كلية الطب – جامعة الفيوم
قسم الأطفال

البحث الثاني

عنوان البحث :

Diagnostic approach of floppy infants: a study in Fayoum University Hospital, Egypt

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Background
Floppy infants represent a large proportion of patients attending the neurology clinic in Fayoum University Hospital. Neuromuscular disorders represent a common cause of floppiness. Electromyography (EMG) is an easy and cheap tool used for the diagnosis of these infants.

Patients and methods
The study classified cases with neuromuscular disorders using different and new diagnostic modalities. It included 42 floppy infants who were subjected to a full assessment of history, anthropometric measurements, laboratory investigations (serum creatine kinase and lactate), and EMG. Echocardiography, muscle and liver biopsy, and molecular study for bi-allelic deletion of exon 7 of the SMN1 gene were carried out when indicated.

Results
Floppy infants were classified as follows: spinal muscle atrophy (SMA): 23 (55%) cases, congenital myopathies: 11 (26%) cases, peripheral axonal neuropathy: two (5%) cases, muscle eye brain disease: one (2.25%) case, glycogen storage type 2 (Pompe disease): one (2.25%) case, arthrogryposis multiplex congenita: one (2.25%) case, and undiagnosed etiology: three (7.25%) cases.

Conclusion
SMA represents the most common neuromuscular cause of floppy infants in this study, followed by congenital myopathy. EMG is considered a cheap diagnostic tool and provides a good clue for the next step for diagnosis. Further, detection of other mutations involved in SMA is crucial for proper genetic counseling and early prenatal detection.