

Cover Page



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Title: Toward prevention of Hemoglobinopathies in Oman

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Stellingen behorend bij het proefschrift:

Toward prevention of hemoglobinopathies in Oman

1. Identifying the mutation spectrum of the Hemoglobinopathies in a specific population is not only essential for primary prevention, but it also gives an opportunity to look into a population's history. This thesis.
2. Being a healthy carrier of sickle cell disease or beta thalassemia can be seen as an evolutionary advantage. The disadvantage (the risk of having affected progeny with an equally carrier partner) seems to be the price to be paid for being protected against child death due to *malaria tropica*. This thesis.
3. Multiple genetic factors may ameliorate or worsen disease severity. Identifying significant determinants that may influence the risk assessment and / or the response to treatment is essential. This thesis.
4. Treatment is not the same as cure. Parents at risk for hemoglobinopathy should be well informed about the burden and the meaning of treatment. This thesis.
5. The actual "state of the art" diagnosis of Hemoglobinopathies needs to be done at the hematological, biochemical and molecular level. Screening must start with the biochemical step (separation and measurement of the Hb fractions) must be supported by the hematological parameters (CBC) while molecular analysis is needed for specific genotype/ phenotype correlation, risk assessment, prevention or tailored treatment. Giordano PC Int J Lab Hematol. 2013 Oct;35(5):465-79.
6. Emerging sequencing technologies may eventually replace the 'golden standard' Sanger sequencing and become affordable for screening and primary prospective prevention. Traeger-Synodinos et al Eur J Hum Genet. 2015 Apr;23(4):560.
7. Discouraging marriages between carriers at risk is in some cultures considered to be the "easy" way out to prevent the birth of severely affected children. Giordano PC et al. Int J Environ Res Public Health. 2014 Jun 11;11(6):6136-46.
8. Although primary prevention methods aim for the same goal, the ways to acceptance relies on cultural and religious differences. Giordano PC et al. Prenat Diagn. 2005 Oct;25(10):885-93.
9. The views and attitudes towards a once unacceptable issue are prone to change with the change of generations.
10. The world population is bound to become much more multi-ethnic in few decades. Genetically speaking the mixture of distant ethnicities is favored by a natural mechanism called the "attraction of the rare phenotype" (Piero Giordano, 2014).
11. 'Impossible' is not a scientific term (Vanna Bonta, The Space Review, 2004).
12. The whole of science is nothing more than refinement of everyday thinking (Albert Einstein, Physics and Reality, 1936).