Case Report

Superficial siderosis presenting as hemiparesis in a paediatric patient with congenital Factor V deficiency – A case report

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Abstract

Superficial siderosis is a rare abnormality caused by deposition of hemosiderin in the subpial or subarachnoid spaces of the brain, cranial nerves, and spinal cord. It results from chronic repetitive bleeding in the subpial/subarachnoid space. Magnetic resonance imaging of the brain is the diagnostic modality of choice. We describe a rare case of an 18-month-old male, a known case of congenital Factor V deficiency, who presented with recurrent vomiting, irritability and right-sided hemiparesis. On Magnetic resonance imaging, there was hypointense coating of the surface of the brainstem and cerebellum with ventricular dilatation. This case emphasizes the fact that superficial siderosis should be specifically looked for when evaluating a patient with a history of previous intracranial bleed followed by a thorough search for the source of the hemorrhage.

Keywords: Superficial siderosis, Paediatric, Magnetic Resonance Imaging, Factor V, Hemosiderin, Intracranial bleed, Subarachnoid bleed.

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Introduction

Superficial siderosis is a rare abnormality caused by deposition of hemosiderin in the subpial or subarachnoid layers of the brain, cranial nerves, and spinal cord. It results from chronic repetitive bleeding in the subpial or subarachnoid spaces which is sometimes difficult to differentiate on imaging. The resulting pathological changes include reactive gliosis, neuronal loss and demyelination which result in a progressive debilitating clinical disorder characterised by a clinical triad of sensorineural hearing loss, cerebellar ataxia and pyramidal signs [1]. Headache and seizures have been reported in the paediatric population.

We present a rare case of superficial siderosis in a child with known congenital Factor V deficiency with a previous history of subarachnoid hemorrhage. The description of the findings associated with the case is followed by a brief account of the pathological changes, imaging findings and management of such patients.

Case summary

An 18-month-old male child, a known case of congenital Factor V deficiency developed recurrent episodes of vomiting and irritability followed by right-sided hemiparesis. On a neurological examination, motor power was grade 3 in all groups of muscles on the right side. The patient was also noted to have a tense anterior fontanelle with increased occipito-frontal circumference of 54 cm which was more than the 97th percentile for the age of the patient. The child was born after a full-term (gestational age of 38 weeks) and normal delivery, without a history of birth trauma or any other perinatal complication. Parents gave the history of the child having a subarachnoid hemorrhage at 2½ months of age which was diagnosed by a Magnetic Resonance Imaging (MRI) of brain and was managed conservatively. The images of this MRI were not available for review.

The patient underwent MRI of the brain at our hospital on a Siemens Avanto 1.5T machine. Axial T2 weighted turbo spin echo (TSE), gradient echo sequence (GRE); Coronal T2 weighted TSE; sagittal T2 weighted TSE, T1 MPRAGE (Magnetization Prepared Rapid Acquisition Gradient Echo) and 3D-fast TSE Sampling Perfection with Application optimized Contrasts using different flip angle Evolution (SPACE) sequences with a slice thickness of 5 mm and gap of 0.5 mm were performed. MRI revealed linear uniform hypointense coating of the surface of the brainstem, base and foliae of cerebellum bilaterally on T2 weighted and GRE images (Red arrows; Figure 1a and 1b). In addition, there was associated dilatation of both lateral ventricles with intraventricular blood CSF levels noted bilaterally (Red arrows; Figure 2a and 2b). There was also dilatation of the third ventricle noted (green arrow Figure 2b). These findings are consistent with superficial siderosis, occurring in this case as a consequence of previous subarachnoid hemorrhage, with associated hydrocephalus. The raised intracranial pressure due to hydrocephalus causing the symptoms, was relieved by ventriculoperitoneal shunting. The patient was managed with fresh frozen plasma infusions to replenish the factor V. The patient was advised to undergo a regular follow-up both with the neurosurgeon and the hematologist.

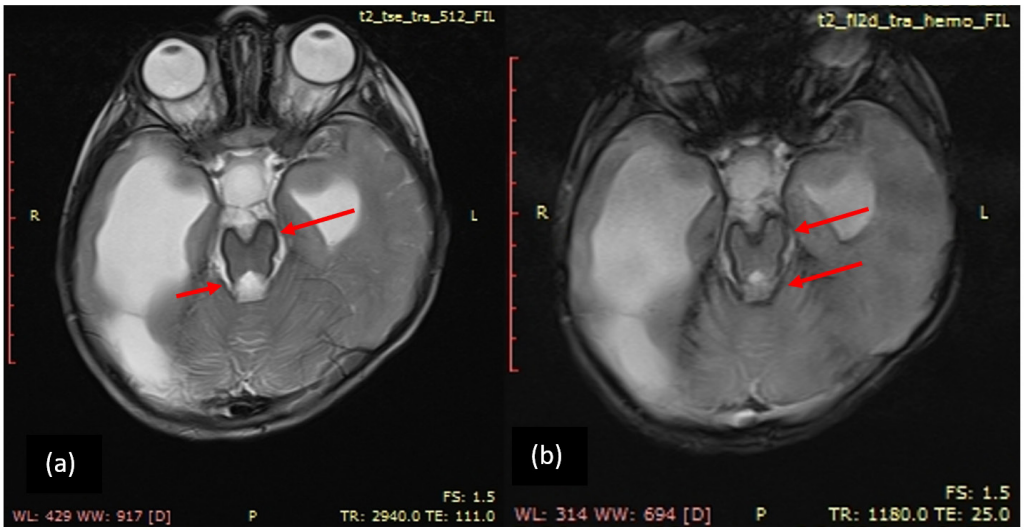

Figure 1. *(a) Axial T2 weighted image and (b) axial GRE image at the level of brainstem and cerebellum shows Hypointensity involving brainstem, base and foliae of cerebellum (red arrows).*

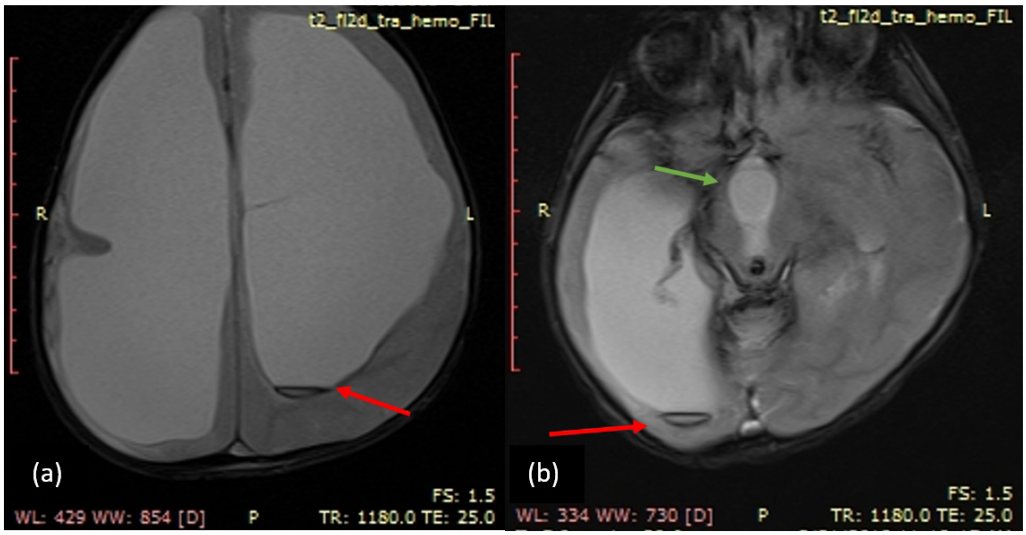

Figure 2. *(a)-(b) Gross dilatation of both lateral ventricles with blood fluid levels in the occipital horns of both the lateral ventricles (red arrows). Dilatation of third ventricle (green arrow).*

Discussion

Superficial siderosis is a rare clinical condition which results from chronic or repeated bleeding in the subpial or subarachnoid spaces. This leads to hemosiderin deposition in the subpial layer of the brain and spinal cord causing gradual and progressive neurologic deterioration. The common conditions that are associated with the development of superficial siderosis include trauma and neurosurgery. Less common conditions are hemorrhagic neoplasms, vascular malformations, hemorrhagic vasculopathies and cerebral amyloid angiopathy [2]. In up to one third of the cases, etiology remains unknown even after extensive relevant diagnostic imaging.

There are two clinical and pathological types - 'Classical' superficial siderosis which primarily affects the infratentorial regions and the spinal cord and 'cortical' superficial siderosis, which primarily affects the supratentorial compartment and cerebral convexities. The classical subtype has predilection for superior cerebellar vermis, cerebellar folia, brainstem, spinal cord, nerve roots, cranial nerves I and VIII. It is characteristically associated with the clinical triad of slowly progressive sensorineural hearing impairment, cerebellar ataxia and pyramidal signs. Cortical superficial siderosis (cSS), on the other hand, predominantly affects the cortical sulci over the cerebral convexities, sparing the brainstem, cerebellum and spinal cord. It is generally seen in association with cerebral amyloid angiopathy and reversible cerebral vasoconstriction syndrome [3].

The intracellular process of formation of hemosiderin is albeit a complex one too. The heme is broken down to free iron by enzyme heme-oxygenase. Free iron is responsible for cell injury. The free iron not only upregulates ferritin production but also the persistent free iron exposure changes the ferritin production from heavy subunit to light subunit which allows long-term iron storage as hemosiderin deposits. Hemosiderin being insoluble, it gets pulled to the subpial surface by natural gravitational force. While storage in the form of hemosiderin is protective in the short term, prolonged exposure which occurs in repetitive episodes of hemorrhage leads to neuronal loss, reactive gliosis and demyelination [4].

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The classical clinical triad consists of sensorineural hearing loss, cerebellar ataxia and pyramidal signs [5]. Symptoms described in paediatric population include seizures, bladder disturbance, headache and extraocular motor palsies while other rare symptoms described in literature are memory loss, backache, bilateral sciatica, anosmia, anisocoria and lower motor neuron signs. However, there is limited literature on the study of the effect of superficial siderosis on neurodevelopmental outcomes. One such retrospective study conducted with 7 infants showed no obvious neurodevelopmental implications of superficial siderosis. However, associated intra/periventricular hemorrhage has been found to be linked with neurosensory impairment and alterations in auditory brain stem responses [6]. The rarity of superficial siderosis in paediatric population is highlighted by limited reported studies and case reports available; however, superficial siderosis developing as a result of subarachnoid bleeding due to factor V deficiency was not encountered on literature research.

Hemosiderin deposition on the surface of neuroparenchyma creates a superficial brownish-yellowish pigmentation, which has shown to have a predilection for superior cerebellar vermis, crests of the cerebellar folia, basal frontal lobe, temporal cortex, brainstem, spinal cord, nerve roots and cranial nerves I and VIII [7]. However, the peripheral nervous system is not involved in superficial siderosis.

Before modern imaging techniques were available, diagnosis could be made on autopsy only. Non-contrast computerized tomography (NCCT) of the head and spine may occasionally point towards the etiology in the form of evidence of trauma or surgery. It may also show atrophy of the cerebellum or rarely hyperattenuated rim along the affected neuroparenchymal surface, which can occur due to calcification at sites of long-standing hemosiderin deposition.

MRI of the brain is the investigation of choice for diagnosing this condition. T2 weighted spin echo images and gradient-echo images (or susceptibility weighted images) show hemosiderin deposition of superficial siderosis fairly well. However, FLAIR images may be useful for identifying acute subarachnoid hemorrhage.

T2-weighted MRI images show a rim of hypointensity around the affected areas predominantly the cerebellum and brain stem. The other preferentially involved areas also show a rim of hypointensity including the cranial nerves. Cerebellar atrophy is often noted with the superior cerebellar vermis and anterior cerebellar hemispheres preferentially involved. A recent episode of sub-arachnoid hemorrhage may appear as a hyperintense rim owing to the presence of methemoglobin. Gradient-echo imaging (GRE) or susceptibility-weighted imaging (SWI) is the most sensitive sequence to look for superficial siderosis which appears as hypointense rim due to magnetic susceptibility effects on these sequences. This effect is also known as blooming artefact, which conspicuously shows the hypointense rim. T1-weighted images may show a subtle partially corresponding hyperintense rim. There is no enhancement noted at the sites of hemosiderin deposition. Tissue damage associated with superficial siderosis may be detectable as atrophy and abnormal signal intensity of the involved regions [8].

The extent and distribution of MRI findings may not correlate with the clinical severity. However, extensive involvement of the brain and spinal cord is usually associated with clinical symptoms.

When superficial siderosis is suspected or diagnosed on MRI, a careful and meticulous search for the source of the hemorrhage should be sought. Hence, in cases where the cause of recurrent bleeding is obscure, additional sequences/diagnostic techniques may be implemented to look for the source. The routine brain MRI protocol includes T1 weighted, T2 weighted, FLAIR and GRE/SWI sequences. Additional sequences/diagnostic techniques include four-vessel cerebral catheter angiography, spinal angiography, CT myelogram, MR angiography, whole spine MRI, MR myelography and MR cisternography.

The imaging differential diagnoses include normal leptomeningeal melanin, brain surface venous plexuses and bounce point artefact which occurs due to phase cancellation of the MR signals in boundary pixels. Rare imaging differential diagnosis includes meningioangiomatosis which is a meningovascular hamartomatous cortical lesion in nodular or gyriform configuration and

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neurocutaneous melanosis which is a rare phakomatosis with meningeal melanocytosis/meningeal melanomatosis on imaging. Normal leptomeningeal melanin appears generally as a variably thick hypointense rim on the ventral surface of the medulla oblongata on T2-weighted spin-echo images [9].

Treatment consists of obliteration of the bleeding site, which may be the cause of future repetitive bleeding. Medical management has been attempted with corticosteroids, chelating agents like desferrioxamine or trientine but with limited success. CSF shunting may be done as it has been thought to reduce prolonged exposure to blood promoting faster clearance of toxins from CSF.

Conclusion

This rare case of superficial siderosis due to congenital factor V deficiency in a paediatric patient reinforces the importance of keeping an eye out for superficial siderosis in any patient with a history of previous subarachnoid bleed. The recognition of the source of bleed has a vital implication in the management of such patients. Accurate radiological diagnosis and early intervention may benefit the patient in the form of reduced morbidity and mortality.

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