

Name of Journal: *World Journal of Obstetrics and Gynecology*

ESPS Manuscript NO: 21530

Manuscript Type: Minireviews

Universal screening for Hemoglobinopathies in today's multi-ethnic societies: How and when

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Abstract

Increasing multi-ethnicity in countries endemic or non-endemic for Hemoglobinopathies has brought fundamental changes to the screening strategies for these traits. While in the past pre-screening on microcytosis was a reasonable method to economize upon follow up analysis, selecting low MCV means today missing all those normocytic carriers of common traits associated with severe conditions. Therefore, blood count should not be considered as a pre-selection tool but as additional information to be used for the interpretation of the provisional results, obtained by routine high throughput separation and measurement of the Hb fractions. Moreover, the moment of screening should be well planned depending on the social and cultural situation. Screening for genetic diseases in a modern multi-ethnic society should be offered to couples seeking progeny when both partners are more likely to be equally concerned with the good health of their children. In several societies screening before marriage and changing partner choice is culturally accepted. However, new generations are bound to disagree with these more or less imposed conditions and may decide not to renounce the choice of their partner asking for other preventive methods. In addition,

a carrier state during pre-marital screening may in some cultures stigmatize the carrier.

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