Benign familial neonatal convulsions: A case report

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Introduction

Benign familial neonatal convulsions (BFNC), also known as benign familial neonatal epilepsy, is a rare dominantly inherited epileptic condition[1]. An incidence of 14.4 per 100,000 live births has been reported from a study in Canada[2]. We report on a family with this epilepsy syndrome.

Case report

A family of three children born to non-consanguineous parents are discussed in this case report (Figure 1).

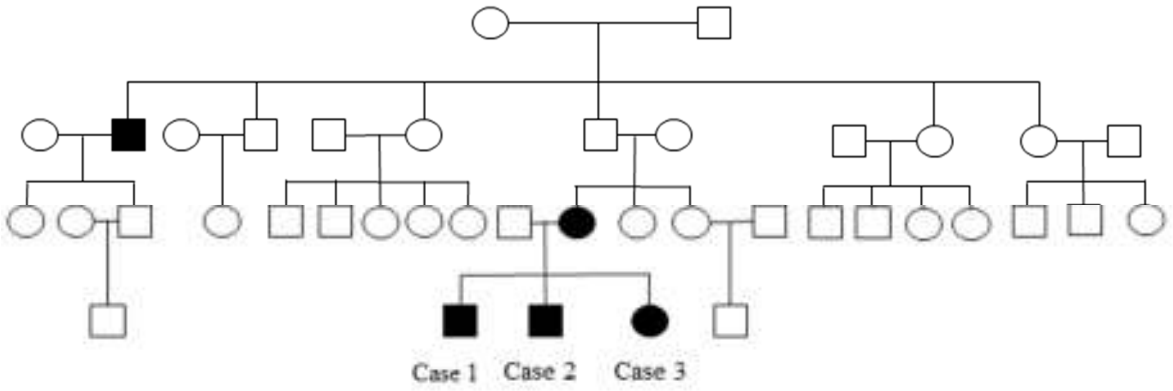

Figure 1: ***Pedigree chart of the family with benign familial neonatal convulsions***

The eldest (Case 1) who is now six years, presented at the age of three months with repeated episodes of focal seizures since day three of life. There was no fever and his neurological examination was normal. Magnetic resonance imaging (MRI) revealed no aetiological cause for the seizure. His inter-ictal electroencephalogram (EEG) was normal. The seizures were easily controlled with initial phenobarbitone followed by sodium valproate therapy. His last seizure was experienced at three months of age. He has been off anticonvulsants since one year of age and remains well, now schooling in grade two.

His brother (Case 2) who is now three years old, also presented on the third day of life with generalised convulsions without any fever or metabolic disturbance following an uncomplicated pregnancy and early neonatal period. Seizures settled with oral phenobarbitone. He had no further seizures till five months of age when he presented again with repeated episodes of focal and generalised convulsions. These were frequent and required intravenous medication as well as addition of levetiracetam to his regular dose of sodium valproate. A subtle deterioration of milestones was noted during recurrence of seizures. After about 5 days, his seizures settled and since then he has remained seizure free. Detailed neuro-imaging and metabolic screening, revealed no abnormalities. Developmental regression normalised gradually and when 12 months seizure free, anti-epileptics were tailed off. Currently he is off medications for over 18 months with normal development.

The third member to be affected was the newborn sister (Case 3) who also presented on the third day of life with multiple episodes of generalised increased tone which was preceded by an abnormal irritable cry. She had an uneventful antenatal period and delivery by elective caesarean section at term with a birth weight of 3300g. Immediate

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postpartum period was uneventful. Examination was completely unremarkable. All her investigations, including imaging, were normal. Genetic testing for epilepsy panel of genes was not performed due to the cost. She was also initially treated with intravenous phenobarbitone but taken off soon due to the benign nature of her epilepsy. She has been followed up for one year now and only once developed a convulsion with fever. She showed normal developmental milestones.

The mother of these three children and her uncle have had a similar type of very early presentation with recurrent multiple seizures during the first week of life. They were multiple but brief and none had any developmental concerns. The mother is now thirty seven years and has not had any further seizures and her uncle who is a retired clerk remains well at the age of sixty.

Discussion

Mutation involving the potassium channel gene KCNQ2, a submicroscopic deletion of chromosome 20q13.3, has been identified as a cause of BFNC in the majority[1]. KCNQ3 located on chromosome 8q24, which is less frequently identified, reveals the heterogenic nature of this condition[3]. BFNC has a characteristic pattern of presentation, occurring mostly among healthy term neonates, with equal sex distribution, an uneventful antenatal history and an Apgar score more than 7 at one minute[4]. There is always an interval between birth and the onset of seizures. Majority of patients had the onset on 2nd or 3rd day of life[5]. Seizures are known to remit around four to 12 months of life. Clinical manifestations could take a wide spectrum of seizure types (tonic or apnoeic episodes, focal clonic activity, or autonomic changes). They are usually brief, lasting between one to two minutes.

A family history of neonatal seizures and exclusion of secondary causes are essential for the diagnosis of BFNC. Miles *et al.* proposed five criteria for the diagnosis of BFNC which are: neonatal or early infancy onset of seizure, a normal neurological examination, a normal neurodevelopmental outcome, negative evaluation for any other seizure aetiology and a positive family history for neonatal or infantile seizures[6]. Our patient fulfilled all these criteria for the diagnosis of BFNC. EEG performed during the ictal phase has shown changes commencing with a symmetrical flattening, followed by asymmetrical spikes and sharp waves for 1-2 minutes. The electro-clinical findings in this study were suggestive of a form of generalised tonic clonic seizure[7].

Treatment of BFNC is controversial as it is known to resolve spontaneously[6]. If anti-epileptics had been started, it is advisable to tail it off by 3 to 6 months' time. Long term prognosis is good. BFNC is a benign condition; though it is a diagnosis of exclusion, a characteristic family history helps prevent unnecessary investigations and interventions.

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