

Haemophilia - a missed diagnosis

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OBJECTIVES

A missed diagnosis of haemophilia can prove disastrous. To determine the extent of wrong/misdiagnosis amongst patients with haemophilia, we analyzed records of patients referred to our Haemophilia Centre and the treatment they were being administered or denied and its implications on the morbidity and mortality.

METHODS

There are current 1,156 haemophilia patients registered in our Haemophilia Centre in New Delhi, India. All the patients referred to our haemophilia centre, irrespective of previous laboratory results, are subjected to fresh investigations after a careful clinical work-up as per our protocol before giving out a confirmed diagnosis of haemophilia. We came across nine instances of wrong haemophilia diagnosis and these are presented in this paper including the impact it had on their treatment and outcome.

OBSERVATIONS AND RESULTS

S.No.	TRUE DIAGNOSIS	MISDIAGNOSED AS	No. of CASES
1.	HAEMOPHILIA-A	HAEMOPHILIA-B	2
2.	HAEMOPHILIA-B	HAEMOPHILIA-A	1
3.	FACTOR-VII DEFICIENCY	HAEMOPHILIA-A	1
4.	VON WILLEBRAND DISEASE	HAEMOPHILIA-A	1
5.	MULTIPLE FACTOR DEFICIENCY	HAEMOPHILIA-A	1
6.	RHEUMATOID ARTHRITIS	HAEMOPHILIA-A	1
7.	HAEMOPHILIA-B with PSEUDOTOMOUR	VILLONODULAR SYNOVITIS WITH SEPTIC ARTHRITIC	1

Two patients of haemophilia-A were wrongly diagnosed as coagulation factor IX deficiency whereas one haemophilia-B was receiving treatment as factor VIII deficiency case. In one, the wrong treatment lasted as long as 25 years of his life. Haemophilia-A was misdiagnosed in one patient each, with coagulation Factor VII deficiency and von Willebrand disease. One patient was being treated as haemophilia-A with factor VIII infusion alone even though he had multiple coagulation factor deficiencies.

One patient of haemophilia-A was wrongly diagnosed as rheumatoid arthritis and treated as such for over a decade including the unwarranted use of methotrexate. One patient of haemophilia-B with pseudotumour was wrongly diagnosed as villonodular synovitis with septic arthritis. There was one instance of a patient with haemophilia-A with old thigh bleeds and hematoma wrongly given a suspected diagnosis of sarcoma. These patients were not responding to the treatment being given to them.

Fig-2
HAEMOPHILIA MISSED DIAGNOSIS
(n-9)



- HAEMOPHILIA-A as HAEMOPHILIA-B
- HAEMOPHILIA-B as HAEMOPHILIA-A
- HAEMOPHILIA-A as FACTOR-VII DEFICIENCY
- VWD as HAEMOPHILIA-A
- MULTIPLE FACTOR DEFICIENCY as HAEMOPHILIA-A
- HAEMOPHILIA-A as RHEUMATOID ARTHRITIS
- HAEMOPHILIA-B with PSEUDOTOMOUR as VILLONODULAR SYNOVITIS WITH SEPTIC ARTHRITIS
- HAEMOPHILIA-A with THIGH SWELLING as SARCOMA

CONCLUSIONS

1. A firm diagnosis of the haemophilia and its type is a *sine qua non* for appropriate treatment.
2. A diagnosis of haemophilia may be considered in patients with chronic multiple joints and/or muscle disabilities do not respond expectedly to the treatment administered.
3. Re-confirm the diagnosis and type of haemophilia if patient does not show the expected response to specific coagulation Factor concentrate.
1. A missed diagnosis of haemophilia may breed avoidable morbidity with denial of right treatment and possibly administration of the wrong one.

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