News
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March 23, 1978

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******Fetal diagnosis first of a kind.

DALLAS--Two scientists at The University of Texas Southwestern Medical School here have performed the first-of-its-kind diagnosis of an hereditary blood fat disease in the fetus of a woman in Belgium.

In an article appearing in the current issue of the British medical journal LANCET, Drs. Michael S. Brown and Joseph L. Goldstein join associates in Dallas and at the University of Leuven in Belgium, in describing tests which showed that the 15-week-old fetus had a severe cholesterol problem.

It was, in fact, the same hereditary disorder which had caused the death of the woman's first child, a boy, at age eight of a heart attack.

The tests involved Belgian doctors drawing amniotic fluid from the sac containing the fetus and mailing the samples to Drs. Goldstein and Brown in Dallas. Fetal cells in the fluid were cultured at Southwestern and then tested for their ability to bind with a type of blood fat known as low density lipoprotein.

This was a crucial element in determining whether the fetus had inherited a disorder known as familial hypercholesterolemia.

Unfortunately, the tests showed the defect had, indeed, been passed on to the fetus and that it could expect only a short, painful existence. The mother decided on an abortion which was performed at 20 weeks. Dr. Petri T. Kovanen of the Southwestern group went to Belgium to return post mortem samples and these have indicated the fetal cells contained approximately nine times the normal amount of cholesterol.

Drs. Brown and Goldstein have received international attention for their work in specifically identifying the genetic defect and the biochemical malfunctions which occur in familial hypercholesterolemia.

In the LANCET article, co-authored with Kamiel Vandenberghe, Jean Pierre Fryns, Roger Eeckels, Herman van den Berghe and Jean Jacques Cassiman of the University of Leuven; and Dr. Kovanen, it was related that the family--described only as "L"--was investigated when the only child--a six-year-old boy--was found to have the FH disease.

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The Dallas scientists' work has shown that there are two forms of the disease--depending on whether the child inherits the faulty dominant genes from one or both of the parents. With defective genes from both parents--the homozygous form--the victim's body cells do not have a certain type of chemical receptor located in various pit-like areas over the cell surface.

In normal persons, the receptors, which coat the walls of small pits in the cell's surface, bind with a form of blood fat known as low density lipoprotein.

Once the chemical bond is acheived, the pits and contents envaginate and move inside the cell where they engage in further chemical reactions which restrain the production of cholesterol so that it stays within normal limits.

But when a genetic defect produces body cells with very few or even no receptors or pits, then cholesterol manufacture goes unchecked.

The authors say that a number of members of the Belgian family suffer from elevated cholesterol levels. The father seems to have a milder form of the disease. The paternal grandfather died at 47 with a heart attack. The mother had elevated cholesterol as did two brothers. Her mother died of a non-specified heart condition at 54.

It is possible, say geneticists, that the Belgian couple could still have a normal child. Chances are 50-50 with this kind of "dominant" genetic disease.

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