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Electro-clinical Profile of Suspected Neuromuscular Diseases in Children Referred to A Tertiary Care Hospital Neurophysiology Laboratory

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Abstract

Background: *There has been a growing interest in childhood neuromuscular diseases due to the advancement of neurophysiological tests in Bangladesh recently. This diagnostic tool is very useful in identifying various neuromuscular disorders. This study aims to observe the electro-clinical profile of patients with suspected neuromuscular diseases referred to a neurophysiology laboratory.*

Methodology: *This cross-sectional study was conducted in the Neurophysiology Laboratory of the Paediatric Neuroscience Department at Bangladesh Shishu Hospital and Institute from May 1st, 2024, to April 30th, 2025. During this period, children with suspected neuromuscular disease underwent nerve conduction studies and, when appropriate, needle electromyography (EMG). Demographic and clinical data were collected from the referral notes and from the parents.*

Results: *Among 115 children with suspected neuromuscular disease, the most common presenting complaints were acute flaccid paralysis (40%), progressive limb weakness (28.7%), and traumatic injury (13%). Other symptoms included limb pain (10.4%), generalized floppiness (5.2%), periodic paralysis (1.7%), and isolated facial nerve palsy (0.9%). Nerve conduction studies (NCS) showed abnormalities in 67.8% in cases. EMG was performed in 26(22.6%). 15.65% participants showed abnormal EMG findings. Guillain-Barré Syndrome was the most common electro diagnosis (40%), followed by traumatic neuropathy (7%), non-inflammatory myopathy (6%), spinal muscular atrophy (5.2%), polyneuropathy (3.5%), inflammatory myopathy (2.6%), motor neuron disease (1.7%), plexopathy (1.7%), and facial nerve palsy (0.9%). Abnormal electrodiagnostic findings were significantly associated with suspected GBS, progressive disease course, and non-specific/symptom-based referral diagnosis, while age and sex showed no significant association.*

Conclusion: *This study highlights the high diagnostic yield of electrodiagnostic testing in children with suspected neuromuscular disorders, with nearly two-thirds demonstrating abnormal NCS findings. Abnormal electrodiagnostic findings were significantly associated with suspected GBS, progressive disease course, and non-specific/symptom-based diagnosis, emphasizing the importance of appropriate clinical diagnosis.*

Key words: *Electro-clinical profile, Neuromuscular disease, Nerve conduction study (NCS), Electromyogram (EMG), Guillain-Barré syndrome (GBS)*

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Introduction

Paediatric neuromuscular disorders (NMD) are conditions that impact a child's peripheral nervous system and skeletal muscles. These commonly present with symptoms such as hypotonic (low muscle tone), muscle weakness, and hyporeflexia (reduced reflexes[1]. These disorders affect the motor unit, which comprises the lower motor neuron (anterior horn), the peripheral nerve, the neuromuscular junction, and the muscle [2].

The muscle weakness associated with these conditions can significantly hinder a child's development, potentially leading to delays in achieving gross motor milestones, gait abnormalities, and, in severe cases, even the loss of independent ambulation.[3]

Diagnosing and managing paediatric neuromuscular disorders typically requires a systematic and multidisciplinary approach[4]. Electrodiagnostic (EDX) studies, specifically nerve conduction studies (NCS) and needle electromyography (EMG), are considered the most effective diagnostic tools for evaluating children with suspected neuromuscular disorders[5]. However, electrodiagnostic results alone are not pathognomonic for a specific disease and cannot provide a definitive diagnosis; they are considered a continuation of the clinical examinations[6].

While performing EDX studies in children can be quite challenging due to poor tolerability, difficulty in cooperation, technical difficulties, and interpretational challenges. Despite these hurdles, EDX studies can provide crucial information to differentiate between neuropathy and myopathy[7].

Routine NCS typically involves testing the motor and sensory fibres of the median, ulnar, and radial nerves. It also assesses the motor fibres of the peroneal and tibial nerves, and the sensory fibres of the superficial peroneal and sural nerves. Less frequently, the facial and

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accessory nerves may be tested. While standard NCS evaluates the distal nerve segments, late responses like H-reflexes and F-wave latencies offer valuable insights into the proximal nerve segments [8].

Needle EMG is performed by inserting a needle containing a recording electrode into the muscle of interest. Abnormal spontaneous activity, the recruitment pattern, the motor unit potential (MUP) duration, shape and amplitude are evaluated with minimal and maximal activation. Repetitive nerve stimulation (RNS) and single-fibre EMG (SFEMG) are utilized when a neuromuscular junction (NMJ) problem is suspected.[7]

In Bangladesh, there's been a growing focus on childhood neuromuscular diseases due to advancements in nerve conduction study (NCS) technology. However, access to this crucial diagnostic tool remains limited. The high cost of equipment and the specialized expertise required restrict its availability to only a few centres across the country. Furthermore, performing nerve conduction studies on children presents unique challenges, making their application in paediatric cases particularly scarce.

Amidst this challenge, Bangladesh Shishu Hospital recently established nerve conduction and EMG services at its neurophysiology laboratory. The hospital's newly established neurophysiology laboratory now offers comprehensive nerve conduction and electromyography (EMG) studies. With a team of experts, Bangladesh Shishu Hospital is successfully performing these vital diagnostic tests on children suspected of having various neuromuscular diseases, significantly improving the landscape of paediatric neuromuscular care in the country.

This study aims to see the clinical and electrodiagnostic profiles of children referred for suspected NMDs to a neurophysiology laboratory in a tertiary care setting and to analyse the correlation between clinical presentation and electrophysiological findings.

Methodology

This is a cross-sectional study conducted in the Neurophysiology Laboratory of Bangladesh Shishu Hospital and Institute over a period of one year, from May 1st, **2024**, to April 30th, **2025**. Children referred with various signs and symptoms suggestive of neuromuscular disorders, such as acute flaccid paralysis, hypotonic/floppiness, polyneuropathy, weakness of limbs, trauma, facial nerve palsy, or plexopathy, were included in the study. Acute flaccid paralysis was defined as an acute onset of focal weakness or paralysis characterized as **flaccid** (reduced tone) without other obvious cause (e.g., trauma) in children < 15 years old [9]. Socio-demographic and clinical information were collected from referral notes. Upper, middle and lower income families were categorized according to household monthly income.[10] Before the procedures, parents or guardians were counselled, and informed consent was obtained. A thorough clinical history and detailed neurological examination were performed by the attending physician before initiating the electrophysiological studies. Nerve conduction studies (NCS) were carried out following standard protocols, tailored according to the limb(s) involved[11]. When clinically indicated, electromyography (EMG) and repetitive nerve stimulation (RNS) tests were also performed. Before EMG, bleeding time (BT), clotting time (CT), and hepatitis B surface antigen (HBsAg) status were checked to ensure safety. For the upper limbs, motor and sensory conduction studies were performed on the median, ulnar, and radial nerves.

In cases where brachial plexopathy was suspected, additional motor nerve conduction studies were conducted on the musculocutaneous, axillary, and suprascapular nerves. In the lower limbs, motor conduction was assessed in the tibial and peroneal nerves, while sensory conduction studies were performed on the sural nerve. For all motor nerves, F-waves were recorded, and H-reflexes were evaluated specifically in the tibial nerve. The parameters analysed included distal latency, compound muscle action potential (CMAP) amplitude, and conduction velocity. EMG findings were interpreted based on motor unit action potential (MUAP) characteristics. Normal NCS and EMG in children were defined as age-appropriate nerve conduction parameters and electromyography findings without evidence of axonal loss, demyelination, spontaneous muscle activity, or abnormal motor unit recruitment, based on established paediatric normative data[11]. Myopathy was diagnosed when low-amplitude, polyphasic MUAPs were observed, along with early recruitment and a full interference pattern[1211]. Neuropathy was identified when high-amplitude, polyphasic MUAPs were present, accompanied by reduced recruitment, decreased interference pattern, and increased insertional activity[11]. Spontaneous activity in EMG refers to abnormal electrical discharges recorded from a muscle at rest, when the patient is completely relaxed. In normal muscle, there should be electrical silence at rest. All Spontaneous activities are normal except potential that occur in the muscle end plate. GBS is defined as an acute, immune-mediated polyradiculoneuropathy characterized by rapidly progressive, symmetrical flaccid weakness with areflexia

or hyporeflexia, reaching nadir within 4 weeks, often following an infection, with supportive evidence from CSF (albuminocytologic dissociation) and nerve conduction studies [12]. SMA is a genetic lower motor neuron disorder caused by SMN1 gene mutation, presenting with progressive symmetrical proximal muscle weakness, hypotonia, muscle atrophy, and areflexia, with preserved sensation[13]. Muscular dystrophy is a group of inherited myopathies characterized by progressive muscle weakness and wasting, elevated serum CK, and structural muscle fibre degeneration with replacement by fat and fibrosis[14]. Myopathy is a primary muscle disorder presenting with symmetrical proximal muscle weakness without sensory loss or significant reflex changes (early), due to structural, metabolic, inflammatory, or toxic muscle pathology[11]. HMSN is a genetically determined chronic peripheral neuropathy characterized by slowly progressive distal muscle weakness and atrophy, sensory loss, reduced or absent reflexes, and characteristic nerve conduction abnormalities[15]. The course of the disease was categorized as either progressive or non-progressive. For Guillain-Barré Syndrome (GBS), classification was based on the Erasmus GBS Respiratory Insufficiency Score (EGRIS). Neuromuscular disorders of suspected genetic origin—such as spinal muscular atrophy (SMA), muscular dystrophies, myopathies, and hereditary motor and sensory neuropathies (HMSN)—were classified as progressive in nature [16]. Patients were divided into two groups: the normal electrodiagnostic group and the abnormal electrodiagnostic group. These two groups were compared with the clinical profile of the patients.

Data were processed and analyzed by SPSS 23 (Statistical Program for Social Sciences. Categorical variables were presented in frequency and percentage. Pearson's chi-square or Fisher's exact test, as appropriate, was applied for qualitative variables. A P-value less than 0.05 was considered to be statistically significant.

Results

This study included a total of 115 children aged 3months to 18 years with suspected neuromuscular disease. Males were 58.3% and females were 41.7%. The majority of children (68.7%) were referred from the outpatient department. Middle-income families were over-represented (49.6%). Among the referred cases, the most common presenting complaint was acute flaccid paralysis (40%). Other significant presenting features included progressive weakness of limbs with wasting or hypertrophy (28.7%), traumatic injury (13%), non-specific pain with weakness of a single limb (10.4%), generalized floppiness (5.2%), periodic paralysis (**1.7%**), and isolated facial nerve palsy (0.9**%**). (Table I)

Table-I

Demographic and clinical/referral characteristics of patients with suspected neuromuscular disease

Variables	Category	Frequency(n)	Percentage (%)
Age	3M-<5Y5Y-10Y>10Y-18Y	443932	38.333.927.8
Sex	MaleFemale	6748	58.341.7
Income	Lower income	30	26.1
	Middle Income	57	49.6
	Higher income	28	24.3
Referral Unit	Outpatient department	79	68.7
	Inpatient Department	36	31.3
Referral diagnosis	i)Specific/Aetiology oriented	70	60.8
	without complications	46	40.0
	Traumatic injury	15	13.0
	Floppiness	6	5.2
	Periodic paralysis	2	1.7
	Isolated facial nerve palsy Acute	1	0.9
	flaccid paralysis with or		
	ii) Non-specific/Symptom-based	45	39.1
	Weakness and wasting of limbs	33	28.7
	Pain of single limbs	12	10.4
Disease course	Progressive	44	38.3
	Non progressive	71	61.7

All the participants underwent NCS. Electromyography (EMG) was performed in 26(22.6%). 15.65% participants showed abnormal EMG findings (Figure-1)

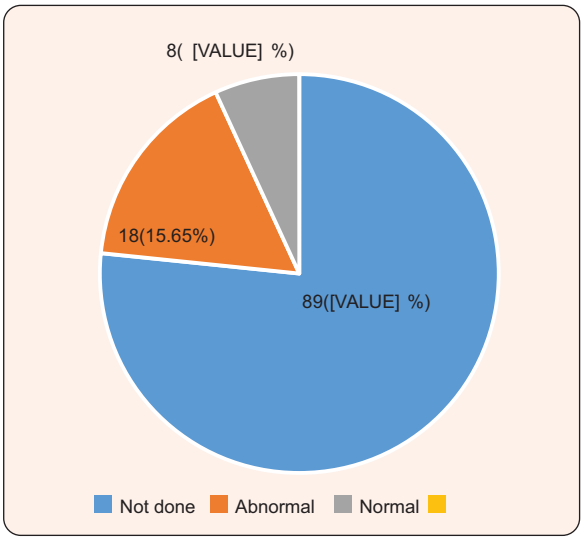

Figure-1: *Distribution of patients according to EMG findings*

Nerve conduction studies (NCS) revealed abnormalities in 67.8% of the children. Various subtypes of Guillain-Barré Syndrome (GBS) was the most frequently identified electrodiagnosis, accounting for 40% of cases. Other electrodiagnostic outcomes included traumatic neuropathy (7%), non-inflammatory myopathy (6.1%), spinal muscular atrophy (5.2%), polyneuropathy (2.6%), inflammatory myopathy (2.6%), motor neuron disease (1.7%), plexopathy (1.7%), and facial nerve palsy (0.9%).

The bar diagram illustrates the number of patients with normal and abnormal NCS findings stratified by reason for referral. Among all categories, 'AFP without complication' showed the highest number of abnormal NCS results (n = 34), followed by 'Weakness of limbs' (n = 14) and 'Traumatic injury' (n = 9). In contrast, 'Weakness of limbs' accounted for the highest number of normal NCS results (n = 19), followed by 'Nonspecific pain' (n = 11) and 'Traumatic injury' (n = 6).(Figure 2)

Figure 3 shows, among patients referred for acute flaccid paralysis (AFP), with or without complications, Guillain-Barré Syndrome (GBS) was the most common diagnosis (34 and 11, respectively), underscoring the high diagnostic yield of NCS in such cases. Referrals for limb weakness revealed a broader diagnostic spectrum, including normal findings (19), non-inflammatory

Table- II

Electro-clinical Diagnosis of suspected neuromuscular disease (n=115)

Electro-clinical Diagnosis	Frequency(n)	Percentage%
Normal	37	32.2
Abnormal	78	67.8
GBS	46	40.0
Traumatic Neuropathy	8	7.0
Non- inflammatory Myopathy	7	6.1
Spinal Muscular Atrophy	8	7.0
Polyneuropathy(hereditary motor sensory neuropathy-HMSN)	3	2.6
Inflammatory Myopathy	3	2.6
Plexopathy	2	1.7
Facial nerve palsy	1	0.9
Total	115	100

myopathy (7), inflammatory myopathy (3), spinal muscular atrophy (n=2), and polyneuropathy (2), reflecting the heterogeneity of underlying conditions. Floppiness was exclusively linked to spinal muscular atrophy (6), highlighting the role of NCS in diagnosing pediatric neuromuscular disorders. Referrals for nonspecific pain mostly yielded normal results (11), indicating a lower diagnostic yield. Rare but specific diagnoses included facial nerve palsy (1) and periodic paralysis, with one case each due to GBS and polyneuropathy. (Figure 3)

Table III shows that all patients clinically diagnosed with GBS showed abnormal NCS findings. This association is statistically significant with extremely higher odds(OR=101.87;p<001), indicating a very strong link between GBS diagnosis and abnormal NCS results. A Progressive course is associated with higher odds of abnormal NCS (OR=3.7; p-0.004). Children referred with a specific/aetiology-oriented clinical diagnosis were much less likely to have abnormal findings compared to those referred with non-specific/symptom-based complaints, and this association was highly statistically significant (0.003). (Table III)

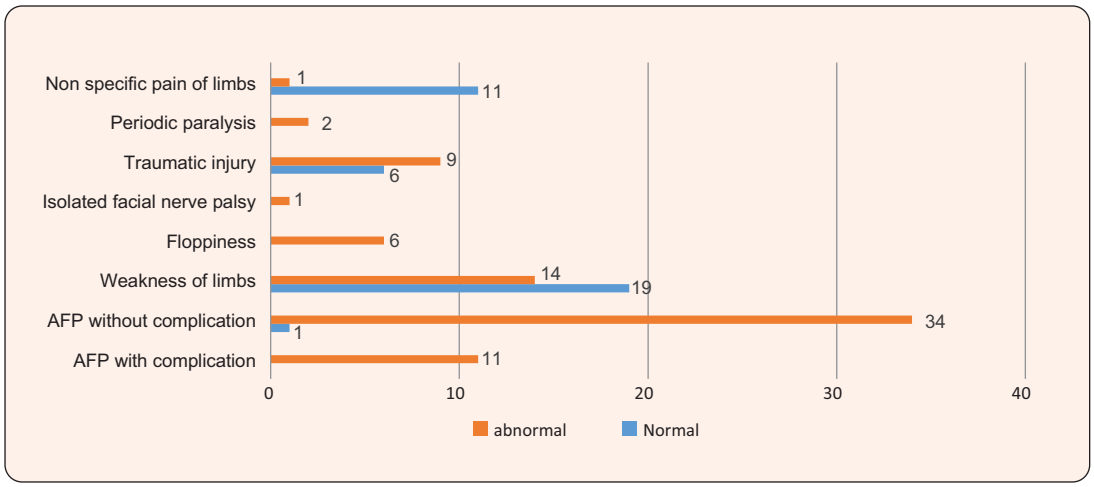

Figure- 2: *The frequency of normal and abnormal electrodiagnostic findings across different referral diagnoses.*

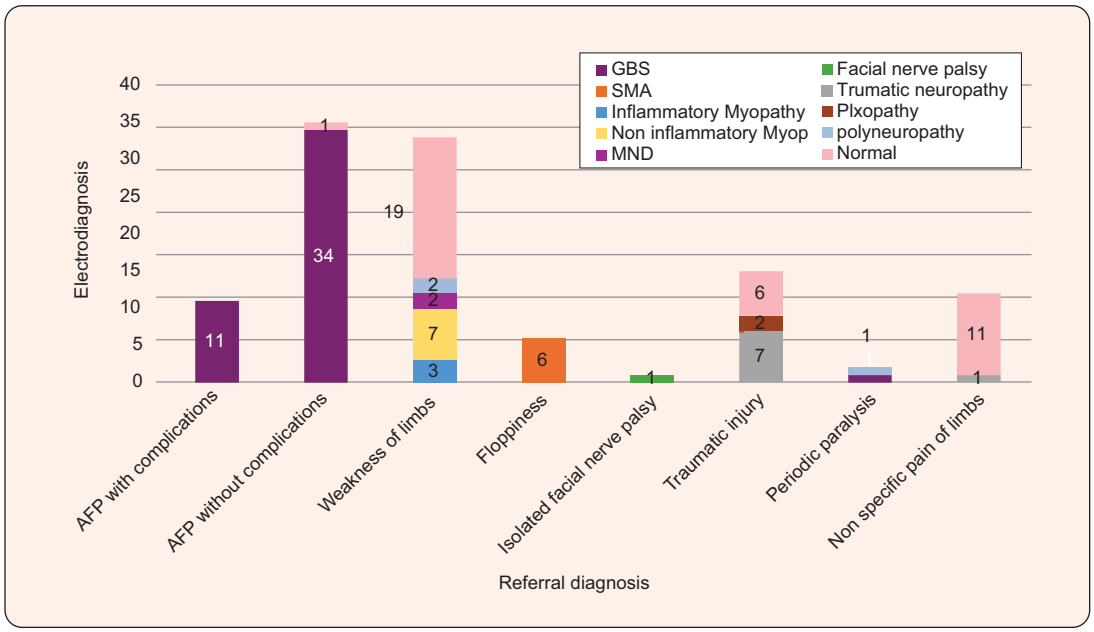

Figure- 3: *The distribution of electroclinical diagnoses across various referral diagnoses.*

Table-III

Association between clinical profile and electro diagnosis

Variables	Abnormal EDX (n_1=78)	Normal EDX (n_2=37)	Odd ratio	95%CI	P value
Age <5 Y	33	11	1	0.21-1.36	0.403[1]
5Y-10Y	24	15	0.53	0.23-1.73	
>10Y	21	11	0.64		
Sex					
Male	48	19	1.69	0.77-3.72	0.228[1]
Female	30	18	1		
Clinical diagnosis					
Suspected GBS	45	0	101.87	6.04-1718.81	<001[2]
Other than GBS	33	37	1		
Course of Illness					
Progressive	37	7	3.78	1.52-9.85	0.003[1]
Non-Progressive	41	30	1.00		
Referral diagnosis					
Specific/Aetiology Oriented	63	7	0.056	0.020-0.150	<0.001[1]
Non-specific/symptom based	15	30			

1-Chi-square test, 2-Fisher exact test

Discussion

In this study, middle-income families were overrepresentation of (49.6%), and outpatient referrals were (68.7%). This suggests that access to electrodiagnostic services may still be limited for lower-income families, warranting policy-level attention to improve accessibility and equity. The findings are dissimilar to Fraser et al, who found that a total of 55.3% (166) of all referrals came from areas of the highest deprivation [17]and Ali Shazima et al found that a majority of the children (81.9%) were from rural areas [18].

In this study, the most common presenting complaint was acute flaccid paralysis (AFP), accounting for 40% of cases. This finding is consistent with Hage et al., who reported AFP in 40.6% of children under 5 years and 37.9% in children aged 5–9 years[19]. Similarly, a national survey conducted in Bangladesh in 2007 found that 46% of pediatric neurology referrals were due to AFP [20]. The next most common complaints in this study were progressive weakness of the limbs (28.7%), traumatic injury (13%), non-specific pain with weakness in a single limb (10.4%), and generalized floppiness (6.9%).

These findings are consistent with Akhlaque U et al who found commonly reported symptoms at the time of electrodiagnostic referral included limb weakness (36%) and lower limb pain (1.7%) [21]. In contrast, a study from East Africa by Zewde et al found motor weakness to be the most frequent reason for referral (40.2%), followed by trauma or iatrogenic complications (10.5%), and a suspected diagnosis of motor neuron disease in 7.7% of cases[22].

In our study, nerve conduction studies (NCS) were performed in all patients, and needle electromyography (EMG) was conducted in 26(22.6%) of cases where clinically appropriate. Electrodiagnostic abnormalities were found in 67.8% of participants, showing a similar pattern to that reported by Akhlaque U et al. (56%), [17] and slightly lower, observed by Zewde et al (73.2%)l[22].

Guillain-Barré Syndrome (GBS) was the most frequent electrodiagnosis in our study, identified in 40% of cases. This is comparable to the findings of Ali et al., who reported GBS in 47% of AFP cases[18]. Similarly, Alshekhlee et al. identified GBS as a leading cause of acute neuromuscular weakness in both pediatric and adult populations[23].

In this study, a diverse spectrum of electrodiagnoses was observed in children presenting with weakness, including inflammatory myopathies (2.6%), non-inflammatory myopathies (6.1%), spinal muscular atrophy (6.9%), and polyneuropathy due to hereditary motor and sensory neuropathy (2.6%). These findings indicate both myopathic and neuropathic causes underlying the clinical presentation. Notably, all children presenting with generalized floppiness were electroclinically diagnosed with SMA, demonstrating 100% concordance between clinical suspicion and electrodiagnosis. These rates are comparable to those reported by Levison et al., where SMA and myopathies together accounted for approximately 10% of confirmed neuromuscular diagnoses in a large pediatric cohort[24]. However, our findings differ from those of Akhlaque et al., who reported a higher prevalence of peripheral neuropathies due to injection neuritis and trauma (14.5%), polyneuropathy (13%), and non-inflammatory myopathy (29%), with 71% of cases diagnosed as inflammatory myopathy[21]. A recent study by Nastasi et al. also reported similar findings, with motor neuron disease accounting for 9% and myopathy for 14% of electrodiagnoses [25].

Furthermore, plexopathy (1.7%) and isolated lower motor neuron facial palsy (0.9%) were identified in this study, further highlighting the diversity of referrals and the broad utility of neurophysiological studies in pediatric practice. Similar findings were reported by Akhlaque et al., who observed facial nerve palsy in only 0.4% of cases, on the other hand plexopathy was more frequent, occurring in 14% which was dissimilar to this study [21].

Finally, a statistically significant association was found between abnormal electrodiagnostic findings and several clinical variables, including suspected Guillain-Barré Syndrome (GBS), a progressive disease course, and the presence of a non-specific/symptom based diagnosis at the time of referral, that highlights the importance of appropriate patient selection and supports the role of electrodiagnostic studies as a valuable tool in evaluating children with unexplained neuromuscular symptoms rather than for routine confirmation of clinically obvious diagnoses.. These findings support previous observations that children with more defined and progressive symptoms are more likely to exhibit electro diagnostic abnormalities, thereby facilitating early and appropriate management [21].

This study had several limitations. It was conducted at a single centre with a relatively small sample size, which may have limited the statistical power and generalizability of the findings. Additionally, cerebrospinal fluid (CSF) analysis, creatine phosphokinase (CPK) levels, muscle biopsy, and genetic testing were not available in most cases, which may have affected the diagnostic accuracy and comprehensive evaluation of neuromuscular disorders.

Conclusion

This study demonstrates a high diagnostic yield of electrodiagnostic testing in children with suspected neuromuscular disorders, with nearly two-thirds showing abnormal nerve conduction study findings. Abnormal electrodiagnostic results were significantly associated with suspected Guillain–Barré syndrome, underscoring the importance of appropriate clinical stratification before testing.

Conflict of interest: None

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