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## **BAKERSFIELD COUPLE ATTRIBUTE BIRTH OF HEALTHY BABY TO PRENATAL DIAGNOSIS AT CEDARS-SINAI MEDICAL CENTER**

LOS ANGELES (April 30, 2004) – At four months, Samuel Maccari is the proverbial “bouncing baby boy.” He is strong, active and alert—and the natural child of a Bakersfield couple who once thought adoption might be their best and only option. State-of-the-art prenatal counseling and diagnosis at Cedars-Sinai Medical Center changed all that.

Dad Dan Maccari remembers the day last May when he and wife Deborah Bechtold learned her prenatal chorionic villi sampling (CVS) showed no sign of the genetic disease that claimed their first baby. “We went from thinking we were never going to have children to that news,” he recalls.

Six years earlier, the couple had endured the painful loss of an infant born with a genetic disorder, spinal muscular atrophy (SMA). Subsequent genetic analysis determined that the couple had a 25 percent risk of recurrence in any future pregnancy. They couldn’t imagine losing another child to SMA.

Daughter Camryn seemed perfectly healthy at birth in 1997, but her two-month check-up determined “something was wrong,” according to Maccari. That “something” was diagnosed as SMA, an invariably fatal disease—95 percent of victims die before age 2—that affects cells along the spinal column, leading to debilitating muscle weakness and deterioration.

“For the next 8 months, we watched Camryn wither away,” explains Maccari, who says neither he or his wife had any family history or known risk factors for the disease.

At the time, the couple lived in Nevada, and there was only one other baby with SMA in the whole state. They found doctors there knew little about the disease. When genetic testing confirmed that both carried the recessive gene responsible for SMA, the couple feared their chances of ever having a healthy baby. “At first, we didn’t want to take a one-in-four chance, and we were pursuing adoption,” says Maccari.

After relocating to California, the couple decided to explore their options and conferred with a genetics counselor at Children’s Hospital in Fresno who told them about CVS. Cedars-Sinai was one of two centers recommended for the test, which involves taking a small tissue sample from the chorion, part of the placenta. The procedure is conducted in the first trimester—between 10 and 13 weeks—using ultrasound to guide a catheter either abdominally or, more commonly, cervically to collect the sample. The primary advantage of CVS over amniocentesis, performed between 16 and 20 weeks, is detection of problems earlier in pregnancy. CVS identifies the same conditions as amniocentesis except for neural tube disorders like spina bifida.

“We chose Cedars-Sinai because of name recognition and reputation—we wanted the best possible,” says Maccari. “We did research and found out Dr. (John) Williams was ‘the guy’ when it comes to CVS, and we wanted to go to ‘the guy.’”

The fact that the doctor’s reputation precedes him is no surprise: John Williams III, M.D., director, Reproductive Genetics, Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, has performed more than 14,000 CVS procedures in nearly 20 years—more than all but one other physician in the nation. In 2004 alone, Dr. Williams expects to complete up to 1,500 CVS tests, a number that has been steadily rising each year.

“Contrary to popular misconception in the community (including many, many obstetricians), it is absolutely not true that CVS is more dangerous than amniocentesis,” Dr. Williams asserts. “There is no significant statistical difference in risk if the procedure is done by a skilled physician, and there is at least one skilled CVS practitioner in each major metropolitan area of the U.S.”

With their “ducks in a row,” Maccari says, the couple moved forward with their plans and pregnancy. The first step was meeting with Nancy Kramer, M.S., a board-certified genetic counselor at the Cedars-Sinai Prenatal Diagnosis Center. Kramer went over their case and discussed the CVS process.

“People are definitely more and more educated about testing for genetic problems, which is good because it helps people like Deborah and Dan have their own child,” explains Kramer, who describes herself as an advocate for the patient.

When Bechtold was 10 to 11 weeks into her pregnancy, she went to Cedars-Sinai for the CVS procedure, which she described as quick and “completely painless—more comfortable than a pelvic exam.” They were overjoyed when they received the news they’d hoped for—“negative.”

The pregnancy continued problem-free, and on Nov. 21, 2004 they delivered 8-pound, 13-ounce Samuel without complications. Whether he is a carrier for SMA can be determined by a blood test, which his parents plan to do sometime in the future. This information can help Samuel make educated decisions about starting his own family when that time comes.

While couples can and do self-refer, many are sent to the Prenatal Diagnosis Center by their own obstetricians, who then continue their patient’s care throughout pregnancy and delivery, says Kramer, adding, “We’re here to help them along the way through a particular concern or problem related to their pregnancy.”

Before any diagnostic procedure, the couple meets with a genetic counselor to discuss family histories, determine risk factors and then decide which, if any, tests are appropriate. In the general population, the risk of a genetic defect is 2 to 4 percent, and it is estimated that most people are carriers of five to seven recessive genetic conditions. “Patients are often surprised to learn that they are a carrier of a recessive condition. Many may not know that they are carriers until they (or a family member) has an affected child or undergoes carrier screening,” Kramer explains.

Genetic screening tests available today have greatly increased couples’ ability to predict the likelihood of genetic disorders in their offspring. Ethnicity is one of the main factors determining risk. For example, families of Ashkenazi Jews (of Eastern European Jewish ancestry) are at risk to be carriers of a number of genetic conditions. A carrier screening panel for nine genetic conditions, including Tay-Sachs and cystic fibrosis, should be considered for any couple of Jewish ancestry who is considering a pregnancy.

Prenatal diagnostic tests can provide additional information and, in many cases, peace of mind. Testing is available to patients in various situations—women over age 35 who are worried about chromosomal problems like Down Syndrome, patients exposed to potentially harmful medications or chemicals, couples with a

family history or previous child with a birth defect, pregnancies where an abnormal ultrasound finding is detected, or couples at increased risk of ethnic-related genetic disorders.

It is important to realize even the newest tests have limitations. “We can rule out the biggies—chromosomal abnormalities like Down syndrome or specific genetic disorders when there is a family history, such as spinal muscular atrophy or cystic fibrosis – but we can’t guarantee everything about the baby is okay,” Kramer cautions. “We know this is what you really want to hear when you’re pregnant.”

Other options include the relatively new first trimester screening for Down syndrome and trisomy 18, chromosomal defects that cause deformity and retardation. First-trimester screening can be conducted as early as 11 to 14 weeks. The screening is comprised of a blood test to analyze two proteins—hCG and PAPP-A—and an ultrasound measuring the thickness of the skin at the nape of fetus’ neck.

“The first trimester test is very good at detecting Down syndrome with 80 to 90 percent accuracy, and is actually better than the second trimester screening, which has about a 60 percent detection rate,” explains Dr. Williams, pointing out that earlier diagnosis allows for greater privacy, peace of mind and opportunity to bond with the pregnancy.

Also available is the second trimester Expanded AFP (“triple screen”) administered at 15 to 20 weeks to identify Down syndrome, trisomy 18, neural tube defects like spina bifida and other malformations. A blood sample from the mother is evaluated to gauge levels of alpha-fetoprotein (AFP), hCG and estriol. Based on the results of these blood tests, other diagnostic procedures like amniocentesis and ultrasound may be recommended. Some laboratories are now offering a “quad test,” that uses a fourth serum marker to up the detection rate of affected pregnancies.

“We now have 3D/4D ultrasound that enables you to see motion and movement in three dimensions,” says Dr. Williams. “Though traditional two-dimensional ultrasound is faster, 3D/4D offers the advantages of seeing better contour and allowing multiplanar images of the fetus. It holds a lot of promise.”

The Prenatal Diagnosis Center also offers antenatal testing, which typically utilizes fetal heart rate monitoring and ultrasound to determine the baby’s well-being in late pregnancy. This may be appropriate for patients who’ve passed their due dates, have diabetes or high blood pressure, or poor fetal growth. The Antenatal Unit also offers a wide variety of sophisticated tests to help patients and their physicians plan for care during and after pregnancy.

Regardless of the specifics of a particular case, the Prenatal Diagnosis Center staff approaches each patient with a similar philosophy, says Kramer, “We try to apply empathy. Reducing anxiety is part of the job. Our focus is on supporting and helping patients like Dan and Deborah have healthy families.”

Cedars-Sinai Medical Center is one of the largest non-profit academic medical centers in the Western United States. For the fifth straight two-year period, Cedars-Sinai has been named Southern California's gold standard in health care in an independent survey. Cedars-Sinai is internationally renowned for its diagnostic and treatment capabilities and its broad spectrum of programs and services, as well as breakthrough in biomedical research and superlative medical education. It ranks among the top 10 non-university hospitals in the nation for its research activities.

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