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CASE REPORT

Delayed Presentation of Hematomyelia as Spastic Quadriparesis: A Rare Presentation in Known Case of Haemophilia

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ABSTRACT

Haemophilia is an inherited bleeding disorder caused by deficiency of coagulation factor VIII (hemophilia A), factor IX (hemophilia B), or factor XI (hemophilia C). Age of presentation and severity of bleeding depends on the factor levels. Majority of central nervous bleeding are intracranial. Intraspinal bleeding is rare with the incidence rate of less than 10% and can have varied presentations. Here we present a case of delayed presentation of hematomyleia as spastic quadriparesis in a known case of hemophilia A.

KEYWORDS

Haemophilia; Coagulation factor VIII; Intraspinal bleeding; Hemophilia A; Hemophilia B; Hemophilia C

INTRODUCTION

Haemophilia is an inherited bleeding disorder caused by deficiency of coagulation factor VIII (hemophilia A), factor IX (hemophilia B), or factor XI (hemophilia C). Based on the residual factor activity level haemophilia is characterised as mild (5% - 40%), moderate (1% - 5%) or severe (<1%). Factor levels correlate with the severity of bleeding and its complications [1].

Patients with severe haemophilia present early on in their life [2]. Newborns present with central nervous system bleeding, bleedings extra cranial sites such as cephalohematoma, and sites of medical interventions including circumcision, heel sticks, and venipunctures [3]. In older children the common sites of bleeding include joints, muscles, central nervous system, and oral or gastrointestinal tract [4].

Majority of central nervous bleeding are intracranial with <10% cases being intraspinal [5,6]. Intraspinal bleeding can have varied presentations with history of trauma not being evident in many cases [7-9]. Epidural hematomas have been reported in literature, but intramedullary bleeding is rare with only few case reports [10-12].

CASE REPORT

A 37-years-old male, domestic help by profession presented with complaints of neck pain and weakness in bilateral lower limbs followed by upper limbs for 1 day. It was associated with bowel and bladder involvement. There was no history of sensory deficit, trauma, loss of consciousness. He had history of non-resolving bleed following trauma 15 years ago for which he required treatment at a hospital. He was told about some bleeding disorder (details were not available).

On examination, the patient was conscious, oriented, vitals were stable. Flexion deformities was present in both knees, and left ankle was plantar flexed. On systemic examination, respiratory, CVS and abdomen were within normal limits. In CNS examination higher mental function, cranial nerves, sensory and cerebellum were normal. Bulk of muscle of both upper limb and lower limb was normal. Tone was increased. Power in bilateral shoulder, elbow and wrist in all range of motion was 3/5 and in bilateral hip, knee, and ankle in all range of motion was 1/5. Deep tendon reflexes were exaggerated. Plantar reflex was equivocal in both the limbs. Fundus examination was normal in both the eyes.

Initial investigations CBC, KFT, PT-INR and apTT was normal. X ray of the chest and cervical spine was normal. NCCT head was suggestive of ill-defined hypodensity in left parietal region s/o glioencephalomalacic changes.

MRI brain with spine was suggestive of altered signal intensity in the spinal cord appearing hyperintense on T2/STIR images at the cervicomedullary junction and at C1-C2 vertebral level and chronic SDH in left parietal region (Figure 1). The altered signal intensity could signify ischaemia/demyelination/ oedema due to resolving bleed.

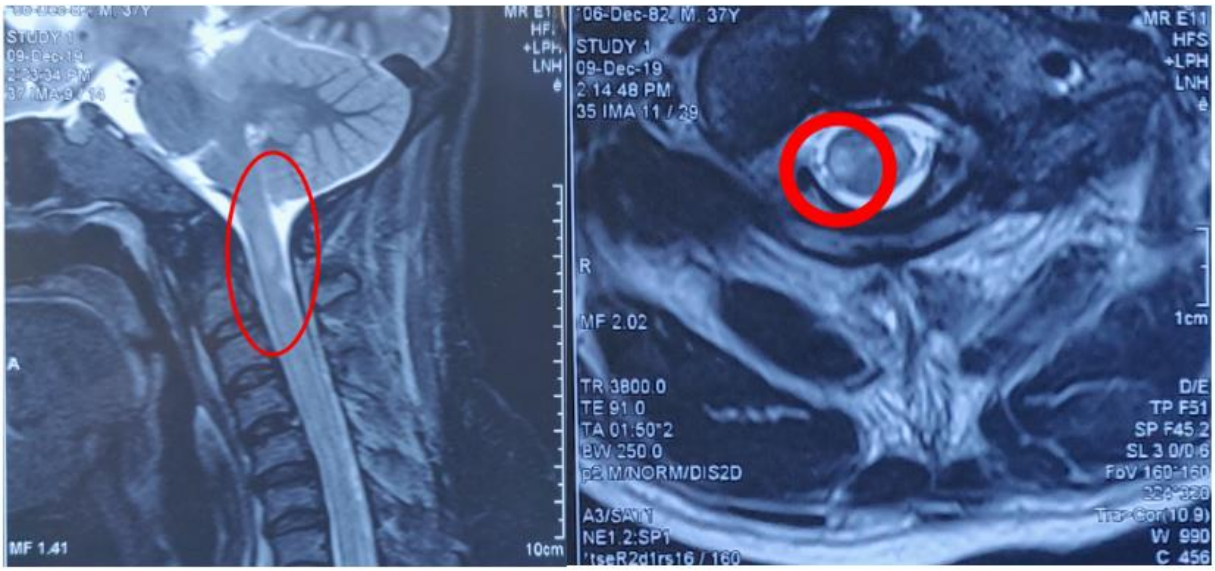

Figure 1: MRI brain and spine cord: Altered signal intensity in the spinal cord appearing hyperintense onT2/STIR images at the cervicomedullary junction and at C1- C2 vertebral level.

The previous records brought by his attendant subsequently, showed that our patient was suffering from Haemophilia A and his factor levels <1%. His X ray knee & ankle was suggestive of haemophilic arthropathy (Figure 2).

Based on the history possibility of resolving intraspinal bleed with oedema was kept as main differential diagnosis. The other possibilities of altered signal intensities like infarction/demyelinating disorders were considered unlikely in view of the history. The main causes of spinal cord infarction are aortic procedures, cardiogenic embolus, vasculitis, systemic hypoperfusion, hypercoagulable state [13]. The workup for hypercoagulable state could not be done in this patient. The common causes of demyelination in spinal cord include multiple sclerosis, NMO spectrum disorder which were ruled out on the basis of history and examination.

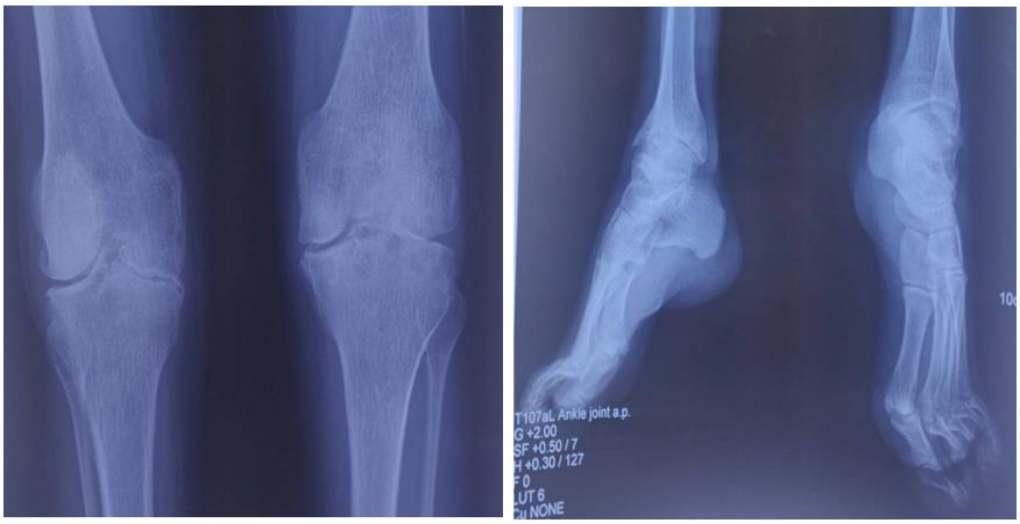

Figure 2: X-ray bilateral knee and ankle showing haemophilic arthropathy.

TREATMENT

The patient was treated with inj. Methylprednisolone for 5 days and 100% factor VIII replacement. The patient showed dramatic improvement by the end of pulse therapy. Power improved to 4/5 in lower limbs & 5/5 in upper limbs.

The patients were further worked upon. 2D echo was normal. HIV, HbsAg, Anti HCV was negative. ANA and serum homocysteine levels were normal.

On follow up, the patient improved significantly requiring only minimal support for mobility. He was subsequently registered in our hospital's Haemophilia Day care centre for further management.

DISCUSSION

The peculiarity of this case was the following:

- Spontaneous haemotomyelia is a rare complication with very few cases reports available.
- Delayed presentation in case of spinal cord bleed.
- Possibility of cord infarction in a case of haemophilia.

Haemophilia is a hypocoagulable state due to deficiency of coagulation factor deficiency. The reported incidence of CNS bleeding in hemophiliacs has ranged from 2.2% to 7.8% [14]. The CNS bleeding occurs at all ages but is seen pre- dominantly in young hemophiliacs.

A study conducted by M. Elaine Eyster et al. in patients of hemophilia retrospectively analysed cases of CNS bleeding. They showed that 53% of study population had a recent history of trauma, 5% of study population had associated hypertension and no etiology was apparent in 38% of study population as in our case [15]. The most frequent presenting symptom of intracranial bleeding was headache, often with vomiting and seizures. Intraspinal hematomas usually presented with backache and/or paralysis.

The delayed presentation of symptoms in our case during resolving stage of bleed was unusual. The possible explanation would be bleed most likely caused cord oedema which caused the symptoms; which was subsequently resolved with steroids. The patient also had evidence of chronic SDH which was not known to the patient. Also, the patient's clinical features with x ray was suggestive of haemophilic arthropathy. These are indicative of the fact the patient used to have subclinical bleed which did not cause any symptoms.

However, the same MRI finding can also be seen in spinal cord infarct presumably due to a hypercoagulable state. Two possible mechanisms been explored for hypercoagulable states in hemophilia.

The hemophilia B Leyden phenotype is characterized by severe hemophilia in childhood that becomes mild after puberty. Mutations in these individuals occur in the factor IX promotor, affecting expression of the gene, rather than the factor IX coding region. The increase in factor IX expression after puberty is likely due to hormonal changes and testosterone sensitivity [16-18].

Other mechanism is the co-inheritance of thrombophilia in a patient. Factor V Leiden mutation, factor II mutation and MTHFR mutations have been shown to be co-occur in haemophilia patient [19,20]. These patients are also prone to thrombosis. Heterozygosity for Prothrombin G20210A has been shown to cause upper limb deep vein thrombosis in a case of haemophilia A [21]. Factor VIII inhibitor bypassing activity (FEIBA) or recombinant activated factor VII (rFVIIa) have also been shown to be important risk factors for thrombosis in cases of Haemophilia [22].

In our case we could not get the work up for thrombophilia done. However, the possibility of thrombosis in anterior/posterior spinal cord artery without thrombosis without involvement of any other arteries/veins was minimal. We could not find any case report of spinal cord infarction in a case of haemophilia.

CONCLUSION

Haemophilia is a hypocoagulable state due to deficiency of coagulation factor deficiency. Spontaneous haemotomyelia is a rare complication of haemophilia and delayed presentation of quadriparesis in resolving stage of haemotomyelia is very unusual.

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