Regarding protecting patients from harm:

1. A couple who had a second child with intellectual disability and multiple birth defects because they were told incorrectly by their pediatric neurologist that they were not balanced carriers of a chromosome abnormality. The doctor had ordered the wrong test for the parents, cytogenomic SNP microarray, thinking that if it found the abnormality in the child then it was correct for parents too. The doctor told the parents they were not carriers based on the normal CMA result. One parent was, in fact, a carrier of a balanced chromosome rearrangement. This would have been detected had the correct test, chromosome analysis, been ordered. The couple had a second child with an unbalanced form of the translocation one parent carried. Incidentally, I doubt the parents pursued any legal action as they were not sophisticated medical consumers and would not likely have access to any recourse.

2. I have had too many conversations with doctors to count regarding why exome analysis will not detect every type of genetic variant, typically after a normal exome analysis performed on a patient whom the doctor strongly believes has a genetic condition. The answer is: Yes, your patient may still have a genetic condition, and this is very possible because even whole genome sequencing does not detect every type of variant. Kudos to all of these doctors who sought help understanding the results of a test that did not exist when they were in training.

Regarding saving health care dollars:

Miller CE, et al. Genetic counselor review of genetic test orders in a reference laboratory reduces unnecessary testing. Am J Med Genet A. 2014 May;164A(5):1094-101.

A retrospective review of orders at a national reference lab found that over 21 months, review of molecular testing orders by genetic counselors who suggested, when indicated, alternative cheaper and/or more appropriate tests saved the referring institutions an average of $792 per misorder. Approximately 26% of all requests for complex genetic tests assessing germline mutations were changed following GC review.