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50 Years Ago in *The Journal of Pediatrics*

RECENT ADVANCES IN GENETICS IN RELATION TO PEDIATRICS

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Five years after Watson and Crick published their hypothesis on the structure of DNA,¹ Fraser opined about the trends in genetics, as well as their potential clinical applications. It is astounding how far the field has come in half a century. We now know that the normal chromosomal complement is 46, not 48, and that DNA is the carrier of genetic information. What is currently called the "central dogma" (DNA→RNA→protein) was a mere "modern concept" 50 years ago. The Human Genome Project, which completed the sequencing of the human genome in 2003, was not even a twinkle in a geneticist's eye in 1958.

From conceptual foundations to practical applications, the role of genetics in medicine has bloomed. Now, molecular testing is the gold standard for diagnosing a multitude of genetic conditions and is the preferred modality for assessing carrier status. State-coordinated newborn screening programs began in the 1960s with testing for phenylketonuria using a dried blood spot. Recently, many states have expanded their panels to screen for more than 40 conditions as a result of the introduction of tandem mass spectrometry into these programs, which has dramatically improved the ability to diagnose and treat inborn errors of metabolism before symptoms develop.

The first genetic counseling graduate program was established a decade after Fraser noted that "the demand for genetic counseling is growing and there is an increasing need for suitably trained counselors." There are now more than 2400 board-certified genetic counselors, and the need for these professionals continues to transcend multiple specialties, including pediatrics, adults, prenatal, cancer, cardiovascular, hematology, and neurology.

Most fascinating is how Fraser's words still ring true today: "Exciting things have been happening in genetics in the past few years, many of them directly or potentially relevant to the practice of medicine." This discipline has progressed greatly in the last 50 years, and there is still plenty of excitement to come. Medical Genetics is now a primary specialty with more than 1000 board-certified clinical geneticists, "personalized medicine" is on the horizon, and genetic testing is being marketed directly to consumers. The questions we now face are not just how to identify genes, but rather what can/should we do with this information. It was surely impossible for Fraser to predict that genetics would be so intimately involved in all aspects of health care, as we now know it today.

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10.1016/j.jpeds.2007.12.038

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