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Case report

A rare case of familial hyperekplexia

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ABSTRACT

Familial hyperekplexiaia a rare autosomal dominant or sporadic disorder characterized by abnormal startle reaction elicited by auditory or somatosensory stimuli. Here we report a 3 months old female child presented with complaints of exaggerated startle response followed by shrill cry to tactile stimuli over the face noticed by mother since birth. There was also a positive family history in her father and father's brother. Neurological examination was normal except for hyperreflexia. Child was evaluated further and investigations like EEG and imaging were done. IEM workup was also done. Investigations were all normal. Hence from the classic diagnostic sign and positive family history, diagnosed as a case of familial hyperekplexia and the child improved after being started on clonazepam.

Introduction

Hyperekplexia is a neurological disorder characterized by pronounced startle response to tactile or acoustic stimuli and rarely with hypertonia. Familial hyperekplexia is a rare autosomal dominant disorder running in families. Usually the onset of symptoms is from neonatal period. The traid of hyperekplexia is generalized stiffness, nocturnal myoclonus and pathologic startle response. Here we report a 3 months old female child with exaggerated startle response and a classical family history suggesting the diagnosis of familial hyperekplexia.

Case report

3 months old female child brought to our hospital with complaints of exaggerated response to tactile stimuli in any form over the face. Mother noticed the symptoms since birth. This baby is first born of non consanguineous marriage. Antenatally there was no history of drug intake, fever, radiation exposure. Quickening felt at fourth month. There had been no history of any decreased or increased fetal movements. Baby was delivered via labour naturalis, cried immediately after delivery. No history suggestive of neonatal seizures or apnoeic episodes. For the complaints of increased startle response, baby was started on some medications which the mother discontinued after one month. Child attained developmental milestones normally till age. On eliciting further history, baby's father and father's brother had similar complaints of exaggerated startle response since birth. In addition they also had momentary stiffening of all four limbs followed by sudden fall attacks. Both were on multiple drugs and the frequency of the attacks decreased.On examination baby recognizes mother, interested in surroundings, no signs suggestive of any cranial nerve palsies. Motor system examination showed hyperreflexia[3]. Spine and cranium examination was normal. The specific diagnotsic sign is, tapping the nose would elicit exaggerared startle response with head retraction. We could even abort the episode with Vigevano manevour. Further evaluation was then done. Serum electrolytes, blood sugar, magnesium, calcium levels were normal. Imaging studies were normal. Electroencephalography was also normal. Other investigations to rule out inborn errors of metabolism were also done (lactate, pyruvate, ammonia, pH, organic acids, amino acid levels, urine for reducing substances, TMS), which turned negative. Hence finally, after ruling out all other causes, and with the clinching diagnostic sign and a positive family history, diagnosis was arrived as familial hyperekplexia syndrome. Child was then started on oral clonazepam and is on regular follow up at our outpatient department. According to the mother, symptoms have improved and number of attacks has decreased.

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Discussion

Familial hyperekplexia is a rare autosomal dominant disorder characterized by abnormal startle reaction[1] elicited by either auditory or somatosensory stimuli, with transitory stiffness during the neonatal period[6] followed later by falling attacks accompanied by momentary generalised muscular stiffness. The minor form is characterized by an inconstant excessive startle response. Neonatal onset may even be fatal as it may precipitate apnea[2] and convulsive hypoxic seizures. These children may have dysphagia, choking, hip dislocation, umbilical, inguinal hernia and delayed motor development. Stiffness may improve by one year of age and may disappear during sleep.The genetic defect is in the alpha beta subunit of strychnine sensitive glycine receptors [4,5]. The triad of hypereklexia though not observed in our case, could be still classified as milder form. The specific diagnostic sign of tapping over the nose causing non-fatiguable startle reflex with head retraction. Other conditions which may even precipitate the event include bathing, sudden awakening, auditory or tactile stimuli. Flexion of neck and hip repeatedly (Vigevano maneuver) may abort the episode. The differential diagnosis includes congenital stiff person syndrome, startle epilepsyyoclonic seizures, neonatal tetany, phenothiazine toxicity and Schwartz-jampel syndrome. The treatment of choice for the same is oral clonazepam.

Conclusion

Hyperekplexia is a seizure mimic which should be diagnosed clinically as early as possible. Treatment should be started immediately to avoid life threatening events. These children should be on regular follow up with frequent modification of drug dosage.

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