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Improving interpretation of newborn screening results

DOI: 10.1038/gim.2012.2

The use of pattern recognition software improves the quality of screening newborns for more than 40 inborn errors of metabolism diseases, according to a study published online this week in *Genetics in Medicine*. This software integrates all results generated as part of the screening panel normally administered to newborns, giving a single score that can be compared to known true positive cases. The authors suggest this approach reduces both false positive outcomes and false negative events.

In traditional laboratory medicine, blood samples from newborns are tested by tandem mass spectrometry and the results are reported to be "normal" or "abnormal" following comparison to cutoff

values set higher or lower than normal values. Without collaboration, comparison to actual cases with informative results may be limited.

Piero Rinaldo and his colleagues collaborated with 154 laboratories in 49 countries to populate a database, which included 767,464 results from 12,721 newborns affected with 60 conditions. First, the authors defined thousands of specific-condition disease ranges for more than 100 biomarkers, and then used the multivariate pattern recognition software to generate tools integrating clinically significant results into a single score. Rinaldo and colleagues suggest this software provides a broad and consistent differential diagnosis by determining what condition best fits a given profile, rather than which marker is individually abnormal.

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