New system for classifying infant lung disease developed

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A new classification system of rare lung diseases in infants is improving diagnosis and treatment. The system clears up considerable confusion about how to classify and treat diseases that are rarely seen by most doctors and pathologists, says Gail H. Deutsch, M.D., lead author of the multi-center study that developed the new guidelines.

“Formerly, doctors used a number of different terms to label the same disease,” explained Dr. Deutsch, who is an assistant professor of pathology at Cincinnati Children’s Hospital and Medical Center. “In some cases, a disease with a favorable prognosis has been confused with a potentially lethal lung disease.”

The research was published in the first issue for December 2007 of the American Journal of Respiratory and Critical Care Medicine, published by the American Thoracic Society.

The study, funded by the National Institutes of Health through its Rare Lung Diseases Consortium, included data from 11 medical centers in North America. Investigators reviewed 187 biopsies of children under the age of 2 who were being evaluated for diffuse lung diseases like interstitial lung disease (ILD), an uncommon disorder that affects the interstitium, or tissue between the air sacs of the lungs. Children with ILD commonly have prolonged respiratory symptoms of fast breathing and low oxygen levels and exhibit diffuse changes on chest radiographs. When the cause of their symptoms is not identified with blood tests or x-rays, a surgical lung biopsy is often needed for diagnosis.

In this study, the researchers were able to classify 88 percent of the 187 lung biopsy cases, and found a diverse spectrum of lung diseases that are largely unique to young children.

One-quarter of the lung diseases studied were grouped together under the label “growth abnormalities.” The best-known is pulmonary hypoplasia, in which prenatal conditions result in deficient lung growth. Another group of diseases was categorized as “surfactant dysfunction disorders,” which refer to genetic abnormalities of surfactant, a fluid, detergent-like substance that plays a critical role in keeping the air sacs of the lungs open.

Dr. Deutsch said that at Cincinnati Children’s Hospital and the other medical centers that participated in the study, the new classification system is helping pathologists diagnose children’s lung disease more accurately, leading in some cases to more appropriate treatment.

For instance, in the past, children with lung growth abnormalities might have been treated as though they had ILD and given steroids, which may not be an effective treatment for them.

The new system also gives doctors more information about an infant’s prognosis. In the past, children with ILD were thought to have a high rate of illness and death. The classification system can help doctors distinguish certain children who may appear very ill, but who have a high chance of recovery (such as children with pulmonary interstitial glycogenosis and neuroendocrine cell hyperplasia of infancy), from those with a particular genetic mutation, known as ABCA3, who are unlikely to recover on their own and may need a lung transplant, Dr. Deutsch said.

“This new system gives clinicians, radiologists and pathologists a structure upon which to base diagnoses in a uniform fashion —we’ll all

be reading from the same book," said Andrew Nicholson, M.D., consultant histopathologist at Royal Brompton Hospital and professor of respiratory pathology at the National Heart and Lung Institute Division of Imperial College School of Medicine in London, who co-authored an editorial accompanying the study with Andrew Bush, M.D.

"The next step is for researchers at other medical institutions and in other countries to test the new system to see how applicable it is to their patients," Dr. Nicholson said. "Pathologists should review other cohorts of cases to see if any disease groups are missing, or if others are over-divided."

"When a classification system for adult diffuse lung disease was proposed by the American Thoracic Society and European Respiratory Society in 2002, pathologists took part in such a validation process," he said. "Before the adult classification system was in place, doctors sometimes used the same term for different diseases, but since 2002, we have been able to use much stricter definitions, which has improved accuracy of diagnosis."

It has also improved physicians' understanding of some disease patterns that were previously viewed as idiopathic. "The new classification for infant lung disease gives us the same opportunity to tighten our definitions and increase the accuracy of diagnosis in children," said Dr. Nicholson.

Source: American Thoracic Society