A project to enlist computers in battle against birth defects

By Richard Saltus
Science Writer

The infant boy had some sort of birth malformation, there was no question about that. But the doctor had no answers to the distraught mother's questions.

"What is it? Will he be able to walk? Will he be retarded?" The pediatrician couldn't say because he had never seen a case like this one.

But instead of sending the mother and child from one specialist to another, or spending long hours in the medical library tracing reports scattered through the literature, the doctor simply made a phone call.

Within minutes, via the type-writer-like terminal of his office computer, he was in touch with the world's largest store of knowledge on more than 1,000 birth defects, from Aarskog Syndrome to Zytomegalie.

After a "conversation" with the data bank in Valley Forge, Pa., the doctor had a tentative diagnosis and a print-out of information on the infant's prognosis to offer his mother.

This vignette is on the verge of reality, National Foundation-March of Dimes officials said yesterday in unveiling the Birth Defects Information System.

More than 400 physicians around the world have pooled their knowledge in a computer bank that will be accessible 24 hours a day to hospitals and doctors, who previously had no central source of defects information.

With more than 250,000 children being born with defects in the United States each year, and new congenital syndromes continually arising, "keeping up to date is essential," said Dr. Marylou Buyse, assistant professor of pediatrics at Tufts University School of Medicine in Boston.

The computer system is operated by the Center for Birth Defects Information Services of Tufts-New England Medical Center.

Because defects often affect several body systems and occur in a wide variety of forms, no one medical specialty or textbook can know them all. The nearest thing to a standard textbook is the March of Dimes' Atlas & Compendium of Birth Defects, first published in 1973 and already out of date.

In its first phase of operation, the computer system simply will serve as a source of accumulated information on defects. It will be constantly updated.

Later, according to John Donovan of the Massachusetts Institute of Technology, which is involved in the system's development, the computer will be programmed to aid in diagnosis.

A physician suspecting a particular congenital syndrome will be able to type the signs, symptoms and laboratory findings on his terminal and the computer will search its data bank for a disorder fitting those factors.

The computer may ask the doctor to consider other possibilities and may inquire about additional symptoms the patient might display.

The results could "add weight to the physician's diagnosis or suggest he look elsewhere," Buyse said.

"Pediatricians are not well educated about medical genetics," she said, "and especially if they've been out of school for 10 years or so, it's hard for them to be up to date on the entire range of birth defects."

Even at specialized centers such as the Tufts facility, she said, as many as 40 percent of defects are unidentified. The computer system will be able to match rare cases from around the world and perhaps lead to identification of additional syndromes, she added.

Farther away are plans to use the system as a tool for research on defects, and finally as an "early warning system" to monitor the occurrence of defects and, it is hoped, prevent disasters like the thalidomide babies of the 1950s.

Donovan said the information retrieval system is one of the most sophisticated ones involving medical diagnosis.

In a demonstration at the annual Birth Defects Conference, which concludes today at the Sheraton-Palace Hotel, Buyse selected a defect from an index that assigns each syndrome a computer code number.

Because it is relatively common she picked Down's Syndrome (formerly called mongolism), and a technician punched its code number, 133, into the terminal.

Almost immediately, the terminal began printing out a "thumbnail sketch" of the disorder — its clinical signs, how frequently it occurs, the characteristic genetic defect, prognosis, treatment and a list of references for further information. A notation showed that the sketch had been updated last month.