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## Science Tracks Down Bearers Of Deadly Cystic Fibrosis

CYSTIC FIBROSIS is the most common of the serious genetic diseases to afflict the children of America. Among Caucasians, about one in 25 persons is a silent carrier of the gene for the disease. Potential mates are rarely aware of their genetic potential.

Hence, by the operation of simple laws of probability, about one in 625 (25 times 25) marriages stand in risk of having the disease in their children. For those marriages, the odds that a given child will be affected are one in four. Half their children will again be unaffected carriers; the remaining fourth will be neither affected nor potential transmitters of the lethal gene.

ALTHOUGH cystic fibrosis is now recognized as a severe pediatric problem, it was not even identified as a distinct disease until 1938. This can be put down to the fact that most of its victims died in their first year of life from secondary complications—lung infections, diarrhea and failure to thrive.

Even then, it was long regarded as a disease of the pancreas, but the lesions found in this organ are now understood to be secondary to biochemical changes involving secretory glands throughout the body. Thus, the difficulties in breathing are correlated with obstructions that develop in the small air passages leading to the lung and probably with changes in the biochemistry of a mucous substances in the secretions.

Until recently, the disease was invariably lethal at an early age. With the help of antibiotics to control infections and supportive measures for the nutrition and salt balance of its victims, affected children can now be helped through the perils of infancy. Their parents must be waiting in agony for further research advances that can deal with the disease at a more fundamental level and give them a better hold on life.

ONE OF THE most troublesome obstacles has been the difficulty in identifying the silent carriers of the disease, the 4 per cent of the population who carry one normal, one abnormal form of the corresponding gene. In a communication to the British medical publication *Lancet*, Drs. B. S. Danes and A. G. Bearn of Cornell University Medical College in New York have now reported a solution to this problem by a microscopic study of skin fibroblast cells.

Minutes fragments of skin were first placed into cell culture in the laboratory and then submitted to special staining techniques. In contrast to normal controls, cells from patients with cystic fibrosis and from their parents invariably gave positive staining reactions.

Besides giving the first reliable method of identifying carriers, this work also reinforces the concept that the abnormal gene influences cells throughout the body to give the special staining reaction. These cells can now be studied directly in the laboratory, for a direct attack on the fundamental biochemical lesion.

THE RECOGNITION of the carriers has a certain value in warning (or reassuring) potential parents about their prospects of transmitting the disease. This information could be used in two ways, depending on contemporary social judgments about rational forethought in family plan-

ning. A couple who are both carriers might decide not to have children when faced with a one-in-four risk of catastrophe. Alternatively, they might decide to attempt to have a normal child, with the help of prenatal diagnosis of the disease and a therapeutic abortion should they lose the gamble.

Another sideline of extraordinary interest is how a genetic disease is maintained in the population when the abnormal genes should be extinguished by the early death of the affected children. The most plausible explanation is that the carriers have had some biological advantage, an issue that can now be precisely studied for the first time.

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