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Duchenne Muscular Dystrophy

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Abstract: *Duchenne muscular dystrophy (DMD) is a severe X-linked neuromuscular disorder caused by mutations in the DMD gene, resulting in little or no dystrophin production and progressive muscle degeneration. This review summarizes contemporary evidence (post-2010) on the epidemiology, clinical progression, treatment patterns, quality of life, and economic burden of DMD. A systematic search of major databases identified 9,850 studies, of which 58 met quality criteria. Findings show that although diagnostic practices have shifted toward genetic testing, diagnosis is still commonly delayed until 4–5 years of age. Prevalence estimates remain inconsistent due to variations in diagnostic methods and incomplete reporting. Despite significant disease burden— including early motor delays, loss of ambulation around 12 years, cardiomyopathy, scoliosis, and respiratory decline—advances such as corticosteroid therapy, improved ventilation, and standardized care guidelines have increased life expectancy.*

Quality-of-life studies reveal substantial psychosocial and physical challenges, with caregiver burden also notably high. Economic analyses indicate that indirect costs exceed direct medical costs, especially in early and late disease stages. Major evidence gaps persist in accurate prevalence measurement, long-term outcomes, and cross- country comparisons. Patient registries, such as TREAT-NMD and CINRG, offer promising opportunities for obtaining more reliable, standardized, and contemporary data. Overall, the review highlights the need for earlier diagnosis, better reporting practices, and expanded research that includes all age groups and stages of DMD to fully understand and address the global burden of the disease.

Keywords: Duchenne muscular dystrophy, Muscle fiber degeneration, Inflammation and fibrosis, Scoliosis, Clinical trials

I. INTRODUCTION

Mutations in the DMD gene, which provides instructions for making the protein dystrophin, cause two related muscle diseases: Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD).[1,2]

DMD is the more severe form, while BMD is milder.

Under normal conditions, dystrophin works like a shock absorber in muscle cells. It connects the internal skeleton of the muscle fibre (actin) to the protective connective tissue that surrounds each muscle fibre.[3,4] This connection helps protect muscles from damage when they contract.

In DMD, mutations either break the reading frame of the gene or create a premature stop signal, which prevents the body from making functional dystrophin. Because dystrophin is missing or non-functional, the link between the muscle cell's internal structure and surrounding support tissue is lost.[5]

As a result, muscle fibres become very fragile and get damaged easily during normal use. [6]Over time, this leads to chronic muscle injury, inflammation, and replacement of healthy muscle with fat and scar tissue, causing a steady decline in muscle strength and function.

Duchenne muscular dystrophy (DMD) is the most common inherited muscle disease seen in children. It is a progressive condition that mainly affects boys because it is passed down through the X-chromosome.[7-10] DMD is caused by changes (mutations) in the DMD gene, which leads to a complete lack or a very low amount of dystrophin—a protein that helps keep muscle cells strong and functioning normally.

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Without enough dystrophin, muscles become weak over time.[11] Globally, DMD affects about 7 out of every 100,000 males,[12] and almost 20 out of every 100,000 male babies are born with this condition. Similar numbers are seen in countries like the USA and the UK.[13]

Muscle damage in DMD begins early in childhood. As a result, children may show:

- Very high levels of creatine kinase (CK) in the blood
- Gradually increasing muscle weakness
- Delays in motor skills (like walking or running)
- Loss of the ability to walk, usually in early adolescence
- Breathing difficulties
- Heart problems

Some children with DMD also have challenges beyond muscle symptoms—such as learning difficulties, speech delays, autism-related traits, or attention-related problems.[14]

The severity of symptoms can differ depending on the type of mutation in the DMD gene.

Unfortunately, complications involving the heart and lungs are the main causes of reduced life expectancy in DMD.[15,16]

New treatments for Duchenne muscular dystrophy (DMD),[17-19] especially those that try to restore dystrophin, have made it more important than ever to diagnose the condition early and confirm it genetically. Doctors are now focusing on spotting the earliest signs of DMD in very young boys.

But despite this, diagnosis is still often delayed. On average, children are not diagnosed until they are 4–5 years old—usually about two years after their first symptoms appear. Unfortunately, this delay has not improved much in the last 20 years, even though health organizations have tried to increase awareness among doctors.

There are many important reasons why diagnosing DMD earlier matters. It allows families to get proper genetic counseling, check carrier status, start the recommended medical and therapeutic care early, and begin corticosteroid treatment at the right time.

Early diagnosis also helps identify the exact gene mutation, which is essential for accessing new mutation-specific therapies. It gives families the chance to join clinical trials, prevents unnecessary or costly tests, and reduces the emotional stress of going through a long and confusing diagnostic process.

II. METHODS

New treatments for Duchenne muscular dystrophy (DMD)—especially those aimed at restoring dystrophin—have made early detection more important than ever. Doctors are now paying closer attention to the earliest signs of DMD in very young boys so the condition can be diagnosed and confirmed through genetic testing as soon as possible.

However, even today, there is still a significant diagnostic delay. Most children are not diagnosed until they are about 4–5 years old, which is nearly two years after parents first notice something is wrong. Sadly, this delay in diagnosis has not improved much over the last two decades, despite efforts to increase awareness among healthcare professionals.

Identifying DMD early makes a big difference. It allows families to receive timely genetic counseling, understand carrier risks, and start proper medical care early—including corticosteroid treatment at the right stage. Early diagnosis also helps pinpoint the exact gene mutation, which is crucial for accessing newer, mutation-specific therapies and joining clinical trials. Most importantly, it prevents families from going through unnecessary tests and helps reduce the emotional strain of a long, uncertain search for answers. In short, better diagnostic timing can greatly improve the path forward for children and families affected by DMD.[20]

This review followed well-established guidelines—such as those from the Cochrane Collaboration and the Centre for Reviews and Dissemination—to make sure the process was reliable and unbiased. To gather information, the researchers searched scientific articles, looked through online sources, and contacted registries and patient organisations.

We carried out a thorough search of the Medline/Medline In-Process and EMBASE databases (details are in Table S1). The search was based on PECOS criteria, which helped us decide what types of studies to include—who the patients

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were, what exposures and comparisons were involved, what outcomes were measured, and what study designs were used.

We included studies published in English from the time each database began (1946) up to November 2018. To be included, a study had to report the ages at which important clinical milestones occur in boys with Duchenne muscular dystrophy (DMD).

To make sure the results were useful and relevant, we focused on observational studies from North America or international studies that included North American patients. We only selected studies with fairly large groups of patients (more than 50 participants) who were treated with corticosteroids.

We did not include animal studies or studies that involved patients with other types of muscular dystrophy.

"We checked how strong and reliable each study was by using the STROBE checklist, which is a standard tool used to judge the quality of observational and non-randomized clinical studies."

III. RESULT

The search process originally found 5,637 articles that might be relevant. After removing duplicates, only four articles were taken out. Then, by reading the abstracts, 5,213 articles (about 92.5%) were found to be not useful for the review. This left 410 articles to look at in full. After reading these completely, 381 more were excluded, leaving only 29 studies that met all the criteria.

These 29 studies included different types of research, such as hospital-based (single or multiple centers) chart reviews and information from DMD patient registries. This included 6 publications from the CINRG registry and 4 from the MD STARnet registry.

Details about corticosteroid treatment—such as when treatment was started, how patients were monitored, and how often side effects were reported—are listed in Additional file 1: Table S2. But the amount of information differed a lot between studies, and only a few studies looked at how differences (for example, starting steroids at an older or younger age) affected how DMD progressed.

Information about cardioprotective (heart-protecting) medications is given in Additional file 1: Table S3. Additional file 1 also contains a summary of the quality of the included studies.

Loss of ambulation:

Six studies reported the average age at which boys lost the ability to walk, ten studies reported the median age, and thirteen studies reported the percentage of boys[21-26] who experienced loss of ambulation (LOA) (Table 2). Two of these studies also gave results for specific subgroups of patients.

For boys with Duchenne muscular dystrophy (DMD) who were treated with corticosteroids, the average age at which they stopped walking ranged from about 9.5 years (based on 112 boys from the MD STARnet registry) to 12.5 years (based on a smaller hospital-based group of 75 boys). These numbers are shown in Figure 2a.

Three other studies included boys who had mixed patterns of steroid use (some taking steroids and some not). Their results were similar: the average age when walking was lost ranged from 9.8 years (in a group of 432 boys from Mexico) to 10.8 years (in 462 boys from MD STARnet).

One important pattern stood out:

• Boys who had been on corticosteroids for 3 years or less lost the ability to walk earlier, around 9.5 years old.

• Those who had been on steroids for more than 3 years were able to keep walking longer, losing ambulation at about 12.3 years old (MD STARnet data).

Scoliosis:

Only one study reported the average age when scoliosis develops, two studies reported the median age, and five studies looked at how many patients had scoliosis at different ages. The way scoliosis was defined varied between studies.[27]

In one single-center study of 56 patients, the average age when spinal surgery was done was 14 years, and 14.5 years among the 20 patients who also had lung-function tests.[28]

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Another study from the MD STARnet group found that, in a mixed group of boys (some using corticosteroids, some not), the median age for scoliosis surgery was 14.6 years (with a range from 10.2 to 20.2 years). In this group of 208 patients, 52.4% eventually needed surgery.[29]

In a different MD STARnet study of 274 boys who were all taking corticosteroids, the median age when scoliosis (defined either as a spine curve greater than 30° or surgery) was identified as 14.2 years (range: 12.5–15.6 years) among the 107 boys who developed scoliosis.[30]

Across all studies, the percentage of boys with scoliosis increased as they got older. According to a long-term MD STARnet study, about 5G% of boys with DMD have scoliosis by age 15, and about 72% by age 20.[31]

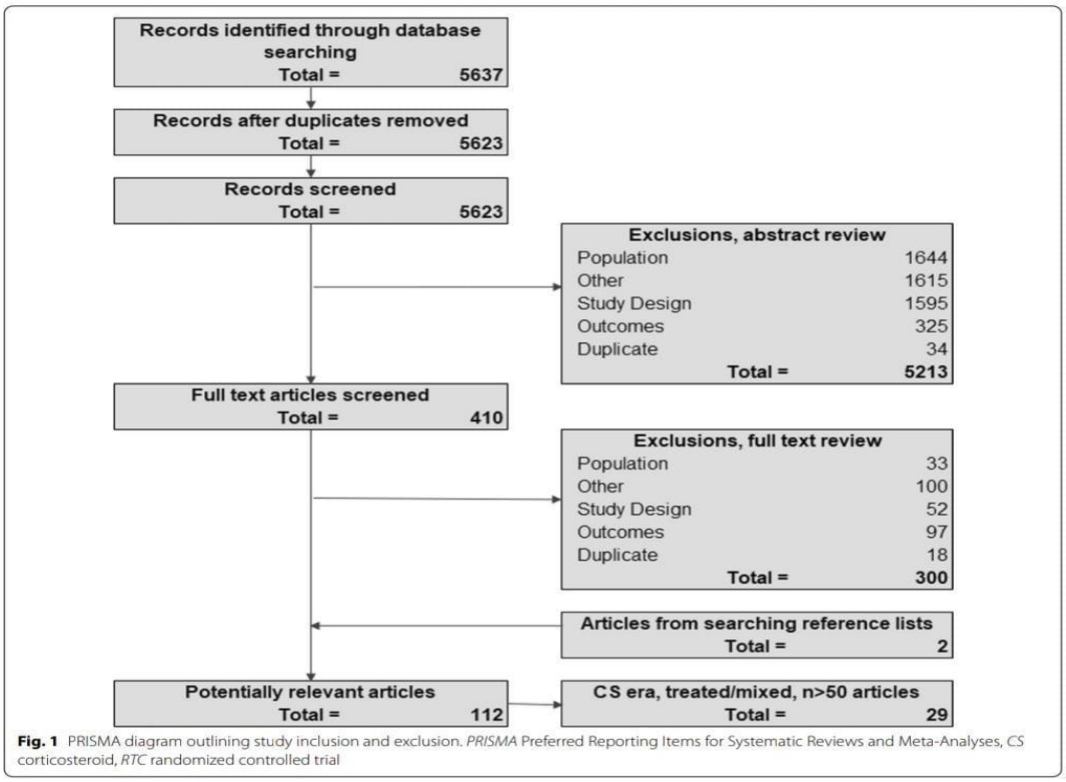

Fig. 1 PRISMA diagram outlining study inclusion and exclusion. *PRISMA* Preferred Reporting Items for Systematic Reviews and Meta-Analyses, *CS* corticosteroid, *RTC* randomized controlled trial

Fig.No.1 Prisma diagram outing study inclusion and exclusion Prisma preffered Reporting Items for Systemic Reviews and Meta analysis CS corticosteroid ,RTC (Randomised controlled trial)

Developmental milestones and early gross motor function in DMD:

Children with Duchenne muscular dystrophy (DMD) usually start showing noticeable movement-related symptoms around 3–4 years of age. These can include difficulty running or jumping, trouble getting up from the floor (often using their hands to push themselves up, known as Gower's sign), struggling to hop or climb stairs, and frequent falls. Their walking style may look unusual—sometimes waddling or walking on their toes. Many also have enlarged calf muscles or complain of muscle pain and cramps. These symptoms typically lead to further medical tests and eventually a diagnosis of DMD.[32]

In recent years, research has shown that many children with DMD also have delays in early developmental milestones—not just physical ones. Compared to other children, they may start speaking later, take longer to form sentences, or be later in learning skills like toilet training or early reading. Studies suggest that about 36% to 67% of children with DMD experience delays in these early developmental skills, even though these are not traditionally seen as typical signs of DMD.[33]

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Other studies that used detailed developmental assessments have also shown that children with DMD begin to show early developmental differences. One study used the Bayley-III test to track motor and cognitive development in boys with DMD who were between 1 month and under 3 years old (average age about 2 years).[34]

The results showed that, on average, these young boys scored lower than typically developing children in overall motor skills, as well as in both gross motor (big movements like sitting, standing, walking) and fine motor skills (small movements like grasping). They also scored lower on thinking skills and language skills—both understanding and speaking.[35]

When the boys were followed over time, their gross motor skills continued to get worse at 6 and 12 months. However, their thinking and language scores stayed about the same, and their fine motor skills actually improved over the year.[36]

Other studies that used detailed developmental tests have also shown that babies and toddlers with Duchenne muscular dystrophy (DMD) start to show developmental delays very early in life.

In one study, researchers used the Bayley-III test—a standard tool for checking a young child's motor, thinking, and language skills—to follow boys with DMD who were between 1 month and 3 years old (average age about 2 years).

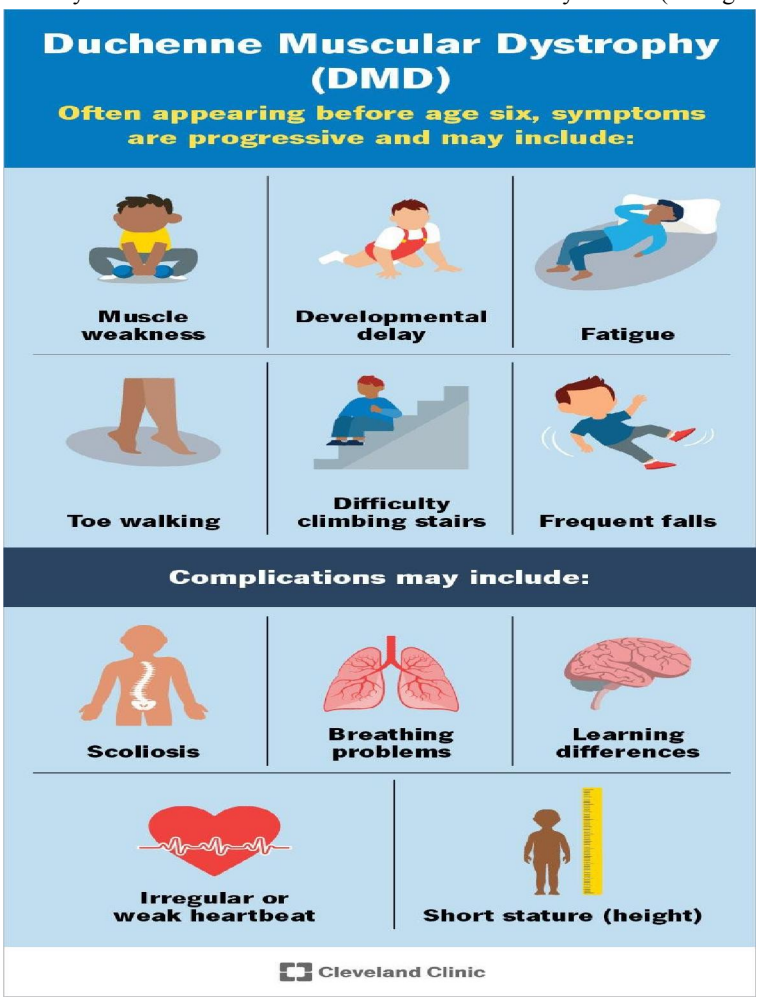

Fig.No.2 Duchenne muscular dystrophy (DMD)

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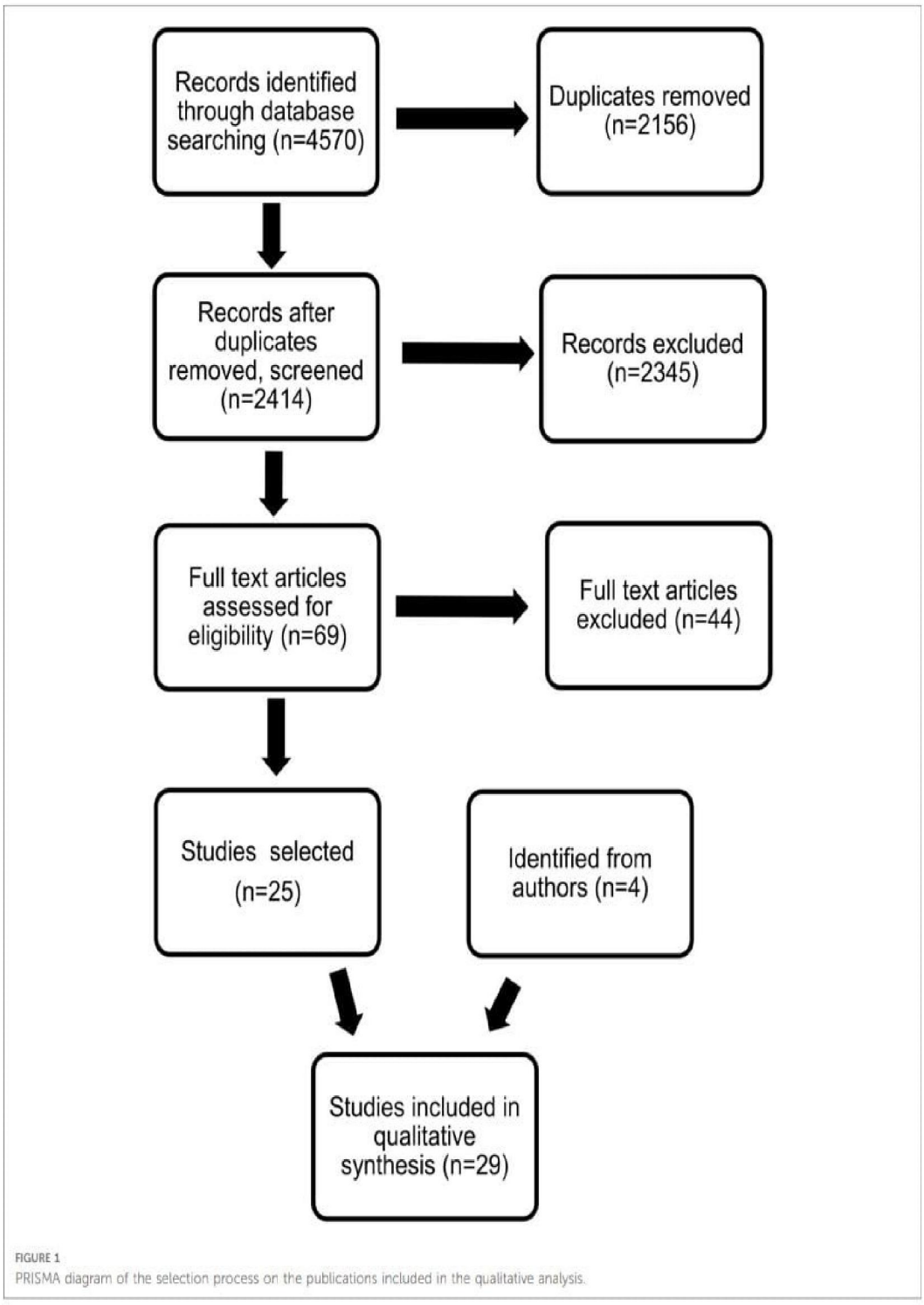

Fig.No.3 PRISMA diagram of the selection process of the publication included in qualitative analysis

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Prevalence:

Our review shows that over time, doctors and researchers have shifted from using both genetic testing and muscle biopsies to diagnose DMD, to mainly relying on genetic testing alone. Because of this, studies done many years ago may have included different types of patients than more recent studies.

We found two studies that reported how often DMD occurs at birth (birth prevalence) and five studies that reported how common it is in the population at a given time (point prevalence). Details of these studies are in the appendix.

When we looked at how well these studies were reported, both birth-prevalence studies were rated as medium quality, but neither gave enough information about who was included in the study. Among the five point-prevalence studies, two were medium quality but again did not clearly describe the participants. The remaining three were rated as low quality because they did not properly explain important aspects of the study—such as the study design, who was eligible to take part, or details about the participants.[37]

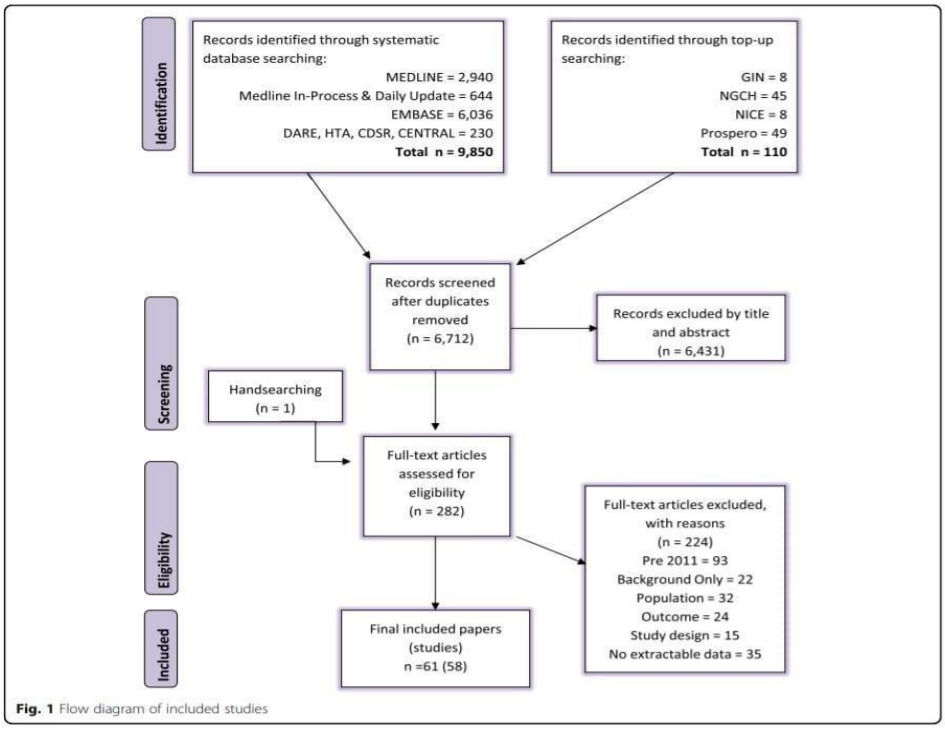

Fig No.4 Flow diagram of included studies

IV. DISCUSSION

We carried out a systematic review looking at recent (post-2010) evidence on the burden of Duchenne muscular dystrophy (DMD), including how common it is, how the disease progresses, its costs, treatment patterns, and current care guidelines.

From our search, we found 9,850 titles. After reviewing them, 58 studies were assessed for quality: 3 were judged high quality, 33 medium quality, and 22 low quality.[38-40]

Prevalence and Diagnosis

Two studies reported birth prevalence using newborn screening, and five studies reported how common DMD is at a given point in time. Over the years, there has been a shift away from using both genetic testing and muscle biopsy toward relying on genetic testing alone. This makes it harder to compare older and newer studies because the diagnostic methods have changed. Many studies also did not clearly describe who was included, adding to the uncertainty.[41]

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Mortality and Life Expectancy

We identified three studies examining death rates in DMD. Overall, people with DMD are living longer than before. This improvement is linked to wider use of corticosteroids, better availability of ventilation, and more detailed care guidelines.[42]

For example, a French study reported that boys born between 1970–1994 had a median survival of nearly 41 years, compared with about 26 years for those born between 1955– 1969. Diagnostic method also appeared to influence survival: patients diagnosed using molecular (genetic) tests had higher reported mortality compared to those diagnosed clinically alone. This could mean that earlier estimates of survival for "true DMD" may never be completely accurate, since reliable genetic testing was not available before 1987. Naturally, this also affects how accurately prevalence can be estimated.[43]

Disease Severity and Progression

We found 41 studies that looked at how severe DMD becomes and how it progresses. Many people with DMD have significant dependence on others: between 22% and 56% had lost the ability to walk, and between 27% and 57% had developed cardiomyopathy. Severity increases with age—loss of walking typically occurs around age 12, and ventilation often begins around age 20.[44]

Some studies highlight how progression differs between subgroups. For example, in one study, boys who could walk further when they first joined the study lost the ability to walk much more slowly over the next 3 years than boys who could walk only short distances. Another French study tracked changes in respiratory function, offering insight into how function and quality of life decline over time.[45]

Comparing studies is difficult because diagnostic methods vary, and most studies did not describe their participants in enough detail.

Treatment Patterns

Fourteen studies reported on treatment use, showing wide variation between countries. The use of corticosteroids, surgery for scoliosis, ventilation support, and physiotherapy differed internationally. One study also found important differences in steroid access between ethnic groups. Again, many studies did not report participant characteristics well enough.[46]

Quality of Life

Thirteen studies reported on health-related quality of life (HRQoL) or utility values. The PedsQL was the most commonly used tool for HRQoL. For utility values, the HUI was used in several countries. Some measures are better at reflecting disease progression than others: for example, generic PedsQL scores do not seem to track disease worsening very well in DMD.[47]

Interestingly, two studies found that self-reported quality of life did not decline much as the condition worsened physically. This may reflect psychological adaptation. Parents, however, tended to provide lower quality-of-life ratings than the boys themselves.

Costs

One study suggested that indirect costs (such as lost productivity) are higher than direct medical or social-care costs in early and late stages of the disease, but not necessarily in the middle stages. This shows the importance of understanding disease stage when planning financial and social support.[48]

Strengths and Limitations of the Review

A key strength of our review was that we used established systematic review methods across multiple areas of disease impact. The main limitation was that we restricted our search to 2011–2015, as more recent studies tend to use better diagnostic techniques and more accurately distinguish DMD from Becker muscular dystrophy (BMD).

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However, we found very few newer studies on prevalence, incidence, or mortality published since 2010—and earlier systematic reviews also identified very few studies from before 2010. This gives us some confidence that we did not miss many relevant studies.[49]

To our knowledge, no previous systematic reviews had covered treatment, progression, severity, or utility in a comprehensive way, making our work the first to do so.

Evidence Gaps and Future Directions

Important gaps remain—especially in prevalence, life expectancy, and treatment patterns. These may be filled as more patient registries emerge. The TREAT-NMD network, launched in 2007, hosts information on 49 national registries worldwide and could become a valuable resource for more accurate and timely prevalence data.

Natural history studies run by the Cooperative International Neuromuscular Research Group (CINRG) are also important sources for future insights.[50]

RECOMMENDATIONS:

We suggest that future research should not only rely on natural history studies but also make greater use of patient registries. Registries can be a valuable source of information for estimating how common DMD is, how it is treated, how effective those treatments are, and how the condition varies in severity, progression, and survival.

Registries have several important advantages. They usually follow standard criteria (sometimes even internationally), they can be updated regularly, and they make it possible to link different types of patient information. However, before relying on registries, it's important to check how complete they are (not all patients may be included) and to ensure good data quality—for example, avoiding duplicate entries and keeping information current as new cases arise or as patients pass away.

We also recommend that any future studies aiming to measure the overall burden of DMD include people of all ages and all disease stages. At present, very few prevalence studies exist, and the ones that do can't be reliably compared because each uses different age ranges to define the population. To improve this, studies should report prevalence in relation to the entire male population, since this better reflects the changing age distribution of people with DMD. This approach would provide a more complete picture of the burden of DMD across a whole country or region and could help increase clinical awareness.[51]

Although we found some good-quality evidence on the costs associated with DMD, only one study compared costs between countries. More research is needed to understand how costs vary across different disease stages. This would help capture the full economic impact of treatments that might slow progression or extend life. In addition, more research is needed to understand how comorbid conditions—such as scoliosis— affect quality of life and healthcare costs. The impact on caregivers' quality of life is also an area that has not been well studied.

Finally, across all types of studies, better standardisation in reporting is needed. Many papers did not clearly describe who was eligible to participate or provide adequate information about the people included in the study.[52]

V. CONCLUSIONS

A review of studies published since 2010 found 58 papers that looked at Duchenne muscular dystrophy (DMD) in terms of how common it is, its costs, impact on quality of life, and care guidelines.

There are still important gaps in our knowledge, especially around how many people have DMD and how long they live. However, people with DMD are generally living longer today. This improvement is likely due to more widespread use of corticosteroids, better access to ventilation, and more detailed care guidelines.[53]

Because people are living longer, studies that only focus on younger patients no longer give a full picture of the disease's impact. Research that includes older patients and those with advanced disease is still limited but increasingly important.

The severity of DMD is high. At any given time, in any country, roughly 22–56% of patients have lost the ability to walk, and 27–57% have heart problems. Disease severity rises with age: most patients lose the ability to walk around age 12, and many start needing ventilation around age 20.

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Comparing studies over time is difficult because the way DMD is diagnosed has changed. Earlier studies often used both genetic testing and muscle biopsy, whereas more recent studies mostly rely on genetic testing alone.[54]

DMD also has significant indirect costs, such as caregiver time and lost productivity, which grow as the disease progresses. Understanding these costs is important for planning health and social care support.

The main takeaway from this review is that using patient registries more widely could help track DMD over time and across countries, making data more comparable and useful.

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