

Controlling sickle-cell syndromes in Portuguese-speaking sub-Saharan African populations: Role of the Portuguese health system in a trans-continental patient mobility context

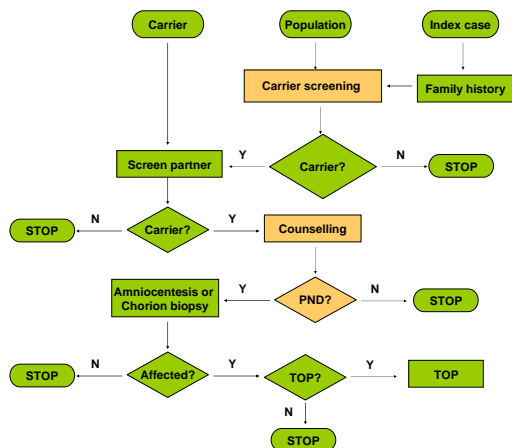
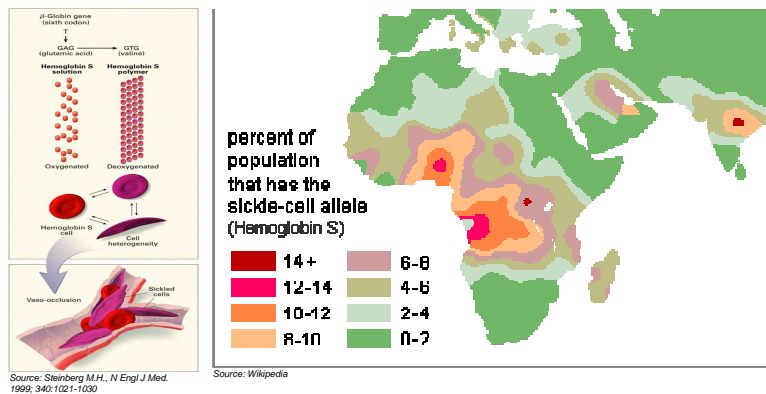
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Background

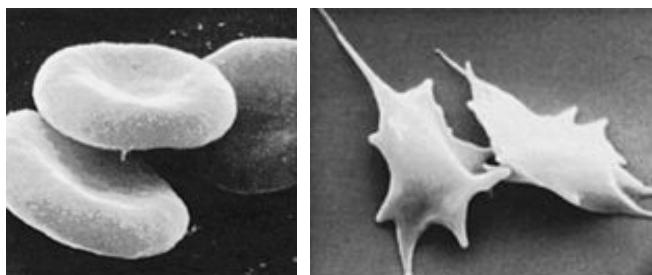
Sickle-cell disease (SCD; OMIM 603903) is a rare autosomal recessive condition characterised by haemoglobin S production, leading to haemolysis (anaemia), vascular disease (thrombophilia) and immunodeficiency (infection). SCD affects predominantly populations of sub-Saharan African and Asian Indian origin and their descendants in Europe and the New World. It displays marked clinical heterogeneity with a wide severity range, possibly in result of a complex gene-gene and gene-environment interplay. Due to the lack of reliable severity predictors and an effective affordable cure, many at-risk couples choose to participate in prevention programmes based on carrier screening, nondirective genetic counselling, prenatal diagnosis (PND) and termination of affected pregnancies (TOP):



Successful programmes have been running for the last two decades in the USA, Latin America and Europe (1-3). However, in the highest prevalence regions this kind of control has not been a priority, despite the fact that thousands of new patients are born every year. This may be due to the religious beliefs, and the political and social attitudes of the affected populations and local health care providers (4).

Following its long-standing relationship with African populations, the Chief Medical Officer of Portugal has issued an official guideline opening the Portuguese national haemoglobinopathy control programme to families coming from African Portuguese-speaking countries (5).

As part of this programme our centre of expertise combines the provision of genetic testing with research activities aiming to decipher the physiopathology of SCD.



Results

Since 1990, sixty-seven sub-Saharan African couples at risk for SCD have been referred to our centre by various medical genetics or obstetrics services for PND. So far, this activity, positively sought after by the affected families, allowed (through an informed choice) the prevention of eleven new SCD cases and the detection of a large number of asymptomatic HbS carriers.

Discussion

The data presented here demonstrate the acceptability of PND by the African at-risk couples (at least in an European setting) and are a successful illustration of the concept that the provision of genetic services should be adapted to the population's needs, socio-cultural background, history and demography, respecting differences, while being equitable and accessible to all.

To fully achieve this goal a comprehensive collaborative endeavour is underway aiming to replicate this intervention model in a large African Portuguese-speaking country where sickle-cell trait reaches one of the highest prevalences world-wide.

This capacity building exercise should include training (of doctors, nurses and laboratory technicians) both locally and in Portugal, and appropriate technology transfer (e.g. patient database, phenotyping and genotyping).

References

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