Genetic analysis offers options for some developmental delay

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(HealthDay)—Genetic analysis can improve diagnosis and management

of intellectual developmental disorder and unexplained metabolic abnormalities, according to research published online May 25 in the *New England Journal of Medicine*.

Clara van Karnebeek, M.D., Ph.D. a pediatrician and biochemical geneticist at BC Children's Hospital and principal investigator with the Center for Molecular Medicine and Therapeutics at the University of British Columbia in Vancouver, Canada, and colleagues focused on 41 children with developmental delays tied to neurometabolic disorders. Using traditional urine and blood tests, the researchers screened the children for 90 known metabolic diseases tied to developmental delay, and found that based on those tests the children didn't qualify for any of the known illnesses. The researchers then performed whole-exome sequencing on samples obtained from the children.

The process led to a diagnosis in 68 percent of cases, and identification of 11 candidate genes newly implicated in neurometabolic disease. The team was also able to change treatment beyond genetic counseling in 44 percent.

The genetic analysis led to "better clinical practice and, most importantly, potential better outcomes for these children who, if they went undiagnosed, would often develop severe impairment and never have an opportunity to improve," van Karnebeek told *HealthDay*.

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