WE'RE WINNING THE BATTLE AGAINST BIRTH DEFECTS

A heartening progress report on the never-ending struggle to prevent babies from being malformed at birth—and on brilliant new methods of correcting the deformities that do occur

By Albert Q. Maisel

"You have a beautiful, healthy, normal baby." Every year nearly four million newly delivered American mothers hear these reassuring words from their doctors. But 250,000 others learn, with fear and heartbreak, that their newborn child has a defective heart, or a clubfoot, a harelip, a damaged brain or some other major illness or deformity.

Until recent years, scientists and physicians could offer parents scarcely a hope of preventing such disasters. They could deride, for instance, the old superstition that viewed malformations as punishment for the parents' transgressions, but in its place they could offer only the "explanation" of heredity. Since we are born with our genes, and pass them on, they said, there seemed to be nothing to prevent one unlucky child out of every 16 from being condemned to the misery of inborn illness or deformity.

Here and there, however, a few indomitable investigators refused to accept so dismal a view. Some birth defects, they proved, were caused not by heredity but by *preventable injuries* to the infant in the womb. Other malformations, they demonstrated, could be corrected by early diagnosis and prompt treatment.

The first clear picture of just how much has been done, and how much

remains to be done, before all 644 types of birth defects can be conquered, emerged last summer in London. There, at the first International Conference on Congenital Malformations, sponsored by the United States' National Foundation, nearly 500 leading geneticists, biologists, physicians and surgeons from 25 nations exchanged knowledge and launched a world-wide research war against birth defects.

The most spectacular victories have been scored by surgeons. Until only 20 years ago, doctors were powerless to help the 35,000 children born each year with congenital defects of the heart or its major arteries. Then Dr. Robert E. Gross, of Children's Hospital, Boston, devised an unprecedented operation. On an infant facing certain death, he tied off and severed a duct that had failed to close at birth and was shunting blood in a futile circle between heart and lungs instead of into the rest of the body. That first operation was followed by hundreds of others equally successful.

But Dr. Gross had worked only on blood vessels *near* the heart. Ten years were to pass before surgeons dared attempt repairs to an infant's heart structure. Then, in the early 1950's the heart-lung machine was perfected. By taking over the functions of the baby's heart and lungs, this device makes it possible for surgeons to perform corrective operations in the heart's interior. Today most types of major heart defects can be corrected by surgery, and thousands of children have been saved from otherwise inevitable lingering deaths.

A common birth defect is the disfiguring "port-wine stain" on a baby's face. Physicians have tried to remove these ugly blotches with radium, X rays, dry ice, bleaches and strong chemicals—frequently with unfortunate results. Now, thanks to such pioneering work as that of Dr. Herbert Conway, chief of plastic surgery at New York Hospital, a normal appearance can often be achieved. Dr. Conway has developed a technique by which flesh colors, blended to match a child's complexion, are tattooed into the lower layers of the skin overlying the stain.

A dozen years ago doctors hesitated to operate on babies born with harelips or cleft palates until they were four or five years old. While waiting for surgery, many of these tykes suffered infections that caused at least partial loss of hearing. And the eventual operation often left them with severe speech defects. Nowadays plastic surgeons repair harelips a few hours after birth and cleft palates at about 18 months. Speech therapy is usually not required.

Similarly, children born with clubfeet were often left untreated until they approached school age, in the unfounded hope that they might outgrow their deformity. When correction was then attempted, it was often followed by a gradual crippling relapse. Today treatment starts while bones and tendons are still pliable. Frequently, with no more than adhesive tape or a plaster slipper, full correction is obtained within six to eight weeks.

One of the most devastating of all congenital malformations is hydrocephalus, or water on the brain. In this condition, the normal drainage of fluid from the brain is blocked, usually causing progressive enlargement of the head, mental retardation, convulsions. Until four years ago, many of these infants died early; most of those who survived spent their lives in institutions for the mentally defective.

In 1956, however, at the Children's Hospital in Philadelphia, Dr. Eugene Spitz, aided by engineer John Holter, devised a new operation using a unique tube and valve to lead the excess fluid into the blood stream.* Performed a few days after birth, this procedure can arrest the enlargement of the head and prevent mental retardation. Among 750 infants so treated at Children's Hospital, the mortality rate has been barely three percent. In all, more than 10,000 operations using the Spitz-Holter valve have been successfully performed.

Quite as awe-inspiring as these surgical advances are the recent achievements of biochemists and physicians in correcting and *preventing* a number of the more than 300 inborn defects of metabolism that often cause early death or mental and physical impairment. The livers of babies born with phenylketonuria, for example, fail to produce the essential enzyme that breaks down the amino acid, phenylalanine. As a result, abnormal chemicals accumulate in the blood stream and begin to affect mental development. Until recently, 98 percent of the infant victims of this malady became severely retarded, largely because doctors could not identify it early enough. Today many hospitals use a simple urine test on all newborn infants to spot this subtle birth defect. Then, victims can be placed on phenylalanine-free diets which prevent the onset of mental retardation.*

New urine tests and dietary treatments are working an equal miracle for many victims of galactosemia, an inability to digest milk sugars, which formerly caused either early death or severe mental deficiency, cataracts and liver damage. Today a special diet, free from milk and lactose, permits these infants to develop normally. Even where limited damage has already occurred, the lactose-free diet has resulted in the resorption of cataracts, resumption of mental growth and improvement of liver function.

Rapid progress is being made, too, in the prevention of congenital deformities caused by prenatal environment. For many years scientists believed that heredity caused almost

*See "A Father's Last-Chance Invention Saves His Son," The Reader's Digest, January '57.

*See "Forgotten Children No Longer," The Reader's Digest, September '60.

all human birth defects. Then, in 1941, an Australian eye specialist, Dr. Norman McAlister Gregg, noted that an unusually large number of babies were being brought to his office suffering from cataracts and other serious eye ailments. Many, in addition, had heart malformations or were deaf. Investigating, Dr. Gregg found that each of the mothers, when less than three months pregnant, had been among the victims of a German measles epidemic. Their babies had been injured in the womb by the German measles virus.

Research has since shown that most women who have German measles during pregnancy can still bear normal, healthy children. Abnormalities occur only if the infection takes place in the early months of pregnancy, when vital organs are being formed; even then, they occur in only 5 to 20 percent of the births.

This first link between birth defects and a virus infection set scientists upon a renewed search for other environmental causes of birth deformities. Over the last few years several more have been identified.

For example, radiation studies with animals had lent support to the theory that diagnostic X rays during pregnancy can cause infant abnormalities. The harmful effects of excessive radiation were clearly shown by studies of Japanese atom-bomb victims. Of 190 children whose mothers were pregnant at the time of the Hiroshima explosion, 33 were born with abnormally small heads and 16 of these developed mental deficiencies. Hence, today, physicians shield the patient's abdominal area, if possible, when X rays are necessary during pregnancy.

A mounting mass of evidence strongly supports the case against still other environmental factors. In laboratory animals, for example, diets markedly deficient in vitamins or other essential nutrients produce a sharp rise in malformations. Though a direct link between human maternal dietary deficiencies and the occurrence of birth defects has not been proved, recent studies in Great Britain show that some severe defects are more common among the very poor—and presumably the most poorly nourished—than among other groups.

Various virus infections in pregnant women—including polio, hepatitis, measles, mumps and influenza—have been indicted by some researchers and exonerated by others as causes of malformed babies. Even if the indictments are eventually confirmed, medical research will still have to determine the culprit—*i.e.* the virus itself, the medications used or some other factor. In the case of flu, the one rational precaution is the use of influenza vaccine for all pregnant women.

Cerebral palsy, epilepsy and a number of mental abnormalities have long been blamed on both heredity and injuries during delivery. Many researchers today believe, however, that deficiencies of the oxygen supply to the fetus account

for a substantial number of these birth defects. This is why some doctors urge pregnant women to avoid abrupt changes to high altitudes, and to postpone, if possible, operations involving general anesthesia.

Only a small percentage of all birth defects can today be ascribed to either environmental causes or genetic causes alone. Such experts as Dr. F. Clarke Fraser, of McGill University, and Prof. Hans Nachtsheim, of the Free University of Berlin, are convinced that the overwhelming majority result from the complex interaction of genetic predisposition *and* environmental factors. Both environment *and* heredity must be studied in determining how to prevent malformations.

Less than two years ago, Drs. Jerome Lejeune and Raymond Turpin, of the Institut de Progenèse Faculté de Médecine, Paris, demonstrated that Mongolian idiocy—which afflicts more than 6500 infants born each year in the United States alone—is a genetic defect. Victims are all born with 47 chromosomes in their cells instead of the normal 46. Since then, at least 25 other types of congenital defects have been linked to chromosome abnormalities.

This is exciting news. For, as Dr. L. S. Penrose, professor of eugenics at the University of London, has pointed out, genetic aberrations may be the result of controllable influences: "hormonal or other chemical factors which affect the germ cells in ways hardly yet seriously considered." And Nobel Prize-winner Edward L. Tatum, of the Rockefeller Institute, even expresses the hope that some day medical men may be able to replace defective genes with normally functioning genetic material.

When that day arrives, the way will be open for the elimination of *all* birth defects and their attendant misery.

Sign Language

A SIGN in a San Francisco saloon announcing free coffee for all customers at closing time is headed: "Safety Belts."

—Herb Caen in San Francisco *Examiner*

ON a large barn near Lancaster, Wis.: "The Weittenhiller Farm. Uncle Sam, Operator."

—AP

OUTSIDE a veterinarian's office: "Hospital Zone—No Barking."

—E. K. H. in *Journal of the American Medical Association*

ON a fence in New York's Greenwich Village: "Karen loves money."

—Contributed by Dorothea Stilphen

IN an airline ticket office: "Why Not Jet Away From It All?"

—Cedric Adams in Minneapolis *Star*