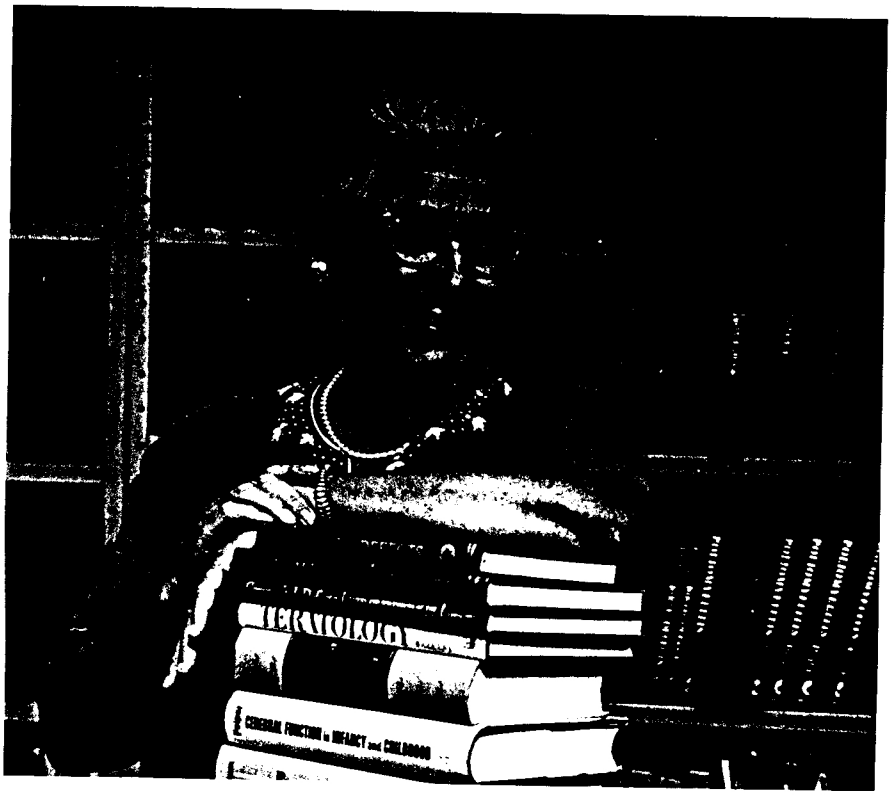

# Apgar on Birth Defects

**Virginia Apgar, M.D.**

For mothers, for fathers, for grandparents, indeed, for all members of every family on earth, there is no greater gift than that of a normal, healthy baby. Yet those of us who spend much of our time in hospitals or in the homes of the physically or mentally ill know only too well that every baby is not born healthy. Many seem condemned from birth to a life that some believe is worse than death.

I address this not to those who hang their heads and think, "How tragic!" when they see a child with multiple deformities, but to those who look such children squarely in the eye and ask, "What can I do to help?"

Some kinds of birth defects can be prevented. Many other conditions which we cannot yet prevent, we can treat. But before medical professionals try to prevent or treat these conditions, they should understand what birth defects are and what causes them.

Birth defects—those 1,000 or more disorders that can kill, cripple or retard—are mistakes in body formation or function that happen when a baby comes into being. They may be caused by heredity, by disease, or by something that happens to the fetus in the womb. Each year, in the United States alone, an estimated 250,000 babies —one almost every two minutes— come into the world seriously defective in some way. Some of these conditions are so slight that the are but minor inconveniences. Col or blindness, for example, usuall means that the individual cannc tell red from green. On the othe hand, the baby born with multipl defects involving many organs ma die almost at once. Between thes extremes there are many cond tions that disable bodies and minds, such as cleft lip, cleft palate, water on the brain, open spine, dwarfism, shortened or miss ing arms, diabetes, cystic fibrosis heart defects, mongolism, and oth- er forms of intellectual impairs ment.

In absolute numbers, more than half a million lives are claimed each year by birth defects. Some 60,000 deaths among children and adults annually can be traced to defective prenatal development. An estimated 500,000 fetuses a year are spontaneously aborted or stillborn because of abnormalities.

These appalling statistics explain why The National Foundation-March of Dimes, after organizing and directing the conquest of polio, turned its attention and its research resources to the fight against birth defects.

What are the causes of birth defects? Let us begin with a few points about some of the things that do *not* cause birth defects. There is a great deal of superstition about birth defects that dates back to antiquity and that even today has a surprising number of believers. For example, when a child was born with a hareli , its mother was often thought to have been frightened by a rabbit during pregnancy. Also, the arrival of a deformed child was sometimes interpreted as God's punishment. The guilt and shame felt by some parents of children born with birth defects has its roots in such ancient beliefs.

Neither the sudden jumping of rabbits nor the wrath of an angry deity causes birth defects. However, we can blame heredity for about 20 per cent of these dread conditions. PKU, as an exa ple, is an inborn error of metabolism

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*Dr. Apgar is vice president for medical affairs, The National Foundation-March of Dimes. She is the creator of the "Apgar Score" which is now standard procedure in many hospitals throughout the world. Made within 60 seconds after birth to determine a baby's overall condition, this vital clinical evaluation serves to alert physicians to the need for emergency procedures.*

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caused by interference in the environment of the developing fetus. Infection of the mother by German measles or use of the drug thalidomide during certain stages of pregnancy are environmental agents which may have disastrous effects on the fetus.

A combination of environmental and hereditary factors working together causes the great majority of birth defects. The mother younger than eighteen or older than forty runs a greater risk of giving birth to a baby with a defect than a woman in her twenties or thirties. The rate of mongolism, which is caused by a chromosomal abnormality, increases greatly among mothers over forty. Just as some individuals, because of inherited sensitivity, are allergic to environmental things like pollens and drugs, so the hereditary makeup of an unborn baby may predispose him to damage from outside influences that would not affect another baby of different genetic constitution.

Once we know something about the causes of birth defects, we can begin to see ways of preventing these conditions from occuring at all. Birth defects prevention should begin before marriage. Proper diet, maintaining normal weight and good hygiene, as every nurse knows, are conducive not only to the good health of parents, but also to that of their potential offspring.

The couple with a family history of hereditary abnormality may want to seek professional counseling on their risk of having a defective child. Certainly all people should realize that marrying a close relative is potentially dangerous because it increases the chances of bringing together potentially harmful genes. Also, tests have been developed for spotting hereditary carriers of a few disease traits such as galactosemia, sickle cell anemia, and the blood deficiency, agammaglobulinemia. It should be noted that there are tests for German measles immunity, and within the next few months we should have a vaccine to prevent the kind of tragedy this "mild" disease can cause when a woman has it in early pregnancy.

*This young victim of a condition which caused him to be born without arms is greeted by nurse Susan Bumpass and physical therapist Shirley C. Fisher at the Birth Defects Center associated with the University of North Carolina School of Medicine, Chapel Hill*

*Dr. Apgar in the delivery room* ►

Perhaps the greatest advancement in birth defects prevention is the Rh vaccine that is now on the market. Rh incompatibility is a condition which once killed or caused brain damage in 10,000 babies born in the United States each year. In recent years about 95 per cent of these babies were saved at birth by means of exchange transfusions. An estimated 40 per cent of stillborn Rh babies could be saved by new techniques of transfusion before birth. But, with the new gamma globulin injections given to Rh negative women within 72 hours after the birth of their first baby, complete prevention of the condition is now possible.

For about nine months, the most important thing an expectant mother has to think about is proper prenatal care. By prenatal care I mean, generally, what is or is not done during pregnancy. The first thing any woman should do when she even suspects she is pregnant is to see a doctor. He will advise her on some of the important aspects of prenatal care such as X-rays (low back and pelvic X-ray pictures are to be avoided); heavy smoking (studies show that when the mother smokes excessively, the baby may weigh less than normal, and if the baby is born prematurely, this difference could affect its survival); and drugs (an expectant mother should not take any drugs—including aspirin and pain killers—without a prescription from a doctor who knows she is pregnant).

When a baby is born with a birth defect there is a great deal medical professionals can do to help. We can control many congenital diseases, repair handicapping conditions, or in other ways restore children to health and a useful life. One of the keys to success is the earliest possible detection of the condition and proper treatment. Here are some examples of what can be done:

**Open Spine.** Little more than a decade ago, the child born with spina bifida, or open spine, was more often than not the victim of fatal infection. Today, surgeons can close and repair the spinal opening in all but the most severe cases. Many of these children who are partially paralyzed below the waist because of spinal nerve damage need the attention of various specialists to see them through years of learning to walk with braces, adapting to special procedures because of loss of bladder and bowel control, and adjusting mentally and physically to family, school, and community life. The results—victory over handicap—are as inspiring and gratifying as similar triumphs by victims of accidents, polio, and other crippling conditions.

**Hydrocephalus.** For the victims

*A nurse at the Birth Defects Center in the Boston City Hospital looks after a "preemie" in an Isolette. Many preemies are victims of some type of birth defect.*

of hydrocephalus, or water on the brain, special tube devices called shunts have been developed to drain excess fluid harmlessly from the head into other parts of the body. This life-saving device can often prevent retardation and abnormal skull expansion.

**Clubfoot.** Babies born with clubfoot twenty or more years ago were, more often than not, left untreated in their early months in hope that spontaneous improvement would occur. Pediatricians and orthopedists now know that the idea of any spontaneous improvement in true clubfoot is wrong—rather, growth and increased weight increase the problem. Today, doctors treat clubfoot immediately by forcing the feet into a normal position thus stretching the shortened muscles and ligaments and aligning the bones into a more natural relationship. In the large majority of cases, casts, braces, and corrective shoes will correct the condition before the child is ready to walk. Occasionally, surgery is also needed.

**Congenital heart defects.** It is possible to correct many congenital heart defects so that children who would otherwise be condemned to invalidism and early death can grow up to lead normal lives.

**Incomplete esophagus, diaphragmatic hernia.** These and other once-fatal malformations of internal organs may now be repaired within hours after birth by pediatric surgeons.

**Genito-urinary defects.** Surgeons have been making great strides in developing techniques for treating abnormalities in kidneys, ureters, bladders, and the whole genito-urinary system.

Other defects can be treated by special diets to either remove a food substance the body cannot handle or to provide extra amounts of a substance the body needs. Such diets do not cure, but they can prevent damage from occurring. Even children with missing or imperfect arms or legs can be helped. Many of these children are now being fitted with artificial limbs almost as soon as they are ready to walk or grasp.

A decade ago many of these conditions seemed hopeless; today improved diagnostic methods and newer medical and surgical techniques have revolutionized the outlook for babies born with birth defects. A great deal of research is being done today on basic causes of these complex conditions. Much of it is being conducted with government funds; some is supported by groups devoted to one or another kind of defects.

The National Foundation-March of Dimes finances a broad, nationwide program of research into the causes of birth defects with an eye toward both prevention and better treatment. Scientists are exploring the nature of the hereditary material, DNA. They are studying cells, chromosomes, body chemistry, body systems, and the numerous problems of diagnosis and treatment of specific conditions.

At the same time it is expanding its nationwide network of Birth Defects Centers, which now numbers more than 100, to provide what we term "the team concept" of treatment for the "total child."

When a baby is born with a birth defect, his parents need almost immediate advice, reassurance, and perhaps financial or other assistance. The baby may need the kind of care that only a number of specialists can provide. This might involve taking him, say, from pediatrician to neurologist to neurosurgeon to orthopedist. It might mean referral from physician to physician, even from town to town.

A Birth Defect Treatment C

*(Above) Arthrogryposis is a birth defect that twists and stiffens the arms and legs. For 3 year-old victim Laura Wagoner, walking does not come easily. Here she is helped at the Birth Defects Center associated with the University of North Carolina School of Medicine, Chapel Hill, by Dr. Loren G. MacKinney.*

*(Right) Joan Rutten, 6, Falls Church, Va., is seriously crippled by multiple birth defects. Nurse Mary A. Jackson removes body corset and leg braces prior to check-up examination at Children's Hospital of the District of Columbia.*

ter provides complete diagnostic services and the best of care in a single setting. Perhaps as many as 16 specialists will examine the child, evaluate his problems, and plan what can be done about them. This is the team concept of medical care.

When we speak of treating the "total child," we mean we are interested in more than the medical aspects of his problem. There are medical social workers on the center teams to help the parents whose problems with a birth defects child do not end when they leave the clinic. It should be noted, also, that these Birth Defects Centers are associated with medical schools or teaching hospitals. Teaching medical students and other professionals to care for birth defects victims is thus an integral part of the center program.

With the expansion of facilities, the blossoming of research, the improvement of medical care, and the awakening of interest in the American people, the victim of birth defects can hope to lead a relatively normal and useful life and certainly a life that is well worth living. Moreover, medical science is on the threshold of new discoveries that will lead to the prevention of many of these dread conditions. Little more than a decade ago there were relatively few professionals or private citizens who had any hope for the victim of birth defects. Today there is more than hope—there is the joy and triumph of progress.