

Frequency distribution of types of Floppy infants and role of Electromyography and Nerve Conduction Studies in their diagnosis

SARFRAZ AHMAD¹, SYEDA RAHAT²

¹Assistant Professor of Physical Medicine & Rehabilitation, The Children's Hospital & Institute of Child Health, Lahore.

²Physiotherapist (CH & ICH)

Correspondence: Dr. Sarfraz Ahmad, Email: dr_sarfraz@hotmail.com Cell: 0300-4232055,

ABSTRACT

Background: Floppy infant is one with hypotonia at birth or in early infancy. Hypotonia is a symptom associated with disorder of brain, spinal cord, nerves and muscles.

Aim: To determine the frequency distribution of types of floppy infants and to determine the usefulness of EMG & NCS study in diagnosis of different types of floppy infants and children.

Study design: Descriptive study

Place and duration of study: The Department of Physical Medicine & Rehabilitation, the Children's hospital & ICH Lahore from June 2017 to June 2018.

Methodology: Sixty infants and children under 02 years of age having floppiness since birth were included in this study. After detailed history, physical examination and investigation, a clinical diagnosis was made and then EMG & NCS was performed on same group of patients and final diagnosis was made and compared it with clinical diagnosis.

Results: Final electrophysiological study showed that 46.6% were diagnosed as SMA, 36.6% were diagnosed as Central Hypotonia and 16.6% were diagnosed as Congenital Myopathy. Of total 60 patients, 30% patients had a final electrophysiological (EMG & NCS) diagnosis different from diagnosis made on clinical grounds and 42 (70%) had similar diagnosis. Chi square test of statistical significance was applied by using SPSS version 25. P value < 0.05 showed the significance of EMG & NCS studies.

Conclusion: We concluded that most common type of floppy infants and children is SMA followed by central hypotonia and congenital myopathy and EMG & NCS is the best diagnostic tool for diagnosis of floppy infants.

Keywords: SMA, EMG & NCS, Congenital Myopathy, Central Hypotonia

INTRODUCTION

A floppy infant is one with hypotonia at birth or in early infancy^{1,2}. Hypotonia or floppiness is demonstrated by low resting muscle tone, diminished stretch reflexes and frequently diminished primitive reflex patterns³.

The first decision point in evaluating floppy infants is to determine whether the site of abnormality is in the central nervous system, in the peripheral nervous system or in both places^{4,1}.

Maintenance of normal tone requires that the central and peripheral nervous systems are intact and that the skeletal muscle is healthy. Therefore, hypotonia is encountered in disease of the brain, spinal cord, nerves and muscles². One anterior horn cell and all the muscle fibres that are innervated make a motor unit. A primary disorder of the anterior horn cell body is a neuronopathy, of the axon or its myelin covering is neuropathy and of the muscle fibre is a myopathy. During infancy and childhood, diseases of the brain is far more common than the diseases of the motor unit^{5,6,4}.

SMA is one of the most common autosomal recessive disease affecting approximately 1 in 10,000 live births and with a carrier frequency of approximately 1 in 50^{7,8}. Timing of diagnosis is crucial for SMA because early diagnosis may lead to early supportive care and reduction in parents and caregiver stress⁹.

The term Central hypotonia is used to encompass all causes of postural hypotonia due to cerebral disease or

disorder. Central hypotonia is a non progressive disorder of the motor system during early development.

The congenital myopathies comprise a group of fairly benign muscle disorders that are apparent at birth or soon after. The affected child is floppy at birth. The presence of typical myopathic faces and paucity of facial expression are common in hypotonic infants¹. In congenital myopathy, unlike other muscle disorders, the weakness is non progressive and muscle strength usually improves¹⁰.

The floppy babies are in danger of developing different postural deformities such as scoliosis, extremity contractures and dislocation of hip^{11,1}. Rehabilitation plan including different stretching exercises of tight structures and strengthening exercises of weak muscles, chest physiotherapy, NDTs, sensory integration therapy and developmental therapeutic techniques are very helpful in improving their quality of life^{12,1,3}.

METHODOLOGY

The study was conducted at The Children Hospital & ICH Lahore in the Department of Physical Medicine & Rehabilitation (PM&R) over a period of one year from June 2017 to June 2018. Informed consent was taken from parents or attendants to take data for research purpose after taking informed consent and approval from ethical committee. Sixty Patients under two years of age with generalized hypotonia since birth were included in the study. Proforma was filled for required variables of interest like birth history (prenatal, perinatal and postnatal), age, sex and family history^{13,1}.

Received on 19-09-2018

Accepted on 29-12-2018

Physical and musculoskeletal examination including muscle tone, deep tendon reflexes and investigation including CPK, CSF and MRI were performed^{1,4,6}. Provisional diagnosis was made on the clinical grounds (history, physical examination and investigations). Floppy babies were categorized into SMA, Central hypotonia and congenital myopathy then EMG & NCS study was done on same group of patients and on these results final diagnosis was made. In EMG & NCS, median and ulnar nerves in the upper limbs and peroneal and tibial in the lower limbs were selected for motor nerve conduction studies. Sensory study was performed on median nerves and sural nerves. EMG of 1st dorsal interosseous, brachioradialis, tibialis and gastrocnemius muscles were performed. EMG is very accurate tool in diagnosis of SMA. In SMA electrodiagnosis revealed normal motor conduction velocities but compound muscle action potential amplitudes decreased markedly. Sensory amplitude and latencies were normal. EMG showed large motor unit potentials with presence of spontaneous potentials such as fibrillation and positive sharp waves. In congenital myopathy, myopathic findings include low amplitude compound muscle action potentials (CMAPs) and small polyphasic motor unit potentials¹⁴. Slow nerve conduction velocity (NCV) and conduction block favoured peripheral nerve involvement. In central hypotonia electrodiagnostic study was normal^{15,16}.

The results based on clinical findings were compared to the results made on EMG & NCS study and p value < 0.05 showing the significance of EMG and NCS studies.

RESULTS

In this study of one year, 60 floppy babies with mean age 6.15 month and standard deviations 5.42 months were included.

Of total 60 patients, 18 (30%) patients had a final diagnosis made on EMG & NCS findings was different from diagnosis made on clinical grounds and 42 (70%) patients had similar diagnosis. Chi. Square test of statistical significance was applied by SPSS version 25. There is significant difference as a whole in diagnosis made on clinical ground to that of EMG & NCS (P value < 0.05) showing EMG & NCS is valuable diagnostic tool for floppy babies.

Table: 1 Comparison of diagnosis made on clinical grounds to Electrophysiological diagnosis

Diagnosis made on Clinical grounds	Diagnosis made on EMG & NCS Study	Diagnosis Concordant	Diagnosis Discordant
SMA (n=31)	SMA (n=23) CH (n=05) CM (n=03)	23	08
Central Hypotonic (n=22)	CH (n=15) SMA (n=04) CM (n=03)	15	07
Congenital Myopathy (n=07)	CM (n=04) SMA (n=01) CM (n=02)	04	03
Total		42 (70%)	18 (30%)

P < 0.05 (significant)

CH: Central hypotonia

SMA: Spinal muscular atrophy

CM: Congenital Myopathy

Table 2, showed frequency of different types of floppy babies based on EMG & NCS findings. In final diagnosis 28 patients (46%) were diagnosed as spinal muscular atrophy, 22 patients (36.6%) were diagnosed as central hypotonia and 10 patients (16.6%) were diagnosed as congenital myopathy. In our study the most common type of floppy babies is SMA followed by central hypotonia and congenital myopathy. In our study male dominance was n=38 (63.3%) seen and most patients n=41 (68.3%) were presented before one year of age^{20,8,18}.

Table 2: Mean Ages and Type

Type	SMA	Central hypotonia	Congenital myopathy
Frequency	28 (46.6%)	22 (36.6%)	10 (16.6%)
Gender	Male	19 (67%)	05 (50%)
	Female	09 (32%)	05 (50%)
Age	≤ 01 year	22 (78.5%)	04 (40%)
	01-02 years	06 (21.4%)	06 (60%)

DISCUSSION

In our study 60 patients under two years of age having floppiness since birth were included. On clinical grounds, 31 patients were diagnosed as SMA while EMG & NCS on these patients revealed that 23 patients were SMA and remaining 08 patients had different diagnosis such as 05 patients were with Central hypotonia and 03 patients were of congenital myopathy. Similarly on clinical ground 22 patients were diagnosed Central hypotonic while EMG & NCS on these patients revealed that 15 patients among them were of central hypotonic and remaining 08 had different diagnosis such as 04 among them were with SMA and 03 were of congenital myopathy. Similarly on clinical grounds 07 patients were diagnosed as congenital myopathy while EMG & NCS on these patients revealed, 04 patients among them were of congenital myopathy and 01 patient was SMA and 02 were of central hypotonic. In general, out of total 60 patients, 18 (30%) had a final electrophysiological (EMG & NCS) diagnosis different from diagnosis made on clinical grounds. Chi. Square test of statistical significance was applied by SPSS version 25. There is significant difference as a whole in diagnosis made on clinical grounds to that of EMG & NCS P < 0.05 showing that EMG & NCS is best diagnostic tool for floppy babies This also relates to the previous study¹⁷.

In our study we observed that depending upon final electrophysiological (EMG & NCS) findings, 28 patients (46%) were diagnosed as SMA and 22 patients (36.6%) were diagnosed as central hypotonia and 10 patients (16.6%) were diagnosed as congenital myopathy¹⁸. In our study the most common type of floppy babies is SMA followed by CH and CM, these results relate to the previous study¹⁹.

In SMA, 22 patients were presented with age less than one year and 06 patients were within range of 1 to 2 years and in CH group, 15 patients were less than 1 year and 07 patients were within range of 1 to 2 years and in congenital myopathy group, 04 patients were less than 1 year and 06 patients were within range of 1 to 2 year. In general we observed major group 41 patients (68.3%) of patients in our study were presented with age less than one

year whereas 19 patients (31.6%) were within range of 1 to two years.

In SMA, 19 patients were male and 9 patients were female, in central hypotonia, 14 patients were male and 8 patients were female and in congenital myopathy, 5 patients were male and 5 patients were female. In general 38 patients (63.8%) were male and 22 patients (36.6%) were female, so male patients were dominant in our study^{20,8,18}.

CONCLUSION

We concluded that the most common type of floppy babies is SMA followed by central hypotonia and congenital myopathy.

We also concluded that EMG & NCS study is the best diagnostic tool for the diagnosis of floppy infants and children. It is recommended to create awareness among doctors about the indication and usefulness of EMG & NCS studies for proper diagnosis and management of patients.

REFERENCES

1. Mollamaduddin Ahmad, Mehran Iqbal, Nahin Hussain. A structured approach to the assessment of floppy neonate. *J Pediatr Neurosci.* 2016 Jan-Mar; 11 (1): 2-6. doi:10.4103/1817-1745.181250.
2. Dimuthu Saraji Wijesekara. Clinical approach to floppy infant. *Sri Lanka Journal of Child Health*, 2013; 41: 211-216.
3. Christopher RP, Gans BM. Rehabilitation of pediatric patients. In: Delisa JA, Gans BM. *Rehabilitation medicine: principles and practice.* 3rd ed. Philadelphia: Lippincott Raven Publishers; 1998: 946.
4. Fenichel GM. The hypotonic infant. In: Bradley WG, Daroff RB, Fenichel GM. *Neurology in clinical practice: Principles of diagnosis and management.* 2nd ed. Washington: Butterworth-Heinemann; 1995: 381-90.
5. Cohen W. Hypotonia and weakness. In: Kleigman RM, Nieder ML, Super DM. *Practical strategies in pediatric diagnosis and therapy.* 1st ed. Philadelphia: W.B. Saunders; 1996: 590-610.
6. Swaiman KF. Muscular tone and gait disturbances. In: Swaiman KF, Ashwal S. *Pediatric Neurology: principles and practice.* 3rd ed. Missouri: Mosby; 1999: 54-57.
7. Walter MC, Stauber AJ. Spinal muscular atrophy- clinical spectrum and therapy. *J Dev Behav Pediatr.* 2017 Sep; 38(7):556-557. doi: 10.1097/DBP.0000000000000497.
8. Madrid Rodriguez A, Martinez Martinez PL, Ramos Fernandez JM, Urda Cardona A, Martínez Antón J. Infantile spinal atrophy: our experience in the last 25 years. *A Fortschr Neurol Psychiatr.* 2018 Sep; 86(9):543-550. doi: 10.1055/a-0621-9139.
9. Lin CW, Kalb SJ, Yeh WS. Delay in Diagnosis of Spinal Muscular Atrophy: A Systematic Literature Review. *Pediatr Neurol.* 2015 Oct; 53(4):293-300. doi: 10.1016/j.pediatrneurol.
10. Wiechers DO. Motor potential in disease. In: Johnson EW. *Practical electromyography.* 2nd ed. Baltimore: Williams & Wilkins; 1988: 47-85.
11. Warren C. hypotonia and weakness. Practical strategies. In: Robert MK. *Pediatric diagnosis and therapy.* Philadelphia: WB Saunders, 1996; 590-610.
12. Steiner N, Torres A, Reddy A, Augustyn M. Congenital Hypotonia in Toddlerhood. *Curr Opin Pediatr.* 2013 Dec; 25(6):676-81. doi: 10.1097/MOP.0000000000000023.
13. Awater C, Zerres K, Rudnik-Schöneborn S. Pregnancy course and outcome in women with hereditary neuromuscular disorders: comparison of obstetric risks in 178 patients. *Eur J Obstet Gynecol Reprod Biol.* 2012 Jun. 162(2):153-9.
14. Sener U, Martinez-Thompson J, Laughlin RS, Dimberg EL, Rubin DI. Needle Electromyography and Histopathologic Correlation in Myopathies. *Muscle Nerve.* 2018 Nov 10. doi: 10.1002/mus.26381.
15. Richard DB. Electrodiagnostic evaluation of peripheral nervous system. In: delisa JA, Gans BM. *Rehabilitation medicine: principles and practice.* 3rd ed. Philadelphia: Lippincott-Raven Publishers; 1998: 321-371.
16. Brazis PW. Approach to the selection of electrodiagnostic, spinal fluid and other ancillary testing. In: biller J. *Practical neurology.* 1st ed. Philadelphia: Lippincott-Raven; 1997: 342-45.
17. Krocza S, Steczkowska M, Kaciński M. Usefulness of electromyography in diagnostics of the neuro-muscular diseases. *Przegl Lek.* 2009; 66(11):913-9.
18. Zahoor Ahmed, Saleem Ilyas, Muhammad Abdullah. Clinical spectrum of spinal muscular atrophy. *Ann Abbasi Shaheed Hosp Karachi Med Dent Coll.* 2005 Jun; 10(1):625-31.
19. Prremasiri MK, Lee Ys. The myopathology of floppy and hypotonic infants in Singapore. *Pathology.* 2003 Oct; 35(5):409-13.
20. Elif Kocasoy Orhan, Leyla Baysal Kiraç, Pinar Yalinay Dikmen, Zeliha Matur, Mustafa Ertaş, A. Emreöge, Feza Deymeer, Jale Yazici, and M. Barış Baslo. Electromyography in Pediatric Population. *Noro Psikiyatr Ars.* 2018 Mar 19; 55(1): 36–39. doi:10.5152/npa.17023