MALARIA JOURNAL

ORAL PRESENTATION Open Access

Epistasis and the sensitivity of phenotypic screens for beta thalassaemia

Bridget Penman[1*], Sunetra Gupta[1], David Weatherall[2]

From Challanges in malaria research: Core science and innovation
Oxford, UK. 22-24 September 2014

Severe forms of alpha and beta thalassaemia have been estimated to affect approximately 68000 births annually. Individuals who carry thalassaemic genes are protected against death from malaria infection; the global distribution of thalassaemic genes thus matches the historical distribution of malaria.

Screening programmes are a vital tool to counter the thalassaemias by:

(i) identifying individual carriers and allowing them to make informed reproductive choices, and (ii) generating population level gene-frequency estimates, to help ensure the optimal allocation of public health resources. For both of these functions it is vital that the screen performed is suitably sensitive.

One popular first-stage screening option for beta thalassaemia in low-income countries is the One Tube Osmotic Fragility Test (OTOFT). Here we introduce a population genetic framework within which to quantify the likely sensitivity and specificity of the OTOFT in different epidemiological contexts. We demonstrate that the co-occurrence of alpha thalassaemia, and other malaria related erythrocyte poly-morphisms such as Southeast Asian Ovalocytosis and glucose-6-phosphate dehydrogenase deficiency, could reduce the sensitivity of OTOFTs for beta thalassaemia to below 70%. Our results highlight a potential hazard of the widespread application of OTOFTs and emphasize the fact that the public health impact of any single genetic adaptation to malaria cannot be considered in isolation.

Authors' details
[1]University of Oxford Zoology Department, Oxford, UK. [2]Weatherall Institute of Molecular Medicine, Oxford, UK.

Published: 22 September 2014

[1]University of Oxford Zoology Department, Oxford, UK
Full list of author information is available at the end of the article

doi:10.1186/1475-2875-13-S1-O13
Cite this article as: Penman *et al.*: Epistasis and the sensitivity of phenotypic screens for beta thalassaemia. *Malaria Journal* 2014 **13**(Suppl 1):O13.