



*Recent advances in the  
prevention, detection,  
and correction of birth  
defects are now benefiting  
thousands of children  
who once would have been  
denied a normal childhood*

## **They're solving the mysteries of birth defects**

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**M**ARK, it's time to put your tricycle away and come in for lunch." . . . "Linda will be here soon—unless she's playing with friends on the way home from school." . . . "Jerry is doing beautifully in kindergarten. He's so proud of his workbook."

These phrases might be any mother's, any time, anywhere. They're ordinary, typical, fondly maternal.

What makes them quotable is that the children involved are able to enjoy the give-and-take of everyday living because they happened to come into the world within the past decade.

Mark and Linda and Jerry were born with severe birth defects. They represent more than a quarter of a million children born each year in the United States with abnormalities that can limit or deny them the simple, pleasurable activities of childhood. But today Mark and Linda and Jerry—and thousands of youngsters like them—are benefiting from the marvelous advances doctors are making in the treatment and prevention of congenital defects.

I'm happy to say that the dread word "hopeless" is fast

disappearing from the vocabulary of specialists in the field of birth defects. Within the last 10 years or so, pediatricians, surgeons, therapists, and other specialists have gained vast knowledge about the correction and cure of abnormalities that were once considered beyond help.

We've learned how to detect and treat many "hidden defects"—internal or metabolic disorders not apparent at birth—before their deadly effects have a chance to cause irreparable damage.

Perhaps most important of all, we've learned that many potential birth defects can be warded off during pregnancy, or even before, by means of simple, common-sense precautions.

"Enjoy him while you can; there's nothing more we can do" are despairing words that were often all a doctor could say to heartbroken parents. Now research findings and clinical experience constantly make the need for that grim sentence *much* less common.

#### HELP FOR MARK'S HYDROCEPHALUS

MEDICINE'S recent great progress is dramatized by the help that was available for Mark when he was born four years ago with *hydrocephalus*, or water on the brain. Before birth something went wrong in his body's ability to circulate and absorb cerebrospinal fluid in the normal way. Excess fluid backed up under his skull. The resulting abnormal pressure usually causes gross enlargement of the head and can lead to blindness, mental deficiency, or even death.

Today Mark, as he rides his tricycle, pays no attention to the thin plastic tube and tiny pump contrivance lying just under the skin above and behind his right ear. He's too busy with the important matters of growing up and enjoying a normal, active life to think about the surgical miracle that made it possible.

#### A LIFESAVING "DETOUR"

THAT miracle is known as "shunt surgery," because the tube forms an artificial bypass, or shunt, around the defective pathway and permits excess fluid to drain harmlessly into the blood stream. This procedure was developed only a few years ago. Progress in the technique has been so rapid that Dr. Eugene B. Spitz, pioneer in its development in the United States, now calls it "one of the safer things we do today in pediatric neurosurgery, with operative mortality less than in appendicitis." (Dr. Spitz is neurosurgeon-in-chief at Children's Hospital, Philadelphia.)

About 20,000 of these dramatic operations have been successfully performed on babies and youngsters throughout the world. This is a far cry from the time when parents of a hydrocephalic child could only wait and hope that the condition would correct itself.

"If our aim is to preserve normal men- [To page 88]



Dr. Virginia Apgar, a noted specialist in the problems of newborn infants, is the creator of the "Apgar score," a clinical evaluation made within 60 seconds after birth to determine normality. Among the honors held by Dr. Apgar is the New York Infirmary's Elizabeth Blackwell Citation (1960) for outstanding service to medicine by a woman.

# They're solving the mysteries of birth defects

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tality," says Dr. Spitz, "I think the vast majority of hydrocephalic infants and children will need some variety of decompressive shunt surgery. This business of waiting to see if the child's head will stop growing usually results in permanent mental defects. Time is of utmost importance in the preservation of brain function."

## EARLY DETECTION IS A MUST

EARLY detection and treatment are an important part of the "new" medical attitude toward birth defects. Doctors now realize that delay often results in permanent disability, mental retardation, or death.

Early detection has always been easy in some of the more familiar types of birth defects. Clubfoot, missing or underdeveloped extremities, open spine, and similar abnormalities are usually immediately apparent at delivery. But until just a few years ago early measures to correct these defects were the exception rather than the rule. Today medical authorities realize that treatment must begin as early as possible after birth to assure maximum correction. Delay often compounds the disability.

With modern surgical and anesthetic techniques, surgery on newborn babies is not the risk it once was. *Harelip* may be repaired in the first few weeks of life instead of several years later. Not only does this early treatment assure better cosmetic results, but it also often prevents many feeding and emotional problems that

would otherwise arise as the child matures.

Complex surgery is involved in the repair of *cleft palate*, and today's operations are much more effective than they were 25 years ago. Early surgery permits the most effective follow-up care by the orthodontists, speech therapists, and other specialists who play an important role in the full correction of the handicap.

Babies born with *clubfoot* 20 or more years ago were, more often than not, left untreated in their early months in the hope that spontaneous improvement would occur. This forlorn hope was seldom realized, and such children faced life with bone and muscle deformities that grew worse as time passed.

Pediatricians and orthopedists now know that there is no such thing as spontaneous improvement of true clubfoot—rather, that growth and weight-bearing increase the problem. So, as soon as possible, they begin forcing clubfeet into a normal position by stretching the shortened muscles and ligaments and aligning the bones into a more natural relationship.

In the large majority of cases, through the use of casts, braces, and corrective shoes, the condition may be corrected before the child is ready to walk. Occasionally surgery also is needed.

## CORRECTING HEART DEFECTS

SOME of the most dramatic advances in successful correction of birth defects have been made in the field of heart surgery. There are now techniques, literally unknown before 1939, for surgical repair of some 16 of the 19 known congenital defects of the heart and so-called great blood vessels. With protective antibiotics and the almost magical heart-lung machine to maintain circulation during delicate surgery, doctors are now able to operate on all the chambers of the heart. They can repair holes, open or close defective valves, even construct new ducts to do the work of blocked ones—often, in the process, literally changing "blue" babies to a healthy pink.

Of the 30,000 babies born each year with abnormalities of the heart, the great majority are now saved from early death or lingering invalidism because doctors know what to look for and—far more vital—what to do when a defect is found.

Many potentially fatal malformations of other body systems, especially in the alimentary tract and the urinary tract, are now being detected and corrected before they can cause irreparable harm.

For example, sometimes a baby is born with the esophagus—the channel from mouth to stomach—ending in a blind pouch instead of extending to the stomach. The child obviously cannot swallow nourishment, and death is inevitable unless the defect is corrected. Within the baby's first 24 hours, surgeons now safely open the tiny chest, connect the esophagus to the stomach, and feed the baby artificially until the connecting stitches heal; within weeks the infant thrives on a regular formula.

## HELP FOR LINDA'S OPEN SPINE

SPECTACULAR headway has also been made in techniques of rehabilitation, called

"habilitation" when disability is present from birth. Linda, now seven, was born with *spina bifida*, or an open spine. Her backbone failed to close properly before she was born, and the spinal cord and nerves leading to the lower body and legs were damaged so severely that it seemed unlikely that Linda would ever be able to walk.

Yet it has been more than a year since Linda's mother "called up everybody I knew—and probably some I didn't know—to tell them about Linda's first steps without crutches or braces!"

Linda still wears her braces for school and roughhouse play—doctor's orders until her legs are a good bit stronger. But she can use her own two feet, and she treasures her independence.

Her success story—one that these days could be told of growing numbers of children—reflects advances in many fields of medicine. Because *spina bifida* is one of the most complex of all structural birth defects, its treatment involves the services of neurosurgeons, orthopedists, urologists, physiatrists, physical therapists, and many other specialists.

The extent to which an open spine can be corrected surgically depends largely on the degree of spinal-cord and nerve damage. If this is severe, much of the child's success in learning to use his legs and other affected portions of his body rests with habilitation experts. Every year they are better able to teach the child how to walk with braces, how to make movements of the upper body compensate for disabled lower extremities.

## ADVANCES IN ARTIFICIAL LIMBS

HOW far habilitation techniques have leaped ahead in recent years is illustrated by progress reports on thalidomide babies born in tragic numbers in Europe during the last year or so. Long before thalidomide and its devastating effects came to public attention, there were babies born with missing or underdeveloped arms and legs—not in such appalling numbers as those caused by thalidomide, but there were enough of them to goad medical scientists to search for ways to replace missing members.

One of the most striking advances is the growing conviction among habilitation specialists that early fitting with artificial limbs (called prostheses) is the key to successful adjustment and use. Nowadays passive "baby mitts"—artificial arms without moving parts—may be fitted to an infant still in the crib, so that from early awareness they will seem to have been part of him.

When he is old enough to manipulate more advanced mechanical prostheses, he is much further along the road to self-sufficiency than similar children who, up to a few years ago, weren't introduced to artificial aids until they were several years old.

Another striking development is pneumatic prostheses—artificial limbs powered by carbon dioxide. In response to a slight pressure from a body muscle they produce a much more refined and controlled motion than earlier devices. Invented a little more than 10 years ago, these pneumatic limbs are being perfected rapidly. Other researchers are trying to develop transistorized devices that may even more closely

approximate the workings of natural arms and hands.

#### HELP FOR JERRY'S PKU

ALONG with advances in surgical correction and rehabilitation techniques, new knowledge of "hidden defects" is making dramatic progress.

"We were convinced that our son was the most perfect newborn in the nursery," Jerry's mother recalls.

To all outward appearances he *was* perfect—healthy, active, off to a good start in life. Only chance alerted his doctor and parents in time to keep him that way.

Jerry was born at a time when the well-baby clinic in his home town was conducting studies of a test of *phenylketonuria* (PKU), a defect in the body chemistry that prevents assimilation of an amino acid (phenylalanine), found in most protein foods. When a child has PKU, the acid is not used as nature intended. Instead it backs up and eventually finds its way to the brain where it causes irreversible damage.

When a certain chemical was put on Jerry's wet diaper, a green ring appeared around the edges of the urine stain. This showed Jerry had a positive tendency toward PKU. He was started on a special diet low in phenylalanine to prevent the build-up of the deadly excess in his system.

Simply by staying on this diet, Jerry has been spared a life of mental deficiency that, only a few years ago, would have been unavoidable.

Effective as the diaper test was in Jerry's case, unfortunately it was not valid until an infant was several weeks old. By that time, such a baby had left the hospital and might or might not be seen again in time to detect PKU before it began to exact its toll.

However, in the past two years Dr. Robert Guthrie, a National Foundation—March of Dimes grantee at the State University of New York in Buffalo, has developed a blood test for this disease. PKU shows up in the blood much earlier than it does in urine. With the Guthrie test, involving a drop of blood from a newborn's heel, PKU babies can be diagnosed and their protective diet prescribed before they ever leave the hospital.

*Galactosemia* is a defect of body chemistry that prevents normal tolerance of milk sugars; if unrecognized, it can result in mental retardation, cirrhosis of the liver, and blindness. Only in recent years has there been a diagnostic test for galactosemia; when it is detected within the first month of life, before appreciable damage has been done, the immediate institution of a milk-free diet usually provides complete protection.

The conquest of mental retardation and other neurological damage caused by *Rh blood factor incompatibility* is taken for granted these days; the first breakthrough in understanding its cause came in 1941. Today these complications are completely preventable when the condition is known before or early in pregnancy and can be carefully watched by obstetrician and pediatrician. If necessary the infant is given an exchange blood transfusion right after birth.

Another hidden defect, *deficiency of*  
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*thyroid hormone*, has been recognized for several years as the cause of cretinism, typified by stunted growth and severe mental deficiency. Now researchers have shown that this condition can be prevented by treating the mother during pregnancy or by giving the thyroid hormone to babies who are born with a defective thyroid gland.

Doctors and medical scientists all over the country are engrossed in the search for defects that have a deadly delayed action. Dr. Sydney Gellis, acting dean of medicine at Boston University, is a crusader for recognition of clues that may indicate a need for extensive examination of a newborn baby before he is considered free of problems.

For example, the normal umbilical cord has three blood vessels, easily seen by the naked eye. Dr. Gellis has observed that when there are only two blood vessels in the umbilical cord of an otherwise apparently normal infant, "the absence of the third suggests that there may be a major defect in the heart, kidneys, gastrointestinal tract, or central nervous system."

The clue in itself is not conclusive—but it does indicate the need for a more thorough examination than might ordinarily be made. If the investigation uncovers an unsuspected abnormality, corrective treatment can be started with the greatest possibility of success.

Recognition of clues like these has mushroomed in the last few years. There are signs that soon many more will be discovered. For example, a clue was just uncovered by a research team in New York City—a test that detects an inborn inability of the body to handle the copper in oysters, chocolate, nuts, mushrooms, and some other foods.

Known as *Wilson's disease*, the defect permits copper to build up in the system. Like the build-up of phenylalanine in PKU, the unassimilated copper of Wilson's disease eventually is associated with severe liver and brain damage. Unlike PKU, which invariably takes its toll in infancy and early childhood, Wilson's disease may lie dormant for months or years.

When it finally shows its symptoms—jaundice, mental retardation in young victims, and loss of muscular control and personality changes resembling schizophrenia in older ones—much of the damage has been done and the course of the disease can be slowed only by diet and medication. But with the new test, involving a single drop of blood, potential inheritors of a tendency to Wilson's disease can be treated before outward signs of the illness develop.

#### SUMMING UP

THESE achievements—in surgery to repair and correct, in diagnostic methods to detect and allay potential damage, in treatment and rehabilitation to modify conditions not wholly correctible—all provide more hope than ever before existed for the nearly 700 infants with significant birth defects born each day in the United States.

Doctors recommend that every potential mother observe the following simple, common-sense precautions, in order to help protect her baby from birth defects:

1. **DIET.** A mother's health affects her baby. Maternal health should be developed from early adolescence by a diet rich in protein, minerals, and vitamins. During pregnancy, do follow the diet prescribed by your doctor.

2. **DRUGS.** Drugs may harm both the mother and her unborn child. *Never* take drugs during pregnancy unless they are prescribed by your doctor.

3. **X RAYS.** X rays may injure your baby. Their danger is greatest in the earliest days of pregnancy. If X rays are absolutely necessary, your doctor can take proper precautions. If you know that you are pregnant, or suspect that you may be, be sure to tell your doctor.

4. **RH FACTOR.** Under certain conditions the Rh factor may cause serious illness in an unborn child; but effective treatment is often possible at birth. *Do* learn, before pregnancy, both your own and your husband's blood type. Discuss this with your doctor.

5. **DISEASES.** German measles in a mother in the first three months of pregnancy sometimes causes malformations of the baby; other viruses are being investigated as possible culprits. *Don't* knowingly expose yourself to infections during pregnancy, but do try to have your daughter exposed to German measles before she finishes high school.

6. **THE EARLY DAYS** and weeks of pregnancy are *even more important* than later weeks to your baby's development. Your doctor's counsel regarding pregnancy and the care of your unborn child is imperative. *Do* consult him as soon as you suspect you are pregnant, and preferably also *before*.

**T**O speed the acquisition of new knowledge and, as soon as possible, extend its benefits to children everywhere in the United States, The National Foundation—March of Dimes began in 1960 to establish a network of centers for the study and treatment of birth defects. Today there are nearly 50 such centers in teaching hospitals and medical centers throughout the country. As funds permit, more of these centers will be opened, for the demonstrated need has outstripped the facilities.

Not only do these centers provide care for the children who need it; they also form an effective communications network for the medical profession. When new techniques are proved effective, the word is quickly sent to all centers. Center staffs convene regularly to compare experience and draw from the others' knowledge.

Perhaps most important, each center *teaches*. Physicians and medical students, nurses, therapists, and other specialists on the center staffs and throughout the communities the centers serve—all are constantly learning what is newest and best in the care and treatment of birth defects.

While more can be done today than ever before to help victims of birth defects, the ultimate goal, of course, is to keep them from occurring the the first place.

To this end, thousands of research scientists—many with grants from the March of Dimes—are studying hereditary and environmental factors that affect the developing embryo. ##