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Research leads to new molecular diagnostic testing

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Computational analysis of epigenetic patterns facilitates diagnosis of unknown hereditary disorders



A new sophisticated computational model, developed by scientists at Lawson Health Research Institute in Canada, is bringing an innovative method of diagnosing rare hereditary conditions.

Children with developmental delay represent a significant portion of referrals made to the Medical Genetics Program at London Health Sciences Centre (LHSC). The challenge is to determine which of the 25,000 genes in the body is responsible for the condition.

The latest in a series of research publications around the theme of epigenomic testing applied the new pattern recognition model to DNA samples of 965 patients with neurodevelopmental and congenital anomalies that did not have a definitive diagnosis despite extensive clinical genetic testing. Their DNA, acquired through blood samples, was examined using the new model, and dozens of new cases were resolved.

This research highlights the value of epigenomic testing in the routine assessment of neurodevelopmental and congenital disorders.

From this research, LHSC will be the first site in the world to offer this type of genetic testing.