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

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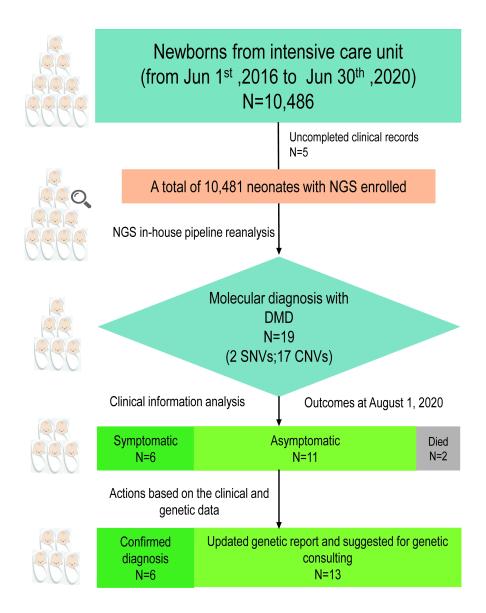
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Abstract

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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Brief report main.pdf available at https://authorea.com/users/361773/articles/483112genomic-screening-for-duchenne-muscular-dystrophy-a-retrospective-study-from-10-481nicu-patients-based-on-next-generation-sequencing-data



neonate_DMD5	CK and AST elevated		
	Preterm infant	3 months ; clinical diagnosis with pseudohypertrophy of the calf and persistent elevated CK	2y, no motor dysfunct
neonate_DMD10	CK and AST elevated		*
	Congenital laryngomalacia Term infant	4 months ; clinical diagnosis with persistent elevated CK	1y, no motor dysfunction
	CK, AST,		
neonate_DMD11	and ALT elevated UTI Term infant	2 months ; clinical diagnosis with pseudohypertrophy of the 10m, no moto calf and persistent elevated CK	r dysfunction
neonate_DMD16	CK elevated Hypotonia		-
	Term infant	1 month ; clinical diagnosis with muscular weakness and persistent elevated CK	3y, no motor dysfund
neonate_DMD18	CK, AST, and ALT elevated Vomiting Term infant		
		6 months ; clinical diagnosis with delayed milestone and persistent elevated CK	3y, no motor dysfund
neonate_DMD19	CK, AST, and ALT elevated Hyperbilirubinemia Term infant		A
		1 month ; clinical diagnosis with persistent elevated CK	3y, no motor dysfund
neonate_DMD1	CK elevated Term infant	4y, no m	otor dysfunction;
		Updated	genetic report and d for further genetic consult
	CK elevated Cardiomegaly,		
neonate_DMD2	Cardiomegaly, Suspected Myocarditis Term infant		n and cardiac dysfunction and suggested for further
neonate_DMD3	CK elevated Term infant		
		3y, no motor Updated gen further geneti	etic report and suggested for
neonate_DMD4	Poor feeding Term infant		
neonale_DMD4		3y, no motor Updated gen further genet	etic report and suggested for
neonate_DMD6	Normal CK	Dead Updated genetic	report and suggested for
		further genetic c	onsulting
neonate_DMD7	CK elevated Preterm infant	9m no motor dysfunction;	
		Updated genetic report and suggested for further genetic consulting	•
neonate_DMD8	CK elevated Term infant		- 🔁
		1y, no motor dysfunction Updated genetic report and s further genetic consulting	uggested for
21/20	CK elevated		*
neonate_DMD9	CK elevated Term infant	1y, no motor dysfunction	
neonate_DMD9		1y, no motor dysfunctio Updated genetic report suggested for further ge	
	Term infant CK elevated	Updated genetic report suggested for further ge	
	Term infant	Updated genetic report	enetic consulting
neonate_DMD12	Term infant CK elevated Asphyxia Term infant Normal CK	Updated genetic report suggested for further ge 9m, no motor dysfunction Updated genetic report ar further genetic consulting	enetic consulting
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