REVIEW OPEN ACCESS

Newborn Screening for Duchenne Muscular Dystrophy

Contemporary Considerations for Implementation of an Early Diagnostic Paradigm

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Abstract

Purpose of Review

The need for earlier diagnosis of Duchenne muscular dystrophy (DMD) is increasingly recognized, prompting consideration of its routine inclusion in newborn screening (NBS) programs and its addition to the Recommended Universal Screening Panel in the United States in December 2025. This review provides an overview of current approaches to DMD NBS, including the role of genomic sequencing for *DMD* variants in NBS algorithms. In an evolving model of care, the challenges and potential factors that may strengthen the practice of family-centered care are considered.

Recent Findings

Contemporary DMD NBS programs have adopted measurement of the muscle isoform of creatine kinase (CK-MM), after US Food and Drug Administration approval of high-throughput assays in 2020. Since then, 7 pilot studies from China, Taiwan, and the United States have reported outcomes using varied screening algorithms. Those incorporating repeat dried blood spot (DBS) testing after an initially elevated CK-MM level increased the positive predictive value compared with single-sample screening. Incorporating molecular genomic analysis of DMD in infants with persistently elevated CK-MM levels further increased the positive predictive value to 100%; however, this multistep approach also highlighted the practical challenges of DBS recollection, with a 17.1% (108/632) attrition rate before confirmatory testing. The choice of screening algorithm also determined the scope of conditions identified. The optimal timing for initiation of disease-modifying therapies is uncertain, and this represents a key priority for future research. The recommended model of care for DMD in early life incorporates genetics; psychosocial support; proactive monitoring and individualized support for cognitive, behavioral, and emotional difficulties; access to educational resources and support networks; and clinical trial readiness.

Summary

Health system preparedness, collaboration across countries, and coordination mechanisms within a multidisciplinary team are important for building effective health care in the context of early diagnosis of DMD through NBS. There is an ongoing need to evaluate screening algorithms using large systematic data sets, assess their cost-effectiveness, explore the potential for earlier initiation of disease-modifying treatments with validated outcome measurement tools, and establish a comprehensive evidence-based model of care to support global implementation of DMD within NBS programs.

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Glossary

BMD = Becker muscular dystrophy; **CK** = creatine kinase; **CK-MM** = muscle isoform of CK; **DBS** = dried blood spot; **DMD** = Duchenne muscular dystrophy; **NBS** = newborn screening.

Introduction

As the commonest form of muscular dystrophy in childhood, affecting 1 in 5,000 live male births,[1] Duchenne muscular dystrophy (DMD) causes progressive muscle weakness, leading to functional motor difficulties with activities such as walking, running, jumping, and climbing stairs.[2] It is an X-linked genetic condition caused by pathogenic variants in the *DMD* gene, which encodes the protein dystrophin.[3,4] The reduction or absence of dystrophin leads to membrane fragility and progressive loss of muscle tissue and function.[3,4] Thousands of different pathogenic variants in *DMD* have been identified in people with dystrophinopathies.[5] While de novo pathogenic variants are common in DMD, maternally inherited variants exist in two-thirds of patients.[6,7]

The benefits of an early diagnosis of DMD through newborn screening (NBS) for affected children and their families have been interrogated for over 4 decades, historically using creatine kinase (CK) as the primary analyte.[8] Studies highlight evidence gaps, including considerations for improving test performance to reduce false-positive and false-negative screening results, ways to facilitate a sustainable model of care that focuses on families' psychosocial needs, and economic implications of screening for a condition without recourse to disease-modifying treatment early in life.

A pipeline of therapies in research and development, alongside the benefits of timely access to multidisciplinary care for the affected child and wider implications of facilitating reproductive choice and economic and social support for families, have reignited the potential of NBS for DMD. In 2020, the Food and Drug Administration (FDA) approved the use of a high-throughput NBS assay based on the measurement of the muscle isoform of creatine kinase (CK-MM) for DMD NBS.[9] The expanding evidence-based care subsequently supported endorsement of DMD for inclusion in the Recommended Uniform Screening Panel in 2025. While the feasibility and performance of DMD NBS have advanced, emerging data highlight scientific and clinical uncertainties regarding optimal pathways for timely diagnosis and possible intervention.

This review provides a contemporary perspective on NBS for DMD. Various current screening approaches are evaluated, including the role of genomic sequencing in DMD variant detection, considerations of false-positive and false-negative results, and international screening experiences. The model of care for DMD in early life is also considered. Taken together, this knowledge serves to build an effective health care pathway for the early diagnosis of DMD.

The Rationale for the Diagnosis of DMD in Early Life: Expansion of Wilson and Jungner Criteria for NBS

Early childhood developmental screening has enabled an early diagnosis of DMD for some children; however, many continue to be referred into clinical services only once gross motor skills have plateaued or started to regress. Awareness of red flags such as motor delay, proximal weakness, abnormal gait, speech delay, and social-communication difficulties may prompt testing for elevated CK levels,[10] and the availability of genetic testing modalities, including chromosomal microarray, enables an earlier diagnosis for children with developmental delay.[11] However, there are fundamental issues with this diagnostic pathway, which are replicated across the global landscape. Parental concerns based on a child's development typically arise at an average age of 2.5 years, while the average age at diagnosis is 4.9 years,[12] with nearly 30% of families consulting ≥4 clinicians before diagnosis.[13] Further disparities are evident for culturally and linguistically diverse families, who experience longer diagnostic pathways and more convoluted health care journeys.[14] These diagnostic delays result in missed opportunities for early intervention, monitoring, support, and planning for future resources and needs, elements that are highly valued aspects of care and management within the DMD community.[15] In addition, subsequent carrier testing allows the identification of heterozygous female individuals and the initiation of cardiac surveillance for dilated cardiomyopathy.[16]

Despite the availability of genetic testing for dystrophin, the incidence of DMD has remained consistent over the past 40 years, with no decrease in theoretically preventable cases (defined as those with either a family history of DMD or a sibling aged older than 6 years), suggesting missed opportunities for genetic counseling and informed reproductive planning for affected families due to the aforementioned diagnostic delays.[17] Indeed, a family's reproductive decision making may also be influenced by an intersection of personal, sociocultural, and socioeconomic factors. These include personal beliefs, deeply embedded cultural norms or religious beliefs, and lack of access to advanced technologies such as preimplantation genetic testing, all of which may alter early management and prevention of DMD.

The directive for NBS in DMD is further accelerated by an emerging repertoire of treatments (Table 1).[18-27] Although the optimal age for initiating disease-modifying therapies is uncertain, earlier intervention may be needed for functional

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benefit, before progressive fibrosis, disruption of architecture, and depletion of muscle regeneration occur.[28] As the primary agent in the therapeutic armory for DMD, prednisolone would be at the forefront of evaluation for its use in early life. A study evaluating the efficacy of prednisolone (5 mg/kg twice weekly) in infancy and early childhood showed a 0.5-point improvement on the Bayley-III gross motor scale in treated infants, compared with a 1.3-point decline in untreated infants.[29] Genotype-specific exon-skipping medications and adeno-associated virus (AAV)–microdystrophin gene therapies may improve outcomes and have received regulatory approvals in some jurisdictions.[18-22] For example, eteplirsen, which produces a truncated dystrophin by skipping exon 51, was reported to be safe and well tolerated in boys aged 6–48 months in a phase 2 study.[30] Nonetheless, horizon scanning for the next generation of therapeutics considering treatment safety and efficacy in younger children and a concurrent pathway that enables early identification of children with DMD remain crucial to support trial readiness in the near future.[31]

These multidimensional benefits, alongside significant advocacy from stakeholders across the DMD community, have challenged the traditional screening principles and provided a modernized interpretation, focusing on a condition's actionability while superseding its treatability.[32,33] DMD fulfills this new preventative paradigm of management, with an emphasis on achieving a timely and equitable diagnosis through NBS to

Table 1 Treatments for Duchenne Muscular Dystrophy (Modified From Parent Project Muscular Dystrophy[61])

Drug name	Mechanism of action	Organization(s) that granted approval	Age criteria	Genetic variant criteria	Administration and frequency
Adeno-associated virus (AAV)–mediated gene therapy					
Delandistrogene moxeparvovec-rokl (Elevidys)[18]	Restoring a shortened dystrophin by delivering a transgene	FDA (June 2023)	Ambulatory individuals who are ≥4 y of age	Contraindicated for pathogenic variants involving exon 8–11 regions	One-time IV infusion
Exon-skipping therapy					
Casimersen (AMONDYS 45)[19]	Restoring a shortened dystrophin by inducing exon 45 skipping during mRNA processing	FDA (February 2021)	Not applicable	Exon 45 skip amenable	1x/week IV infusion
Eteplirsen (Exondys 51)[20]	Restoring a shortened dystrophin by inducing exon 51 skipping during mRNA processing	FDA (September 2016)	Not applicable	Exon 51 skip amenable	1x/week IV infusion
Golodirsen (Vyondys 53)[21]	Restoring a shortened dystrophin by inducing exon 53 skipping during mRNA processing	FDA (December 2019)	Not applicable	Exon 53 skip amenable	1x/week IV infusion
Viltolarsen (Viltepso)[22]	Restoring a shortened dystrophin by inducing exon 53 skipping during mRNA processing	FDA (August 2020)	Not applicable	Exon 53 skip amenable	1x/week IV infusion
Glucocorticoid					
Deflazacort (Emflaza)[23]	Reducing inflammation and promote muscle fiber growth	FDA (June 2019)	≥2 y of age	Not applicable	Oral once daily
Prednisone/ prednisolone[24]	Reducing inflammation and promote muscle fiber growth	Not applicable (prescribed off-label in DMD)	[a]	Not applicable	Oral once daily, alternate d, weekend-only or intermittent[a]
Vamorolone (Agamree)[25]	Reducing inflammation and promote muscle fiber growth	FDA (October 2023), EMA (December 2023), MHRA (January 2024)	≥2 y of age (FDA), ≥4 y of age (EMA and MHRA)	Not applicable	Oral once daily
Histone deacetylase (HDAC) inhibitor					
Givinostat (Duvyzat)[26]	Reducing inflammation and promoting muscle regeneration by blocking HDACs	FDA (March 2024), MHRA (December 2024)	≥6 y of age	Not applicable	Oral once daily

Abbreviations: EMA = European Medicines Agency; FDA = Food and Drug Administration; MHRA = Medicines and Healthcare products Regulatory Agency.
Note: The treatments listed in this table have been approved by specific regional authorities and are not applicable or available globally. Current as of April 1, 2026.
[a] No consensus guidelines on the dosing regimen or the optimal age to commence.

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enable proactive care and support for affected children and their families.

Contemporary and Evolving Strategies for Early Diagnosis of DMD Through NBS

International DMD NBS programs have adopted measurement of CK-MM as the primary analyte after approval of the Genetic Screening Processor (GSP)® Neonatal CK-MM kit in 2020,[9] with outcomes reported from 7 programs across 3 countries, encompassing a total of 187,992 newborns screened.[34-40] Among these, 139 were notified to have abnormal screening results and 18 individuals with DMD were confirmed on diagnostic testing; however, there was variability in study methodologies (setting, recruitment, sex (both or male only), consenting process, total number screened), screening algorithms, and technical details (CK-MM cutoff value, timing of recollection of a second (or third) dried blood spot (DBS), use of molecular genetic analyses as part of NBS, and reporting of off-target conditions) (Table 2).

Across studies, the incidence of DMD ranged from 1:3,398 to 1:11,067 live male births, and all adopted an opt-in approach. While the participation rates were not reported in all studies, potentially positively skewing findings, participation rates of ≥80% were reported in New York and MA.[36,39] One study approached parents prenatally and provided electronic education and consent,[38] whereas other studies did not specify the consent method. None of these studies has thus far ascertained the etiology for the lower participation rate, in comparison with an uptake of 95%–99% in routine NBS programs across health care jurisdictions.[41-43] National NBS programs for DMD should recognize and evaluate its underlying cause so as not to affect public confidence in the NBS program as a whole.

Inclusion criteria varied among pilot studies, with 3 of 7 undertaking male-only screening.[34,35,37] However, none specified the process for determining sex (assigned sex at birth or use of an X counter that evaluated the number of X chromosomes on DBS). For programs that used CK-MM as a first-tier screening test and reported elevated CK-MM results as NBS positives, restricting screening to male individuals may have differentially altered the diagnostic pathway for autosomal recessive neuromuscular disorders affecting both sexes, resulting in earlier diagnosis and intervention for male individuals but not female individuals.

CK-MM cutoff values for first DBS varied widely (360–4000 ng/mL), with samples collected between 24 hours and 7 days after birth. The studies in North Carolina and Ningxia used elevated CK-MM levels on a single DBS,[37,38] with North Carolina reporting a positive predictive value of 4.08%; however, diagnostic testing was declined by the caregivers of 14 of 84 screen-positive babies.[38] The implementation of specific CK-MM cutoffs (300 and 900 ng/mL) for low-birthweight and older newborns improved screening performance in NC.[38] In NY, newborns with borderline CK-MM concentrations underwent repeat DBS collection, whereas those with high CK-MM on first DBS testing or persistently elevated CK-MM on repeat DBS testing were referred directly for diagnostic evaluation.[36] Reliance on elevated CK-MM as the sole screening analyte resulted in varying downstream diagnostic pathways, with consent models encompassing different combinations of repeat CK or CK-MM testing, *DMD* gene testing, and next-generation sequencing for neuromuscular disorders. The latter also identified off-target conditions, carriers, and variants of uncertain significance among genes associated with neuromuscular disorders, introducing additional challenges for result interpretation, counseling, and clinical follow-up.[38] The New York pilot calculated that a redefined positive screen requiring both an elevated CK-MM result and the presence of a *DMD* variant (pathogenic/likely pathogenic or variant of uncertain significance) would have increased the positive predictive value from 14.7% to 83% and reduced the 0.9% recall rate for borderline and referred newborns.[36] Accordingly, reflex genetic analysis of the *DMD* gene after an elevated first-tier CK-MM result was proposed to streamline the diagnostic pathway by avoiding additional consent, reducing turnaround times, and minimizing loss to follow-up; however, this model prevented the broader detection of other neuromuscular disorders beyond DMD.

The screening algorithm in Guangzhou and Henan incorporated recollection of a second DBS in those with an initial elevation of CK-MM for repeat analysis, increasing the positive predictive value to 66.7% and 75%, respectively.[34,35] The influence of age was also relevant to the CK-MM threshold, which was adjusted for recollected samples, with ranges from 300 to 472 ng/mL beyond day 14 of life.[35,40,44] Nonetheless, recollecting a repeat DBS to identify persistent elevation of CK-MM presented challenges, with between 5.5% and 14.8% of screen-positive individuals (based on first DBS) declining.[34,36] In Taiwan, newborns with a persistently elevated CK-MM on a third DBS underwent molecular genetic analysis for the target *DMD* gene and other neuromuscular related genes using a whole-exome sequencing muscle panel, yielding a positive predictive value of 100%.[40] The program adopted a relatively high first-tier recall rate (1.2%) to minimize false negatives, informed by the earlier Wales experience,[45] and to avoid missing infants with milder phenotypes, including Becker muscular dystrophy (BMD). However, maintaining complete participation through multiple recall stages was also challenging, with 524 of 632 newborns (82.9%) with an elevated CK-MM level on the initial DBS undergoing second DBS recollection.[40] This study suggested that the specificity of the algorithm could be improved by adding a second-tier whole-exome sequencing test when the initial DBS identified an elevated CK-MM level; however, the add-on cost was considered not acceptable. This approach was investigated in Massachusetts, with an

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Table 2 Newborn Screening Studies for Duchenne Muscular Dystrophy Using Creatine Kinase-MM as a First-Tier Analyte

Setting	Duration of pilot study	Recruitment and participation	CK-MM cutoff values (ng/mL)	Number of newborns screened	Number of elevated CK-MM cases on first DBS	Number who declined recollection of DBS	Number of screen-positive individuals who declined recall for diagnostic testing	Number of DMD cases and characteristics of the pathogenic variants	Other findings	Reported incidence of DMD
Creatine kinase only										
Guangzhou, China[34] (two-tiered)	June 2019–October 2019, May 2021–September 2021	Hospital-based; opt-in consent (rate not specified)	First DBS: ≥800 Second DBS: ≥400	2019: 21,506 male individuals, 18,258 female individuals 2021: 22,762 male individuals only	164	Lost to follow-up: 9	DMD genetic testing: 1	4 Frameshifting: one exon 45, 54–60 duplication, 1 exon 8–9 duplication, 1 exon 45–50 deletion, 1 exon 48–50 deletion	1 other muscular developmental disorder	1 in 11,067 male births (1 in 15,638 total births)
Henan, China[35] (two-tiered)	June 2019–July 2019	Region-based; opt-in consent (rate not specified)	First and second DBS: ≥472	13,110 (male individuals only)	27	Not reported	DMD genetic testing: 1	3 Frameshifting/ premature stop codon: one exon 3–7 deletion, 1 exon 52 deletion, 1 nonsense variant (c.9568C>T [p.R1390[a]])	Not reported	1 in 4,370 male births
NY, United States[36] (risk stratified: repeat DBS for borderline CK-MM on 1st DBS, direct referral for elevated CK-MM on 1st DBS)	October 2019–September 2021	Hospital-based; opt-in consent (87.10%)	First and second DBS: age at collection 0–47h: ≥1990 (borderline), ≥4,000 (referral) 48–71 h: ≥1,430 (borderline), ≥4,000 (referral) 72–167h: ≥571 (borderline), ≥860 (referral) ≥168 h: ≥571 (referral)	36,781 (18,654 male individuals, 17,993 female individuals)	Borderline: 296 Referral: 42 (direct referral: 40, referral after persistently high CK-MM on second DBS: 2)	Borderline first DBS: 17 Lost to follow-up: 2	DMD genetic testing: 6 NM panel after DMD genetic testing: 4 Lost to follow-up: 2	4 Frameshifting: one exon 18 duplication, 1 exon 51 deletion, 1 exon 3–43 deletion Frame maintained: one exon 48–49 deletion	1 female DMD carrier, 2 carriers of AR *LAMA2* MD, 2 carriers of AR limb girdle MD, 1 carrier of MD dystroglycanopathy, 22/42 with VUS in genes of neuromuscular panel but no DMD PV	1 in 4,664 male births (1 in 7,356 total births)
Ningxia, China[37] (one-tiered[a])	January 2021–December 2021	Region-based; opt-in consent (rate not specified)	First DBS: ≥400	10,000 (male individuals only)	Not reported	Not applicable	Not reported	2 Frameshifting/ premature stop codon: one exon 48–50 deletion, 1 small deletion (c.3347_3350del [p.K1116Mfs[a]36])	Not reported	1 in 5,000 male births

Continued

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Table 2 Newborn Screening Studies for Duchenne Muscular Dystrophy Using Creatine Kinase-MM as a First-Tier Analyte *(continued)*

Setting	Duration of pilot study	Recruitment and participation	CK-MM cutoff values (ng/mL)	Number of newborns screened	Number of elevated CK-MM cases on first DBS	Number who declined recollection of DBS	Number of screen-positive individuals who declined recall for diagnostic testing	Number of DMD cases and characteristics of the pathogenic variants	Other findings	Reported incidence of DMD
NC, United States[38] (one-tiered)	November 2020–October 2022	Region-based; opt-in consent after 13 wk of gestation or up to 4 wk postpartum (rate not specified)	First DBS: ≥1,626 (≥2032 on retest) (age ≤72h, BW > 1500g, GA > 28 wk), ≥900 (age ≥48 and <72 h), ≥360 (age ≥72 h or BW ≤ 1500g)	13,354 (6,795 male individuals, 6,354 female individuals, 205 unknown)	142 (84 on retest)	Not applicable	NM panel + latent total CK (on 2nd blood sample): 11 Latent total CK only (on 2nd blood sample): 11 NM panel only: 3	2 Frameshifting/ premature stop codon: one exon 18–26 duplication, 1 nonsense variant (c.1267C>T [p.Q423[a]])	10/68 with non-DMD PV/LPV in genes of neuromuscular panel (2 AD MD: 1 *COL6A1*, 1 *CAPN3*; 1 AD myotonia congenita: 1 *CLCN1*; 6 carriers of AR MD: 1 *AN05*, 3 *LAMA2*, 1 *SELENON*, 1 *GNE*; 1 carrier of AR metabolic disorder: 1 *PYGM*)	1 in 3,398 male births (1 in 6,677 total births)
Creatine kinase and genetic testing										
MA, United States[39] (two-tiered)	June 2021–September 2021	State-based; opt-in consent (80%)	First DBS: ≥958 (female individuals), ≥1,080 (male individuals)	1,379	29	Not applicable	0	0	1 Chr 16 microdeletion syndrome (Chr 16p11.2)	Not reported
Taiwan[40] (three-tiered)	February 2021–December 2021	Region-based (35% babies born in Taiwan); opt-in consent (rate not specified)	First DBS: ≥750 (full-term), ≥650 (premature) Second DBS: ≥300	50,572 (26,130 male individuals, 24,442 female individuals)	632	Second DBS: 5 died of unrelated reasons, 4 unable to do re-screen Third DBS: not applicable	Not applicable	3 Frameshifting/ premature stop codon: one exon 44–51 deletion, 1 nonsense variant (c.9337C>T [p.R3113[a]]) Frame maintained: one exon 20,600-bp deletion	1 PV *COL12A1*, 8 positive Pompe disease with screening CK-MM levels <99th centile	1 in 8,710 male births (1 in 16,857 total births)

Abbreviations: AD = autosomal dominant; AR = autosomal recessive; BW = birthweight; CK = creatine kinase; CK-MM = muscle isoform of CK; DBS = dried blood spot; DMD = Duchenne muscular dystrophy; GA = gestational age; GNE = glucosamine; MD = muscular dystrophy; NM = neuromuscular; LPV = likely pathogenic variant; PV = pathogenic variant; VUS = variant of uncertain significance.
[a] The authors clarified that a second consent for diagnostic DMD genetic testing occurred after identification of elevated CK-MM of the first DBS.

estimated cost of USD 8 per infant tested (CK-MM assay and reflex genomic testing). However, no *DMD* variants were identified among the initial 1,379 newborns.[39]

Thus far, the feasibility of a three-tiered algorithm, with the addition of rapid aneuploidy testing of sex chromosomes to identify male individuals (46XY) among those with an elevated CK-MM level and a targeted *DMD* next-generation sequencing panel on a single DBS, has not been investigated. This approach has been proposed for a scoping study in NSW, Australia.[13] The most efficient screening algorithm and inclusion of genetic analysis within the NBS program are not yet established.

Considerations for False Positives, False Negatives, and Unexpected Findings

The major consideration for reducing false positives lies in the chosen screening algorithm, which depends on factors including the CK-MM cutoff threshold, number of DBS samples needed for repeat CK-MM testing, and incorporation of genomic testing. Transiently elevated CK-MM in the newborn period has been reported to be a significant cause of false-positive referrals. Thus, challenges remain in determining a CK-MM threshold that considers postnatal age, gestational term, birthweight, and sex.[46] For example, older and heavier newborns and male individuals have higher CK levels after birth.[47] Furthermore, because CK-MM is an indirect marker of DMD, it can also be elevated when muscle damage or traumatic birth events are noted, including the use of forceps, vacuum extraction, fractures, or a nuchal cord.[48] This usually occurs in the first 24–48 hours of life, when DBS is typically collected, making it difficult to distinguish between newborns with DMD and those recovering from birth trauma. The inclusion of these clinical variables in setting CK-MM thresholds varies between DMD NBS programs.

With multiple factors influencing CK-MM concentration in newborns, the observed age-related decrease in CK-MM by 72 hours of life establishes the rationale to perform repeat DBS collection after 72 hours of age.[48] Repeat CK-MM testing helps address the transient nature of nongenetic hyperCKemia and differentiate newborns with DMD experiencing sustained hyperCKemia to reduce false positives. However, screening algorithms relying on multiple sample collections have associated challenges, including the follow-up of patients (due to changes of contact details, failure to receive phone call or text messages, or refusal to perform blood recollection).[39] Further considerations include high labor intensity and the extended screening duration of sample collection and laboratory testing, which leave parents in a psychologically demanding period during which the diagnosis remains uncertain.[39]

As a solution, the incorporation of genomic sequencing into NBS workflows has been shown to minimize false positives, suggesting its potential inclusion as a second tier (instead of repeat DBS collection), given its improved screening specificity and clinical utility in determining future treatment pathways.[39] Yet, heterogeneity remains in the modalities of molecular genetic analysis and in the variant curation parameters used to determine screen positivity. For *DMD* sequencing alone, identification of sex and the "reading frame rule" are key determinants of whether female carriers of a dystrophinopathy or children affected by BMD, respectively, will be identified. However, the reading frame rule is not absolute,[49] thereby complicating the interpretation of screen positives and reporting of pathogenic variants within NBS programs. In contrast, genomic testing approaches that incorporate a curated neuromuscular panel of genes, as explored in some scoping studies, can identify causes of elevated CK-MM beyond DMD. These include autosomal dominant or recessive heterozygous pathogenic variants for other neuromuscular conditions such as limb girdle and congenital muscular dystrophies and heterozygous carriers. Among studies to date, such findings account for 14.71%–16.67% of infants with elevated CK-MM on DBS screening and lead to additional challenges in counseling families, clinical care, and management.[36,38]

To date, the duration of clinical follow-up of DMD NBS using CK-MM as the primary analyte is not yet sufficient to ascertain the number of false-negative cases, or to fully assess the associated risks and potential harms. A historical review of 10 DMD NBS programs conducted between 1975 and 2011, all of which used single-tier total CK-based approaches, identified 21 false-negative cases among 1.8 million newborns screened.[50] In 18 of these cases, the CK levels were below the program specific cutoff thresholds. In addition to the importance of setting a CK threshold to optimize sensitivity and specificity, further explanations for false negatives included infants with low birthweights or who were small for gestational age.[45] These findings highlight the importance of ensuring robust pathways for education, care, and support of affected children and families not identified by NBS. Taken together, there are multiple factors to consider when designing and implementing DMD NBS, pertinent to the subsequent clinical and diagnostic approach for screen-positive individuals, and other implementation outcomes (feasibility, cost, and sustainability in health practice) (Table 3). Indeed, the screening algorithm determines both the scope and performance of a DMD NBS program. It influences whether the program targets DMD alone or also identifies BMD, affected female individuals, carriers, and other neuromuscular disorders. It also determines key performance metrics, including the positive predictive value, false-positive rates, recall requirements and attrition, diagnostic complexity, and overall program feasibility. Recollection of a second DBS after an elevated CK-MM result improves the positive predictive value but introduces additional recall steps and potential loss to follow-up. In contrast, genomic NBS offers a substantially higher positive predictive value and a streamlined diagnostic pathway with greater clinical utility for treatment planning but

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Table 3 Factors to Consider When Implementing Newborn Screening for Duchenne Muscular Dystrophy

Factors	Domains	Considerations
Setting		
Who should be included in DMD NBS?	• Full population • Specific populations	• Operational costs • Accessibility and equity • Variability in screening practices
Cost		
How should it be charged?	• Government-funded • Out-of-pocket	• Family willingness due to availability of financial support • Access equitability to services • Duration of waiting time
Consent method		
When should consent be obtained?	• Prenatally • Postpartum	• Amount of time to understand information and prepare mentally • Stress level during pregnancy • Risk of missed or delayed screening
How should consent be obtained?	• Written consent • Electronic consent	• Convenience and efficiency • Data privacy and cybersecurity concerns
How should the program be designed as?	• Opt-in • Opt-out	• Participation rate • Ethical considerations regarding parental autonomy and decision making • Degree of health literacy required
What educational materials should be provided?	• Online modules, videos, or virtual events • Educational brochures or information sheets	• Convenience of information access • Ease of information comprehension • Cost-effectiveness
Participants		
Who should be screened?	• Male only • Both sexes	• Screening specificity • Equity in identifying autosomal recessive neuromuscular disorders • Detection of female carriers • Overall cost • Follow-up monitoring and model of care for symptomatic female carriers • Unnecessary anxiety for asymptomatic female carriers
How should sex be determined?	• Clinical examination • Rapid aneuploidy testing	• Cost-effectiveness and efficiency • Test sensitivity and specificity
Screening algorithm		
What primary analyte should be used?	• CK-MM	• Elevated in birth trauma, infection, asphyxia, DMD, BMD, other neuromuscular disorders, and carriers of DMD • Different cutoffs for different ages at collection of DBS, e.g., 0–47h, 48–71 h, 72–167 h, and ≥167 h are suggested[48] • Varies with gestational age, birthweight, and sex
If repeat CK-MM testing is conducted, how many samples should be collected?	• Second DBS • Third DBS • Another venous sample along with genetic testing	• Duration of screening • Parental acceptance and dropout rate • False-positive rate and positive predictive value • Sample adequacy
Which molecular genetic analysis and modality should be included?	• No molecular genetic assay • *DMD* NGS panel • Neuromuscular NGS panel	• Cost-effectiveness and efficiency • Specificity for detecting DMD • Genomic coverage and identification of other neuromuscular disorders • Interpretation and report complexity
What conditions should be reported? (Variant curation)	• DMD only • DMD and other genetic conditions (e.g., BMD, carriers of DMD, neuromuscular diseases, and other genetic disorders)	• Ethical concerns regarding patient autonomy • Parental anxiety and psychological burden • Time to initiate clinical monitoring, supportive care, and management • Reproductive planning • Strain on health care system and demand on resources

Abbreviations: BMD = Becker muscular dystrophy; CK-MM = muscle isoform of CK; DBS = dried blood spot; DMD = Duchenne muscular dystrophy; NBS = newborn screening; NGS = next-generation sequencing.

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introduces increased laboratory steps, costs, and variant interpretation challenges.

A Current Understanding of Family and Clinician Experiences and Perspectives of NBS for DMD

Parents of boys with DMD diagnosed after presentation with clinical signs and symptoms are generally supportive of an early diagnosis, despite a lack of targeted treatments for all variants and potential treatments to prevent the most serious symptoms.[51,52] Early diagnosis through NBS is valued for its clinical utility for the child, such as access to multidisciplinary care, early intervention, specialist services, and clinical trials[51,53,54]; personal utility for the affected individuals, including earlier planning for education needs[53]; and benefits to the wider family and society, including adjusting parenting expectations, informed reproductive decision making, and financial planning.[51,53,54] Despite these benefits, some caregivers of boys receiving a later diagnosis of DMD have expressed uncertainty about NBS, perceiving risks of heightened emotional distress and adverse parent-child bonding in the absence of an immediate care plan.[53,55] This echoes with certain perspectives among the Australian health care professionals, who identify the current lack of early disease-modifying therapies as a barrier to implementation.[54]

Parental experiences after false-positive results in NBS programs were also assessed in North Carolina.[56] Although this diagnostic journey was stressful, none reported regret about participating in screening because it provided them with the opportunity to perceive more information about their child's health and prepare for potential outcomes. However, parents reported confusion surrounding initial CK-MM screening results, suggesting the need for clarifying medical terms clearly and using more visual aids or statistics to facilitate understanding.

These concerns highlight the need for co-developing a model of care with stakeholders, including families with lived experience, to ensure that the NBS program for DMD meets the support, care, and information needs of affected children and their families. Addressing the barriers and enablers will be critical if high trust and public acceptability of national NBS programs are to be maintained.[54]

Enablers of a Model of Care for Children Diagnosed Early in Life With DMD

While guidelines for early-life care in children with DMD are lacking, several workshops have focused on developing an optimal model of care, bringing together experts, health care practitioners, and people with lived experience across regions.[15,57] From this consensus-based evidence, a coordinated postdiagnostic management of DMD seems essential, best provided in a multidisciplinary setting (Figure). This includes focusing on genetics (including genetic counseling, carrier cascade testing, and reproductive planning), medical management, clinical trial readiness, psychosocial support, and access to educational resources and support networks.

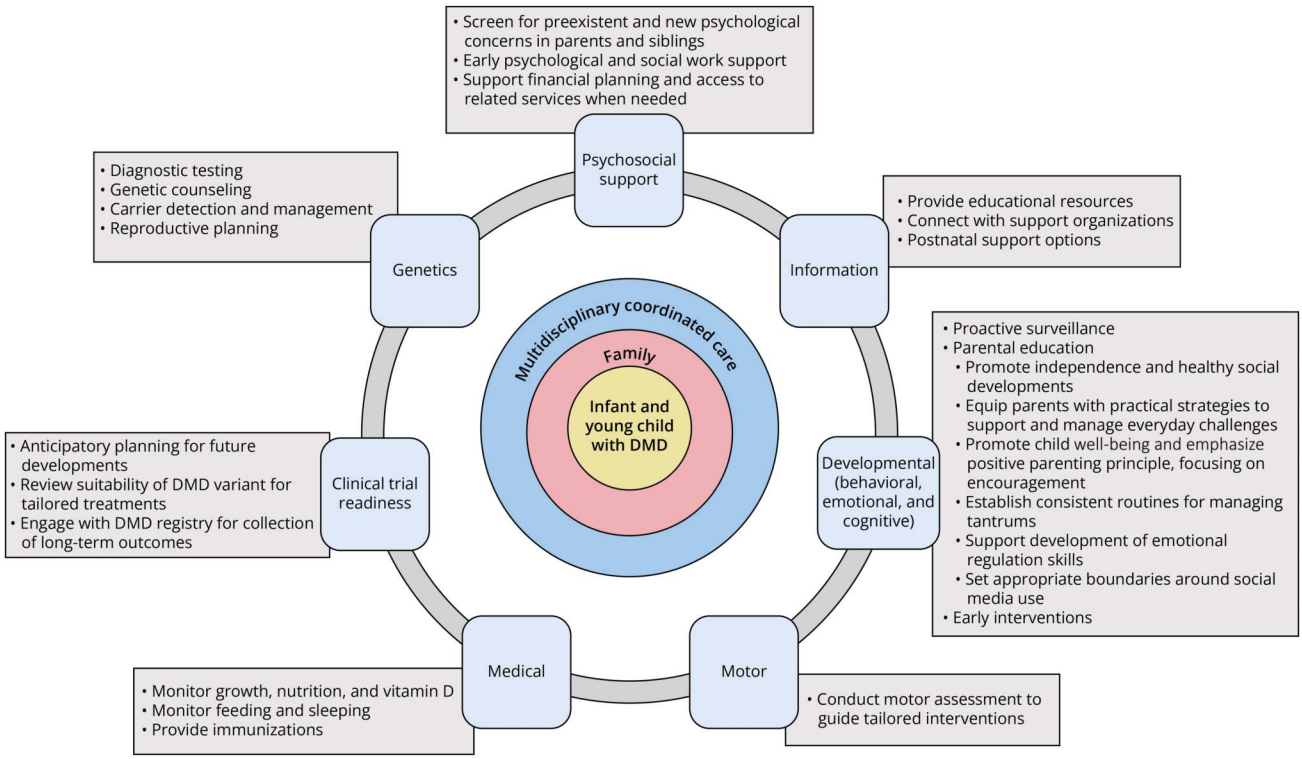
Speech delays are common, and one-third of DMD-affected individuals have cognitive impairment.[3] Consequently, proactive monitoring, individualized support, and referrals to early intervention for speech, language, social communication, sensory processing difficulties, and motor delays are important in mitigating emotional and behavioral sequelae associated with DMD, while supporting participation and inclusion in educational settings.[15] Likewise, instructing families about activities and interactions that promote a child's development and well-being through home-based therapies and environmental modifications are valuable.[15] This also includes equipping parents with practical strategies to support and manage everyday challenges, for example, establishing consistent routines for managing behavioral difficulties and supporting development of emotional regulation skills.

Evidence for early initiation of pharmacotherapies remains limited, and there is variability in access and approach. Establishing validated and meaningful outcome measurement tools is vital to generating evidence because most are not validated in infants. The European Medicines Agency recently approved the 95th centile stride velocity, quantified with a wearable device, as an outcome measure for individuals with DMD older than 4 years, and ongoing studies are determining its reliability and discriminatory ability in younger children.[58] The mainstay of medical care in infancy includes immunizations and monitoring growth, nutrition, and vitamin D, together with ongoing consideration of clinical trial and therapy readiness.[57]

The diagnosis of DMD in a child profoundly affects families, and their feedback has highlighted the power of language and sensitive communication for those experiencing a presymptomatic or early diagnosis.[57] In discussions with families, the changing therapeutic landscape, availability of upcoming clinical trials, and potential for new medications can also provide hope in a very challenging time. The serious and life-limiting aspect of DMD should be balanced without applying catastrophizing words such as "fatal" or "lethal," because these may risk traumatizing families while they adapt to a new diagnosis. It is important to reframe the diagnosis of DMD, acknowledging that many individuals live into their 30s or beyond, and emphasize quality of life and participation. People living with DMD can adapt to their changing functional abilities and develop new goals, values, and expectations. Families should be reassured of ongoing care and support, and co-designed resources are needed to improve strength-based communication after early diagnosis to facilitate best practice.

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Figure Interdisciplinary Management of Duchenne Muscular Dystrophy in Early Life



DMD = Duchenne muscular dystrophy.

Limitations and Controversies

To date, several limitations and data gaps remain unaddressed in current studies. One key issue is the cost-effectiveness of implementing NBS for DMD, which is a core requirement of the Wilson and Jungner[59] criteria. None of the 7 recent pilot studies have included a health economic analysis, limiting their utility in informing policy decisions, particularly regarding allocation of public budget.

In addition, these studies used different methodologies and screening algorithms, without systematic recruitment processes and standardized data sets, making comparisons difficult and further hindering evaluation of the overall effectiveness of NBS. There is also a lack of consensus on the intended outcomes of NBS—specifically, whether to identify only DMD or also BMD, other neuromuscular diseases, or carriers of DMD. This uncertainty complicates decisions around follow-up care and the level of training required for health care professionals.

While existing studies have primarily focused on evaluating the screening program quantitatively, there is limited qualitative research exploring parental experiences after a positive NBS result. There have been concerns regarding the negative impact on mother-child bonding, which arise from clinicians and parents of children diagnosed later in life with clinical signs and symptoms, but these perspectives have not been reported thus far for children identified by NBS. Indeed, only 1 study has examined attitudes toward and the impact of NBS for DMD among families with confirmed DMD identified through screening.[60] Although this study reported no long-term impact on bonding, it was conducted over 30 years ago, with an NBS algorithm solely based on total CK screening. There is a need for more evidence to better understand parents with lived experience in contemporary settings.

Future Directions and Conclusion

As the therapeutic landscape continues to evolve and accelerate, DMD is increasingly aligned with contemporary screening paradigms that focus on the actionability of a condition[33] and the personal, familial, and societal impact made through facilitating a diagnosis in early life. To investigate the effectiveness of NBS for DMD, further scoping studies are required to explore the possibilities of different algorithms, with outcomes reported in a systematic approach alongside comprehensive health economic analysis. It is important to note that a significant knowledge gap remains regarding the best practice from a management perspective for children identified through NBS for DMD and how to facilitate equitable access to care and support for the child and family unit, in an economically viable way.

Evaluating and co-developing an evidence-based model of care with consumers is essential to meet the specific needs and

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preferences of children and families diagnosed through NBS for DMD so that medical management, early intervention, genetic counseling, education, and psychosocial support are cohesively integrated. Future work also rests on the generation of evidence for the safety and effectiveness of early treatments in infants and children with DMD, with validated outcome measures required to interrogate end points within a newborn and infantile period of life. Cumulatively, as the international landscape moves toward NBS for DMD, this review synthesizes the key considerations, barriers, and enablers required to implement NBS for DMD programs that maximize benefits of an early diagnosis and mitigate potential risks.

Author Contributions

U.T.E. Yau: drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; analysis or interpretation of data. D. Lorentzos: drafting/revision of the manuscript for content, including medical writing for content. D. Kariyawasam: drafting/revision of the manuscript for content, including medical writing for content; study concept or design; analysis or interpretation of data. M.A. Farrar: drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; analysis or interpretation of data.

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