Hemoglobinopathies in Iran : molecular spectrum, prevention and treatment.

Yavarian, M.

Stellingen

1. The survival rate emerging from this study reveals that improvement in transfusion and chelation therapy is needed. Moreover, improved social measures could reduce mortality. The process toward a better management of β-thalassemia in Iran is still in need of great attention (this thesis).

2. Although the Xmn-1 polymorphism seems to be the most important modulating factor involved in Hydroxyurea therapy response, other elements could play a role and more studies in this field are needed (this thesis).

3. In absence of direct DNA sequencing DGGE is the most sensible method to detect β-thalassemia mutations in multiethnic areas where a broad spectrum of mutations occurs (this thesis).

4. The provinces of Hormozgan and Mazandaran, with a carrier frequency of 9.7% represent the Iranian areas with the highest prevalence and in highest need of prevention (this thesis).

5. The cheapest prevention measure in Iran is to adapt the choice of the partner when a future couple at risk is diagnosed. This is however only acceptable in case of traditionally arranged marriages (this thesis).

6. To date the average life expectation for well managed β-thalassemia major patients is 28 years. Improvement can be expected with better transfusion and chelation therapy (this thesis).

7. In 1998 was calculated that the economic burden for treatment of the 20,000 thalassemia patients living in Iran is 220 million US$ per year, 11,000 US$ per patient per year (The Iranian Thalassemia Association).

8. Disregarding inflation, over 300,000 US$ are needed to keep each born patient alive. Bone marrow transplant and improved survival by better chelation therapies are not included in this figure (The Iranian Thalassemia Association).

9. Screening at the premarital stage the 12,000 couples that marry each year in Hormozgan would cost 10,000 US$. Spending an additional 600,000 US$ in prenatal diagnosis for the 600 couples at risk would prevent the expected 150 affected children. One-year treatment of the same 150 patients would cost 1.7 million US$ (The Iranian Thalassemia Association).

10. Accepting to remain childless is usually no option for Iranian couples. Prenatal diagnosis in the 11th week of gestation and selective abortion will be also in Iran the most acceptable prevention measure. Iranian authorities allow abortion for medical reasons up to the 17th week of gestation (The Iranian Thalassemia Association).

11. Pre-implantation genetic diagnosis (PGD) represents an alternative for couples with infertility problems or with moral objections against terminating an affected pregnancy (Sermon et al, Lancet 2004)

12. Gene therapy could provide a permanent correction of defects in hematopoietic stem cells. The complexities of the beta-globin gene and its regulation have impeded progress on this field (Puthenveetil & Malik P. Curr. Hematol. Rep. 2004)

13. Pre-implantation HLA matching has recently emerged as a tool for couples desiring to conceive a potential donor progeny for transplantation in a sibling with a life-threatening disorder (Fiorentino et al., Mol Hum Reprod. 2004)

14. Science and everyday life cannot and should not be separated. Science, for me, gives a partial explanation of life. In so far as it goes, it is based on fact, experience and experiments. “Rosalind Franklin, 1940: a letter to her mother”.