

Metabolic Tests at Birth

By Amy Bentley

NEWBORN METABOLIC SCREENINGS

"In the body there are thousands of chemical reactions happening all the time. When one of those steps is not working, you have a buildup of toxic metabolites that can make you sick," says CHOC metabolic disorder specialist Dr. Jose Abdenur. "These metabolic tests are done on newborns with very small blood samples that are taken at birth from every baby in California. They can test for over 60 conditions and 40 metabolic disorders," he says.

PROGNOSIS AND OUTLOOK

With early diagnosis and treatment, most babies with these disorders will get the right treatment and go on to have healthy lives, says Dr. Abdenur. "At birth the baby can look healthy, so the purpose is to take these tests before they become sick and before there is neurological damage later on that we can not reverse."

1 in 10,000-15,000 babies

ARE BORN IN THE U.S. EACH YEAR WITH PKU

1 in 17,000 people

SUFFER FROM MCAD DEFICIENCY IN THE U.S.

WHEN AND HOW METABOLIC TESTS ARE DONE

Babies get their newborn screenings before they leave the hospital when they are generally a day or two old, says Dr. Abdenur. The metabolic tests are done with a blood sample taken from a small prick to the baby's heel. The blood is collected on a special paper and sent to a lab for testing. If the results are positive for a metabolic disorder, the testing lab will connect with CHOC's Metabolic Center to move forward and help the patient and family, says Dr. Abdenur.



50 YEARS OF EXPERT CARE

For over 50 years, CHOC Children's physicians, nurses and staff have been devoted to improving the lives of children and families in our community. As home to the region's only dedicated pediatric emergency department and other world-class services and programs, from cardiology and neurosurgery to neonatology and orthopedics, CHOC is committed to being the leading destination for children's health by providing exceptional and innovative care.

For information on the expert care we provide and the health plans we accept, visit choc.org



Dr. Jose Abdenur
CHOC Metabolic Disorder Specialist

Dr. Abdenur heads the division of metabolic disorders at CHOC and is the director of CHOC's Metabolic Laboratory. Dr. Abdenur completed fellowship training in pediatric endocrinology, metabolism and nutrition at North Shore University Hospital, Cornell University Medical College in New York. He completed fellowship training in biochemical genetics at the Denver Children's Hospital and The Biochemical Genetics Laboratory at the University of Colorado Health Sciences Center, and in Clinical Genetics at Mount Sinai School of Medicine in New York. Dr. Abdenur has coauthored more than 150 abstracts, presentations, book chapters and journal articles, and he has received numerous awards.

Dr. Abdenur's philosophy of care: "Our program is much more than one person. We need all team members, including the physician, the nurse practitioner, the metabolic dietitian, the nurse/case manager, the social worker and the genetic counselor working together with the lab, the family and the patient to achieve success."

EDUCATION:

Medical degree from the Universidad de Buenos Aires, Argentina

BOARD CERTIFICATIONS:

Clinical Biochemical Genetics from the American Board of Medical Genetics

About 1 in 3,000 newborns

HAS A METABOLIC DISORDER AT BIRTH

