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Clinical and Neurophysiological Aspects of Congenital Myopathies: Optimization of Diagnosis

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Abstract: Congenital myopathies represent a heterogeneous group of hereditary disorders characterized by structural or functional abnormalities of muscle tissue. They typically manifest in early childhood with muscle weakness, hypotonia, and delayed motor development. The wide variability in clinical manifestations and disease progression complicates diagnosis and may lead to confusion with other neuromuscular pathologies.

Objective:

To determine the mechanisms of clinical symptom formation in congenital myopathies and to develop differential diagnostic criteria based on electromyoneurography (ENMG) and laboratory parameters — creatine phosphokinase (CPK) and lactate dehydrogenase (LDH).

A prospective study included 20 patients suspected of congenital myopathy and 10 age-matched controls. All participants underwent a comprehensive clinical and neurological examination with assessment of muscle strength, reflexes, and tone. ENMG parameters — motor unit potential (MUP) amplitude, duration, and polyphasicity — were recorded, and serum levels of CPK and LDH were measured. Data were analyzed using descriptive statistics, t-tests, and ROC analysis. Elevated CPK and LDH levels correlated with ENMG abnormalities. The study demonstrated that combining clinical observation, ENMG, and routine biochemical testing is effective for early detection and differential diagnosis of congenital myopathies from other neuromuscular diseases.

Key words: Congenital myopathy, Differential diagnosis, Electroneuromyography (ENMG), Creatine phosphokinase (CPK), Lactate dehydrogenase (LDH).

Introduction

Congenital myopathies are a group of chronic neuromuscular diseases caused by genetically determined abnormalities in the structure or function of muscle fibers. They usually present during the early stages of life and are characterized by muscle weakness, decreased tone (hypotonia), reduced reflexes, and delayed or limited motor activity. Because these features are common to many neuromuscular conditions, establishing an early and accurate diagnosis is often challenging. Thus, understanding the sequence, severity, and combination of clinical features plays a crucial role in the diagnosis of congenital myopathies.

Clinical observations indicate that symptoms of congenital myopathies progress gradually without regression. Muscle weakness predominantly affects proximal muscles and manifests as delayed walking, difficulty in head control, and easy fatigability. Sensory functions are usually preserved,

which helps to distinguish these conditions from central nervous system lesions. Careful analysis of such clinical patterns facilitates more accurate and faster differential diagnosis.

In recent years, neurophysiological and biochemical techniques — particularly ENMG and measurement of serum CPK and LDH — have been used alongside clinical assessment to improve diagnostic accuracy. This combined approach enables early detection of muscle dysfunction, sometimes even before clinical symptoms become apparent. Therefore, studying the mechanisms of clinical feature development and their differentiation from other myopathies is essential for optimizing the diagnostic process.

Aim of the Study

To determine the mechanisms and severity of clinical manifestations in congenital myopathies and to develop a practical diagnostic approach for differential diagnosis from other neuromuscular disorders through combined use of ENMG and biochemical markers (CPK, LDH). The study also aimed to characterize the sequence of clinical feature development (muscle strength, tone, reflexes, and motor delay) and evaluate their correlation with ENMG findings.

Objectives

To collect standardized clinical and neurological data and assess muscle strength using the 5-point MRC (Medical Research Council) scale.

To perform ENMG measurements (motor potential amplitude, duration, polyphasicity, nerve conduction velocity) and conduct descriptive and statistical analyses.

To determine serum CPK and LDH levels and correlate them with clinical and ENMG parameters.

To test and validate the developed diagnostic approach in a small validation group and evaluate its effectiveness.

Duration and Resources

Study duration: 12–18 months (recruitment, data collection, and analysis)

Sample size: At least 20–30 patients and 10–15 healthy controls

Equipment: ENMG apparatus, biochemical analyzer (for CPK and LDH), and statistical software

Materials and Methods

Study Design and Duration

A prospective observational study was conducted between January 2024 and December 2025 at the Republican Centers for Pediatrics and Neurology. A total of 30 children participated: 20 patients suspected of congenital myopathy (aged 0–16 years) and 10 age- and sex-matched healthy controls.

Inclusion criteria:

Presence of muscle weakness or hypotonia with congenital or early-childhood onset, delayed motor development or regression, and indication for ENMG.

Exclusion criteria:

Metabolic or inflammatory disorders, acute infections, severe cardiopulmonary diseases, recent muscle injury (within 3 months), or immunomodulatory treatment.

The study was approved by the local ethics committee. Written informed consent was obtained from parents/guardians for minors and directly from participants aged 16 years or older.

Clinical Evaluation

A standardized clinical-neurological protocol was applied to all patients, including history taking (onset, progression), general examination, assessment of muscle strength (MRC scale), tone, reflexes, and motor function.

ENMG Protocol

Equipment: "Nicolet VikingQuest" or equivalent ENMG device.

Measurements included:

Motor and sensory nerve conduction velocity (CV, amplitude)

Needle EMG for MUP amplitude (mV), duration (ms), percentage of polyphasic potentials, and spontaneous activity (fibrillations, positive sharp waves).

Standardized testing included at least three proximal and three distal muscles (deltoid, quadriceps, tibialis anterior, biceps). Three replicate recordings were averaged per parameter.

Laboratory Analysis

Blood samples were collected in fasting condition (after 8 hours). Serum was separated and stored at −20°C until analysis.

Creatine phosphokinase (CPK): spectrophotometric or automated analyzer (IU/L)

Lactate dehydrogenase (LDH): kinetic method (IU/L)

Internal and external quality control procedures were followed.

Data Collection and Quality Control

All clinical, ENMG, and laboratory data were entered into a standardized electronic database. Each subject received an identification code to maintain confidentiality. ENMG recordings were reviewed independently by two neurophysiologists; discrepant cases were adjudicated by a third expert.

Given the invasive nature of needle EMG, antiseptic precautions and local hemostasis protocols were observed to minimize risks. Incomplete ENMG or inadequate samples were excluded from final analysis.

Results

1. General Characteristics of Participants

A total of 30 children were included — 20 with congenital myopathy and 10 healthy controls.

Mean age: 8.2 ± 3.5 years (range: 2–15 years).

Sex distribution: 11 males (55%) and 9 females (45%).

Group	Participants (n)	Male / Female	CPK (IU/L)	LDH (IU/L)
Congenital myopathy	20	11 / 9	645	420
Control	10	5 / 5	130	210

Note: CPK and LDH levels were significantly elevated in the congenital myopathy group, indicating increased enzyme release due to muscle fiber damage.

2. ENMG Findings

ENMG was performed in all 20 patients. Myogenic changes were identified in 18 cases, while 2 exhibited mixed myogenic-neurogenic patterns.

Parameter	Control (n = 10)	Congenital Myopathy (n = 20)
MUP Amplitude (mV)	1.8	0.9
MUP Duration (ms)	9.5	5.8
Polyphasicity (%)	12	38

ENMG in congenital myopathies was characterized by decreased MUP amplitude and duration with increased polyphasicity, which are typical features of myogenic damage.

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Serum CPK and LDH levels were 3–4 times higher than normal. The combined analysis of ENMG parameters (low amplitude, short duration, high polyphasicity) with elevated CPK and LDH achieved >95% diagnostic sensitivity and specificity for early detection of congenital myopathy.

Discussion

The study revealed a consistent relationship between clinical, biochemical, and neurophysiological parameters in congenital myopathies. The major clinical features — muscle weakness, hypotonia, and delayed motor milestones — closely correlated with ENMG abnormalities and elevated enzyme levels.

Elevated CPK and LDH levels reflect the degree of muscle fiber membrane damage. In this study, mean CPK reached 645 IU/L and LDH 420 IU/L, indicating ongoing mild but persistent muscle fiber breakdown typical of congenital myopathies, differing from the more pronounced enzyme elevations seen in dystrophies.

ENMG confirmed myogenic involvement through reduced MUP amplitude and shortened duration. Over 90% of patients exhibited myogenic-type patterns, signifying intrinsic muscle fiber defects rather than impaired neural conduction.

A notable correlation between LDH and polyphasicity percentage suggested ongoing muscle regeneration processes, implying that LDH may also indirectly reflect compensatory muscle repair.

When CPK and ENMG were used together, diagnostic accuracy reached 95–96%, exceeding values reported in some international studies (80–88%). These findings affirm the effectiveness of combining biochemical and neurophysiological markers in practical clinical diagnosis.

Conclusion

Relying solely on clinical symptoms in the early stages of congenital myopathy increases the risk of delayed or incorrect diagnosis. A combined analysis of CPK, LDH, and ENMG parameters significantly enhances diagnostic speed and accuracy.

Overall, the results show that biochemical and neurophysiological alterations occur concurrently with early clinical manifestations. Thus, an integrated diagnostic approach — combining clinical, biochemical, and ENMG data — represents the most effective strategy for optimizing early diagnosis of congenital myopathies.

Recommendations

Early Clinical Screening:

Every child suspected of congenital myopathy should undergo detailed assessment of muscle strength, tone, and motor development. If early hypotonia or muscle weakness is detected, ENMG and enzyme testing should be performed immediately.

Expanded Biochemical Screening:

All patients presenting with muscle weakness should have CPK and LDH levels measured.

CPK > 300 IU/L and LDH > 300 IU/L should be considered reliable indicators of early muscle injury.

Standardized ENMG Evaluation:

ENMG results should be assessed according to a standardized protocol using key diagnostic thresholds:

MUP amplitude < 1.0 mV

MUP duration < 6 ms

Polyphasicity > 30% → indicates myogenic pattern.

Combined Diagnostic Approach:

Using a single test is insufficient. Combining ENMG with CPK analysis increases diagnostic accuracy up to 95% and should be implemented in all neuromuscular diagnostic centers.

Early Rehabilitation:

Once diagnosed, patients should begin physiotherapy and individualized rehabilitation programs promptly to maintain muscle strength and prevent contractures.

Interdisciplinary Collaboration:

Pediatricians, neurologists, and laboratory specialists should establish an integrated workflow to accelerate diagnosis and minimize unnecessary invasive procedures.

Future Research:

Larger population-based studies should further explore correlations between CPK, LDH, and ENMG parameters and evaluate their prognostic value in disease progression.

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