Ref: Ro J Neurol. 2023;22(4)
DOI: 10.37897/RJN.2023.4.9

Electromyography reveals the etiology of floppy infant in developing country

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ABSTRACT

Background. Floppy infants are correlated with the extensive differential diagnosis. It can make diagnostic approach become more challenging.

Case report. We report a 10-months-old infant referred from Pediatrician to electromyography (EMG) laboratory presenting with floppy and developmental delays. The central and motor neuron manifestations also increased of CPK levels bring ambiguity for the diagnosis. EMG may distinguish the cause from myopathy, anterior horn cell, neuromuscular junction (NMJ) or central origin when genetic testing is not routinely done in developing country.

Conclusion. EMG helps clinician to distinguish the diagnosis of floppy infant.

Keywords: floppy infant, electromyography, child health

List of abbreviations

CMAP – compound muscle action potential
CNS – central nervous system
CP – cerebral palsy
CPK – creatine phosphokinase
EMG – Electromyography
MUAP – Motor unit action potential
NCS – nerve conduction studies
NMJ – neuromuscular junction
PNS – peripheral nervous system
SMA – spinal muscular atrophy
SNAP – sensory nerve action potential

INTRODUCTION

Infant hypotonia (floppy infants) is a significant challenge for clinicians to identify the underlying cause. A baby with low muscle tone (hypotonia), low muscle strength (weakness), or ligamentous laxity with enhanced range of movement is referred to a "floppy infant" [1]. Further evaluation should be done to distinguish the etiology, suitable management, home program, and prognostic education regarding to specific diseases [2]. Pathology in the central nervous system (CNS), peripheral nervous system (PNS), neuromuscular junction (NMJ), myopathies, neurometabolic disease, and systemic sickness are possible causes of floppy presentation [3–5].

According to several studies, PNS causes for 15–30% of hypotonia and CNS causes for 60–80% of them [4,6]. On the contrary, research by Debnath et al. (2002) revealed 18.05% of hypotonia were central, 70.83% were peripheral, and 11.11% were mixed or unclassified. The primary causes of central hypotonia were cerebral palsy (CP), followed by brain abnormalities, hereditary, and metabolic. This motor development abnormality is linear with disturbance in maturation of cerebral connectivity, thought to foundation of adaptive ability [7]. Prevalence of CP is between 1.5 to 4 per 1000 live births, with an average of 2 per 1000 live births [8]. Myopathy and spinal muscular atrophy (SMA) were the two most common

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Article History:
Received: 20 December 2023
Accepted: 29 December 2023

diagnoses for peripheral causes. Electromyography (EMG) is one of the frequent used diagnostic tools to confirm the diagnosis of SMA. It measures electrical signals that associated with muscle-neural activity, as well as the timing and sequence of muscle activity [9]. The evaluation requires a well-organized comprehensive approach. In order to identify the cause, the history and clinical examination should be taken into consideration [1].

CASE REPORT

We present a case of a 10-months-old male, a floppy baby in whom SMA or muscular dystrophy was suspected. Informed consent was obtained from the parents. He was referred from the department of Pediatrics to the EMG laboratory of Physical Medicine and Rehabilitation (PMR) department (Figure 1). He was unable to raise his head yet and lying on bed the entire day. He also exhibited delay in other developmental domains, including language, fine motor, and interpersonal skill. He could smile and make eye contact. His left upper and lower extremities were more active than right. His developmental age was equivalent to 2 months. Feeding was through a nasogastric tube since birth due to mastication and swallowing difficulties.

Anamnesis and physical findings included weakness, weak crying, poor eating, and respiratory impairment may indicate SMA type 1 or an alternative central cause. On the contrary, this patient showed signs of spasticity and clonus that were linear with CNS signs, so the central origins cannot be ruled out. The most prevalent and severe kind of weakness is SMA type 1, which accounts for 45% of cases and mostly manifests after delivery until before 6 months old [10]. However, the supporting examination revealed that the creatine phosphokinase (CPK) levels had climbed to twice until three times of normal levels, therefore muscular dystrophy couldn't be excluded. Molecular genetic testing for diagnostic specificity is often difficult to obtain in developing countries. In such uncertainty of etiology, nerve conduction studies (NCS) and EMG data can provide objective information to support diagnosis [1,11]. These electrodiagnostic studies may help to classify the diagnosis as myopathy, neuropathy, anterior horn cell, NMJ disorder, or central hypotonia [1].

In this instance, the compound muscle action potential (CMAP) and sensory nerve action potential (SNAP) had normal latency, amplitude, and nerve conduction velocity. F-waves showed normal latency and persistency. Needle EMG findings showed normal insertional activity, no spontaneous activity, normal amplitude and duration, no polyphasic potential, and reduced interference pattern (Figure 2). The decreased of MUAPs might be related to the lower intensity of firing. This patient's normal results indicate the central abnormality as its cause [1].

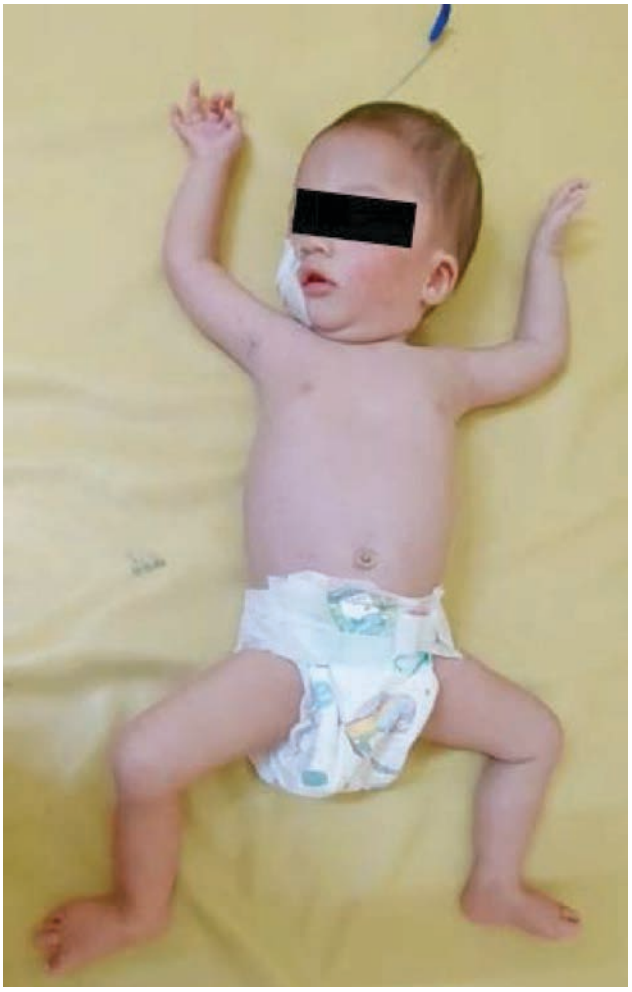

FIGURE 1. Floppy posture of the infant (Author's documentation)

DISCUSSION

The structural integrity of PNS can be evaluated using NCS. It aids in locating lesion, classifying it as axonal or demyelinating nature, determining the lesion's course and severity. The disorders of CNS are associated with normal EMG-NCS result. In demyelinating illness and axonal neuropathies, abnormalities in CMAP latency can occur [11]. The myopathic EMG pattern will exhibit low amplitude, polyphasic, brief MUAPs, and early recruitment. Disorder in anterior horn cell exhibits spontaneous fibrillation at rest, long-duration polyphasic MUAPs, diminished interference pattern along with normal latency and SNAPs in sensory NCS. In Congenital Myasthenic Syndrome (CMS), a decrement response at 2-3 Hz rates of stimulation in at least one muscle was seen as evidence of faulty neuromuscular transmission. EMG-NCS has a sensitivity of 80% for SMA and a sensitivity of 75% for myogenic case [1,12]. In underdeveloped nations, EMG might be a better test than genetic or muscle immunohistochemical research.

CPK is an intracellular enzyme found in the brain, myocardium, and skeletal muscle. CPK maintains

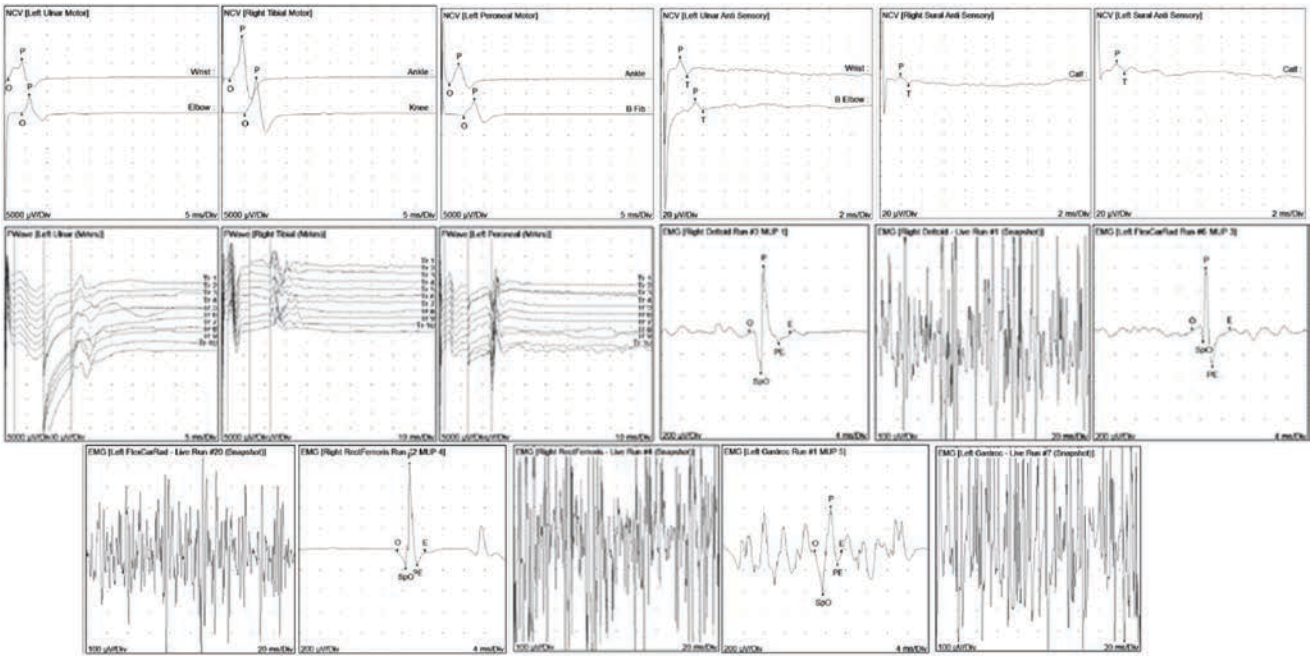

FIGURE 2. Findings from the NCS-EMG test (Author's documentation)

muscular mass. The cell membrane which damaged by hypoxia or any other kind of injury causes the release of CPK from the cytosol into the bloodstream. Duchenne muscular dystrophy is a degenerative muscle illness brought on by altered dystrophin protein, and high CPK levels can be used as an important biomarker for this disease. Exercise, minor trauma, and post-prolonged seizures are a few examples of physiological factors other than pathology that may contribute to a rise in CPK levels [13,14]. In SMA and congenital myopathies, it may also be modestly raised [1].

CONCLUSION

In summary, it is reasonable that the patient's condition is caused by CNS origin since the EMG-NCS results reveal no signs of motor unit disruption. This finding showed that EMG may become a useful diagnostic testing for floppy infant in the developing countries.

Conflicts of interest:
No potential conflict of interest relevant to this article was reported.

Author's contributions:
Conceptualization: PTP, SMW. Data curation: PTP, RDS, SMW. Methodology: PTP, SMW. Project administration: PTP. Visualization: PTP. Writing - original draft: PTP. Writing - review and editing: PTP, SMW, RDS. All authors have read and approved the submitted manuscript. The manuscript has not been submitted elsewhere nor published elsewhere in whole or in part.

Funding:
The authors did not receive financial support for the manuscript and or for publication.

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