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Early warning devised for rare disorders

Simple blood test could screen for enzyme diseases in newborns.

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A simple blood check that can detect some rare but devastating disorders is being trialled in New York state as a screening test for newborns.

The test, described at the American Chemical Society meeting in Atlanta, Georgia, on 28 March, is for progressive, genetic diseases known as 'lysosomal storage disorders'. These conditions can cause symptoms from gross enlargement of the liver to mental retardation. Some 40-50 of the disorders are known, and in total they affect 1 in 5,000 people. Gaucher disease, one of the more common afflictions, affects a little more than 1 in 1,000 Ashkenazi Jews.

The researchers, who are sponsored by the US National Institutes of Health and the Cambridge-based company Genzyme in Massachusetts say their screening method can now detect seven of these diseases. Genzyme, which sells treatments for three of the diseases, hopes to see the screen made a routine part of newborn testing.

Their first trial run in New York is checking for two diseases in a few thousand newborns in the state.



Early start

Lysosomes are small compartments within cells that help to recycle waste cellular material within the body. Enzymes within the lysosomes do this job by breaking up waste materials into smaller pieces. If one of these enzymes is not functioning, usually because of a problem in the gene that codes for it, then the breakdown stops and half-digested matter clogs up the cell with disastrous consequences.

Newborn babies with these diseases can be healthy, because there has been little time for this material to build up. Blood tests are available for cases where lysosomal diseases are suspected. But doctors are keen to find a way to diagnose the disorders before problems begin, so that treatment, if it exists for the specific condition, can start early.

"Once you get damage, there is no way back," says FrantiÅ¡ek Turecek, a mass spectrometry expert from the University of Washington in Seattle who presented the technique at the meeting. "Either you get going on treatment early, or you can die. And it is not a good death."

Twin peaks

Turecek, along with paediatrician and enzymologist colleagues, devised a test in which a newborn baby's enzymes are put through their paces1.

A tiny bit of blood taken from a routine heel-prick is treated so that the enzymes pop out of the cell membranes, then is presented with doses of the kinds of molecules the enzymes were born to cleave. The result is run through a mass spectrometer in tandem with a mixture of molecules of known sizes. A healthy baby's blood should produce a clean set of twin peaks, one for the enzyme product and one for a matching molecule of known similar size. If a twin peak is missing, the baby is probably lacking an enzyme.

The methodology isn't very complicated and should be quite cheap: the group estimates that a single machine should be able to process 85,000 screenings a year. This would be enough to cover the state of Washington, for example. A screening, which could look for up to 20 different enzymes at a time, should cost as little as five cents after the purchase of a suitable mass spectrometer, they say.

References

Lee Y., et al. Clin. Chem., 50. 1723 - 1724 (2004).