Gene analysis helps identify basis of rare infant heart disorder

Écrit par Allen Press, Inc. Mercredi, 04 Janvier 2012 15:02 -

A study reported in the <u>current issue</u> of the journal *Pediatric and Developmental Pathology* compares cardiac tissue from 12 patients with HC and 12 age-matched controls. Researchers found

differences in gene expression

that

could indicate a predisposition for HC

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HC typically occurs in the first 2 years of life. It affects females three times more often than males, leading to the theory that it is an X chromosome-linked disorder with prenatal death is occurring in males. Eighty percent of the cases occur in whites; 15 percent in blacks; and 3 percent in Latin Americans. It is very rare in those of Asian heritage.

A worldwide registry of HC was started in 1999, attempting to identify the gene responsible for this fatal disorder. These data have shown a family tendency for the disease.

The gene analysis undertaken in the current study found decreased protein expression, or downregulation, in two sets of genes aligned sequentially along the genome. This offers several genes as candidates for the mutation, possibly predisposing individuals to HC.

The downregulation of a particular gene could result in reduced survival of cardiac myocytes, leading to cardiac failure during a baby's development. These candidate genes now will offer a starting point for further research for inherited patterns of mutation. Utilizing the HC registry, researchers will seek to confirm the findings of this study and to collect and analyze blood from parents and siblings of HC patients in an effort to further expose the inheritance pattern.

Full text of the article, " <u>Identification of Candidate Genes for Histiocytoid Cardiomyopathy (HC)</u> <u>Using Whole Genome Expression Analysis: Analyzing Material from the HC Registry,</u>

Pediatric and Developmental Pathology , Vol. 14, No. 5, 2011, is available at http://www.pedpath.org/doi/full/10.2350/10-05-0826-OA.1

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About Pediatric and Developmental Pathology

Pediatric and Developmental Pathology – Researchers are closing in on a rare genetic disorder causing a heart condition in infants. Histiocytoid cardiomyopathy (HC) often causes sudden death before a child reaches 2 years of age. Gene analysis is helping to narrow the many theories surrounding the genetic basis of HC.