Anesthesiologists as Genetic Counselors?

To the Editor:

We wish to comment on the editorial by Drs. Lee and Raja “Should Anesthesiologists be Equipped as Genetic Counselors”?

Although Drs. Lee and Raja highlight pharmacogenetics and molecular genetic factors that might influence patients’ response to pain medication, the omission of a discussion of certain pharmacogenetic disorders specifically related to anesthesiology is baffling and a significant oversight. In particular, there are two classic pharmacogenetic disorders of special interest to anesthesiologists: the response to succinylcholine due to mutations in the gene that elaborates pseudocholinesterase (butyrylcholinesterase), and malignant hyperthermia syndrome.

Anesthesiologists who are expert in understanding malignant hyperthermia already use molecular genetic testing to guide patients in the selection of anesthetics and to determine the risk of developing malignant hyperthermia. A sophisticated knowledge of the significance of the more than 200 mutations associated with the ryanodine receptor gene is necessary for advising patients with malignant hyperthermia. In fact, at one of the two DNA testing laboratories for malignant hyperthermia (the University of Pittsburgh, Division of Molecular Genetics, Pittsburgh, Pennsylvania) a genetic counselor is employed to help evaluate and advise patients.

The issue of understanding let alone counseling patients on the direct-to-consumer tests for evaluating a patient’s risk for disease or response to medication, is exceedingly complex because phenotype may be influenced by several genes and gene products. The Food and Drug Administration at recent hearings has cited concerns for risks to public health imposed by the trend toward increasingly complex tests brought to market primarily through the Internet and without Food and Drug Administration review.

As the authors point out, the functional significance of a mutation is complicated by the genetic background of the patient as well as environmental factors. The question concerning these tests is not “Are people buying them?” but rather how does one interpret these tests and provide meaningful advice to patients?

Genetic counseling has become increasingly complex as the collaboration between pathogenic mutations and contributing genetic variants generates sometimes unpredictable phenotypes and patterns of heredity. Without in-depth education and training, anesthesiologists should not be giving advice on the response to pain and pain medication based on a DNA profile. Physicians in all specialties should develop a working relationship with centers that have medical genetics divisions or departments that include board-certified genetic counselors to provide the necessary genetic counseling.

It is entirely appropriate for anesthesiologists to focus research activities on the molecular genetic basis of drug response and take an active interest in education concerning molecular genetic research. This is a far cry from being “equipped” as a genetic counselor.

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References


In Reply:

We thank Rosenberg et al. for their interest in our editorial on the potential role that anesthesiologists may have to play as genetic counselors in light of developments in the direct-to-consumer genetic testing market. We also appreciate these researchers noting the importance of malignant hyperthermia when discussing genetics in anesthesiology. The focus of the editorial was on pain genes; it was not meant to be comprehensive.

It is undeniable that most anesthesiologists have little to no formal training in genetics and genetic counseling; therefore, it would be to the patient’s benefit to seek consultation with a genetic counselor for answers concerning his or her genetic predispositions. We agree that all specialists, including anesthesiologists, should develop a working relationship with a medical genetics department to respond appropriately to patients’ concerns based on their genetic profile and to provide optimal care to their patients.

Unfortunately, such communication between anesthesiologists and genetic counselors is not the norm. For example, although patients with a suspected malignant hyperthermia crisis or a suspected susceptibility to malignant hyperthermia are advised to undergo in vitro contracture testing and gen-

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