The cookies on Karen's plate include a scientific compound responsible for her progress.

A New Life for Karen

This little girl's victory over a mentally crippling disease spells hope for millions of other children

By SHIRLEY SIROTA ROSENBERG

Playing with her friends.

Karen spins a record for her dad.

Laughing with her parents.

Reading to her mother.

EIGHT-YEAR-OLD Karen Lee Egerter of Lanham, Md., has a pixie haircut, shining blue eyes, and a nose that tilts just the way a little girl's should, only better.

She reads, writes, and loves to listen to music. And like other little American girls, she jumps with joy each time an astronaut completes another trip to outer space—though perhaps she jumps a little higher because her father, Richard (Rick) Egerter, works for the National Aeronautics and Space Administration, which is responsible for these trips.

But there was a time when Karen knew neither how to jump nor feel any joy. She would clasp her hands over her ears at the sound of a musical toy, the way others wince at the screech of chalk on a blackboard. There was certainly no hope that she would ever be able to read or write. And although she was almost four, she could not even talk.

Karen was a mentally retarded child.

Today, however, this delightful youngster has escaped from her affliction and is living a near-normal life. To the scientific world, she represents the first major breakthrough in the treatment of mental retardation. To all mankind, she proclaims that mental retardation is not an unalterable fact of life; Karen has been cured, and countless generations will some day follow her to normalcy.

Karen was born with a disorder of the metabolic system which is so rare—but so significant—that more researchers have been involved in its cure than there are patients being treated. Known variously as phenylketonuria, phenylpyruvic oligophrenia, or most simply, PKU, it strikes only one child in 20,000. But circumventing this disease has opened a way toward eradicating the blight of mental retardation which, in the United States alone, afflicts 5,500,000 persons.

In many ways, PKU is similar to some cases of diabetes. Both are defects of metabolism which result in a chemical imbalance. In diabetes, the pancreas fails to secrete a hormone which helps digest sugar. In PKU, the liver cannot manufacture the enzyme which breaks down phenylalanine, a basic constituent of protein.

Here is where the resemblance ends. In diabetes, the missing secretion can be injected directly into the body. But science still does not know how to tempt the human body into accepting foreign enzymes.

Two Doctors Solve the Riddle

The immediate problem then was to eliminate phenylalanine from the diet, just as sugar is forbidden to a diabetic. But phenylalanine is found in every protein, and protein is basic to life itself. In April, 1954, three months after Karen's birth, two scientists, Dr. Marvin Armstrong and Dr. Eugene E. Howe, undertook to develop a protein compound from which it would be possible to remove most of the offending phenylalanine. They came up with Ketonil, a brownish powder whose miraculous future belied its offensive taste and smell. Other researchers soon developed a similar low-phenylalanine powder called Lofenalac.

But in January, 1954, when Karen was born, there were no low-phenylalanine powders available and newborn infants were not even being tested for PKU. Karen was brought home from the hospital, a perfectly normal infant. But the phenylalanine she got from her formula kept building up in her body until it reached 20 to 30 times the normal level. Somewhere along the line—no one knows at just what point—the abnormal accumulation of phenylalanine began to poison her growing mind, leaving intact only those brain cells which were already fully developed.

Karen's retardation soon began to betray itself to her anxious mother. Finally, in April, 1957, after Karen had passed her third birthday still not speaking, the Egerters went to an out-of-town clinic for retarded children. There, the simple identification test for PKU was performed; a few drops of 10 percent ferric chloride were placed on Karen's wet diaper—and it turned olive-green. She definitely had PKU. The prescription: institutionalization.

What Betty and Rick Egerter—and seemingly the clinic—did not know was that, at that very moment, field trials were being run on the new PKU diet. However, the parents instinctively refused to part with their first-born. For the next six months, they tried to cope with a child who could not talk, who would never learn to think, and whose soul contorted in rage as more and more phenylalanine was unleashed into her nervous system.

Finally, the Egerters appealed for help to an outpatient clinic for retarded children run by the Georgetown University Hospital in nearby Washington, D.C.

Miraculously, the Egerters had applied to a clinic which only three months before had started testing the new low-phenylalanine powders. Karen was brought in for immediate treatment, an irascible four-year-old in diapers who spoke three unintelligible words and tried to live on frankfurters and potato chips.

The clinic could promise nothing. Since the accumulated phenylalanine attacks only growing brain cells—and the brain reaches full physical development by the age of six—it was suspected that three-fourths of Karen's potential had already been destroyed.

Karen Comes Close to Starvation

The dietary regime was unbelievably difficult. Infants readily accept the low-phenylalanine powders in a formula. Karen fought every mouthful. When the powder was mixed with the few natural foods she was allowed, it immediately turned them olive-green, bad-tasting, and odorous. She was allowed only five grams of protein a day. A frankfurter has seven grams. Karen wound up in the hospital, close to starvation. But there, under controlled conditions, she finally accepted her new diet. Some children never do.

Karen came home to days of forced feeding. Meanwhile, her mother spent three weeks desperately trying to develop a tasty cooky which would contain all the low-phenylalanine powder the child needed. She used Ketonil, Lofenalac, baking chocolate, vanilla, water, sugar, butter, salt, and a gluten-free flour supplied free of charge by the Huron Milling Division of the Hercules Powder Co., a manufacturer of explosives. At first, some cookies were so runny they practically slid into the trash by themselves: others looked like burned oatmeal.

But one day, on her 12th try, Mrs. Egerter saw before her eyes nine perfect giant-sized cookies. From that day on, Karen's regular meals no longer had to be mixed with the obnoxious raw powders.

Slowly, the effects of the diet began to show. First, Karen's blonde hair began to change at the roots to brown, the color nature had intended it to be. Until now, it had been held in check by the chemical mix-up in her body.

Her temper tantrums decreased and finally disappeared. Within a period of three weeks, she discarded her diapers. One bright afternoon she followed a musical toy down the street. And then came the day when she danced into the kitchen and said: "Please, Mommy, may I have just one potato chip, just one?"

Her mother succumbed. She shouldn't have—but once she had prayed: "Oh God, let her say anything. I don't care what it is, as long as it's a word. Just let her talk."

Karen had checked into the Georgetown clinic with an IQ of 57 (normal is 100). Two years later she reached 85. Karen stayed at that point for the next 18 months. Once she had reached six years of age, however, no further brain development could have been expected. It appeared to the Egerters that the low-phenylalanine food had accomplished as much as it ever would. Since no setback was expected, the parents decided to free the child from the rigors of the diet. The past 3½ years had cost them $4,200.

She Can Have a Near-Normal Future

Today at the age of eight, Karen is completing her second year in a special education class at a public school. Last year she was the only one in her group pushed ahead to the next level of achievement. Some day she may go into a regular class, but even if she must remain in a situation geared to her learning abilities, she will be able to complete high school.

She can become self-supporting. She can get married. And if her husband neither has nor carries PKU, her babies will have the same chance for normalcy as the 4,000,000 others born each year in the U.S.

No one knows exactly what would have happened to Karen if she had never been put on the special powders; the relationship between IQ and diet is still conjectural. But her doctor points out that certainly the low-phenylalanine powders soothed her outraged emotions and pushed up her rate of development. On the day she took her first spoonful of powder, Karen was operating at one-half her normal capacity. By the time she swallowed her last mouthful, she had regained almost nine-tenths of her birthright.

Karen was born a little too soon. Had she come into this world three years later, she could have been put on the low-phenylalanine powders by the time she was six weeks old, before any brain damage had taken place. By the age of six, she would have been taken off the diet, for no further damage can occur once the brain is fully developed.

The progress in the treatment of PKU has spurred great optimism. Today there is no doubt that men of science, working from basic biochemical principles, can make good on some of the tragic mistakes of nature. A dozen other causes of mental retardation have already been linked to "inborn errors of metabolism" similar to Karen's.

Maple-sugar urine disease (so named for its distinctive odor) has been pinpointed and is slated for an eventual cure. The dread scourge of cretinism can now be prevented if the thyroid hormone itself is given early enough to children born with a deficient thyroid gland.

Even in those disorders of the nervous system where no retardation occurs, it is now felt that one small defect in metabolism may be at fault. This is the big hope in epilepsy, and there is even some preliminary evidence that certain speech disorders are due to chemical imbalance.

Thus, through Karen's victory over PKU, millions of others have gained hope for deliverance from a wide range of disorders.

PHOTOGRAPHS BY ARTHUR SCHATZ