A Neonate with Respiratory Distress and Hypotonia- A Case on Congenital Myasthenia Gravis

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Abstract: Congenital myasthenia gravis, an autosomal recessive disorder, affecting neuromuscular transmission is a rare condition presenting as generalized hypotonia, weak cry and ptosis in neonatal period. We are reporting a case of congenital myasthenia gravis in a neonate who came to us on first day of life in respiratory distress and had generalized hypotonia and bilateral ptosis. After ruling out other possible causes of hypotonia, neostigmine test was performed with suspicion of congenital myasthenia gravis which proved out to be positive.

Keywords: Congenital myasthenia gravis, Neonate

INTRODUCTION:
Myasthenia Gravis is neuromuscular disorder affecting synaptic transmission at motor end plate. It is characterized by abnormal muscle fatigability and can be either genetic or acquired. Myasthenia Gravis occurring in neonates is divided into three forms i.e. transient myasthenia gravis, congenital myasthenia gravis and juvenile myasthenia gravis. Congenital myasthenia gravis is autosomal recessive disorder affecting neuromuscular transmission. The prevalence of CMG is estimated to be 1 in 500,000 in Europe and CMG is much more uncommon than autoimmune myasthenia [1]

CASE REPORT:
Full term male newborn, weighing 3.5 kg, delivered by spontaneous vaginal delivery at home by Dai was transferred to neonatal intensive unit 6 hours after birth with complain of difficulty in breathing. He cried immediately after birth. Breastfeeding was initiated 1 hour after birth, but the baby had poor sucking, and while mother continued to attempt oral feeding, he developed cyanosis and subsequently showed respiratory difficulty. Mother, 27 years of age, G5P5, was not a booked case and prenatal ultrasound scans were also not done. There was also no history of fever or any history of drug ingestion prenatally. There is also no history of weakness or ptosis on examination in mother. This baby is the 5th product of consanguineous marriage. All other 4 siblings died within 4-8 hours after birth and they also had same presentation.

General physical examination revealed a neonate in severe respiratory distress having sub costal recessions looking lethargic and having bilateral ptosis and weak cry. Weight, Length and head circumference are appropriate for gestational age.

Vital signs show tachypnea, with respiratory rate of 78 breaths/min and Oxygen saturation of 86% at room air. Head to toe examination show bilateral ptosis and no tongue fasciculation’s.

Chest auscultation revealed decreased breath sounds on right upper chest. Bilateral conducted sounds on auscultation. Heart and abdominal examination was normal. Spine and extremities normal and genital exam is also normal with bilaterally descended testes and normal scrotum, anus patent. Neurological examination show decrease tone in all limbs. Head lag present. Shoulder and ventral suspension show hypotonia. Moro’s absent and sucking, rooting and grasp poor. Deep tendon reflexes are diminished.

Bubble CPAP was given to baby due to respiratory distress and complete blood count and differential count, assessment of calcium, magnesium, blood sugar, C - reactive protein and blood culture was done which revealed normal result. Chest x-ray showed right upper lobe patch most probably due to aspiration. Creatinine Kinase, TSH, T4 also normal. Ultrasound Brain was done which showed normal parenchyma, no solid or cystic lesion no ventricular dilatation and no hemorrhage. Ant acetylcholine receptor antibodies sent which showed negative result.

Neostigmine diagnostic test was performed by giving injection neostigmine 0.04mg/kg intramuscularly 30 minutes after dose baby exhibit significant clinical improvement. Baby was then kept on injection neostigmine after which he improved clinically, bubble
cpap was weaned off and syrup pyridostigmine was then started and patient was then discharged with improved suckling ability. Parents were trained regarding chest physiotherapy and suctioning of baby through suction bulb as baby was hypotonic so there was risk of pooling of secretions. Parents were also trained about feeding of baby through Nasogastric tube.

Parents were asked to follow-up weekly so as to check for compliance of drug and general condition of patient and progression of disease.

Fig-1: Showing mouth held open and frog leg position in baby

Fig-2: Showing bilateral ptosis

Fig-3 Showing improved ptosis after injection neostigmine
DISCUSSION:

Myasthenia gravis (MG) is a disease that affects neuromuscular junction. The cardinal features are weakness and easy fatigability of striated muscles. The weakness increases with activity and improve with rest. Three clinical types are described in childhood: congenital MG, Transient neonatal MG and juvenile MG. Juvenile MG is clinically identical to autoimmune adult form of MG and present in late infancy. This form is associated with antibodies directed against nicotinic acetylcholine receptor (AChR) in 85% of patients. Transient neonatal MG occurs in neonates who are born to mothers with autoimmune MG. It develops in around 10-20% of neonates born to mothers with MG.

Congenital MG occurs in neonates of mothers who do not have myasthenia gravis and it is nearly a permanent disorder without spontaneous remission. Its onset is at birth or in early infancy with myotonia, ophthalmoplegia, ptosis, dysphasia, weak cry, easy muscle fatigue and sometimes respiratory insufficiency or failure. The diagnosis is based on history, clinical examination, negative acetylcholine receptor antibodies and positive neostigmine test. Repetitive nerve stimulation test to check for detrimental response can be performed but it may be absent in cases of CMS caused by mutations in Chat or rapsyn [2]. Vincent et al. stated that absence of acetylcholine receptor antibodies is a prerequisite for diagnosing congenital myasthenia gravis. [3] The serum Creatine Kinase (CK) level is normal. Role of muscle biopsy is limited.[4] The diagnostic bedside test is neostigmine test, administered IM at a dose of 0.04mg/kg. The peak effect is seen in 20-40 min. Yin J et al. concluded that neostigmine test has the highest sensitivity in diagnosing myasthenia gravis. [5] Edrophonium test (Tensilon) is not recommended for use in neonates or infants because its effect is too brief for objective measurement and an increased incidence of acute cardiac arrhythmias is reported especially in neonates.[6] Demonstration of the antibodies and documentation of the response to neostigmine is sufficient to make the diagnosis. [7] Once the diagnosis of MG is established, the primary therapeutic agents for long-term treatment are cholinesterase-inhibiting drugs. [8]

We are reporting first case of congenital myasthenia gravis diagnosed in a neonate in Pakistan. There was a case reported earlier of congenital myasthenia gravis but it was diagnosed in a two and a half year old child. [9]

CONCLUSION:

The diagnosis of congenital myasthenia gravis is often difficult to ascertain as there is frequent absence of a family history of the disease, and there is pre-eminence of the myopathy signs compared with myasthenia signs. The early onset of the first symptoms, demonstration of a neuromuscular block and the cholinesterase inhibitor test all enable the rectification of the diagnosis and the proposal of an effective treatment and genetic counseling.

Abbreviations:
MG: Myasthenia Gravis
CMG: Congenital Myasthenia Gravis
CPAP: Continues positive airway pressure

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