# Genomic screening for Duchenne muscular dystrophy: a retrospective study from 10,481 NICU patients based on next generation sequencing data

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**Abstract**

Newborn creatine kinase screening can identify patients at risk for Duchenne muscular dystrophy. However, it is unclear whether the next-generation sequencing-based screening can identify patients early and guide care. Herein, this study investigates clinical utility of next-generation sequencing-based DMD screening. A total of 19 (0.18%, 19/10481) newborns were identified with pathogenic variants of DMD gene, including 4 (21.1%, 4/19) duplications,13 (68.4%,13/19) deletions, and 2 (10.5%, 2/19) nonsense mutations. Six of them were symptomatic after regular follow up. Therapeutic strategies for these patients were modified. Two neonates died, and the remaining 11 newborns were asymptomatic at August 1, 2020. These 13 families were informed the updated genetic report and suggested for further genetic consulting. Genomic screening for DMD would identify patients who might not come to clinical attention prior to disease manifestation. Early targeted intervention of DMD have the positively impact the clinical decision and the potential to improve outcomes.

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