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PERSPECTIVE

Nothing About Us Without Us: Guidelines for Genetic Testing

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Guidelines for genetic testing developed by professional societies may be more centered on the concerns of society members than the needs of the individuals and families that may undergo testing. This could be considered appropriate because it will be the healthcare provider who will assess whether to test; when to test; whom to test; and how to provide the right support, interpretation, and follow-up to the individual or family tested. Genetic testing requires guidelines because of the complexity of testing for the individual and his or her family. It also requires flexibility because context matters a great deal in genetics.

For this very reason, multiple stakeholders should be involved in guideline development. Professional societies have a deliberative process for their members, and those closely associated with their practice, to engage in discourse that leads to guidelines. We believe that the testing guidelines should reflect more than the professionals' perspective. Ultimately, the ramifications of testing guidelines do not exist in isolation from the whole healthcare experience of the individual. This means that the perspectives of other providers, the individuals, and families should be a part of the deliberative process. One might ask why the advocacy organizations don't establish guidelines themselves. We believe that multiple stakeholder groups should be involved.

A striking example of where two groups of professionals create difficulties for each other and the people they serve is the guidelines for spinal muscular atrophy (SMA) carrier screening. Obstetricians disagree with geneticists about carrier testing for this condition. Geneticists believe that carrier testing for SMA should be offered to the general population, and obstetricians do not believe it should be offered broadly. In statements made by the two societies, the contrast is clear, but from the point of view of the patient, it is confusing. The American College of Obstetricians and Gynecologists' Committee on Genetics concludes: "[The committee] agrees that preconception and prenatal screening for SMA is not recommended in the general population at this time" (American College of Obstetricians and Gynecologists, 2009). The American College of Medical Genetics, on the other hand, concludes: "Because SMA is present in all populations, carrier testing should be offered to all couples regardless of race or ethnicity. Ideally, the testing should be offered before conception or early in pregnancy. The primary goal is to allow carriers to make informed reproductive choices" (Prior 2008).

As a result of these two guidelines, obstetricians, midwives, geneticists, nurses, nurse practitioners, pediatricians, genetic counselors, and potential parents do not have a clear guideline on which to base a decision. It does not appear that either professional society engaged the other stakeholders from the "medical home" to any great degree, particularly in the guideline articulation. It is not apparent that either society invited the very individuals whose lives are affected by the decision into the process of creating guidelines.

This case of SMA carrier screening is also a good example because of the complexity of the issue for parents as well. There are parents of children with SMA who desperately wish carrier screening had been available to them before they had to witness their baby die of the condition. There are also parents of children with the condition, albeit a milder form, who are very concerned that carrier screening might limit the number of children born with the condition. This might decrease appreciation for these children and perhaps reduce research activity and funding for the condition.

The American College of Medical Genetics recently released guidelines for genetic testing in children (American College of Medical Genetics, 2013). This is a revision of a statement they made more than 10 years ago, so it was a welcome addition to current considerations for all of the stakeholders. It is not apparent that the deliberative process included parents of children affected by genetic conditions. It did not include advocates of the various disease advocacy organizations. Although it would be much more difficult to come to consensus with multiple stakeholders, doing so is a worthy endeavor.

The silos that exist in medicine, from basic discovery through translational science to delivery of services, severely limit progress. The learning healthcare system (Institute of Medicine, 2011) requires that all stakeholders be involved in the entire process. Starting with clinical questions emerging from clinical care and the lived experience of individuals and families with disease will help accelerate diagnostic and treatment development. Engaging clinicians (of all specialties) and citizens in formulating basic research plans will help to make discovery more relevant to translational science. Empowering communities, whether geographic, ethnic, or disease affinities, to be responsible for characterizing cohorts will stitch together a national clinical research network of engaged communities (Seifer S, 2013). Learning from the people who

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benefit from interventions will provide relevant outcome measures. Connecting patients to the process by which their care is delivered and subsequently improved will dramatically improve patient-reported outcomes. The cycle will be a healthy and robust one if only our scientific and medical communities will open their doors to the full continuum of interested parties. Progress will accelerate if people reclaim their own health and pathways to improve it.

Engaging a variety of stakeholders in guideline development is a small but important stepping stone on the way. We call for the next set of deliberations to include representatives of many stakeholder groups, for the good of us all.

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