

SOUTHWESTERN NEWS

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DOWN SYNDROME CLINIC A PLACE FOR HOPE, RESEARCH

DALLAS -- October 12, 1994 -- Dr. Golder Wilson and Lisa Maberry have been looking for answers to Down syndrome. Wilson is a pediatric geneticist at The University of Texas Southwestern Medical Center at Dallas, and Maberry is the mother of a 10-year-old with the puzzling genetic disorder.

After two years of working together at the Down Syndrome Clinic at Children's Medical Center of Dallas, Wilson and Maberry are beginning to find some answers.

Wilson, a professor of pediatrics at UT Southwestern and holder of the Mary McDermott Cook Distinguished Professorship in Pediatric Genetics, wants to know what causes this form of mental retardation that occurs in approximately one of every 800 births and affects about one-quarter of a million families in the United States. "The occurrence of Down syndrome is largely random," Wilson explained. "There are no clues why this should happen."

The only consistent indicator of the risk of Down syndrome is the age of the mother. The risk rises dramatically as the mother gets older. For example, Wilson said, the odds of a mother age 25 or younger giving birth to a child with Down syndrome is about 1 out of 2,000. At age 35 the odds increase to 1 out of 200, and at age 45 the odds are about 1 out of 50.

Researchers have looked at a number of variables to try and isolate the cause or causes of the disorder. But, Wilson said, age is the only one they've uncovered so far, and it is not a completely reliable predictor. The problem, he said, is that "there's no easy experimental system in place in the lab to mirror the cellular process that produces Down syndrome."

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Children with the disorder carry three copies of chromosome 21 instead of the usual two. Down syndrome produces mental retardation and, quite often, multiple birth defects -- particularly heart disease. "The children have a characteristic appearance, and their muscle tone is often very weak," Wilson said. Children with Down syndrome also generally develop language skills much more slowly than other children. "That's when the disease becomes most obvious," Wilson added.

Wilson and his colleagues at the Down Syndrome Clinic believe early intervention can significantly improve the general health and quality of life for a child with the disorder. They also believe that because they routinely see so many children with Down syndrome -- about 150 patients a year, making it one of the busiest clinics of its kind in the nation -- they are more likely to recognize the unique medical needs of these patients. "Nearly 90 percent of the children we saw in the clinic's first year were missing some essential part of medical management," Wilson said.

Bridging that gap is what prompted Maberry and other parents in the local Down Syndrome Guild to work with Wilson and other UT Southwestern faculty to help start the clinic at Children's in 1992. "We have a wonderful pediatrician," she said, "but children with Down syndrome like our son, Luke, have some things that need special attention. There are just some times when your pediatrician may be stumped by something. The doctors in this clinic catch things that your doctor may miss."

The clinic is a resource for the parents and doctors of children with Down syndrome, Maberry said. "It's just done wonders to have a regional center like this one available, where you can get a multidisciplinary approach to the child's health care."

In addition to heart disease, children with Down syndrome are more likely to have vision and hearing problems, gastrointestinal disease and

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thyroid disease. The children also have varying levels of mental retardation ranging from mild to severe. Maberry said the clinic is set up so parents can see other specialists who are part of the child's treatment team during their visit to the Tuesday morning clinic. A social worker and counselor also are on staff to help the families in addition to a member of the Down Syndrome Guild.

Maberry is a member of the guild and works as a clinic volunteer.

"We're here to talk to other parents about nonmedical issues like school or just family life with a child with Down syndrome." That component is vitally important to the clinic's success, Maberry believes. "We're here to listen," she said. "While having a child with Down syndrome is not that difficult, some aspects of raising the child — school, day care and the like — are difficult to manage. It's a devastating blow.

"No one expects to have less than a perfect child," she said. "It's a big adjustment to parents. But the clinic has shown many of them what their kids can do."

To help the children achieve their potential, Wilson and other researchers at UT Southwestern use a medical-management checklist for children with Down syndrome. The checklist, which has been adopted for use by the American Academy of Pediatrics, was developed by the parents of children with Down syndrome. "The parents were reporting the problems they saw in their children," Wilson said. "Now that we have the list, we want to test it further and try to improve it." Wilson is currently performing an outcome analysis of the checklist to determine if following the list significantly improves the medical care of the children.

Wilson said his long-term goal is to perfect clinical strategies that improve the quality of life for children who have Down syndrome.

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