
eFigure 1. Comparative Immunohistochemical Analysis of Dystrophin Protein Expression in Patients With IF or OOF DMD Deletions Around Exons 44 and 45

eFigure 2. Quantitative RT-PCR Analyses of Dystrophin mRNA Expression

eFigure 3. Comparative Immunohistochemical Analysis of Dystrophin-Associated Protein Expression in Patients With IF or OOF DMD Deletions Around Exons 44 and 45

This supplementary material has been provided by the authors to give readers additional information about their work.
eFigure 1. Comparative Immunohistochemical Analysis of Dystrophin Protein Expression in Patients With IF or OOF DMD Deletions Around Exons 44 and 45

Transverse muscle sections were immunolabelled for β-spectrin, MANDYS106, Dys2 and MANEX50. Protein expression was quantified relative to control muscle in 40 muscle fibres and normalised to β-spectrin expression. Patients were grouped according to corresponding exon skipping models for DMD: control (Ctrl, black), exon 44 or 45 skippable OOF deletions (red) and IF deletions mimicking exon 45 skipping (IF, blue). Lines represent means.
eFigure 2. Quantitative RT-PCR Analyses of Dystrophin mRNA Expression

*DMD* mRNA transcript levels were quantified using three separate TaqMan assays targeting exon boundaries 19-20, 53-54 and 73-74. The mean PCR efficiency (E) per amplicon (table 2) and the mean Ct value per sample were used to calculate the starting concentration (N0) using the equation \( N_0 = N_t / E^{C_t} \) where \( N_t \) is the fluorescence threshold. Data was normalised to myotilin and dystrophin expression (n0) presented relative to control ±SD.
eFigure 3. Comparative Immunohistochemical Analysis of Dystrophin-Associated Protein Expression in Patients With IF or OOF DMD Deletions Around Exons 44 and 45

Transverse muscle sections were immunolabelled for β-spectrin, β-dystroglycan, neuronal nitric oxide synthase (nNOS) and utrophin. Expression was quantified relative to control muscle in 40 muscle fibres and normalised to β-spectrin expression. Patients were grouped according to corresponding exon skipping models for DMD: control (Ctrl, black), exon 44 or 45 skippable OOF deletions (red) and IF deletions mimicking exon 45 skipping (model 45 IF, blue). Lines represent means.