

An Inherited Disorder: Sickle Cell Anemia

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DESCRIPTION

With its numerous forms, sickle cell anemia mimics a multitude of illnesses. In a review of 214 cases of sickle cell anaemia, McGavack found that 97 cases, or 45.3 percent, had an incorrect diagnosis. Of these 97 cases, 39 were misdiagnosed as acute abdominal conditions, and 16 of these patients underwent surgery. The incorrect diagnoses included appendicitis, cholecystitis, obstructive ileus, and peritonitis caused by perforated ulcer.

In this series, it was also discovered that sickle cell anaemia was frequently misdiagnosed as rheumatic fever, osteomyelitis, syphilis, pneumonia, encephalitis, arthritis, and other causes of hemolytic jaundice. A partial list of illnesses that can mimic sickle cell anaemia includes rheumatic heart disease, congenital heart disease, brain tumour, meningitis, cerebral embolism, malaria, and other causes of anaemia.

This is especially true when a baby is young since symptoms are much stranger. It's crucial to keep in mind that this disease's early symptoms, despite being atypical, can and do happen. Patients with severe stomach discomfort, muscle spasms, no bowel noises, vomiting, fever, and leucocytosis present a unique diagnostic challenge in life. When dealing with patients known to have sickle cell anaemia, it may be difficult to distinguish between acute surgical abdomen and these cases; as a result, extreme caution must be used. Of course, one should fully recognise that an acute surgical abdominal condition may develop in these patients as well as the possibility of a sickle cell

anaemia. Every black patient who exhibits clinical symptoms or test results consistent with the condition in the first two to four months of life should have sickle cell anaemia considered as a possibility. The presence of high levels of foetal haemoglobin appears to interfere with the bidding phenomenon. As a result, sickle cell preparation tests and tests based on the various solubilities of sickle haemoglobin may not be positive for sick cell anaemia.

According to a report, 20% of infants with sickle cell anaemia who were identified in their first year of life showed negative sickle cell preparation at first. Ballard et al. found no positive results among 33 neonates with a commercial differential solubility test in three infants aged 4, 5 and 7 weeks; however, the solubility test was essentially positive when read at 3 minutes and unequivocally positive at 24 hours; clearly positive reactions were obtained by the time these infants were 4 months old.

There have been reports of haemoglobin C disease and sickle cell trait in cord blood samples. The conclusive diagnosis of sickle cell anaemia is typically made *via* haemoglobin electrophoresis. Distinct electrophoresis techniques have varying degrees of sensitivity and ability to separate different haemoglobin with identical mobility, and some don't allow quantification of the detected haemoglobin. The failure to detect that various haemoglobin have similar mobility to haemoglobin under specific conditions and the failure to quantify certain haemoglobin. It indicates pitfalls in the diagnostic diagnosis of sickle cell illness with haemoglobin electrophoresis. Family studies should support laboratory investigations.

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