Preconceptional diagnosis for giant axonal neuropathy before wedding engagement decision

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INTRODUCTION

Study question: What are the possibilities and the benefits of preconception and premarital genetic diagnosis in genetic disorders particularly those of drastic prognosis?

Summary answer: In conservative societies, premarital screening that offers a crucial health assessment of soon-to-be married couples with genetic risk factors is not usually possible.

What is known already: Preconceptional genetic diagnosis help couples of genetic disorders carrier risk making an informed reproductive decision. Generally, physicians and general population thought that preconception genetic diagnosis or screening is not necessary if a couple have no known family history of a genetic disorder. However, in common genetic disorders like cystic fibrosis (in Europe) and Crigler-Najjar syndrome (in Tunisia), 90% of carriers have no known family history of the condition. Moreover, the risk is considerably higher for consanguineous couples, which is a frequent condition in some ethnicities.

Premarital screening can also offers a crucial health assessment of soon-to-be married couples with genetic risk factors based on specific family history.

However, such approach is not usually easy to manage in conservative societies, particularly when the affected family refuse to deliver the necessary information about the genetic condition considered as a taboo.

OBJECTIVES

This was a retrospective study about the preconceptional diagnosis inquiries in our genetic counselling and a case report of a Tunisian consanguineous couple who was referred to us by their own for seeking a premarital screening for an unknown neuropathy before wedding engagement decision.

MATERIALS AND METHODS

A Tunisian consanguineous couple was referred to us by their own before wedding engagement decision because of a familial history of a severe neuropathy that was noted in the offspring of a shared cousin.

The couple was unable to bring us more information about the genetic condition because of the familial repugnance.

After investigations, the disease was defined as a giant axonal neuropathy as well as the familial mutation of the GAN gene coding for gigaxonin protein.

Molecular analysis was made using extracted genomic DNA according to standard phenol-chloroform procedures.

Direct sequencing of the targeted DNA sequence harboring the mutation of the shared cousin was carried out.

RESULTS

The molecular diagnosis using the direct sequencing of the exon 9 of the GAN gene revealed a normal status for the male and a carrier status for the female with the 1447C>T mutation.

During the genetic counselling, the couple was informed about the options that can be offered to avoid having affected children (prenatal and pre-implantation genetic diagnosis in particular).

CONCLUSION

Although our study is limited at the genetic level, it could be socially interesting because it showed the negative attitudes of the general population towards the genetic conditions and the familial responsiveness, as well as the reticence of physicians towards genetic preconceptional and premarital carrier diagnosis.

Our results emphasizes the importance of education regarding the benefits of preconceptional genetic diagnosis, particularly in conservative societies. Keywords: Giant axonal neuropathy, preconception diagnosis, neuro-genetic diagnosis

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