John Marius Opitz Biography

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Contents

John Marius Opitz Biography.....1

Contents.....2

Biography.....3

Biography

John M. Opitz is renowned as one of the founders of clinical **genetics**. The Opitz G/BBB, Opitz-Kaveggia, and Smith-Lemli-Opitz syndromes all bear his name because of his research into the genetic roots of these pediatric abnormalities.

Opitz received his training in pediatrics and clinical genetics at the State University of Iowa College of Medicine and the University of Wisconsin. He held faculty positions with Montana State University and the University of Wisconsin, Madison, before joining the University of Utah as professor of pediatrics, human genetics, and obstetrics and gynecology.

Early research conducted by Opitz in the 1950s, along with Emil Witschi, focused on **embryology**, endocrinology, **sex determination** and sex **differentiation**. Other work involved the study of hereditary kidney disease and the role of endocrines in carbohydrate metabolism. Opitz was also part of the research group that completed the first successful immunologic analysis of the human growth hormone.

In his later work, Opitz sought to determine the genetic roots of **birth defects** and mental retardation. He discovered the genetic link to several pediatric abnormalities, describing three syndromes, which were consequently given his name: Opitz-G/BBB (otherwise known as Opitz syndrome or Opitz-Frias syndrome), Opitz-Kaveggia, and Smith-Lemli-Opitz. Opitz first described Opitz-G/BBB, an extremely rare syndrome characterized by a disproportionate skull, wide-set eyes, a cleft palate, as well as numerous other physical deformities, in 1969. It was originally thought to be two separate disorders, but since discovered to be one disorder with separate genetic origins--one caused by a **mutation** on the **x chromosome**, and the other by a lack of specific genetic material on **chromosome** 22.

Opitz and his team of researchers at the University of Utah also studied **pleiotropy**, or the ability of one genetic mutation to cause several malformations. An example of this can be found in Smith-Lemli-Opitz syndrome, a disorder caused by the body's inability to metabolize cholesterol. The syndrome can lead to severe mental retardation, cardiac defects, a cleft palate, and webbed toes.

Other research conducted by Opitz and his team included genetic influences on mental retardation, sex determination and differentiation, and skeletal abnormalities. Additional areas of study included morphology, embryology, and human development.

Opitz founded and served as editor-in-chief of the *Journal of Medical Genetics,* and was a founding member of the American Board of **Medical Genetics**. Opitz has authored some 400 textbooks and textbook chapters on the subject of clinical genetics.