

Hereditary haemorrhagic telangiectasia in a black adult male: case report

Abstract:

Hereditary haemorrhagic telangiectasia, (HHT) or Rendu-Osler-Weber disease is a genetic autosomal dominant disorder that is characterised by telangiectasias, (small vascular malformations), in mucocutaneous tissues and arterial venous malformations, (AVMs), in various internal organs. Although HHT is relatively common in whites, the disorder has been reported to be rare in people of black African descent. Majority of HHT patients present with recurrent epistaxis, which in a significant proportion of patients is severe, warranting repeated blood transfusions and iron supplementation. Telangiectasias are most frequent on the tongue, hands, nose, lips and the gastrointestinal tract (GIT). AVMs occur in internal organs, particularly the lungs, brain, and the liver. Early and correct diagnosis of HHT is crucial as patients derive benefit from certain specific treatment modalities. Besides, AVMs which occur in various organs pose serious complications that may lead to death and therefore require early detection. We report a 55 year old black African male with HHT who presented with severe recurrent epistaxis and haematochezia leading to severe anaemia requiring repeated blood transfusions. His son, daughter and a maternal uncle experience milder recurrent epistaxis. The management of this patient and a brief review of the clinical features and management of HHT is presented. Our aim is to raise awareness of the occurrence of HHT in Kenya, in order to enhance early diagnosis and appropriate management