



Заключение

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Обследован в лаборатории селективного скрининга

Заключение:

Методом массового параллельного секвенирования на приборе Ion S5 проведен анализ 82 генов, ответственных за метаболические миопатии и поясно-конечностные мышечные дистрофии: ACAD9, ACADL, ACADM, ACADVL, AGL, AHCY, ALDOA, AMPD1, ANO5, AUH, B3GALNT2, B4GAT1, CAPN3, CAV3, CLPB, COQ2, CPT2, DNAJC19, DOLK, DPM1, DPM2, DPM3, DYSL, ENO3, ETF, ETFB, ETFDH, FKBP, FKTN, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, HTRA2, ISCU, ISPD, LAMP2, LDHA, LPIN1, MYH3, OPA1, OPA3, PFKFB3, PGAM2, PGK1, PGM1, PHKA1, POLG, POLG2, POMGNT1, POMK, POMT1, POMT2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SERAC1, SGCA, SLC6A1, SLC6A7, SLC6A8, SLC6A12, SLC6A14, SLC6A15, SLC6A16, SLC6A17, SLC6A18, SLC6A19, SLC6A20, SLC6A21, SLC6A22, SLC6A23, SLC6A24, SLC6A25, SLC6A26, SLC6A27, SLC6A28, SLC6A29, SLC6A30, SLC6A31, SLC6A32, SLC6A33, SLC6A34, SLC6A35, SLC6A36, SLC6A37, SLC6A38, SLC6A39, SLC6A40, SLC6A41, SLC6A42, SLC6A43, SLC6A44, SLC6A45, SLC6A46, SLC6A47, SLC6A48, SLC6A49, SLC6A50, SLC6A51, SLC6A52, SLC6A53, SLC6A54, 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