
Fact Sheet

Mental Retardation

Thirty Years Ago

- **Haemophilus Influenzae Type B (Hib) was the leading cause of acquired mental retardation.** In the mid-1970s, no means existed to prevent infection from Hib, the cause of meningitis — a serious infection of the membrane surrounding the brain and spinal cord. The disease strikes children under 7 years of age, with most cases occurring in children from six months to two years old. By the late 1980s, roughly 15-20,000 cases of Hib meningitis occurred each year. Antibiotics could treat Hib infection, but couldn't prevent its devastating consequences. On average, 1 in 10 infected children died from Hib meningitis, 1 in 3 became deaf, and 1 in 3 was left with mental retardation.
- **More than 10 million children had blood lead levels high enough to affect their cognitive functioning.** It was not known in the early 1970s that exposure to even small amounts of lead in the environment — from paint and from automobile exhaust — could have an adverse effect on the developing brain.
- **Many children of women with the metabolic disorder phenylketonuria (PKU) were born with severe mental retardation — even though they did not have PKU themselves.** PKU is a genetic inability to process the nutrient phenylalanine. The disorder occurs once in every 10,000 to 20,000 births, affecting 250 children each year in the United States. Without treatment, a child will suffer irreparable brain damage and require a lifetime of care in a nursing home facility. In the 1960s, a blood test for PKU was developed and children with the disorder were identified at birth. A low phenylalanine diet spared them from brain damage. Because the diet is difficult to adhere to, many children, including those that would go on to be mothers, discontinued the low phenylalanine diet at approximately age 7 when the dangers of retardation are past. Unfortunately, by the late 1970s, it was

apparent that children carried by moms with PKU were born with mental retardation.

- **Infants lacking thyroid hormone were destined to a life of mental retardation.** In the mid 1970s, more than 1000 U.S. children each year became mentally retarded shortly after birth, because of hypothyroidism — failure to produce sufficient amounts of thyroid hormone. Thyroid hormone is essential for growth, especially of the brain. Although the hormone could be supplied artificially, diagnosis of the condition was usually not made until after an infant's brain was permanently damaged.

Today

- **Meningitis from Hib has virtually been eliminated.** In the 1970s, the search began for a vaccine to prevent Hib meningitis. The Hib bacterium could hide from a young child's immature immune system by means of a protective sheath, or capsule, which shields its outer surface. In their first attempt at a vaccine to prevent the infection, researchers at NIH isolated a complex polysaccharide — a sugar molecule — from the bacterium's covering. By itself, the polysaccharide was not enough to prime the immune system to eliminate the Hib bacterium. The researchers then chemically combined, or conjugated, the sugar molecule to a protein that was easily recognized by the immune system. The protein and sugar "conjugate" became the basis for a new vaccine, which virtually eliminated Hib meningitis from the developed world. In the United States, there are now fewer than 10 cases of Hib meningitis each year.
- **Lead is no longer an ingredient in paint and gasoline.** In 1979, researchers funded by NIH showed that children whose baby teeth contained relatively high amounts of lead fared poorly on a standard intelligence test when compared to children whose teeth contained much lower amounts of lead. The finding eventually led to Federal laws that banned lead as an ingredient in paint in 1974 and as an additive in gasoline in 1978. As a result, the number of children

with elevated blood lead levels fell from 10 million in the 1970s to 434,000 in 2001. Although the two most common sources of environmental lead exposure have been eliminated, many children are still exposed to such sources of lead as paint in older homes, and contaminated soils.

- **Children of women with PKU can be protected from brain damage.** In the 1960s, children with PKU typically discontinued the low phenylalanine diet by the time they reached 7 years of age. The diet's special protein formulations are expensive, and many find the diet difficult to stick with. To test whether a low phenylalanine diet would prevent mental retardation in the children of women with PKU, the NIH began a large study. The study, which took 18 years to complete, enrolled women from more than 120 clinics in the United States, Canada, and three foreign countries. The study was completed in 2003 and found that limiting phenylalanine in the diets of women with PKU beginning before pregnancy and continuing through pregnancy nearly eliminated mental retardation in their children. Subsequent studies have shown that people with PKU score higher on intelligence tests if they remain on the low-phenylalanine diets throughout their lifetimes, rather than discontinuing it in childhood.
- **Infants who lack thyroid hormone can be identified in time to help them.** Researchers funded by the NIH developed a test to identify newborns that have insufficient thyroid hormones. A large study funded by NIH in the early 1970s showed that hypothyroidism could be easily detected, and treated within two weeks, before any brain damage resulted. Soon, every State required thyroid hormone screening along with PKU screening. Each year in the United States, roughly 1000 cases of mental retardation due to insufficient thyroid hormone are prevented.

Tomorrow

- **The NIH is supporting the development of new DNA microarray chips and other technologies for newborn screening.** The goal is to develop a fast, reliable, cost effective means to screen newborns for a multitude of genetic conditions, including not only causes of mental retardation, but of immune deficiency, blood disorders, nervous system disorders and muscle disorders. Such a screening test would make it possible to begin treatment early, when chances for success are greatest. Large

numbers of infants who have disorders lacking effective treatments could also be identified easily. Although treatment might not yet be available for their conditions, they could be offered a chance to participate in studies of new treatments, so that eventually new therapies could be developed for their disorders as well.

- **NIH-funded researchers hope to develop a drug that may one day treat the symptoms of Fragile X Syndrome.** The condition affects one in 6000 births, resulting in mental retardation, sleep problems, attention deficit disorder, aggression, and compulsive behavior. NIH-funded scientists working with mice having the same genetic mutation found in Fragile X Syndrome learned that the mice have increased activity in the metabotropic glutamate receptor (mGluR), which sits atop brain cells. Studies in mice and fruit flies show that chemically blocking the mGluR receptor results in the animals displaying more normal behaviors. Researchers hope that drugs that block the mGluR receptor might one day be used to lessen the disorder's effects in humans.
- **Researchers have prevented brain damage in newborn infants deprived of oxygen at birth by lowering body temperature.** Accidents of birth — compression of the umbilical cord, or rupture of the uterus, for example — can deprive an infant's brain of blood and oxygen. Survivors of such accidents may suffer life long brain damage and disability. Known scientifically as hypoxic ischemic encephalopathy, or HIE, oxygen deprivation during birth is estimated to occur from 0.5 to 1 times per every thousand births. Researchers in an NIH network were able to reduce the amount of death and disability of a group of infants with HIE, by lowering the infants' body temperature. The cooling treatment, known as hypothermia therapy, consisted of placing the infants on a soft plastic blanket through which cool water circulates. When the infants were examined at 18 to 22 months of age, 44 percent of those given hypothermia treatment had developed a moderate to severe disability or had died, as compared to 62 percent of infants receiving standard treatment for HIE. Because minor fluctuations in an infant's body temperature could result in serious harm, the hypothermia treatment requires personnel trained in life support and the use of the cooling blanket. Researchers in the network are working to refine the therapy so that it may one day be used routinely in newborn intensive care units.