EXAGGERATED STARTLE A CASE OF NEONATAL HYPEREKPLEXIA

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Abstract :
Hyperekplexia in neonates is a rare condition. A 2 day old boy baby presented with rigidity in all muscle groups and exaggerated, non-habituating startle response along with increased hyper tonicity with each startle response. Sepsis screen, serum electrolytes, Amplitude integrated electroencephalogram (aEEG) and MRI brain were normal. He was started on clonazepam that helped in controlling the symptoms but did not eliminate them completely.

Keyword : hyperekplexia, neonates, non-habituating startle response

Introduction:
Neonatal hyperekplexia is a rare condition, often inherited, but sometimes presenting sporadically with three cardinal features: generalized stiffness immediately after birth which increases on handling and decreases during sleep, excessive startle response following unexpected stimulus with no change in level of consciousness and finally generalized stiffness soon after the startle response. (1) The other important symptoms of this condition include tonic neonatal cyanotic attacks and non-habituation of exaggerated startle response following nose tapping test. Spasms could be severe enough to cause apnea, bradycardia and death.

Case report:
A 2 day old boy baby was born of a consanguineous marriage, delivered at term to a G2P1L1 mother by vaginal delivery with no antenatal pregnancy related risk factors. There was no history of central nervous system disorder in the family. APGAR scores were 8 and 8 at 1 and 5 minutes respectively, with only response to stimulus being decreased. Baby was referred to our hospital with intermittent ‘stridor’ and probable ‘tonic seizures’. Examination revealed a comfortably lying baby saturating well in room air, with vital signs being stable. Central nervous system examination revealed rigidity in all muscle groups and exaggerated, non-habituating startle response along with increased hyper tonicity with each startle response. Deep tendon reflexes were normal. Sepsis screen, CSF examination, serum electrolytes and arterial blood gas were normal. Amplitude integrated electroencephalogram
(aEEG) did not reveal any features of seizures with background being continuous with normal sleep wake cycle. MRI brain done was also normal. In view of the presence of all the cardinal features a diagnosis of hyperekplexia was considered. The baby was started on a therapeutic trial of clonazepam at 0.01mg/kg which was graded up to 0.02mg.kg. Baby tolerated the dose well and the symptoms had decreased as compared to baseline by the time of discharge. He continues to have exaggerated startle on follow up.

Discussion:
Hyperekplexia also known as Rutter's syndrome is a neurologic disorder manifested by the presence of three cardinal features: generalized stiffness at birth, excessive startling and temporary generalized stiffness after being startled. (1) Hyperekplexia has been noted without family history i.e., sporadic hyperekplexia. (2) Along with the cardinal symptoms, there are other features also noted like tonic neonatal cyanotic attacks which can be stopped with the 'vigevano' manoeuvre consisting of forced flexion of head and legs towards trunk. (3) Other features described are exaggerated head retraction reflex which consists of brisk, involuntary backward jerk of head following a light tap on the root of nose or middle portion of upper lip - considered by some authors to be a pathognomonic sign. (4) Other frequently mentioned features are periodic limb movements during sleep and hypnagogic myoclonus. (5) Hyperekplexia is an inheritable disorder with mutations in different parts of inhibitory glycine receptor. The alpha 1 subunit of the glycine receptor is the main gene for hyperekplexia and is affected in 80 % cases. (6) The diagnosis is essentially clinical and EMG, if done, may show shortened latencies with continuous activity at rest. (7) Muscle biopsy is usually normal. The condition needs to be differentiated from various other causes of hypertonia in newborn period like neonatal tetanus, perinatal asphyxia and Schwartz-jample syndrome. Clonazepam is the treatment of choice showing most consistent effects. (8) An increased risk of death from apnea associated with severe spasms has been reported. The generalized stiffness present at birth normalizes in the first few years of life. The excessive startle reflex that is present at birth persists throughout life. The short lasting temporary generalized stiffness often causes patients to fall forward ‘as stiff as a stick’ while fully conscious. (9) Delayed motor milestones could be present while cognitive function remains unaffected. (10) Baby described in our case is now 16 months old and his motor milestones are delayed (he is able to walk with support at 16 months). His cognitive functions are good. On oral clonazepam he continued to have exaggerated startle reflex.

References:
4 Shahar E, Brand N, Uziel Y, Barak Y. Nose tapping test inducing a generalized flexor spasm: a hallmark of hyperekplexia.


