muscular dystrophy 1, dystonia 27** |
| 18 |  ***CRYAB*** | **dilated cardiomyopathy-1II, multiple types of cataract 16, myofibrillar myopathy-2, fatal infantile hypertonic myofibrillar myopathy** |
| 19 |  ***DAG1*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A,9, limb-girdle muscular dystrophy-dystroglycanopathy type C9** |
| 20 |  ***DES*** | **limb-girdle muscular dystrophy type 2R, dilated cardiomyopathy 1I, myofibrillar myopathy-1, neurogenic scapuloperoneal syndrome type Kaeser** |
| 21 |  ***DNAJB6*** | **limb-girdle muscular dystrophy type 1E** |
| 22 |  ***DNM2*** | **axonal Charcot-Marie-Tooth disease type 2M, dominant intermediate B Charcot-Marie-Tooth disease, lethal congenital contracture syndrome 5, centronuclear myopathy** |
| 23 |  ***DYSF*** | **Miyoshi muscular dystrophy 1, distal myopathy with anterior tibial onset, limb-girdle muscular dystrophy type 2B** |
| 24 |  ***EMD*** | **X-linked Emery-Dreifuss muscular dystrophy 1** |
| 25 |  ***FHL1*** | **X-linked myopathy with postural muscle atrophy, childhood-onset X-linked reducing body myopathy, severe early-onset X-linked reducing body myopathy, X-linked Emery-Dreifuss muscular dystrophy 6, X-linked dominant scapuloperoneal myopathy** |
| 26 |  ***FKRP*** | **congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A5, congenital muscular dystrophy-dystroglycanopathy with or without mental retardation type B5, limb-girdle muscular dystrophy-dystroglycanopathy type C5** |
| 27 |  ***FKTN*** | **dilated cardiomyopathy 1X, congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A4, congenital muscular dystrophy-dystroglycanopathy without mental retardation type B4, limb-girdle muscular dystrophy-dystroglycanopathy type C4** |
| 28 |  ***FLNC*** | **distal myopathy-4, myofibrillar myopathy-5** |
| 29 |  ***GMPPB*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 14, muscular dystrophy-dystroglycanopathy (congenital with mental retardation) type B 14, muscular dystrophy-dystroglycanopathy (limb-girdle) type C 14** |
| 30 |  ***GNE*** | **autosomal recessive inclusion body myopathy, Nonaka myopathy, Sialuria** |
| 31 |  ***HNRNPDL*** | **Limb-girdle muscular dystrophy type 1G** |
| 32 |  ***HNRNPA2B1*** | **Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2** |
| 33 |  ***ISCU*** | **Herediary myopathy with lactic acidosis** |
| 34 |  ***ISPD*** | **congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A7, limb-girdle muscular dystrophy-dystroglycanopathy type C7** |
| 35 |  ***ITGA7*** | **congenital muscular dystrophy due to ITGA7 deficiency** |
| 36 |  ***KBTBD13*** | **Autosomal dominant nemaline myopathy-6** |
| 37 |  ***KLHL40*** | **autosomal recessive nemaline myopathy-8** |
| 38 |  ***KLHL41*** | **Nemaline myopathy 9**  |
| 39 |  ***LAMA2*** | **Merosin-deficient congenital muscular dystrophy, congenital muscular dystrophy due to partial LAMA2 deficiency** |
| 40 |  ***LAMP2*** | **Danon disease** |
| 41 |  ***LARGE*** | **congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A6, congenital muscular dystrophy-dystroglycanopathy with mental retardation type B6** |
| 42 |  ***LDB3*** | **dilated cardiomyopathy 1C with or without LVNC, hypertrophic cardiomyopathy 24, left ventricular noncompaction 3, myofibrillar myopathy-4** |
| 43 |  ***LIMS2*** | **Limb-girdle muscular dystrophy type 2W** |
| 44 |  ***LMNA*** | **Dilated cardiomyopathy 1A, Charcot-Marie-Tooth disease type 2B1, Emery-Dreifuss muscular dystrophy 2 AD, Emery-Dreifuss muscular dystrophy 3 AR, Slovenian type heart-hand syndrome, Hutchinson-Gilford progeria syndrome, familial partial lipodystrophy 2, Malouf syndrome, mandibuloacral dysplasia, lethal restrictive dermopathy, limb-girdle muscular dystrophy 1B, congenital muscular dystrophy** |
| 45 |  ***LMOD3*** | **Nemaline myopathy 10**  |
| 46 |  ***MEGF10*** | **Early-onset myopathy, areflexia, respiratory distress and dysphagia, early-onset myopathy, areflexia, respiratory distress and dysphagia mild variant** |
| 47 |  ***MTM1*** | **X-linked myotubular myopathy** |
| 48 |  ***MYF6*** | **Centronuclear myopathy 3** |
| 49 |  ***MYH2*** | **Proximal myopathy and ophthalmoplegia**  |
| 50 |  ***MYH7*** | **dilated cardiomyopathy-1S, hypertrophic cardiomyopathy-1, Laing distal myopathy, left ventricular noncompaction 5, autosomal dominant myosin storage myopathy, autosomal recessive myosin storage myopathy, myopathic type scapuloperoneal syndrome** |
| 51 |  ***MYOT*** | **Limb-girdle muscular dystrophy type 1A, myofibrillar myopathy 3, spheroid body myopathy** |
| 52 |  ***NEB*** | **Autosomal recessive nemaline myopathy-2** |
| 53 |  ***PABPN1*** | **Oculopharyngeal muscular dystrophy**  |
| 54 |  ***PLEC*** | **Epidermolysis bullosa simplex with nail dystrophy, epidermolysis bullosa simplex with pyloric atresia, Ogna type of epidermolysis bullosa simplex, muscular dystrophy with epidermolysis bullosa simplex, limb-girdle muscular dystrophy type 2Q** |
| 55 |  ***POMGNT1*** | **Congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A3, congenital muscular dystrophy-dystroglycanopathy with mental retardation type B3, limb-girdle muscular dystrophy-dystroglycanopathy type C3** |
| 56 |  ***POMGNT2*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies type A 8**  |
| 57 |  ***POMK*** | **Muscular dystrophy-dystroglycanopathy (limb-girdle) type C 12, muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 12** |
| 58 |  ***POMT1*** | **congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A1, congenital muscular dystrophy-dystroglycanopathy with mental retardation type B1, limb-girdle muscular dystrophy-dystroglycanopathy type C1** |
| 59 |  ***POMT2*** | **congenital muscular dystrophy-dystroglycanopathy with brain and eye anomalies type A2, congenital muscular dystrophy-dystroglycanopathy with mental retardation type B2, limb-girdle muscular dystrophy-dystroglycanopathy type C2** |
| 60 |  ***RYR1*** | **Central core disease, King-Denborough syndrome, minicore myopathy with external ophthalmoplegia, congenital neuromuscular disease with uniform type 1 fiber, susceptibility to malignant hyperthermia 1** |
| 61 |  ***SEPN1*** | **Rigid spine muscular dystrophy 1, congenital myopathy with fiber-type disproportion** |
| 62 |  ***SGCA*** | **limb-girdle muscular dystrophy-2D** |
| 63 |  ***SGCB*** | **Limb-girdle muscular dystrophy type 2E** |
| 64 |  ***SGCD*** | **dilated cardiomyopathy 1L, limb-girdle muscular dystrophy type 2F** |
| 65 |  ***SGCG*** | **limb-girdle muscular dystrophy type 2C** |
| 66 |  ***SMCHD1*** | **digenic fascioscapulohumeral muscular dystrophy 2** |
| 67 |  ***SPEG*** | **Centronuclear myopathy 5**  |
| 68 |  ***SYNE1*** | **Autosomal dominant Emery-Dreifuss muscular dystrophy 4, autosomal recessive spinocerebellar ataxia 8** |
| 69 |  ***SYNE2*** | **autosomal dominant Emery-Dreifuss muscular dystrophy-5** |
| 70 |  ***TCAP*** | **hypertrophic cardiomyopathy 25, limb-girdle muscular dystrophy-2G** |
| 71 |  ***TMEM43*** | **Arrhythmogenic right ventricular dysplasia 5, autosomal dominant Emery-Dreifuss muscular dystrophy 7** |
| 72 |  ***TMEM5*** | **Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies) type A 10** |
| 73 |  ***TNNT1*** | **Amish type nemaline myopathy 5** |
| 74 |  ***TNPO3*** | **limb-girdle muscular dystrophy type 1F** |
| 75 |  ***TPM2*** | **distal arthrogryposis multiplex congenita type 1, distal arthrogryposis type 2B, autosomal dominant nemaline myopathy-4, CAP myopathy-2** |
| 76 |  ***TPM3*** | **Congenital myopathy with fiber-type disproportion, autosomal dominant or recessive nemaline myopathy 1, CAP myopathy 1** |
| 77 |  ***TRAPPC11*** | **Limb-girdle muscular dystrophy type 2S** |
| 78 |  ***TRIM32*** | **Bardet-Biedl syndrome 11, limb-girdle muscular dystrophy type 2H** |
| 79 |  ***TTN*** | **Dilated cardiomyopathy-1G, familial hypertrophic cardiomyopathy-9, limb-girdle muscular dystrophy type 2J, early-onset myopathy with fatal cardiomyopathy, proximal myopathy with early respiratory muscle involvement, tardive tibial muscular dystrophy** |
| 80 |  ***VCP*** | **amyotrophic lateral sclerosis-14 with or without frontotemporal dementia, inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1** |

**Supp. Table S3.**

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Family ID** | **Sample ID** | **Sample Name** | **Total data (Mb)** | **Raw data >= Q30 (%)** | **Alignment (%)** | **Duplicate (%)** | **DMD Gene Coverage (%)** | **Average Depth (X)** |
| **FM-1** | M-1 | DMD2-M | 616.24 | 95.53 | 99.9594 | 13.8004 | 94.89174205 | 239.9998846 |
| **FM-1** | S-1 | DMD2-S | 459.22 | 95.475 | 99.9459 | 10.8242 | 94.88548569 | 174.5052257 |
| **FM-1** | P-1 | DMD2 | 579.78 | 95.46 | 99.9415 | 12.104 | 79.8758763 | 140.4494965 |
| **FM-2** | GM-2 | DMD22-GM | 622.36 | 95.415 | 99.7512 | 12.1446 | 94.89200273 | 232.7753287 |
| **FM-2** | M-2 | DMD22-M | 646.3 | 95.385 | 99.9233 | 13.0104 | 94.885225 | 227.586477 |
| **FM-2** | P-2 | DMD22 | 412.16 | 94.61 | 99.9356 | 8.1673 | 73.736882 | 97.36237591 |
| **FM-3** | GM-3 | DMD12-GM | 700.8 | 94.84 | 99.9246 | 8.4269 | 94.88646324 | 260.1294977 |
| **FM-3** | B-3 | DMD12-B | 588.78 | 95.385 | 99.923 | 10.3766 | 94.88470364 | 167.8053247 |
| **FM-3** | M-3 | DMD12-M | 923.72 | 95.8 | 99.9292 | 16.1218 | 94.88587671 | 372.8456717 |
| **FM-3** | P-3 | DMD12 | 700.84 | 88.845 | 99.8838 | 8.1542 | 89.01505894 | 176.9567659 |
| **FM-4** | GGM-4 | DMD21-GGM | 737.72 | 90.1 | 99.9256 | 10.3212 | 94.89584779 | 285.8800075 |
| **FM-4** | GM-4 | DMD21-GM | 688.48 | 89.26 | 99.9281 | 7.5331 | 94.8971512 | 263.5321607 |
| **FM-4** | M-4 | DMD21-G | 554.38 | 89.425 | 99.9381 | 9.4969 | 94.79359531 | 226.2813768 |
| **FM-4** | P-4 | DMD21 | 273.14 | 89.13 | 99.9241 | 8.9085 | 94.85009811 | 82.71054921 |
| **FM-5** | GM-5 | DMD3-GM | 203.54 | 94.185 | 99.9338 | 9.3354 | 94.8799462 | 74.44783983 |
| **FM-5** | M-5 | DMD3-M | 332.12 | 93.945 | 99.943 | 6.6558 | 94.88496432 | 117.0340535 |
| **FM-5** | P-5 | DMD3 | 199.94 | 93.31 | 99.8247 | 8.7824 | 91.26415747 | 50.30468895 |
| **FM-6** | GM-6 | DMD1-GM | 179.6 | 94.345 | 99.9035 | 6.9397 | 94.8755146 | 63.40615874 |
| **FM-6** | M-6 | DMD1-M | 472.54 | 89.07 | 99.4265 | 3.9185 | 94.87564494 | 190.5725084 |
| **FM-6** | B-6 | DMD1-B | 195.86 | 88.78 | 93.7172 | 3.4731 | 94.82631089 | 57.99058743 |
| **FM-6** | P-6 | DMD1 | 269.18 | 95.22 | 99.8841 | 7.9338 | 91.05424335 | 73.30130595 |
| **FM-7** | GM-7 | DMD17-GM | 348.06 | 95.395 | 99.3512 | 8.5707 | 94.86632556 | 126.5361295 |
| **FM-7** | M-7 | DMD17-M | 236.06 | 91.315 | 99.1837 | 5.255 | 94.86156812 | 75.42283 |
| **FM-7** | P-7 | DMD17 | 890.36 | 90.555 | 99.4234 | 5.5233 | 94.77756337 | 249.8065219 |
| **FM-8** | GGM-8 | DMD15-GGM | 366.52 | 88.81 | 99.1506 | 1.9999 | 94.77091598 | 141.1786245 |
| **FM-8** | GM-8 | DMD15-GM | 332.3 | 88.57 | 99.2502 | 1.8521 | 94.85837477 | 128.5095674 |
| **FM-8** | M-8 | DMD15-M | 476.16 | 89.775 | 99.5033 | 2.5347 | 94.8697796 | 177.5589757 |
| **FM-8** | P-8a | DMD15 | 328.88 | 89.78 | 99.4766 | 2.7569 | 93.19470268 | 92.72466644 |
| **FM-8** | P-8b | DMD16 | 686.08 | 95.985 | 99.9615 | 8.9691 | 93.24266816 | 170.1386039 |
| **FM-10** | M-10 | DMD36-M | 242.88 | 93.115 | 97.7913 | 3.9908 | 94.85928715 | 82.43431369 |
| **FM-10** | P-10 | DMD36 | 265.84 | 94.03 | 98.6719 | 5.9279 | 89.50064421 | 61.60802952 |
| **FM-11** | M-11 | DMD38-M | 536.82 | 91.01 | 98.9455 | 3.7251 | 94.89167688 | 202.1455596 |
| **FM-11** | P-11 | DMD38 | 814.46 | 90.455 | 99.2182 | 5.493 | 88.67545556 | 217.8007647 |
| **FM-12** | M-12 | DMD40-M | 671.26 | 89.325 | 99.5744 | 3.2374 | 94.88476881 | 263.4188129 |
| **FM-12** | P12 | DMD40 | 414.04 | 89.225 | 97.2542 | 3.068 | 75.80923818 | 97.46815021 |
| **FM-13** | GGM-13 | DMD8-GGM | 268 | 89.715 | 99.1233 | 1.6169 | 94.88157546 | 102.8710328 |
| **FM-13** | GM-13 | DMD8-GM | 2298.56 | 93.285 | 99.932 | 42.2006 | 94.88392159 | 586.7483976 |
| **FM-13** | M-13 | DMD8-M | 2882.52 | 93.42 | 99.9287 | 41.2517 | 94.88809251 | 744.8692889 |
| **FM-13** | B-13 | DMD8-B | 1017.02 | 92.275 | 99.8533 | 29.6446 | 94.84182146 | 191.8869944 |
| **FM-13** | P-13 | DMD8 | 2300.62 | 92.89 | 99.9212 | 32.5248 | 93.63760128 | 476.6825989 |
| **FM-14** | GGM-14 | DMD20-GGM | 2122.26 | 93.27 | 99.9239 | 35.268 | 94.88561603 | 631.5439709 |
| **FM-14** | GM-14 | DMD20-GM | 353.3 | 89.715 | 98.0939 | 4.1612 | 94.86502215 | 120.2366392 |
| **FM-14** | M-14 | DMD20-M | 2602.7 | 93.4 | 99.9478 | 40.7355 | 94.8845733 | 791.0553649 |
| **FM-14** | P-14 | DMD20 | 400.46 | 96.77 | 98.1195 | 23.4477 | 94.87401568 | 96.92320375 |
| **FM-15** | GM-15 | DMD6-GM | 541.02 | 96.77 | 99.6938 | 23.4708 | 94.87395051 | 163.7359338 |
| **FM-15** | M-15 | DMD6-M | 504.2 | 96.905 | 99.7875 | 24.3976 | 94.87590563 | 174.8572473 |
| **FM-15** | S-15 | DMD6-S | 1326.12 | 91.195 | 99.9074 | 32.6692 | 94.87740455 | 354.9732697 |
| **FM-15** | P-15 | DMD6 | 896.52 | 90.01 | 99.9445 | 29.7042 | 90.69130893 | 158.4958516 |
| **FM-16** | GGM-16 | DMD5-GGM | 686.38 | 96.36 | 99.7475 | 20.5906 | 94.75462336 | 244.4608557 |
| **FM-16** | GM-16 | DMD5-GM | 1215.46 | 93.305 | 99.8571 | 23.5421 | 94.86645591 | 266.7045607 |
| **FM-16** | M-16 | DMD5-M | 1431.2 | 90.82 | 99.9392 | 27.9926 | 94.88555086 | 396.1309614 |
| **FM-16** | B-16 | DMD5-B | 781.38 | 93.555 | 99.931 | 26.6641 | 94.85029363 | 154.1912728 |
| **FM-16** | P-16 | DMD5 | 1253.76 | 92.955 | 99.9313 | 27.1232 | 92.74268021 | 246.1972222 |
| **FM-17** | M-17 | DMD4-M | 479.22 | 93.31 | 99.9169 | 25.518 | 94.86971443 | 143.1185477 |
| **FM-17** | S17 | DMD4-S | 1625.94 | 92.68 | 99.9093 | 31.4273 | 94.86678176 | 476.9069828 |
| **FM-17** | P-17a | DMD4 | 1109.62 | 92.35 | 99.8595 | 32.5724 | 79.12876188 | 183.6667775 |
| **FM-17** | P-17b | DMD7 | 1101.12 | 93.65 | 99.958 | 23.2386 | 77.42331552 | 222.9311989 |
| **FM-18** | GM-18 | DMD14-GM | 1987.04 | 93.21 | 99.9093 | 37.5393 | 94.87075716 | 523.8385994 |
| **FM-18** | M-18 | DMD14-M | 1941.38 | 94.04 | 99.9627 | 20.911 | 94.86130744 | 642.1905155 |
| **FM-18** | S-18 | DMD14-S | 1787.78 | 93.825 | 99.9549 | 23.6956 | 94.84899022 | 562.3562968 |
| **FM-18** | P-18 | DMD14 | 1420.56 | 92.65 | 99.8817 | 35.4337 | 93.61088139 | 244.4519977 |
| **FM-20** | GM-20 | DMD11-GM | 1267.4 | 92.24 | 99.9436 | 29.1402 | 94.8845733 | 388.1242944 |
| **FM-20** | M-20 | DMD11-M | 1515.16 | 92.96 | 99.9327 | 29.7981 | 94.88691944 | 449.6148587 |
| **FM-20** | P-20 | DMD11 | 596.06 | 93.22 | 99.8052 | 24.8723 | 86.46695824 | 113.9391594 |
| **FM-21** | GM-21 | DMD9-GM | 1000.28 | 93.845 | 99.9385 | 31.9625 | 94.76922155 | 294.7718401 |
| **FM-21** | M-21 | DMD9-M | 974.06 | 91.805 | 99.7647 | 20.6374 | 94.88151029 | 262.560777 |
| **FM-21** | B-21 | DMD9-B | 691.16 | 93.695 | 99.8847 | 26.0988 | 94.75410199 | 139.820325 |
| **FM-21** | P-21 | DMD9 | 930.62 | 93.035 | 99.9251 | 25.0032 | 93.69723227 | 168.1326676 |
| **FM-22** | GM-22 | DMD10-GM | 1138.82 | 93.015 | 99.9209 | 32.2722 | 94.87310329 | 335.8623274 |
| **FM-22** | M-22 | DMD10-M | 926.9 | 93.415 | 99.915 | 24.2284 | 94.87584045 | 263.0187274 |
| **FM-22** | P-22 | DMD10 | 262 | 89.725 | 99.2486 | 9.6864 | 92.89693875 | 63.23886872 |
| **FM-23** | M-23 | DMD82-M | 983.64 | 93.255 | 99.8779 | 24.3421 | 94.87681801 | 224.1856792 |
| **FM-23** | P-23 | DMD82 | 628.5 | 92.89 | 99.897 | 29.2968 | 86.87740194 | 114.6860621 |
| **FM-24** | GM-24 | DMD18-GM | 1665.36 | 93.1 | 99.9256 | 30.924 | 94.88085858 | 502.396346 |
| **FM-24** | M-24 | DMD18-M | 1913.72 | 91.65 | 99.9445 | 26.9243 | 94.86704244 | 629.6313136 |
| **FM-24** | S-24 | DMD18-S | 1405.34 | 92.4 | 99.9318 | 29.2779 | 94.88131478 | 431.6946509 |
| **FM-24** | P-24 | DMD18 | 807.64 | 92.885 | 99.8153 | 26.5219 | 94.8572017 | 159.086839 |

**Supp. Table S3**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Fam ID** | **Sam ID** | **Sam Name** | **Align %** | **DMD CaptureRegionSize (bp)** | **Exons affected** | **DEL Start** | **DEL End** | **DEL Len (bp)** | **Tool for DEL prediction** |
| **TRUPNL** | **MGM DMD Panel** | **TRUPNL** | **MGM DMD Panel** | **TRUPNL** | **MGM DMD Panel** | **Exon coords for ENST00000357033** | **TRUPNL** | **MGM DMD Panel** | **Brkpt in Exon / Intron** | **TRUPNL** | **MGM DMD Panel** | **Brkpt in Exon / Intron** | **TRUPNL** | **MGM DMD Panel** | **TRUPNL** | **MGM DMD Panel** |
| **FM-1** | P-1 | DMD2 | 99.87% | 99.93% | 15622 (0.69 %) | 1534437 (68.44 %) | 46-55 | 46-55 | 31645790-31950344 | 31645790 | 31588767 | Intron | 31950344 | 31954409 | Intron | 304555 | 365643 | BEDTools | SoftSV |
| **FM-2** | P-2 | DMD22 | 99.81% | 99.93% | 15622 (0.69 %) | 1534437 (68.44 %) | 33-45 | 33-45 | 31986456-32404582 | 31951085 | 31951085 | Intron | 32404576 | 32404576 | Exon | 453492 | 453492 | SoftSV | SoftSV |
| **FM-5** | P-5 | DMD3 | 99.87% | 99.81% | 15622 (0.69 %) | 1534437 (68.44 %) | 46-48 | 46-48 | 31893305-31950344 | 31893308 | 31877001 | Intron | 31950344 | 31982603 | Intron | 57037 | 105603 | BEDTools | SoftSV |
| **FM-6** | P-6 | DMD1 | 99.83% | 99.87% | 15622 (0.69 %) | 1534437 (68.44 %) | 46-49 | 46-49 | 31854835-31950344 | 31854835 | 31847041 | Intron | 31950344 | 31963099 | Intron | 95510 | 116059 | BEDTools | SoftSV |
| **FM-7** | P-7 | DMD17 | 99.80% | 99.39% | 15622 (0.69 %) | 1534437 (68.44 %) | SNV |   |   |   |   |   |   |   |   |   |   |   |   |
| **FM-8** | P-8a | DMD18 | 99.72% | 99.46% | 15622 (0.69 %) | 1534437 (68.44 %) | 51 | 51 | 31792077-31792309 | 31792077 | 31789297 | Intron | 31792309 | 31826726 | Intron | 233 | 37430 | BEDTools | SoftSV |
| **FM-13** | P-13 | DMD19 | 99.81% | 99.88% | 15622 (0.69 %) | 1534437 (68.44 %) | 46-47 | 46-47 | 31947713-31950344 | 31947713 | 31940865 | Intron | 31950344 | 31979547 | Intron | 2632 | 38683 | BEDTools | ExomeDepth |
| **FM-14** | P-14 | DMD20 | 99.81% | 97.56% | 15622 (0.69 %) | 1534437 (68.44 %) | SNV |   |   |   |   |   |   |   |   |   |   |   |   |
| **FM-15** | P-15 | DMD21 | 99.82% | 99.92% | 15622 (0.69 %) | 1534437 (68.44 %) | 18-29 | 18-29 | 32456358-32536248 | 32456358 | 32446399 | Intron | 32536248 | 32542070 | Intron | 79891 | 95672 | BEDTools | ExomeDepth |
| **FM-17** | P-17a | DMD22 | 99.84% | 99.79% | 15622 (0.69 %) | 1534437 (68.44 %) | 45-52 | 45-52 | 31747748-31986631 | 31747748 | 31728909 | Intron | 31986631 | 32130344 | Intron | 238884 | 401436 | BEDTools | ExomeDepth |
| **FM-18** | P-18 | DMD23 | 99.84% | 99.82% | 15622 (0.69 %) | 1534437 (68.44 %) | 51 | 51 | 31792077-31792309 | 31792077 | 31778073 | Intron | 31792309 | 31813704 | Intron | 233 | 35631 | BEDTools | ExomeDepth |
| **FM-20** | P-20 | DMD24 | 99.74% | 99.74% | 15622 (0.69 %) | 1534437 (68.44 %) | 48-52 | 48-52 | 31747748-31893490 | 31747748 | 31743342 | Intron | 31893490 | 31944322 | Intron | 145743 | 200980 | BEDTools | ExomeDepth |
| **FM-21** | P-21 | DMD25 | 99.68% | 99.90% | 15622 (0.69 %) | 1534437 (68.44 %) | 49-50 | 49-50 | 31838092-31854936 | 31838092 | 31834919 | Intron | 31854939 | 31864242 | Intron | 16848 | 29323 | BEDTools | ExomeDepth |
| **FM-22** | P-22 | DMD26 | 99.77% | 99.17% | 15622 (0.69 %) | 1534437 (68.44 %) | 49-50 | 49-50 | 31838092-31854936 | 31838092 | 31807504 | Intron | 31854939 | 31856691 | Intron | 16848 | 49187 | BEDTools | ExomeDepth |

**Supp. Table S4**

|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Family ID | **Sample ID** |  Sample Name | **MLPA** | **mPCR** | **Exons** | **Variant Caller** | **Mutation** | **Zygosity** | **Inherited/ De novo** | **Reported in Databases** |
| FM-2 | GM-2 | DMD22-GM |   |   |   |   |   |   |   |   |
| FM-2 | M-2 | DMD22-M |   |   | 33-45 | SoftSV | chrX:31951084-32404576 | Heterozygous |   |   |
| FM-2 | P-2 | DMD22 | 33-45 | Not Done | 33-45 | SoftSV | chrX:31951084-32404576 | Hemizygous | Inherited | No |
| FM-3 | GM-3 | DMD12-GM |   |   |   |   |   |   |   |   |
| FM-3 | B-3 | DMD12-B |   |   |   |   |   |   |   |   |
| FM-3 | M-3 | DMD12-M |   |   | 46-50 | SoftSV | chrX:31798830- 31950822 | Heterozygous |   |   |
| FM-3 | P-3 | DMD12 | 46-50 | Not Done | 46-50 | SoftSV | chrX:31798830- 31950822 | Hemizygous | Inherited | Yes |
| FM-4 | GGM-4 | DMD21-GGM |   |   | 7 | HC | chrX:32827676G>A (p.Arg195Ter) | Heterozygous |   |   |
| FM-4 | GM-4 | DMD21-GM |   |   | 7 | HC | chrX:32827676G>A (p.Arg195Ter) | Heterozygous |   |   |
| FM-4 | M-4 | DMD21-G |   |   | 7 | HC | chrX:32827676G>A (p.Arg195Ter) | Heterozygous |   |   |
| FM-4 | P-4 | DMD21 | Negative | Not Done | 7 | HC | chrX:32827676G>A (p.Arg195Ter) | Hemizygous | Inherited | Yes |
| FM-5 | GM-5 | DMD3-GM |   |   |   |   |   |   |   |   |
| FM-5 | M-5 | DMD3-M |   |   | 46-48 | SoftSV | chrX:31877000-31982603 | Heterozygous |   |   |
| FM-5 | P-5 | DMD3 | 46-48 | Not Done | 46-48 | SoftSV | chrX:31877000-31982603 | Hemizygous | Inherited | Yes |
| FM-6 | GM-6 | DMD1-GM |   |   | 46-48 | SoftSV | chrX:31847040-31963099 | Heterozygous |   |   |
| FM-6 | M-6 | DMD1-M |   |   | 46-48 | SoftSV | chrX:31847040-31963099 | Heterozygous |   |   |
| FM-6 | B-6 | DMD1-B |   |   |   |   |   |   |   |   |
| FM-6 | P-6 | DMD1 | 46-49 | Not Done | 46-49 | SoftSV | chrX:31847040-31963099 | Hemizygous | Inherited | Yes |
| FM-8 | GGM--8 | DMD15-GGM |   |   |   |   |   |   |   |   |
| FM-8 | GM-8 | DMD15-GM |   |   |   |   |   |   |   |   |
| FM-8 | M-8 | DMD15-M |   |   | 51 | SoftSV | chrX:31789296-31826726 | Heterozygous |   |   |
| FM-8 | P-8a | DMD15 | 51 | Positive | 51 | SoftSV | chrX:31789296-31826726 | Hemizygous | Inherited | Yes |
| FM-8 | P-8b | DMD16 | 51 | Positive | 51 | SoftSV | chrX:31789296-31826726 | Hemizygous | Inherited | Yes |
| FM-11 | M-11 | DMD38-M |   |   | 45-50 | SoftSV | chrX:31829985-31993786 | Heterozygous |   |   |
| FM-11 | P-11 | DMD38 | 45-50 | Not Done | 45-50 | SoftSV | chrX:31829985-31993786 | Hemizygous | Inherited |   |
| FM-17 | M-17 | DMD4-M |   |   | 45-52 | SoftSV | chrX:31728694-32130548 | Heterozygous |   |   |
| FM-17 | S-17 | DMD4-S |   |   |   |   |   | Heterozygous |   |   |
| FM-17 | P-17a | DMD4 |   |   | 45-52 | **ED** | **chrX:31728909-32130344** | Hemizygous | Inherited | Yes |
| FM-17 | P-17b | DMD7 | 45-52 | Not Done | 45-52 | SoftSV**ED** | chrX:31728694-32130548**chrX:31728909-32130344** | Hemizygous | Inherited | Yes |
| FM-22 | GM-22 | DMD10-GM |   |   | 49-50 | ED | chrX:31807843-31856691 | Heterozygous |   |   |
| FM-22 | M-22 | DMD10-M |   |   | 49-50 | ED | chrX:31810391-31856451 | Heterozygous |   |   |
| FM-22 | P-22 | DMD10 | 49-50 | Not Done | 49-50 | ED | chrX:31807504-31856691 | Hemizygous | Inherited | Yes |
| FM-23 | M-23 | DMD82-M |   |   | 48-54 | ED | chrX:31662823-31924904 | Heterozygous |   |   |
| FM-23 | P-23 | DMD82 | 48-54 | Not Done | 48-54 | ED | chrX:31662823-31924904 | Hemizygous | Inherited | Yes |
| FM-14 | GGM-14 | DMD20-GGM |   |   |   |   |   |   |   |   |
| FM-14 | GM-14 | DMD20-GM |   |   |   |   |   |   |   |   |
| FM-14 | M-14 | DMD20-M |   |   | 51 | HC | chrX:31792270\_31792271insC (p.Val2327GlyfsTer3) | Heterozygous |   |   |
| FM-14 | P-14 | DMD20 | Negative | Not Done | 51 | HC | chrX:31792270\_31792271insC (p.Val2327GlyfsTer3) | Hemizygous | Inherited | Yes |
| FM-1 | M-1 | DMD2-M |   |   |   |   |   |   |   |   |
| FM-1 | S-1 | DMD2-S |   |   |   |   |   |   |   |   |
| FM-1 | P-1 | DMD2 | 46-55 | Not Done | 46-55 | SoftSV | chrX:31588766-31954409 | Hemizygous | De novo |   |
| FM-7 | GM-7 | DMD17-GM |   |   |   |   |   |   |   |   |
| FM-7 | M-7 | DMD17-M |   |   |   |   |   |   |   |   |
| FM-7 | P-7 | DMD17 | Positive | Not Done | 21 | HC | chrX:32503176delG (p.Glu765AsnfsTer3) |  Hemizygous | De novo | No |
| FM-10 | M-10 | DMD36-M |   |   |   |   |   |   |   |   |
| FM-10 | P-10 | DMD36 | 1 | Not Done | 1 | SoftSV | chrX:33122034-33245415 | Hemizygous | De novo |   |
| FM-12 | M-12 | DMD40-M |   |   |   |   |   |   |   |   |
| FM-12 | P-12 | DMD40 | 35-45 | Not Done | 35-45 | SoftSV | chrX:31959962-32394772 | Hemizygous | De novo |   |
| FM-13 | GGM-13 | DMD8-GGM |   |   |   |   |   |   |   |   |
| FM-13 | GM-13 | DMD8-GM |   |   |   |   |   |   |   |   |
| FM-13 | M-13 | DMD8-M |   |   |   |   |   |   |   |   |
| FM-13 | B-13 | DMD8-B |   |   |   |   |   |   |   |   |
| FM-13 | P-13 | DMD8 | 46-47 | Not Done | 46-47 | ED | chrX:31940865-31979547 | Hemizygous | De novo | Yes |
| FM-15 | GM-15 | DMD6-GM |   |   |   |   |   |   |   |   |
| FM-15 | M-15 | DMD6-M |   |   |   |   |   |   |   |   |
| FM-15 | S-15 | DMD6-S |   |   |   |   |   |   |   |   |
| FM-15 | P-15 | DMD6 | 18-29 | Not Done | 18-29 | ED | chrX:32446399-32542070 | Hemizygous | De novo | No |
| FM-16 | GGM-16 | DMD5-GGM |   |   |   |   |   |   |   |   |
| FM-16 | GM-16 | DMD5-GM |   |   |   |   |   |   |   |   |
| FM-16 | M-16 | DMD5-M |   |   |   |   |   |   |   |   |
| FM-16 | B-16 | DMD5-B |   |   |   |   |   |   |   |   |
| FM-16 | P-16 | DMD5 | 8-9 | Not Done | 8-9 | ED | chrX:32675098-32721781 | Hemizygous | De novo | No |
| FM-18 | GM-18 | DMD14-GM |   |   |   |   |   |   |   |   |
| FM-18 | M-18 | DMD14-M |   |   |   |   |   |   |   |   |
| FM-18 | S-18 | DMD14-S |   |   |   |   |   |   |   |   |
| FM-18 | P-18 | DMD14 | 51 | Not Done | 51 | ED | chrX:31778073-31813704 | Hemizygous | De novo | Yes |
| FM-20 | GM-20 | DMD11-GM |   |   |   |   |   |   |   |   |
| FM-20 | M-20 | DMD11-M |   |   |   |   |   |   |   |   |
| FM-20 | P-20 | DMD11 | 48-52 | Not Done | 48-52 | ED | chrX:31743342-31944322 | Hemizygous | De novo | Yes |
| FM-21 | GM-21 | DMD9-GM |   |   |   |   |   |   |   |   |
| FM-21 | M-21 | DMD9-M |   |   |   |   |   |   |   |   |
| FM-21 | B-21 | DMD9-B |   |   |   |   |   |   |   |   |
| FM-21 | P-21 | DMD9 | 49-50 | Not Done | 49-50 | ED | chrX:31834919-31864242 | Hemizygous | De novo | Yes |
| FM-24 | GM-24 | DMD18-GM |   |   |   |   |   |   |   |   |
| FM-24 | M-24 | DMD18-M |   |   |   |   |   |   |   |   |
| FM-24 | S-24 | DMD18-S |   |   |   |   |   |   |   |   |
| FM-24 | P-24 | DMD18 | Positive | Not Done | 46 | HC | chrX:31950337C>A (p.Glu2200Ter) | Hemizygous | De novo | Yes |