2011 William Allan Award Introduction: John M. Opitz[1]

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As a member of the awards committee of the American Society of Human Genetics, it is my privilege, great honor, and pleasure to introduce to you the winner of the 2011 William Allan Award: Dr. John M. Opitz.

Dr. Opitz is unarguably a pioneer in the field of medical genetics. The year 2012 will mark 50 years of working in this field to which he has contributed over 500 publications in peer-reviewed journals. Dr. Opitz is a rare scholar and polyglot who speaks several languages in addition to his native German. He has used this knowledge to read texts in their original language and include pertinent references and ideas into his writings on embryology, fetal pathology, developmental biology, and medical genetics.

Dr. Opitz is probably best known for working with patients and their families in order to observe and delineate many of the now classical genetic syndromes. He very well might be the single one geneticist who has described the most novel syndromes. In order to honor the patients rather than to call attention to the identifying clinician, Dr. Opitz named these syndromes after the first initials of the families that they affected, such as G syndrome,[1] BBB syndrome,[2] FG syndrome,[3] C syndrome,[4] KBG syndrome,[5] and many others. However, some of them now bear his name as in Smith-Lemli-Opitz syndrome[6] and Opitz syndrome (for GBBB syndrome). Not only did Dr. Opitz delineate novel syndromes in the 1960s, 70s, and 80s, but he also helped to understand the constellation of findings in the context of normal and abnormal embryological development. It was Dr. Opitz who rediscovered the concept of developmental field defect in humans,[7,8] a concept that was previously described in amphibians by the German Nobel Laureate Hans Spemann. Over the past two decades, Dr. Opitz has been actively involved in the identification of the underlying genetic bases of the various syndromes that he had previously described.[9–11] The disease-associated genes of two of these syndromes, Bohring-Opitz syndrome (*ASXL1*)[12] and KBG syndrome (*ANKRD11*),[13] were recently described.

Dr. Opitz was the founding editor and, for 25 years, the editor-in-chief of the American Journal of Medical Genetics (1976–2001). Not only did he create a forum to exchange ideas between clinicians and basic scientists, but he was also a mentor to a generation of geneticists.

Allow me to indulge in two brief personal examples. My interactions with Dr. Opitz began in 1987 when, as a clinical-genetics fellow at the Children's Hospital of Philadelphia, I had submitted my first manuscript on patients with holoprosencephaly.[14] Along with the reviewers' comments, I also received Dr. Opitz's feedback. I was reminded of the children's riddle, "What is black and white and re(a)d all over?" In the US, every parent of a 5–8-year-old child knows, of course, that the answer is "a newspaper." After Dr. Opitz's edits with his famous "red pen," the answer to "What is black and white and re(a)d all over?" was then "A manuscript edited by Dr. Opitz."

Another form of mentoring came when Dr. Opitz invited me to review several manuscripts on the topic of holoprosencephaly. When I was still in clinical-genetics training, he asked me to also write a review article to accompany the other holoprosencephaly manuscripts.[15] This was the push that I needed to start investigating the underlying causes of normal and abnormal brain development. And that is what my lab has focused on for the past two decades. So, it is no exaggeration to state that my career was shaped and defined by Dr. Opitz's early

[1]This article is based on the address given by the author at the meeting of the International Congress of Human Genetics on October 13, 2011 in Montreal, Quebec, Canada. The audio of the original address can be found at the web site of the American Society of Human Genetics.

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encouragement. I know of several other colleagues who had a similar career boost from Dr. Opitz.

In closing, let me tell you about Dr. Opitz's love of nature and, most of all, his dedication to his patients and their families. I have witnessed this personally at a weekend with a support group for parents who had children with various intellectual and/or physical disabilities. His warmth, caring, and compassion had a healing effect on the families even when little else could be offered from the medical field.

Join me in celebrating a giant in the field of medical genetics, Dr. John Opitz, the winner of the 2011 William Allan Award!

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