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Pediatrician and Geneticist Dr. Harvey Levy Receives 2020 David L. Rimoin Lifetime Achievement Award in Medical Genetics from the ACMG Foundation for Genetic and Genomic Medicine

BETHESDA, MD – March 18, 2020 | Internationally acclaimed clinical geneticist and pediatrician Harvey Levy, MD, FACMG, is the recipient of the 2020 ACMG Foundation for Genetic and Genomic Medicine's **David L. Rimoin Lifetime Achievement Award in Medical Genetics.**

Dr. Levy, senior physician in medicine and genetics at Boston Children's Hospital and professor of pediatrics at Harvard Medical School, is being honored for his many years of groundbreaking work with patients who have genetic metabolic diseases including phenylketonuria (PKU), homocystinuria, cobalamin metabolic disorder, and others; as well as for his training and mentoring of the next generation of genetics service providers; and for his major contributions to the development of newborn screening in the United States and around the world.

Dr. Levy's medical career spans more than 60 years. He has mentored over 60 medical genetics fellows; published more than 400 research articles, reviews, book chapters, and proceedings from research meetings; written 2 books and created 2 educational videos for patients and clinicians; served on editorial boards and as a reviewer for numerous prominent research journals; and founded and formerly directed both the Maternal PKU Program and the Inborn Errors of Metabolism/PKU Program at Boston Children's Hospital.

"Harvey Levy is a physician scientist who has been instrumental in the development of newborn screening programs for metabolic diseases," said former ACMG Executive Director Dr. Michael S. Watson, FACMG. "Of particular importance has been his melding of knowledge of clinical genetics, population genetics and metabolic diseases to identify critical issues in the transition from a disease-based understanding of particular metabolic diseases to a population-based prevention program that has had enormous impact on hundreds of newborns in the United States."

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"We take newborn screening for granted now," said Gerard Berry, MD, director of the Metabolism Program and professor of pediatrics at Harvard Medical School. "There are laws in different states that babies need to be screened for certain diseases. But when Harvey began, this was uncharted territory. People didn't understand the power of newborn screening and how it could change lives by allowing someone to get on a diet or a medication that they need to take for life in order to be healthy. Harvey played a major role in allowing all of this to come to fruition. These same individuals, who might have been institutionalized years ago because of severe intellectual disability, are now students in elite colleges. Harvey possesses insight and super-ability to understand what is really important for healthcare. Newborn screening is one of the major healthcare successes of the previous century…maybe the *most* important healthcare success. And Harvey was part of a group of unique individuals who helped to see that through."

The news that he had received the David L. Rimoin Lifetime Achievement Award came to Dr. Levy as a delightful surprise. "This means a great deal to me because of the ACMG, where I've been an active member for a number of years," Levy shared. "It's a wonderful organization full of outstanding individuals, so to be in that company is particularly gratifying. And it's very, very nice to be appreciated."

"The Rimoin family is proud to recognize Dr. Harvey L. Levy, whose outstanding work includes studies that formed the basis for newborn metabolic screening, the discovery of the first human vitamin B12 defect and the establishment of cobalamin defects, and the development of Maternal PKU programs," said Dr. Ann Garber, David Rimoin's surviving spouse. "Based on his scientific accomplishments, along with his remarkable integrity, empathy and collaboration, our family is pleased to honor Dr. Levy with the David L. Rimoin Lifetime Achievement Award."

Beyond his list of academic achievements and leadership positions, the nominations for Dr. Levy to receive this award stressed his abounding generosity of time, knowledge and skill while working with patients, families and the broad range of clinical providers and researchers who have collaborated with him.

"He's dedicated himself to the study of PKU and metabolic disorders with an energy and intellect and soul that is extraordinary," said neuropsychologist Susan Waisbren, PhD, a professor of psychology at Harvard Medical School and Dr. Levy's long-time collaborator at Boston Children's Hospital Metabolism Clinic. "One of the qualities I've always found striking is the respect he has for professions outside of medicine. In his mind, every member of the clinical team is important. He truly feels this and it shows in his academic as well as clinical work. He has included as co-authors psychologists, dieticians, social workers, genetic counselors, nurses, administrators, secretaries, and parents.

"The patients adore him, always," she added, "and they recognize a certain compassion and ability to see the whole person, not just the metabolic disorder."

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"Harvey is one of those special individuals who one may encounter once in a lifetime," said Dr. Levy's collaborator at Boston Children's Hospital, Dr. Berry. "He's much more than an accomplished geneticist and investigator. First and foremost, he's a very endearing individual with a wonderful bedside manner, and he's beloved by patients and families whom he's cared for over the years. Harvey goes out of his way to make things better for patients and their families."

As an example, Dr. Berry, who has known Dr. Levy for several decades, recalled a case around 15 years ago, when a baby had been born with PKU in a suburban hospital outside of Boston. "Without telling anyone, Harvey drove to the hospital just to say hello to the new parents and to see the baby," Dr. Berry recounted. "He didn't need to do that. Everything was already in place, people were already taking care of what needed to be done, but he felt compelled to drive out there on a Friday evening to say hello."

Harvey L. Levy was born in Augusta, Georgia in 1935, the eldest of three sons. His father owned a one-room mercantile that supplied clothing to families of the surrounding area, which comprised mostly farmland during that period. His mother, who was a homemaker, graduated from Hunter College and served as a technician in a research laboratory in New York before her marriage. He credits her with some of his initial interest in research.

"I was a guy who was looking for answers to things, so I was always interested in science. And I particularly liked chemistry," Dr. Levy recalled. "My mother was a very intelligent person and very interested in education and music and arts, and also interested in science. I talked with my mother quite a bit about science. So, I think she had a feeling that maybe it would be a good idea for me to be a doctor."

Dr. Levy began studying history as an undergraduate student at Emory University and then switched to an early admission program at the Medical College of Georgia. One of his medical school professors, the famous Dr. Victor Vaughan, headed the department of pediatrics and had a profound influence on the direction of Dr. Levy's career. "I was always interested in pediatrics because of its developmental aspects," explained Dr. Levy. "I felt that if I was going to do something in terms of disease, preventing or helping patients in a significant way, I had to start early, and the earlier the better."

After completing his medical degree in 1960, Dr. Levy served an internship in pediatrics at the Boston City Hospital under Dr. Sydney Gellis, a renowned teacher of pediatrics. Following the internship he moved to New York and the Columbia-Presbyterian Medical Center, where he spent a year under Dr. Dorothy Anderson, the discoverer of cystic fibrosis. Then, as world events escalated toward the start of the Vietnam War, he was drafted and served 2 years in the Unites States Navy as a medical officer stationed in the Philippines.

His introduction to genetics came when he returned to his medical training in 1964 as a second-year pediatrics resident at Johns Hopkins University, where he met the pioneering pediatric clinical geneticist Dr. Barton Childs. What he learned from Dr. Childs about DNA triggered memories of an earlier time, and brought forth questions that further defined Dr. Levy's future career.

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"If I go back to my childhood, my upbringing, I had three cousins from one of my father's brothers, whose family we were very close to, and all of these cousins were developmentally disabled," Dr. Levy said. " No reason was given for their disability and I always thought if I got into genetics, then maybe I could discover the causes of brain disease, particularly intellectual disability, and maybe I could influence the prevention of it."

Dr. Levy returned to Boston, where he served as Chief Resident in Pediatrics back at the Boston City Hospital. During that year he heard a lecture by Dr. Mary Efron, director of the Amino Acid Laboratory at Massachusetts General Hospital, in which she described her studies on metabolic disorders and their enzymatic defects as well as how newborn screening was helping clinicians to identify infants with these disorders so they could receive immediate preventive treatment.

"I became so fascinated with that. It was just absolutely the thing that I really wanted to do," recalled Dr. Levy. "Here was chemistry, biochemistry, genetics, and the prevention of disease! So I asked Dr. Efron if I could do a fellowship with her, which resulted in an NIH-funded fellowship at Massachusetts General Hospital. And that began the journey that has continued to this day."

One cold, fateful Friday afternoon while he was working in Dr. Efron's lab, a telephone call came from Dr. Robert MacCready, director of the Massachusetts Newborn Screening Program. Dr. MacCready asked if someone could come to the screening lab to look at an unusual screening result. Dr. Efron was ill, so Dr. Levy rode his bicycle seven miles across town to the State Laboratory Institute, where he recognized the unusual spot on the paper chromatogram test as a high level of methionine, the hallmark of a genetic disorder he had recently learned about called homocystinuria.

"I called the baby's doctor and asked if I could see the baby at the Massachusetts General Hospital the following Monday," Dr. Levy recalled. "The family and baby came that Monday and I confirmed that the infant indeed had homocystinuria. I asked if they had other children, and was told, 'Yes, we have a daughter.' And I asked if she was ok, and they said she was fine. I asked to see her and she was brought to the next visit, where I immediately recognized that she was developmentally delayed and had other features of homocystinuria that had only recently been described. She was born before screening for homocystinuria had begun. So that launched me into the field of methionine metabolism and some very interesting new areas of research." Much of this research was in collaboration with the late Dr. Harvey Mudd of the NIH, who was the world's foremost authority on methionine and on sulfur amino acid metabolism in general.

Dr. Efron passed away and Dr. Levy assumed Dr. Efron's position as consultant to the Massachusetts Newborn Screening Program and, in 1972, was appointed Director of the program. Four years later, he became Chief of Biochemical Genetics for the New England Newborn Screening Program, a position he held until 1997. Throughout this period, Dr. Levy collaborated with the famed, late microbiologist Robert Guthrie, MD, PhD, of Buffalo, New York, who had established newborn screening with his invention of the PKU test. During this time, he also continued to conduct research and to diagnose and treat patients with metabolic disorders at the Massachusetts General Hospital. An extraordinary influence for Dr. Levy during this time, and continuing to the present, is the internationally famous Canadian biochemical geneticist Dr. Charles Scriver, with whom Dr. Levy has often collaborated.

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Toward the end of the 1970's Dr. Levy moved to Boston Children's Hospital, where he transformed the PKU Clinic it into a larger, comprehensive clinic—the Inborn Errors of we've had to rely on complicated diets that alter the lives of patients so they cannot enjoy a normal meal with their family or their friends, they have to only be able to eat this very difficult diet, and also the fact that we still discover diseases for which we have no treatment, " he explained, "these are the issues that trouble me. There are still individuals we discover during newborn screening or we discover later on because we didn't screen for their disorder, and they have severe disorders for which we have no treatments. There are still metabolic diseases that are not being prevented." Metabolism clinic—that now sees patients and families from around the world who are affected by a range of diseases: PKU, galactosemia, histidinemia, methylmalonic acidemia, problems with vitamin B12 metabolism and many other disorders. The hospital recently named the metabolic program after Dr. Levy.

At Boston Children's Hospital Dr. Levy became concerned about infants born to mothers who have genetic metabolic disease. "Before we began newborn screening girls who had PKU became delayed in their mental development, so very few bore children," Dr. Levy explained. "But now that we were treating them from infancy, they were bearing children. Even though their babies were genetically normal, they would be born with multiple severe problems if the mothers were not strictly treated for PKU during the pregnancies. So, with an extraordinary group of very talented professionals, including psychologists, nutritionists, a nurse, and a social worker, as well as physicians, we organized the New England Maternal PKU Program and followed these women on very strict dietary treatment throughout their pregnancies. We found that this regimen prevented many of these problems that the babies would otherwise have."

Today Dr. Levy is considered one of the foremost proponents worldwide for newborn screening. He led a successful effort in Massachusetts to expand newborn metabolic screening with new technology so that 20 to 30 disorders of amino acid, organic acid and fatty acid metabolism could be included rather than only 5 or 6 disorders previously screened. Within the ACMG, Dr. Levy led the effort to develop "ACT Sheets," one-page synopses of the newborn screened metabolic disorders so that physicians caring for infants can easily read an explanation of the biochemical, clinical and treatment characteristics of the disorders when contacted by a newborn screening program about an abnormality. As part of a contract funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), Dr. Levy began and led the Newborn Screening Translational Research Network of the ACMG.

Though many of his contemporaries have retired, Dr. Levy continues to lead research efforts that examine the long-term outcomes of expanded newborn screening using tandem mass spectrometry—including the medical, biochemical and neuropsychological outcomes in relation to early treatment. He is also involved with clinical trials to develop new therapies for PKU and homocystinuria. Dr. Levy is driven to continue his work because there is still much work to do. "The fact that we've had to rely on complicated diets that alter the lives of patients so they cannot enjoy a normal meal with their family or their friends, they have to only be able to eat this very difficult diet, and also the fact that we still discover diseases for which we have no treatment, " he explained, "these are the issues that trouble me. There are still individuals we discover during newborn screening or we discover later on because we didn't screen for their disorder, and they have severe disorders for which we have no treatments. There are still metabolic diseases that are not being prevented."

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Dr. Levy still spends time communicating face-to-face with patients. "If you have a new baby, in a room with the family, you have to present this very complicated story, and the family has no idea what this is about," he explained. "So, we spend a great deal of time explaining the biochemistry, the genetics, the problems that can occur and the treatments that can prevent these problems. Early on, we just thought about biochemistry. But today we become more involved in talking about the genes, because we think it's important for families to understand the origin of these disorders since at some point we are likely to talk to them about the possibility of gene therapy, actually introducing the normal gene into the child. So, they need to understand where the disorder comes from. It's a complicated and long process. The family will take in as much information as they can, but as you can imagine, a lot of what we tell them will be forgotten or not understood. So, we go over everything with them again, and for as many times as they need."

One of the most pleasing aspects of Dr. Levy's career, he recounted, has been working with wonderful and dedicated individuals—psychologists, nutritionists, dieticians, nurses, social workers, coordinators, administrators—and within the community of clinicians and researchers who study metabolic genetic disorders, a "relatively small, cohesive group of delightful, brilliant people" as he describes them. "It's been an extraordinarily wonderful professional life, as gratifying as any professional life I could ever dream of," reflected Dr. Levy. "Little did I know when I started that I would have this kind of life and little did I know that I would be awarded with the awards and certainly nothing comparable to the David L. Rimoin Lifetime Achievement Award."

The David L. Rimoin Lifetime Achievement Award is the most prestigious award given by the ACMG Foundation. A committee of past presidents of the American College of Medical Genetics and Genomics selects the recipient following nominations, which come from the general membership.

About the ACMG Foundation for Genetic and Genomic Medicine

The ACMG Foundation for Genetic and Genomic Medicine, a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics and Genomics (ACMG) mission to "translate genes into health." Through its work, the ACMG Foundation fosters charitable giving, promotes training opportunities to attract future medical geneticists and genetic counselors to the field, shares information about medical genetics and genomics, and sponsors important research. To learn more and support the ACMG Foundation mission to create "Better Health through Genetics" visit www.acmgfoundation.org.

Note to editors*:* To arrange interviews with experts in medical genetics, contact ACMG Senior Director of Public Relations Kathy Moran, MBA at kmoran@acmg.net.

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