

Diagnosis: Testing Is Developed for Rare Diseases in Infants

By ERIC NAGOURNEY
Published: April 4, 2006

Researchers say they have developed a relatively simple technique for screening newborns for a variety of rare, sometimes fatal diseases that are easy to overlook until it is too late to do much about them.

The researchers, who presented their findings last week at a meeting of the American Chemical Society, said the test can pick up Tay-Sachs, Gaucher, Krabbe, Pompe, Nieman-Pick, Fabry and Hurler syndromes.

The lead author of the study, Frantisek Turecek of the University of Washington, said one of the test's big selling points was that it could be done using the blood that is routinely taken from newborns' heels for other tests.

"The sample is available no matter what," Dr. Turecek said. "That's a big bonus." While some of the diseases can also be detected through [amniocentesis](#), the procedure poses a risk to the fetus, and many women do not have the test done.

In the new test, the blood is examined using a process known as tandem mass spectrometry for the enzyme deficiencies that characterize the syndromes. Although the diseases are not curable, some are more treatable than others. In all cases, the key is to act quickly, if only to minimize the damage.

Infants with the syndromes tend to appear normal in the first months of life then begin to deteriorate.