Cyanotic breath holding spell in a neonate. *A rare entity*

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Breath holding spells (BHS) are a common disease entity in children and mostly occur between 6-18 months of age and terminate by 3-4 years.[1] Onset below 6 months of age is seen in 13-18% of patients.[1,2] However, onset in the neonatal period is very rare and only a single case has been reported who had BHS from day 3 of life in the background of a strong family history.[3] We present a neonate who had cyanotic breath holding spells from day 2 of life without any family history. A 15-day-old male child born to a primiparous mother at 38 weeks of completed gestation presented to the out patient department (OPD) of Sheri-Kashmir Institute of Medical Sciences, Srinagar, India with a history of BHS from day 2 of life. Antenatal history was uneventful. Mother's hemoglobin was 13.5 grams/dl. Duration of labor was 12 hours and duration of rupture of membranes was 8 hours. The baby cried immediately after birth, had Apgar score of 8/10 and was breast-fed from 2 hours of life. The baby and mother were discharged home after 24 hours of stay. The neonate was all right until 42 hours of life when he started crying followed by bluish discoloration of whole body and loss of consciousness for a brief moment. There was no history of incontinence, tonic posturing, or deviation of eyes during the episode. The baby immediately turned pink and started sucking at the breast. A local pediatrician was consulted who reassured the parents. He remained all right for 4 days when he again developed a similar type of cyanotic spell. There were 4 such spells until day 15 of life when he presented to our OPD. During the spell, which was observed in the hospital, the baby started crying followed by noiseless expiration and bluish discoloration of the whole body and losing consciousness for a brief moment. There was no family history of seizures or BHS. On examination, the neonate was healthy, pink, and alert with admission weight of 3300 grams, length of 49.5 cms and head circumference of 33.5 cms. The rest of the physical and systemic examination was normal. Neurologically, tone was normal with intact neonatal reflexes. Investigations revealed hemoglobin of 13.2 grams/dl (normal 13-20 grams/dl), hematocrit of 44.8% (normal 42-66%), total leukocyte count of 8006 cells/mm^3 (normal 5000-21000/mm^3). There were no band cells or toxic granules. Septic screen was negative, and lumber puncture revealed no meningitis. A chest x-ray, electrocardiogram, and echocardiogram were normal. Computerized tomography of the head was normal and electroencephalogram (EEG) revealed no epileptiform focus. The patient was started on oral iron at the dose of 4 mg/kg body weight from day 21 of life for 12 weeks. The patient was evaluated weekly during the first month and then every 2 weeks until 12 weeks. The frequency of BHS diminished, and he had only 2 spells in the next one month, and from 3 months of age there were no spells.

Breath holding spells are generally benign, but may cause intense parental concern. In 1967 Lombroso and Lerman[1] in a study of 5000 patients identified 225 patients with severe breath holding spells amounting to a prevalence of approximately 4.6%. Breath holding spells commonly occur between the ages of 6 months and 2 years. The age of onset of BHS in most of the studies has been in the first 12 months of life.[2] In a previous study, 76% of patients had onset within the first 12 months of life, with 13% of patients having onset before 6 months of age.[3] However, the onset of BHS in the neonatal period is quite rare, and only one case has reported the age of onset on day 3 of life.[4] Breath holding spells in this neonate occurred in the background of a strong family history. However, our patient started having BHS from day 2 of life (42 hours of life). The neonate was fully investigated for other conditions that could have mimicked a BHS. All the investigations were normal except for the hemoglobin that was in the lower normal zone. The patient was put on oral iron at the dose of 4 mg/kg/day. There was a gradual improvement with the mother reporting only 2 episodes in the next one month, and subsequently there were no episodes for the next 4 months. During follow up, the infant had normal development with social smile noticed at 2 months and head holding at 4 months of age.

The pathophysiological mechanisms of BHS remain controversial. The presence of an underlying dysfunctional autonomic nervous system with cerebral anoxia or cerebral ischemia resulting from vagally mediated cardiac arrest may be responsible for the condition.[5,6] What provoked our patient to have BHS at such an early age remains unknown. Although BHS can occur in neonates with a family history, the same was absent in our patient. The hemoglobin and hematocrit in the patient on day 15 of life were in the lower range of normal. Subclinical anemia could have precipitated the condition in this neonate with a dysfunctional autonomic nervous system. This is emphasized by the response to the iron therapy shown by the infant. In most patients with BHS, spontaneous recovery occurs by 36-42 months of age.[2,3] The early termination of BHS in this neonate is unlikely to have occurred spontaneously, however, the possibility cannot be ruled out.

In conclusion, BHS can present as early as day 2 of life and needs to be considered in the differential

diagnosis of any neonate presenting with cyanotic spells. Iron therapy may be considered in the termination of BHS in neonates.

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