

CC-08

**CARRIER SCREENING – WILL IT BE POSSIBLE TO
ELIMINATE AUTOSOMAL RECESSIVE DISORDERS?**

José Carlos Ferreira, MD, PhD

Medical University of Warsaw

Warsaw, Poland

ZEC1962@gmail.com

In a future not far away, anyone can have his/her exome/genome sequenced for a reasonable price. Lots of debate will deal with the utility of doing this. But, at least, a likely useful use of that type of data will be carrier screening. To find out, prior to conception, if a couple carries recessive disease causing mutations in the same gene, will make it possible, using preimplantation or prenatal genetic diagnosis, to identify embryos or fetuses affected. Current screening methods, for the vast majority of diseases, rely on family history. The main drawback of such methods is the need for the birth of an affected individual – a proband – to identify carriers. Universal preconceptional screening for all/most recessive disease causing mutations would reduce dramatically this drawback.

But is it worth the cost? Are there other more cost reasonable alternatives to exome sequencing based screening? Are there other more cost reasonable alternatives to universal screening? That is, population targeted screening?

In this new age of genomics, where exome based screening is already being offered to anyone who can afford it, it may be worth to revisit the discussions about carrier screening.

Where there is no screening program, is it reasonable to jump directly to the exome based screening, wait until it gets more affordable, or consider the implementation, even temporary, of other more limited technologies and programs already tested. Will it be worth? Which perspective should be taken? The public health perspective or the individual perspective? Are there differences between the two perspectives?

The history of carrier screening programs and the discussions that preceded them may be good starting points in the search for answers to these questions. In the beginning, carrier screening was based on family history. Later population targeted preconceptional screening for targeted diseases was added – sickle cell disease in African ancestry, thalassemias in Mediterranean ancestry, Tay-Sachs disease in Ashkenazi Jewish (AJ) ancestry. Then several expansions of number of diseases in AJ ancestry and universal screening for Cystic Fibrosis occurred. And more recently, the offer of universal genomic based carrier screening to anyone who can afford it is the last development in this process. What were the problems in the implementation of those programs? What was accomplished? What can be learned?

This presentation will probably raise more questions than answer them. Discussion will be expected.