Findings indicate digestive disorder in infants may be genetic

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In a study that includes nearly 2 million children born in Denmark, researchers have found that there is a higher rate of occurrence of the digestive tract disorder pyloric stenosis among twins and siblings, suggesting that this is a genetic and inherited disorder, according to the report in the June 16 issue of *JAMA*.

Pyloric stenosis is a severe and potentially fatal condition in which apparently healthy infants, typically 2 to 8 weeks old, develop an inability to pass food from the stomach into the duodenum (part of the small intestine). "Today pyloric stenosis is the most common condition requiring surgery in the first months of life. Among white individuals, pyloric stenosis is relatively common, with an incidence of 1.5 to 3 per 1000 live births," the authors write. "Case reports have suggested familial aggregation [the occurrence of more cases of a given disorder in close relatives of a person with the disorder than in control families], but to what extent this is caused by common environment or inheritance is unknown."

Camilla Krogh, M.D., of the Statens Serum Institut, Copenhagen, Denmark, and colleagues examined the familial aggregation of pyloric stenosis from monozygotic twins (derived from a single fertilized egg) to fourth-generation relatives in the population of Denmark. The population-based cohort study included 1,999,738 children born in Denmark between 1977 and 2008 and followed up for the first year of life, during which 3,362 children had surgery for pyloric stenosis.

Among the findings of the researchers, 46 percent of children with an affected monozygotic twin were diagnosed as having pyloric stenosis. In dizygotic twins (derived from two separately fertilized eggs), 7.7 percent with an affected twin had pyloric stenosis.

"This nationwide study documented strong familial aggregation of pyloric stenosis, with a nearly 200-fold increase among monozygotic twins and 20-fold increase among siblings. Familial aggregation of pyloric stenosis was pronounced even in more distant relatives," the authors write.

"Our findings argue for a hereditary component of pyloric stenosis: (1) predominance in boys; (2) familial aggregation in first-, second-, and third-degree relatives; (3) high concordance rate in monozygotic twins; (4) similar degree of aggregation in dizygotic twins and siblings; (5) difference in risk for siblings vs. maternal half-siblings (as well as cousins vs. half-cousins); and (6) heritability of 87 percent."

"… with a heritability estimate of 87 percent, it seems that familial aggregation is primarily explained by shared genes that may affect responses to postnatal factors. The high rates for twins and siblings should be considered in counseling families with affected children," the researchers conclude.

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