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Pediatricians Take New Stand on Genetic Testing

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The Human Genome project is producing new information about genes on a rapid basis. Because of this, the American Academy of Pediatrics (AAP) recently issued a new policy statement about genetic testing in children, which it recommends all members follow.

The statement addresses three distinct situations. The first situation has to do with newborn screening, tests that are performed in the hospital when an infant is



born. Dr. David Horwitz, pediatrician at NYU Medical Center explains, "In New York, these tests are mandatory with mandatory notification to the parents." This differs from state to state and the AAP now recommends that all newborn testing be done after informed and voluntary consent is given by parents so that they are more involved and informed in the decision-making process. Dr. Horwitz says this recommendation is "controversial" and states he is not sure this can be effective because of the time necessary to explain every single test performed.

The AAP's position, according to Dr. Jeffrey Botkin at the University of Utah in Salt Lake City, is that "the academy would like to find better ways to educate parents before the baby is born and work together to protect babies. The system can work better." He does point out that the recommendation that doctors consult with parents about these tests is "radical" in the pediatric community. Dr. Harry Ostrer, professor of pediatrics, medicine and pathology and the Human Genetic

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Program Director at NYU School of Medicine says that "education is of paramount importance and there is a role for improving that."

The AAP further recommends that future newborn tests be introduced in a careful way that allows for evaluation of the risks and benefits of screening. The March of Dimes has stated that it believes all newborns across the country should receive the same tests and that even if a test is for a very rare disease, it should be given regardless of cost-effectiveness. "When newborn screening tests for all treatable conditions are universally available and the quality of tests is assured, it may well turn out this effort will be economically beneficial to health insurers," says Dr. Jennifer Howse, March of Dimes National President. "Nevertheless, the March of Dimes believes the primary consideration should always be the health of babies."

Dr. Robert Nelson of Children's Hospital in Philadelphia, and spokesperson for the AAP on this policy, takes issue with this. He points out that all tests are not appropriate in all areas. "Tests should be more uniform, but pockets of population make it worth screening for certain things only in certain areas." Some diseases are carried only by people of certain types of descent and while the disease may be quite rare in the general population, when a large group of people with common descent is clustered in an area, it makes sense to test for the illness there, but not other places. Dr. Botkin points out that this does raise "serious questions about justice...and cost issues". Dr. Horwitz says, "Public health screening does have to take into effect cost-effectiveness" but points out that there should be newborn screening only when there is some treatment available.

The second recommendation in the AAP policy statement is that children should not be tested for carrier genes. A carrier gene is an inherited gene that means the patient cannot contract the genetic disease and merely carries the gene for it, but that he or she could pass it on to his or her children who could contract the disease. Sickle cell anemia is an example of a disease that can be carried without actually having it. The AAP recommends that children not be tested for carrier genes because this is a decision the patient should make as an informed adult and is only a concern when a person reaches childbearing years. Since the child's health is not at risk, there is no reason to test during childhood. The AAP also points out that discrimi-

nation and mental anguish are possible effects of identifying children who carry genes for these diseases.

The AAP does recommend that when carrier gene status is found while testing for the actual disease itself, or accidentally, that the information should be disclosed to the parents. With regard to carrier status, Dr. Ostrer says that "genetic testing should be done when the results are useful to the recipient of the test" and points out that parents ask for tests out of curiosity when it is not useful to them. He also relates an anecdote of a male friend who was tested for the BRCA1 (breast cancer) gene and was a carrier. This result had a devastating effect on his family because he had a young daughter who then had a 50 percent likelihood of contracting breast cancer herself based on this. The family felt incredible stress and obsessed about the child's diet and ways to reduce her risk.

The third part of the AAP statement recommends that pediatricians not test children for late onset diseases, unless there is a treatment available to prevent children from developing the disease or if the child exhibits symptoms. Late onset diseases are illnesses that do not cause symptoms until adulthood. Dr. Nelson says, "It is important to be clear about this. We are not talking about diagnostic testing." Diagnostic testing is done to determine the cause of symptoms a patient is experiencing, so if a child exhibits symptoms of a late onset disease, testing would be done to diagnose it under the policy.

The policy recommends that tests for late onset diseases not be done until the patient is old enough to make the choice him or herself. Dr. Horwitz points out that identification of the gene "can lead to misinterpretation, social discrimination and personal anguish. In practice this is correct. You don't know the depths that people can sink to when they learn this information, and it can be devastating and very disruptive for a person." Dr. Botkin believes that the policy does not interfere with a parent's or child's right to choose but "reinforces caution with certain kinds of choices." He says the statement "doesn't take away the choice but suggests the choice should be rare or seriously considered." Dr. Ostrer recommends that parents "really think about what is in the best welfare of their children" and points out that it is up to the professionals to establish a standard of care and to provide guidelines without public input. ♦