Tropical distal renal tubular acidosis: clinical and epidemiological studies in 78 patients

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†The senior author Professor Oliver Wrong died 24 February 2012 while completing this paper.

Received 6 May 2012 and in revised form 1 July 2012

Summary

Background: Distal renal tubular acidosis (dRTA) caused by mutations of the SLC4A1 gene encoding the erythroid and kidney isoforms of anion exchanger 1 (AE1 or band 3) has a high prevalence in some tropical countries, particularly Thailand, Malaysia, the Philippines and Papua New Guinea (PNG). Here the disease is almost invariably recessive and can result from either homozygous or compound heterozygous SLC4A1 mutations.

Methods: We have collected and reviewed our own and published data on tropical dRTA to provide a comprehensive series of clinical and epidemiological studies in 78 patients.

Results: Eight responsible SLC4A1 mutations have been described so far, four of them affecting multiple unrelated families. With the exception of the mutation causing South-East Asian ovalocytosis (SAO), none of these mutations has been reported outside the tropics, where dRTA caused by SLC4A1 mutations is much rarer and almost always dominant, resulting from mutations that are quite different from those found in the tropics. SLC4A1 mutations, including those causing dRTA, may cause morphological red cell changes, often with excess haemolysis. In dRTA, these red cell changes are usually clinically recessive and not present in heterozygotes. The high tropical